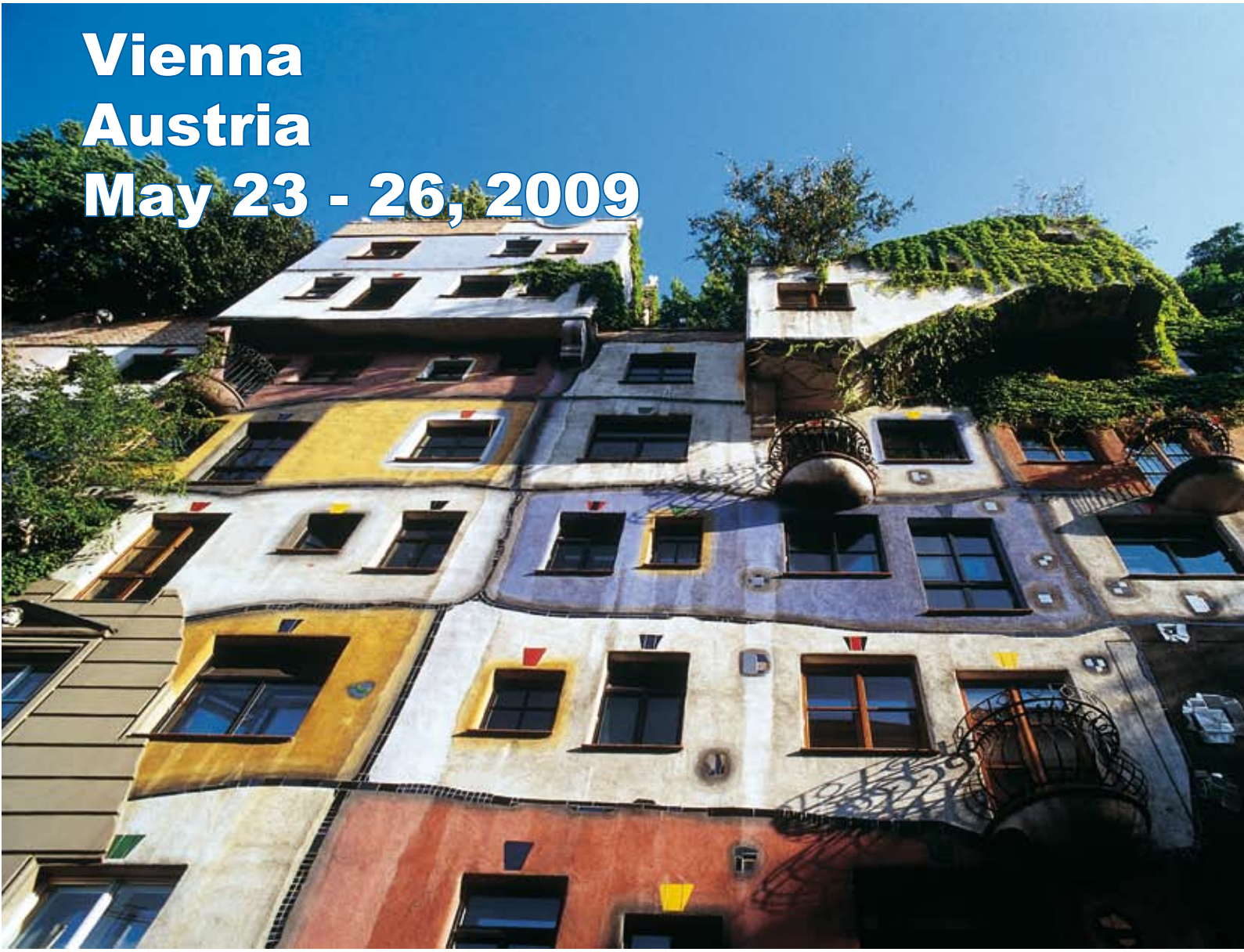
European Society of Human Genetics

HUMAN GENETICS

Conference 2009



Final Programme

www.eshg.org

European Society of Human Genetics

EUROPEAN HUMAN GENETICS CONFERENCE 2009

Austria Center Vienna
Vienna, Austria

Saturday, May 23 – Tuesday, May 26, 2009

Final Programme

www.eshg.org/eshg2009

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Dear Colleagues,

We cordially welcome you to 41st European Human Genetics Conference (ESHG 2009) in Vienna, Austria. This meeting will continue in the successful tradition of excellent conferences that cover the latest developments in the field of human genetics that are of interest for both clinicians and research scientists.

The city of Vienna, located in the heart of Europe, has been the traditional bridge from East to West. Its cultural and scientific heritage makes it one of the most attractive cities for holding meetings and conferences, thus the decision to return to the capital of Austria, after the International Congress in 2001, seemed a logical one.

2009 is the year of the 200th birthday of Charles Darwin. The Scientific Programme Committee (SPC) will endeavour to make Evolutionary Genetics a main topic at the conference, but also to cover the entire spectrum of hot topics in human and medical genetics.

We sincerely hope that you will enjoy the conference in Vienna, both from the scientific and from the social point of view.

Willkommen in Wien!

Hans-Christoph Duba
Local host ESHG 2009

European Society of Human Genetics

ESHG Office

European Society of Human Genetics
Ms. Karin Knob
c/o Vienna Medical Academy
Alserstrasse 4
1090 Vienna
Austria
Tel: +43 1 405 13 83 20
Fax: +43 1 405 13 83 23
Email: office@eshg.org
Website: www.eshg.org

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Further information on structure and organisation can be found on the website **www.eshg.org**

European Conference of Human Genetics 2009

Conference Organisation, Abstract Management

ESHG 2009 Secretariat
c/o Vienna Medical Academy
Mr. Jerome del Picchia
Alser Strasse 4, A-1090 Vienna, Austria
Tel: +43 1 405 13 83 22
Fax: +43 1 407 82 74
conference@eshg.org
www.medacad.org

Exhibition, Sponsoring and Commercial Satellites

Rose International
Exhibition Management and Congress Consultancy bv
Ms. Jantie de Roos
P.O. Box 93260, NL-2509 AG
The Hague, The Netherlands
Tel: +31 70 383 8901
Fax: +31 70 381 8936
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Mondial Congress & Events
Mondial GmbH & Co. KG
Mr. Patrick Barabas
Operngasse 20B
1040 Vienna Austria
Tel. +43 1 588 04 - 143
Fax. +43 1 588 04 - 185
eshg2009@mondial-congress.com
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Acknowledgements

The European Human Genetics Conference gratefully acknowledges the support of the following institutions, companies and organisations (list correct as per date of printing):

- Abbott Molecular
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- Nature Publishing Group
- Roche Applied Science
- Roche NimbleGen
- Rosetta Biosoftware
- Sistemas Genómicos

Future European Human Genetics Conferences

European Human Genetics Conference 2010
June 12 – 15, 2010
Gothenburg, Sweden

European Human Genetics Conference 2011
May 28 - 31, 2011
Amsterdam, The Netherlands

CME Credits

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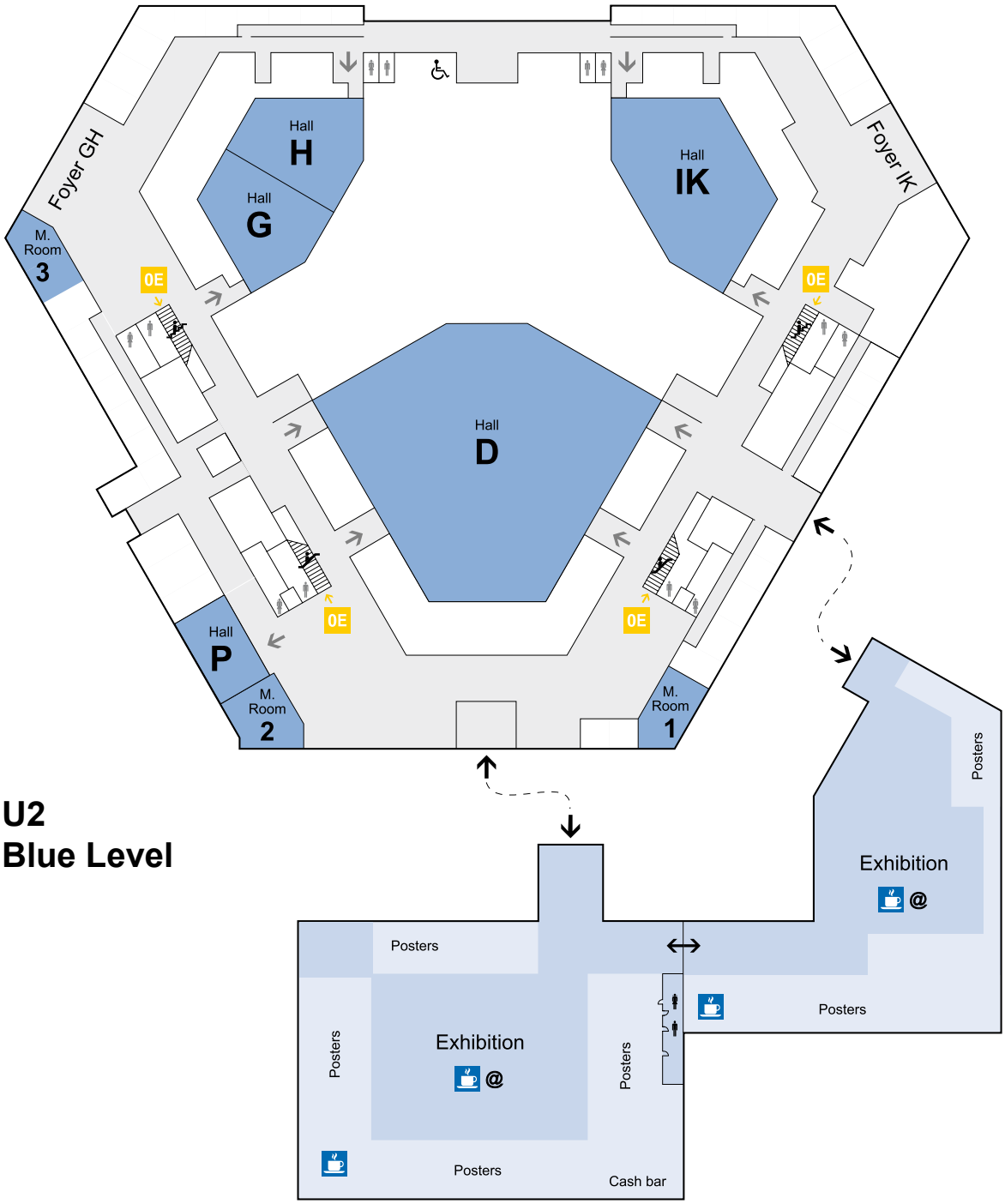
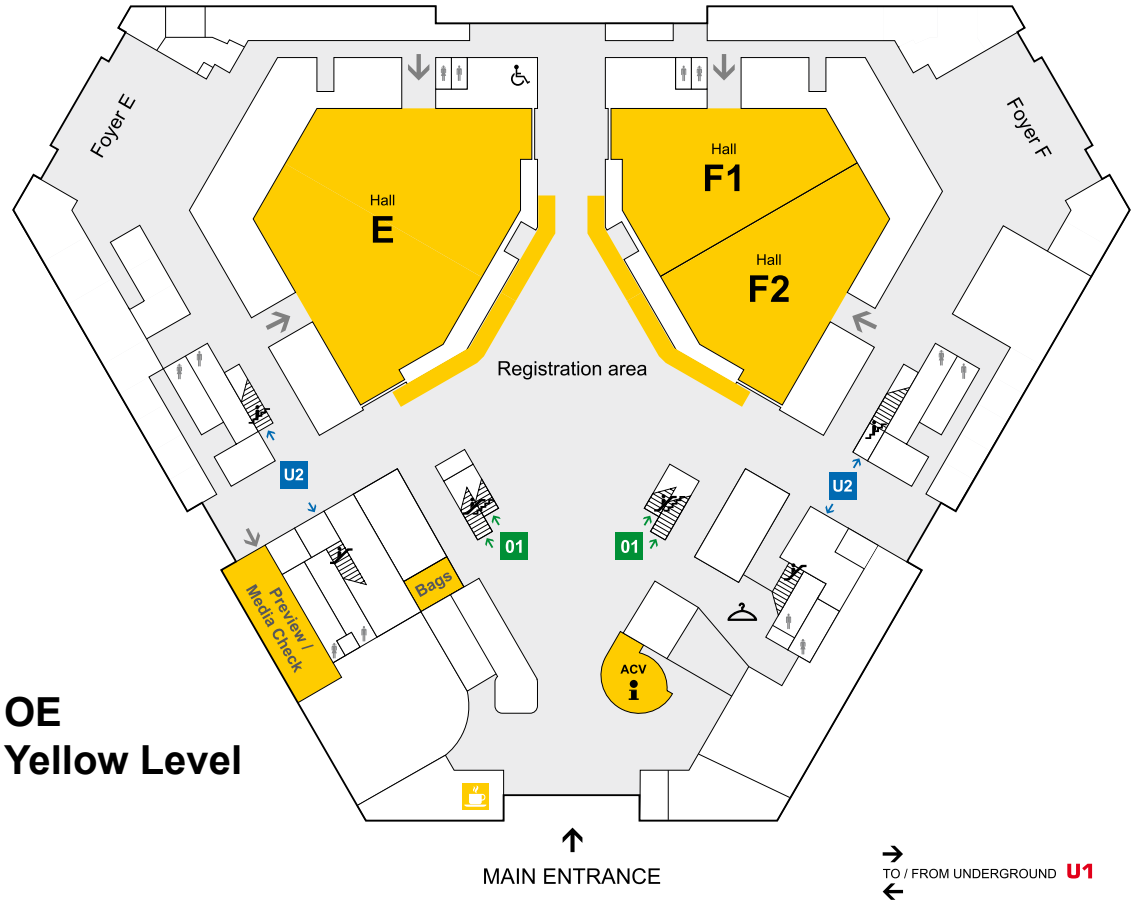


U2
Blue Level

U2 - Blue Level (this page)
Lecture rooms D, G, H, IK, P
Meeting rooms 1, 2 & 3
Posters, Exhibition, Coffee, Lunch boxes, Cash Bar, Internet Café, WiFi

OE - Yellow Level (opposite page)
Main Entrance - Access to Subway U1 (red line)
Lecture rooms E, F1, F2
Preview/Media Check
Registration, Message Boards, Jobbing Mall, Cloakroom

O1 - Green Level (opposite page)
Lecture room LM
Lounge 7-8



OE
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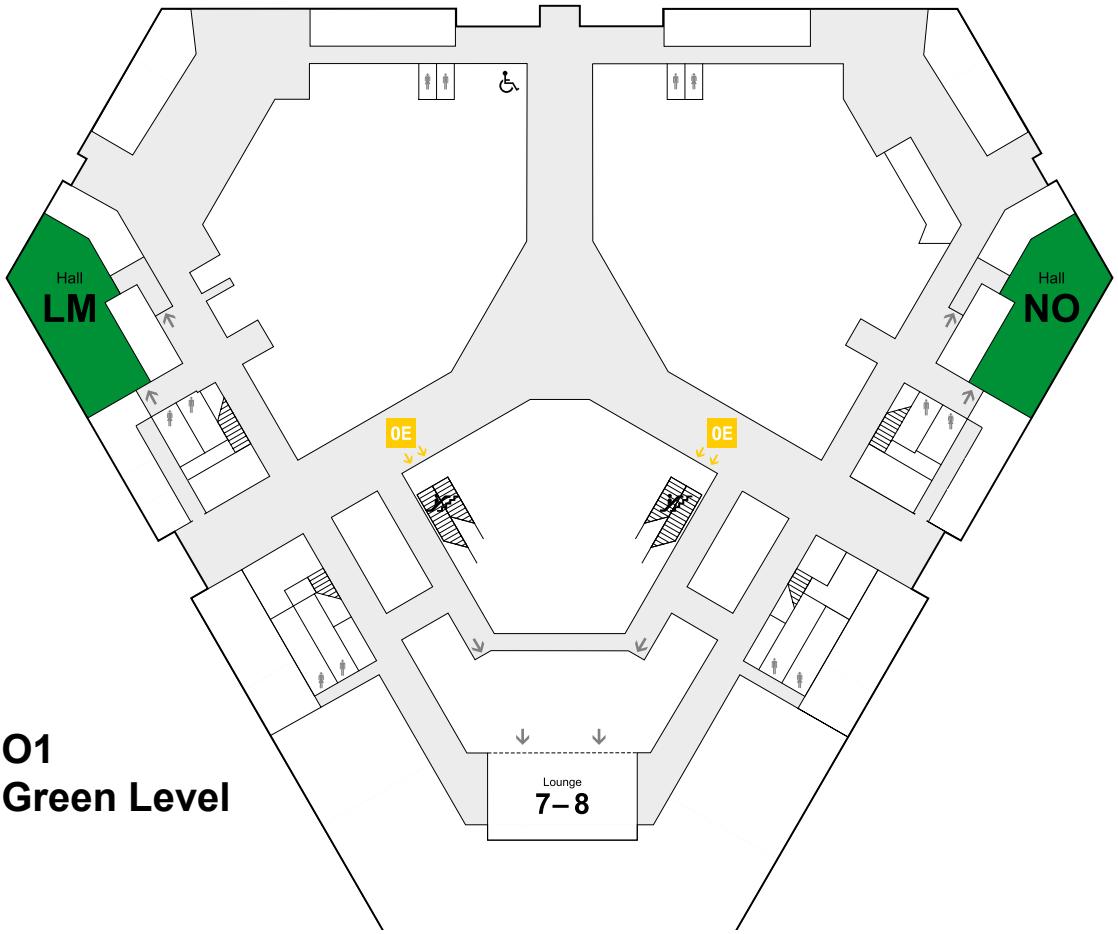

O1
Green Level

Saturday, May 23, 2009

Time	D	F1	F2	IK	G	H
14.00 – 15.30		ES1 What to do with unclassified variants	ES2 The changing scene in prenatal diagnosis	ES3 Genetic epidemiology	ES4 Molecular cytogenetics	ES5 Cardiogenetics in Clinical Practice
15:45 – 16.30	Opening Welcome Addresses EJHG Awards ESHG Education Award DNA-Day 2009 Contest					
16.30 – 18.00	Plenary Session 1					
18.00 – 18.30	*Coffee Break – Foyer D*					
18.30 – 20.00	PL 2 What's new?					
20.00	*Welcome Reception in Foyer D, Level U2*					

Sunday, May 24, 2009

Time	D	E	F1	F2	IK	LM	G	H
08.45 – 10.15	S01 Genomic Variation	S02 RNA in health and disease	S03 Counselling in a multidisciplinary cardiogenetics team: a luxury or a need	S04 Therapy for genetic disease				
10.15 – 10.45	*Coffee / Poster Viewing / Exhibition*							
10.45 – 11.15	Free Poster viewing							Applied Biosystems Satellite
11.15 – 12.15	Poster discussion with presenters (odd poster numbers)							
12.15 – 13.15	*Lunch / Poster Viewing / Exhibition*							
13.15 – 14.45	WS1 Direct to consumer genetic tests: confronting the issues for Europe	WS2 Dysmorphology I	WS3 Quality control	WS4 Genetic education	WS5 Using arrays in a diagnostic setting	WS6 Preimplantation and Prenatal diagnosis	Affymetrix Satellite	Roche Applied Science Satellite
15.00 – 16.30	C01 Next Generation Sequencing	C02 Clinical Cytogenetics	C03 Cerebellar disorders	C04 Molecular basis of skeletal and facial abnormalities	C05 Statistical genetics	C06 Cancer genetics		
16.30 – 17.15	*Coffee / Poster Viewing / Exhibition*							
17.15 – 18.45	S05 Modern cytogenetics	S06 Genomewide association studies	S07 Trinucleotide repeat diseases	S08 Inherited cancers				
18.45								Illumina Satellite
19.00 – 19.45				ESHG Membership Meeting				

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Satellite Symposium

Monday, May 25, 2009

Time	D	E	F1	F2	IK	LM	G	H
08.45 – 10.15	**PL 3 The Darwin legacy**							
10.15 – 10.45	*Coffee / Poster Viewing / Exhibition*				**„Innovative Techniques in Genome Diagnostics“** Eurogentest Satellite	*Coffee / Poster Viewing / Exhibition*		
10.45 – 11.15	Free Poster viewing						**Abbott Molecular Satellite**	**Agilent Technologies Satellite**
11.15 – 12.15	Poster discussion with presenters (even poster numbers)							
12.15 – 13.15	*Lunch / Poster Viewing / Exhibition*							
13.15 – 14.45	**WS7 Community Genetics**	**WS8 Dysmorphology II**	**WS9 Diagnostic Cytogenetics**	**WS10 Genetic counselling and predictive testing: a dynamic perspective**	**WS11 To GWAS, or not to GWAS. How to spend your money wisely**	**WS12 Debate: Why test for moderate or low genetic risk of cancer anyway?**	**Roche NimbleGen Satellite**	**Applied Biosystems Satellite**
15.00 – 16.30	**C07 Reproductive genetics and prenatal diagnosis**	**C08 Molecular dysmorphology**	**C09 Evolutionary and population genetics and Genetic epidemiology**	**C10 Complex genetics I**	**C11 Molecular basis of Mendelian disorders**	**C12 Pathophysiology and therapy for genetic disorders**		
16.30 – 17.15	*Coffee / Poster Viewing / Exhibition*							
17.15 – 18.45	**S09 Prenatal and preimplantation diagnosis**	**S10 Neurodegeneration**	**S11 Metabolic diseases**	**S12 Stem cells**				
18.45	*Bus departure to the Congress Party at the Jugenstiltheater in the Otto Wagner Hospital*							

Tuesday, May 26, 2009

Time	D	E	F1	F2	IK	LM
08.45 – 10.15	**S13 Epigenetics**	**S14 Aging**	**S15 Development and pathogenesis**			
10.15 – 10.45	*Coffee / Poster Viewing / Exhibition*					
10.45 – 12.15	**C13 Cardiac Genetics**	**C14 Neurogenetics**	**C15 From genome to phenome**	**C16 Cytogenetics**	**C17 Complex genetics II**	**C18 Genetic counseling and services**
12.15 – 13.15	*Lunch / Poster Removal / Exhibition*					
13.15 – 14.00	**P4 ESHG Award Lecture**					
14.00 – 15.30	**P5 Journal, Young Investigator & Poster Awards** **Special Lecture** **Closing**					

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Satellite Symposium

Time	F1	F2	IK	G	H
14.00 - 15.30	**ES1. What to do with unclassified variants**	**ES2. The changing scene in prenatal diagnosis**	**ES3. Genetic epidemiology in the post-GWAS era**	**ES4. Molecular cytogenetics**	**ES5. Cardiogenetics in Clinical Practice** Chair: Ulf Kristofferson and Irene van Langen
	What is the problem? ***R. Sijmons***	**Targeted rapid aneuploidy detection in prenatal diagnosis: changing scene in Europe** ***T.H. Bui***	***J. Witte***	***M. Speicher***	**Introduction: The Cardiogenetics Outpatient Clinic** ***I.M. van Langen***
	What can the lab do for you? ***R. Hofstra***				**Cardiomyopathies** ***W.J. McKenna***
	In silico prediction can help ***S. Tavtigian***	**Non-invasive prenatal diagnosis of chromosomal aneuploidy by massively parallel genomic sequencing** ***R. Chiu***			**Primary arrhythmias** ***A.A.M. Wilde***
	Panel discussion moderated by: *Diana Eccles*				**Psychosocial aspects of Cardiogenetics** ***E.M.A.Smets***
					Panel discussion

Time	D
15.45 - 16.30	**Opening Ceremony** Chair: J.J. Cassiman, H.C. Duba
	Welcoming addresses by Jean-Jacques Cassiman, President of the ESHG and Hans-Christoph Duba, Local host, President of the Austrian Society of Human Genetics EJHG-Nature Awards ESHG Education Award presented to A. Schinzel European DNA-Day Essay Contest 2009
16.30 - 18.00	**Opening Plenary Session P1** Chair: J.J. Cassiman, H.C. Duba
16.30	**PL1.1. Genetic epidemiology of lipid-associated disorders** ***F. Kronenberg***
17.00	**PL1.2. Clinical significance of embryoscopy for early intrauterine deaths** ***T. Philipp***
17.30	**PL1.3. Immunology** ***J. Penninger***
18.00 - 18.30	*Coffee Break*

Time	D
18.30 - 20.00	**Plenary Session P2: What's new?** Chair: H. Brunner, B. Wirth
18.30	**PL2.1. Genetics meets metabolomics: a genome-wide association study of metabolite profiles in human serum** ***C. Gieger****, L. Geistlinger, E. Altmaier, M. Hrabé de Angelis, F. Kronenberg, T. Meitinger, H. Mewes, H. Wichmann, K. M. Weinberger, J. Adamski, T. Illig, K. Suhre*
18.45	**PL2.2.* Massive parallel sequencing of ataxia genes after array-based enrichment** ***A. Hoischen****, C. Gilissen, W. van der Vliet, P. Arts, N. Wieskamp, S. Vermeer, R. Meijer, M. Buckley, B. Kremer, N. van Slobbe-Knoers, J. Veltman, H. Scheffer*
19.00	**PL2.3. The SLC29A3 gene is mutated in Pigmented Hypertrichosis with Insulin Dependent Diabetes Mellitus syndrome and interacts with the insulin signalling pathway** *S. T. Cliffe, J. M. Kramer, K. Hussain, J. H. Robben, E. K. de Jong, A. P. de Brouwer, E. Nibbeling, E. Kamsteeg, M. Wong, J. Prendiville, C. James, R. Padidela, C. Becknell, H. van Bokhoven, P. M. T. Deen, R. C. M. Hennekam, R. Lindeman, A. Schenck, T. Roscioli,* ***M. F. Buckley***
19.15	**PL2.4.* Duplication of conserved non-coding sequence elements - a novel mechanism in the pathogenesis of congenital malformations** ***E. Klopocki****, K. Dathe, A. Brehm, K. W. Kjaer, C. Ott, I. Kurth, S. Mundlos*
19.30	**PL2.5.* Mitosis updated - PICH and the anaphase threads** ***T. Schwarzbraun****, L. Wang, P. Ulz, E. A. Nigg, M. R. Speicher*
19.45	**PL2.6. Genome-wide association reveals master eQTL regulators of microRNA expression variation in human fibroblasts** ***C. Borel****, S. Deutsch, A. Letourneau, M. Gagnebin, C. Gehrig, E. Falconnet, Y. Dupré, S. E. Antonarakis*
20.00	Welcome Reception in Foyer D, Level U2

Presentations highlighted by a * and a gray background are from Young Investigator Award Finalists.

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08.45 - 10.15	**S01 Genomic Variation** Chair: C. Wijmenga, F. Kronenberg	**S02 RNA in health and disease** Chair: B. Wirth, K. Wimmer	**S03 Counselling in a multidisciplinary cardiogenetics team: a luxury or a need** Chair: T. Clancy, G. Evers-Kiembooms		**S04 Therapy for genetic disease** Chair: O. Riess, F. Laccone
08.45	**S01.1. Genome variation, gene regulation, and human disease** ***S. McCarrol***	**S02.1. Non-coding antisense RNAs epigenetically regulate transcription in human cells** ***K. V. Morris***	8.45	**S03.1. Predictive Genetic Testing for Cardiovascular Diseases: Impact on Carrier Children.** ***E. M. A. Smets****, T. M. Meulenkamp, M. M. H. Stam, A. Tibben, E. D. Mollema, I. M. van Langen, A. Wiegman, G. M. de Wert, I. D. de Beaufort, A. A. M. Wilde*	**S04.1. Targeted treatment of tuberous sclerosis using mTOR inhibitors** ***J. R. Sampson***
09.15	**S01.2. New methods for detecting rare variants associations** ***A. Kong***	**S02.2. The role of microRNAs in brain tumors** *D. Beier, J. Y. Zhu, A. Eichner, C. Beier,* ***G. Meister***	9.05	**S03.2. Uptake of genetic counselling and predictive DNA testing in hypertrophic cardiomyopathy** *I. Christiaans*	**S04.2. Genetic aetiology defines optimal treatment in monogenic diabetes** ***S. Ellard***
09.45	**S01.3. Rare and common variants in human disease** ***D. Goldstein***	**S02.3. mRNA splicing and disease** ***U. Fischer***	9.25	**S03.3. Genetic testing in familial cardiomyopathies: the cardiologists view** *J. van Cleemput*	**S04.3. MicroRNAs: Functions in metabolism and therapeutic opportunities** ***M. Stoffel***
			9.40	**S03.4. Peer Support: A Critical Resource** *A. Cox*	
10.15-10.45	*Coffee Break / Poster viewing / Exhibition*				
11.15-12.15	**Poster viewing with presenters (odd poster numbers)**				
12.15-13.15	*Lunch / Poster viewing / Exhibition*				

Time	D	E	F1	F2	IK	LM
13.15 - 14.45	**WS1 Direct-to-consumer genetic tests: confronting the issues for Europe** P. Borry, H. Howard	**WS2 Dysmorphology I** D. Donnai, J. Clayton-Smith	**WS3 Quality control** E. Dequeker, M. Morris	**WS4 Genetic education** D. Coviello, P. Farndon	**WS5 Using arrays in a diagnostic setting** N. de Leeuw	**WS6 Preimplantation and Prenatal diagnosis** I. Liebaers, M. Macek Jr.

Information on Workshops (if submitted by the organisers) can be found in the separate programme sheet in the conference bag.

Time	D	E	F1	F2	IK	LM
15.00 - 15.45	**C01 Next Generation Sequencing** Chair: B. Wirth, T. Schwarzbraun	**C02 Clinical Cytogenetics** Chair: M. Soller, H. Rehder	**C03 Cerebellar disorders** Chair: T. Ozcelik, P.M. Kroisel	**C04 Molecular basis of skeletal and facial abnormalities** Chair: K. Avraham, A.R. Janecke	**C05 Statistical genetics** Chair: F. Clerget-Darpoux, H.G. Kraft	**C06 Cancer genetics** Chair: A. Cambon-Thomsen, C. Fonatsch
15.00	**C01.1. mRNA-Seq Whole Transcriptome Analysis of a Single Cell** ***K. Q. Lao***, *F. Tang, C. Barbacioru, Y. Wang, E. Nordman, C. Lee, N. Xu, X. Wang, J. Bodeau, A. Surani*	**C02.1*. Phenotypic and genomic evaluation of 52 subjects with a Smith-Magenis-like phenotype: identification of new syndromic regions associated with altered gene dosage** ***S. R. Williams***, *S. Girirajan, D. Tegay, N. Nowak, E. Hatchwell, S. Elsea*	**C03.1. Joubert syndrome and related cerebellar disorders among Egyptian patients: Clinical and genetic heterogeneity** ***M. S. Zaki***, *A. K. Abdel Aleem, G. M. H. Abdel Salam, S. L. Bielas, J. L. Silhavy, D. Swistun, S. E. Marsh, J. G. Gleeson*	**C04.1. Spondyl-ocheiro dysplastic form of the Ehlers-Danlos Syndrome - A novel recessive entity caused by mutations in the zinc transporter gene SLC39A13** ***C. Giunta***, *C. Bürer-Chambaz, N. H. Elçioglu, B. Albrecht, G. Eich, A. R. Janecke, M. Kraenzlin, H. Yeowell, M. Weis, D. R. Eyre, B. Steinmann*	**C05.1. Combined analysis of 19 common validated type 2 diabetes susceptibility gene variants show moderate discriminative value and no evidence of gene-gene interaction** *T. Sparso, N. Grarup, C. Andreasen, A. Albrechtsen, J. Holmkvist, G. Andersen, T. Jørgensen, K. Borch-Johnsen, A. Sandbæk,* ***T. Lauritzen***, *S. Madsbad, T. Hansen, O. Pedersen*	**C06.1. Variants of the Xeroderma Pigmentosum Variant gene (POLH) are associated with melanoma risk** ***N. Soufir***, *J. Di Iucca, M. Guedj, J. Lacapère, M. Fargnoli, A. Bourillon, V. Descamps, C. Lebbe, N. Basset- Seguin0, K. Peris, B. Grandchamp, MelanCohort*
15.15	**C01.2. Integrated analysis of high-resolution transcriptomics data reveals new insights into the differentiation state-dependent control of transcript isoform abundance** ***P. A. C. ,t Hoen***, *M. S. Hestand, Y. Ariyurek, A. Klingenhoff, M. Scherf, M. Harbers, W. van Workum, G. J. B. van Ommen, J. T. den Dunnen*	**C02.2*. Further delineation of the 15q13.3 microdeletion and duplication syndromes: A clinical spectrum varying from non-pathogenic to a severe outcome** ***B. W. M. van Bon***, *H. C. Mefford, B. Menten, A. Sharp, J. W. Innis, C. van Ravenswaaij, N. de Leeuw, A. Kurg, L. Willatt, S. Knight, J. Vermeesch0, C. Romano, J. C. Barber, G. Mortier, L. A. Pérez-Jurado, F. Kooy, H. G. Brunner, E. E. Eichler, T. Kleefstra, B. B. A. de Vries, for the collaborative q. study group*	**C03.2. CC2D2A mutations in Meckel and Joubert syndromes, a strong genotype phenotype correlation** *S. Zerelli, S. Thomas, E. Szenker, S. Audollent, S. Romano, C. Babarit, M. Gonzales, R. Salomon, P. Loget, Y. Hillion, J. Roume, S. Khung, R. Bouvier, J. Martinovic, M. C. Gubler, N. Boddaert, A. Munnich, F. Encha-Razavi, E. M. Valente0, A. Saad, S. Saunier, M. Vekemans,* ***T. Attié-Bitach***	**C04.2. Frontorhiny, a distinctive presentation of frontonasal dysplasia caused by recessive mutations in the ALX3 homeobox gene** ***S. R. F. Twigg***, *S. L. Versnel, G. Nurnberg, M. M. Lees, M. Bhat, P. Hammond, R. C. M. Hennekam, J. M. Hoogeboom, J. A. Hurst, D. Johnson, A. A. Robinson, P. J. Scambler, D. Gerrelli, P. Nurnberg, I. M. J. Mathijssen, A. O. M. Wilkie*	**C05.2. Joint re-analysis of twenty-nine correlated SNPs supports the role of PCLO/Piccolo as a causal risk factor for major depressive disorder** ***Z. Bochdanovits***, *A. van der Vaart, M. Verhage, A. Smit, E. de Geus, D. Posthuma, D. Boomsma, B. Penninx, W. Hoogendijk, P. Heutink*	**C06.2*. Identification of novel genes involved in colorectal cancer predisposition** ***R. Venkatachalam***, *M. J. L. Ligtenberg, E. J. Kamping, E. Hoenselaar, M. Voorendt, H. Görgens, H. K. Schackert, A. Geurts van Kessel, N. Hoogerbrugge, R. P. Kuiper*
15.30	**C01.3*. Genomic variation detection by DNA selection and high throughput sequencing** ***S. Nikolaev***, *C. Iseli, D. Robyr, A. Sharp, J. Rougemont, C. Gehrig, L. Farinelli, S. Antonarakis*	**C02.3. Incomplete penetrance and variable expressivity in a series of 11 French patients with 15q13.3 recurrent microdeletion detected using array-CGH.** ***A. Masurel-Paulet***, *J. Andrieux, C. Le Caignec, P. Callier, M. P. Cordier, M. Beri, B. Doray, E. Flori, O. Boute, B. Delobel, B. Isidor, S. Jaillard0, S. Odent, C. Thauvin-Robinet, C. Bidon, B. Aral, F. Mugneret, P. Jonveaux, D. Sanlaville, L. Faivre*	**C03.3. CA8 mutations cause a novel syndrome characterized by ataxia and mild mental retardation with predisposition to quadrupedal gait** ***S. Türkmen***, *G. Guo, M. Garshasbi, K. Hoffmann, A. Alshalah, K. Kahrizi, A. Tzschach, A. Kuss, A. Kuss, H. Najmabadi, H. Ropers, N. Humphrey, S. Mundlos, P. Robinson*	**C04.3. ALX4 dysfunction disrupts craniofrontonasal and hair follicle development** ***N. A. Akarsu***, *H. Kayserili, E. Uz, C. Niessen, I. Vargel, Y. Alanay, G. Tuncbilek, G. Yigit, O. Uyguner, S. Candan, H. Okur, S. Kaygin, S. Balci, E. Mavili, M. Alikasifoglu, B. Wollnik*	**C05.3. Unified framework for epistasis detection in (un)relateds** ***K. Van Steen***, *T. Cattaert, M. Calle*	**C06.3*. TheTRIM8 gene is a novel player of p53 pathway** ***L. Micale***, *M. F. Caratozzolo, A. M. D'Erchia, M. G. Turturo, B. Augello, C. Fusco, P. Malatesta, E. Sbisà, A. Tullo, G. Merla*

Time	D	E	F1	F2	IK	LM
15.45 - 16.30	**C01 Next Generation Sequencing** Chair: B. Wirth, T. Schwarzbraun	**C02 Clinical Cytogenetics** Chair: M. Soller, H. Rehder	**C03 Cerebellar disorders** Chair: T. Ozcelik, P.M. Kroisel	**C04 Molecular basis of skeletal and facial abnormalities** Chair: K. Avraham, A.R. Janecke	**C05 Statistical genetics** Chair: F. Clerget-Darpoux, H.G. Kraft	**C06 Cancer genetics** Chair: A. Cambon-Thomsen, C. Fonatsch
15.45	**C01.4. Adult human brain samples deep sequencing of small-RNAs reveals specific expression profiles in different brain areas** *E. Martí, L. Pantano, M. Bañez-Coronel, E. Miñones, E. Mateu, S. Porta,* ***X. Estivill***	**C02.4*. Interstitial 18q21 microdeletions and a microduplication including the TCF4 gene causing Pitt Hopkins syndrome** ***I. Feenstra****, I. Rayen, G. Houge, D. Koolen, S. Kant, C. Romano, S. Price, M. Fichera, S. Reitano, M. Breuning, C. Ruivenkamp, L. Vissers, J. Veltman, H. Brunner, C. van Ravenswaaij-Arts, B. de Vries*	**C03.4*. Search for genes implicated in new forms of recessive ataxia** ***M. Assoum****, M. A. Salih, N. Drouot, D. H'Mida-Ben Brahim, C. Lagier-Tourenne, A. Aldriss, S. A. Elmalik, T. S. Ahmed, M. Z. Seidahmed, M. M. Kabiraj, M. Koenig*	**C04.4. TRPS1, a regulator of chondrocyte proliferation and differentiation, interacts with the activator form of GLI3** ***F. J. Kaiser****, M. Wuelling, L. A. Buelens, D. Braunholz, R. Depping, G. Gillessen-Kaesbach, A. Vortkamp*	**P08.40. Statistical properties of tests of association performed on mixtures of singletons and related individuals: effects of the nonorthogonality of linkage and LD parameters on type I error and power** ***T. Hiekkalinna****, L. Peltonen, J. Terwilliger*	**C06.4*. Identification of Low Penetrance Genes associated to thyroid cancer susceptibility using a two-step case-control approach** ***I. Landa****, S. Ruiz-Llorente, C. Montero-Conde, L. Leandro-García, S. Leskelä, E. López-Jiménez, A. Maliszewska, L. Inglada-Pérez, L. De La Vega, G. Pita, M. Alonso, J. Maravall, V. Andía, C. Álvarez-Escolá, A. Meoro, J. Caballero, C. Blanco, J. Díaz-Pérez, J. Serrano0, D. Mauricio, A. Cascón, C. Rodríguez-Antona, A. González-Neira, P. Santisteban, M. Robledo*
16.00	**C01.5*. Estimation of MUTYH variant frequencies in pooled DNA with massive parallel sequencing** ***A. A. Out****, I. J. H. M. van MInderhout, Y. Ariyurek, C. M. J. Tops, M. van Galen, J. J. Goeman, P. E. Taschner, K. Schneeberger, S. Ossowski, M. H. Breuning, G. J. B. van Ommen, J. T. den Dunnen, P. Devilee, F. J. Hes*	**C02.5. Another new microdeletion syndrome due to 11q13.2q13.4 cryptic deletion mediated by segmental duplications.** ***A. Wischmeijer****, P. Magini, M. Gnoli, D. Niedrist, R. Ciccone, I. Cecconi, E. Franzoni, G. Romeo, O. Zuffardi, A. Schinzel, M. Seri*	**C03.5. tRNA Splicing Endonuclease mutations cause Pontocerebellar Hypoplasia** ***Y. Namavar****, P. Kasher, B. S. Budde, P. G. Barth, B. Poll-The, K. Fluiter, E. Aronica, A. J. Grierson, P. van Tijn, F. van Ruissen, M. Weterman, D. Zivkovic, P. Nürnberg, F. Baas*	**C04.5. Homozygous disruption of an extracellular matrix component cause Temtamy preaxial brachydactyly syndrome** *Y. Li, S. Temtamy, K. Laue, M. Aglan, B. Pawlik, G. Nürnberg, P. Nürnberg, M. Hammerschmidt,* ***B. Wollnik***	**C05.5. Development of molecular pathways analysis of GWAS data: application to schizophrenia and bipolar disorder** *C. T. O'Dushlaine, E. Kenny, International Schizophrenia Consortium, M. Gill,* ***D. W. Morris****, A. P. Corvin*	**C06.5*. Detection of tumor-specific somatic mutations by transcriptome sequencing of a cytogenetically normal acute myeloid leukemia** ***S. H. Eck****, P. A. Greif, A. Benet-Pagès, H. Popp, A. Dufour, T. Meitinger, T. M. Strom, S. K. Bohlander*
16.15	**C01.6. Next-Generation-Sequencing as a promising diagnostic tool in heterogeneous genetic conditions: the example of Hypertrophic Cardiomyopathy** ***J. L. Blouin****, C. Iseli, D. Robyr, A. Munoz, S. E. Antonarakis, S. Fokstuen*	**C02.6. Retrospective external quality assessment: the french ACLF online experience** ***M. Doco-Fenzy****, D. Sanlaville, C. Sarraustre de Menthière, C. Cartier, M. Combrisson, S. Dahoun, A. Moncla, F. Mugneret, L. Taine, S. Tapia, F. Vialard0, I. Luquet, F. Thepot, C. Terre, J. Dupont*	**C03.6. Retinal neurone remodelling induced by polyglutamine toxicity in a SCA7 mouse model** ***Y. Trottier****, M. Yefimova, N. Messaddeq, C. Jacquard, C. Weber, L. Jonet, J. Jeanny*	**C04.6. Duplication of the EFNB1 gene in familial hypertelorism: imbalance in ephrin-b1 expression and abnormal phenotypes in humans and mice** ***C. Babbs****, H. Stewart, L. Williams, L. Connell, A. Goriely, S. R. F. Twigg, K. Smith, T. Lester, A. O. M. Wilkie*	**C05.6*. A Comparison of Methods for Testing Association Between Uncertain Genotypes and Quantitative Traits** ***Z. Kutalik****, T. Johnson, M. Bochud, V. Mooser, P. Vollenweider, G. Waeber, D. Waterworth, J. S. Beckmann, S. Bergmann*	**C06.6*. Ikaros is a frequently affected hematopoietic differentiation factor in pediatric relapse-prone precursor B-cell acute lymphoblastic leukemia** ***E. Waanders****, M. W. M. te Loo, F. N. van Leeuwen, V. H. J. van der Velden, S. V. van Reijmersdal, J. de Vries, S. T. M. Keijzers-Vloet, J. Y. Hehir-Kwa, E. Sonneveld, J. J. M. van Dongen, A. Geurts van Kessel, P. M. Hoogerbrugge, R. P. Kuiper*
16.30	*Coffee / Poster Viewing / Exhibiton*					

Presentations highlighted by a * and a gray background are from Young Investigator Award Finalists.

Time	D	E	F1	F2
17.15 - 18.45	**S05 Modern cytogenetics** Chair: M. Speicher, G. Webersinke	**S06 Genomewide association studies** Chair: F. Kronenberg, I.M. Heid	**S07 Trinucleotide repeat diseases** Chair: B. Kerem, F. Laccone	**S08 Inherited cancers** Chair: R. Seruca, H.C. Duba
17.15	**S05.1. Structural genomic variation** ***C. Lee***	**S06.1. Genetics of human autoimmune disease** ***D. Hafler***	**S07.1. Modeling genetic disorders with human embryonic stem cells: The case of Fragile X syndrome** ***N. Benvenisty***	**S08.1. RET as a diagnostic and therapeutic target in MEN2** ***R. M. W. Hofstra***
17.45	**S05.2. Array-CGH in clinical practice: Fascination and frustration.** ***K. Devriendt***	**S06.2. Genome-wide association studies of obesity and height: What have we learned?** ***J. Hirschhorn****, for the GIANT Consortium*	**S07.2. Tissue-Specific Instability of Diseased-Associated Trinucleotide Repeats.** ***C. E. Pearson****, J. D. Cleary, S. Tomé, L. Foiry, H. Sroka, D. Chitayat, I. Paradis, R. Drouin, G. Gourdon*	**S08.2. Li Fraumeni syndrome: The role of DNA copy number variation in cancer susceptibility** ***D. Malkin***
18.15	**S05.3. Mitotic recombination in leukaemia** ***B. D. Young***	**S06.3. Low-penetrance genes for colorectal cancer predisposition** ***I. Tomlinson***	**S07.3. Myotonic dystrophy: Complex repeats in a complex disorder** *C. Braida, J. Couto, F. Morales, P. Cuenca, T. Ashizawa, A. Wilcox, D. E. Wilcox, J. Mandel, H. Radvanyi, F. Niel, M. Koening, C. Lagier-Touren, C. Faber, H. J. M. Smeets, P. A. Hofman0, C. E. M. de Die-Smulders, F. Spaans,* ***D. G. Monckton***	**S08.3. Familial gastric cancer** ***C. Caldas***
	F2			
19.00 - 19.45	ESHG Membership Meeting			

Time	D
08.45 - 10.15	**PL3 The Darwin legacy (1809-2009)** Chair: M. Rocchi, C. Fonatsch
08.45	**PL3.1. Weird animal genomes, sex and dumb design** ***J. Graves***
09.15	**PL3.2. Genetics and sociobiology of microbes** ***K. Foster***
09.45	**PL3.3. Language as Kluge** ***G. Marcus***
10.15 - 10.45	*Coffee Break / Poster viewing / Exhibition*
10.45 - 11.15	*Free Poster viewing / Exhibition*
11.15 - 12.15	**Poster viewing with presenters (even poster numbers)**
12.15 - 13.15	*Lunch / Poster viewing / Exhibition*

Time	D	E	F1	F2	IK	LM
13.15 - 14.45	**WS7 Community Genetics** M. Cornel, U. Kristofferson	**WS8 Dysmorphology II** D. Donnai, J. Clayton-Smith	**WS9 Diagnostic Cytogenetics** R. Hastings, M. Rodrigues de Alba	**WS10 Genetic counselling and predictive testing: a dynamic perspective** A. Tibben, H. Skirton	**WS11 To GWAS, or not to GWAS. How to spend your money wisely** C. Wijmenga	**WS12 Debate: Why test for moderate or low genetic risk of cancer anyway?** J. Lubinski, C. Janssens

Information on Workshops (if submitted by the organisers) can be found in the separate programme sheet in the conference bag.

Time	D	E	F1	F2	IK	LM
15.00 - 15.45	**C07 Reproductive genetics and prenatal diagnosis** Chair: A. Bloch-Zupan, B. Streubel	**C08 Molecular dysmorphology** Chair: N. Akarsu, P.M Kroisel	**C09 Evolutionary & population genetics & Genetic epidemiology** Chair: P.F. Pignatti, H.G. Kraft	**C10 Complex genetics I** Chair: V. Kucinskas, A. Brandstätter	**C11 Molecular basis of Mendelian disorders** Chair: J. Beckman, K. Rötzer	**C12 Pathophysiology and therapy for genetic disorders** Chair: J. Sequeiros, J. Neesen
15.00	**C07.1. Mutations of the SYCP3 gene in women with recurrent pregnancy loss** ***H. Kurahashi***, *H. Bolor, T. Mori, S. Nishiyama, H. Inagaki, H. Kogo, M. Tsutsumi, T. Ohye*	**C08.1*. Nicolaides-Baraitser Syndrome - Delineation of the Phenotype** ***S. B. Sousa***, *O. A. Abdul-Rahman, A. Bottani, V. Cormier-Daire, A. Fryer, G. Gillessen-Kaesbach, D. Horn, D. Josifova, A. Kuechler0, M. Lees, K. MacDermot, A. Magee, F. Morice-Picard, E. Rosser, A. Sarkar, N. Shannon, I. Stolte-Dijkstra, A. Verloes, E. Wakeling, L. Wilson, R. C. M. Hennekam,*	**C09.1*. Genetic risk model for coeliac disease helps identify high-risk individuals.** ***J. Romanos***, *C. C. van Diemen, I. M. Nolte, G. Trynka, A. Zhernakova, J. Fu, M. T. Bardella, D. Barisani, R. McManus, D. A. van Heel, C. Wijmenga*	**C10.1*. A genome-wide association study identified novel susceptibility loci for type 2 diabetes mellitus in Chinese population residing in Taiwan** ***C. F. Yang***, *J. Y. Wu, F. J. Tsai, C. C. Chen, P. Chen, C. H. Chen, Y. M. Liu, C. F. Shiu, C. S. J. Fann, Y. T. Chen*	**C11.1*. An aCGH screening study in 150 patients identifies a novel dosage-sensitive gene, TAB2, which is disrupted in multiple patients with cardiac defects** ***B. Thienpont***, *J. Breckpot, L. Zang, L. Tranchevent, P. Van Loo, K. Møllgård, N. Tommerup, I. Bache, Z. Tümer, D. Waggoner, M. Gewillig, H. Peeters, Y. Moreau, J. R. Vermeesch, L. A. Larssen, K. Devriendt*	**C12.1. DPF3 - a Novel Epigenetic Regulator of Cardiac Muscle Development and Function** *M. Lange, B. Kaynak, U. B. Forster, M. Tönjes, J. J. Fischer, J. Schlesinger, J. Gobom, S. Abdelilah-Seyfried,* ***S. Sperling***
15.15	**C07.2. Copy number changes in patients with disorders of sex development** ***S. White***, *H. Daggag, T. Ohnesorg, A. Notini, K. Roeszler, L. Gordon, E. Vilain, A. Sinclair*	**C08.2. Severe Non-Lethal Recessive Type VIII OI: Clinical, Histological and Radiographic Features** ***J. C. Marini***, *W. Chang, F. H. Glorieux, T. E. Hefferan, F. Rauch, M. Abukhaled, P. A. Smith, D. Eyre*	**C09.2. Identity by descent within and between human populations** *A. Gusev, P. Palamara, A. Darvasi, P. Gregersen,* ***I. Pe'er***	**C10.2*. Genome-wide association analysis and expression analysis from adipose tissue reveals coagulation factor XIII as a novel candidate gene for low HDL-cholesterol** ***P. P. Laurila***, *J. Naukkarinen, S. Söderlund, J. Saharinen, S. Ripatti, I. Lindqvist, M. Gentile, M. Jauhiainen, M. Taskinen, L. Peltonen*	**C11.2*. Shox2 mediates Tbx5 activity by regulating Bmp4 in the sinus venosus of the developing heart** ***S. Puskaric***, *S. Schmitteckert, A. D. Mori, A. Glaser, K. U. Schneider, B. G. Bruneau, R. J. Blaschke, H. Steinbeisser, G. Rappold*	**C12.2*. Neuropathology of Alpha-Synuclein and Synphilin-1 transgenic Mouse Models of Parkinson's Disease** ***S. Nuber***, *E. Petrasch-Parwez, T. Franck, U. Schumann, B. Winner, J. Winkler, H. Wolburg, S. V. Hörsten, T. Schmidt, J. Boy, H. Ngyuen, P. Teismann, J. B. Schulz, M. Neumann, C. Holzmann, I. Schmitt0, W. Kuhn, A. Bornemann, F. Zimmermann, A. Servadio, B. Pichler, O. Riess*
15.30	**C07.3. Afamin deficiency in mice leads to reversible infertility** *G. Wietzorrek, S. Olscher, G. Wakonigg, K. Pfaller, P. Grzmil, I. Adham, W. Engel,* ***H. Dieplinger***	**C08.3. Aicardi-Goutières syndrome and other disorders associated with intracranial calcification** ***Y. J. Crow***	**C09.3. Control of meiotic recombination in the human genome** ***M. C. Ergoren***, *I. L. Berg, P. Donnelly, A. J. Je*	**C10.3*. Loci on chromosome 19 and 20 are associated with age at natural menopause: a meta-analysis of 10,399 women** ***L. Stolk***, *J. M. Murabito, N. Franceschini, A. V. Smith, N. Glazer, G. Zhai, J. R. B. Perry, P. F. McArdle0, A. Arnold, E. Boerwinkle, A. Burri, L. Ferrucci, V. Gudnason, A. Hofman, D. Karasik, A. R. Shuldiner0, E. Streeten0, A. Murray, T. D. Spector, B. McKnight, T. B. Harris, E. Demerath, A. G. Uitterlinden, K. L. Lunetta*	**C11.3. Pharyngeal ectoderm to neural crest signalling is disrupted in a mouse model of DiGeorge syndrome** ***P. J. Scambler***, *A. Calmont, S. Ivins, K. Lammerts van Beuren*	**C12.3*. Rescue of a Lethal Murine Model of Methylmalonic Acidemia using rAAV8 Mediated Gene Therapy- One Year Post-Treatment** ***R. J. Chandler***, *C. P. Venditti*

Time	D	E	F1	F2	IK	LM
15.45 - 16.30	**C07 Reproductive genetics and prenatal diagnosis** Chair: A. Bloch-Zupan, B. Streubel	**C08 Molecular dysmorphology** Chair: N. Akarsu, P.M Kroisel	**C09 Evolutionary & population genetics & Genetic epidemiology** Chair: P.F. Pignatti, H.G. Kraft	**C10 Complex genetics I** Chair: V. Kucinskas, A. Brandstätter	**C11 Molecular basis of Mendelian disorders** Chair: J. Beckman, K. Rötzer	**C12 Pathophysiology and therapy for genetic disorders** Chair: J. Sequeiros, J. Neesen
15.45	**C07.4. The challenge of prenatal and preimplantation genetic diagnosis of mitochondrial DNA disorders** ***S. Monnot**, N. Gigarel, L. Hesters, P. Burlet, A. Benachi, Y. Dumez, G. Tachdjian, A. Rötig, R. Frydman, A. Munnich, N. Frydman, J. P. Bonnefont, J. Steffann*	**C08.4. Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a novel Rab-6 interacting golgin** ***U. Kornak**, H. Hennies, H. Zhang, J. Egerer, X. Zhang, W. Seifert, J. Kuehnisch, B. Budde, M. Naetebus, F. Brancati, W. R. Wilcox, D. Mueller, P. B. Kaplan, A. Rajab, B. Dallapiccola, W. Newman, J. Clayton-Smith, M. Tassabehji, B. Steinmann0, F. A. Barr, P. Nuernberg, P. Wieacker, S. Mundlos*	**C09.4. New evidences about MHC-based patterns of mate choice** ***M. Bicalho**, J. da Silva, J. M. Magalhães, W. Silva*	**C10.4*. Genome-wide association scan for bilirubin levels in a Sardinian population** ***S. Sanna**, F. Busonero, A. Maschio, P. F. McArdle, G. Usala, M. Dei, S. Lai, A. Mulas, M. Piras, L. Perseu, M. Masala, M. Marongiu, L. Crisponi, S. Naitza, R. Galanello, G. R. Abecasis, A. R. Shuldiner, D. Schlessinger, A. Cao, M. Uda*	**C11.4. Positive and negative feedback regulates the transcription factor FOXL2 in response to cell stress: evidence for a regulatory imbalance induced by disease-causing mutations** ***B. A. Benayoun**, F. Batista, J. Auer, A. Dipietromaria, D. L'Hôte, E. De Baere, R. A. Veitia*	**C12.4. Novel Enzyme Replacement Therapy for Gaucher Disease: On Going Phase III Clinical Trials with Recombinant Human Glucocerebrosidase Expressed in Plant Cells** ***D. Aviezer**, E. Almon-Brill, Y. Shaaltiel, R. Chertkoff, S. Hashmueli, A. Zimran*
16.00	**C07.5. Prenatal oligo-based arrayCGH with custom made focused design: first experiences** *A. Lott, M. Kuhn, **H. Gabriel**, M. Gencik*	**C08.5. Familial cases with hypomethylation of the imprinted IGF2-H19 domain in Silver-Russell Syndrome (SRS)** ***D. Bartholdi**, M. Krajewska-Walasek, K. Õunap, H. Gaspar, K. H. Chrzanowska, H. Ilyana, H. Kayserili, I. W. Lurie, A. Schinzel, A. Baumer*	**C09.5. Genomic tests for identification of authenticity of historical remains in case of Romanov family.** ***E. I. Rogaev**, A. P. Grigorenko, Y. K. Moliaka, G. Faskhutdinova, A. Goltsov, E. L. W. Kittler, I. Morozova*	**C10.5*. Identification of a Shared Genetic Susceptibility Locus for Coronary Heart Disease and Periodontitis** ***A. Schaefer**, G. M. Richter, B. Groessner-Schreiber, B. Noack, M. Nothnagel, N. El Mokhtari, B. G. Loos, S. Jepsen, S. Schreiber*	**C11.5*. Loss-of-function mutation in the dioxygenase-encoding, obesity-associated FTO gene causes severe growth retardation and multiple malformations** ***S. Boissel**, O. Reish, K. Proulx, H. Kawagoe-Takaki, D. Meyre, F. Molinari, G. Yeo, B. Sedgwick, V. Saudek, S. Farooqi, P. Froguel, T. Lindahl, S. O'Rahilly, A. Munnich, L. Colleaux*	**C12.5*. Trans-splicing gene therapy in autosomal dominant skin disease** ***V. Wally**, U. Koller, H. Hintner, J. W. Bauer*
16.15	**C07.6. Trend analysis of invasive prenatal diagnosis before and after the introduction of a new prenatal screening policy in the Netherlands.** ***K. D. Lichtenbelt**, B. Z. Alizadeh, P. G. Scheffer, G. C. Page-Christiaens, P. Stoutenbeek, G. H. Schuring-Blom*	**C08.6*. Germline mutation in NLRP2 (NALP2) in a familial imprinting disorder (Beckwith-Wiedemann Syndrome).** ***D. Lim**, E. Meyer, S. Pasha, L. J. Tee, F. Rahman, J. R. W. Yates, C. G. Woods, W. Reik, E. R. Maher*	**C09.6. Dissecting the genetic make-up of Central Eastern Sardinia using a high density set of sex and autosomal markers** ***L. M. Pardo**, P. Rizzu, G. Piras, K. der Gaag, D. Sondervan, Z. Bochdanovits, M. Monne, A. Gabbas, N. Bradman, P. de Knijff, A. Ruiz-Linares, P. Heutink*	**C10.6*. Meta-analysis of genome-wide scans identifies three novel loci influencing central obesity including one with a women-specific association with waist-hip-ratio** ***I. M. Heid**, C. M. Lindgren, C. Lamina, J. Randall, V. Steinthorsdottir, E. K. Speliotes, L. Qi, the GIANT consortium*	**C11.6. Microvillus inclusion disease results from loss of myosin Vb function** ***A. R. Janecke**, F. M. Ruemmele, G. Utermann, M. W. Hess, T. Müller, L. A. Huber*	**C12.6*. Ciliary beating recovery in deficient human airway epithelial cells after lentivirus ex vivo gene therapy** ***B. Chhin**, D. Nègre, O. Merrot, J. Pham, Y. Tourneur, D. Ressnikoff, M. Jaspers, M. Jorissen, F. Cosset, P. Bouvagnet*
16.30	*Coffee / Poster Viewing / Exhibiton*					

Presentations highlighted by a * and a gray background are from Young Investigator Award Finalists.

Time	D	E	F1	F2	
17.15 - 18.45	**S09 Prenatal and preimplantation diagnosis** Chair: I. Libaers, H.C. Duba	**S10 Neurodegeneration** Chair: P. Heutink, M. Auer-Grumbach	**S11 Metabolic diseases** Chair: D. Toncheva, J. Zschocke	**S12 Stem cells** Chair: R. Seruca, M. Speicher	
17.15	**S09.1. Molecular karyotyping: From postnatal to preimplantation genetic diagnosis?** ***J. Vermeesch***	**S10.1. Alzheimer disease** ***P. St.George Hyslop***	**S11.1. Novel insights and challenges from the inborn errors of vitamin B12 metabolism** ***D. S. Rosenblatt***	17.15	**S12.1. Using naturally-occurring mutations to identify stem cell niches, trace cell lineage and the origins of cancer in humans** ***N. Wright***
17.45	**S09.2. Prenatal diagnosis and fetal treatment using fetal RNA in maternal body fluids** ***D. Bianchi***	**S10.2. Pathways to Parkinsonism** ***M. R. Cookson***	**S11.2. Clinical, biochemical and genetic aspects of the methylmalonic acidaemias** ***M. R. Baumgartner***	17.35	**S12.2. Mapping mRNA expression QTLs in hematopoietic stem cells and their progeny** ***G. de Haan***
18.15	**S09.3. The challenge of prenatal and preimplantation genetic diagnosis of mitochondrial disorders** ***C. de Die-Smulders***, *H. Smeets*	**S10.3. Amyotrophic Lateral Sclerosis** ***L. H. van den Berg***	**S11.3. Homocysteine/folate and neural tube defects** ***H. J. Blom***	17.55	**S12.3. Stem cells as common ancestors in a colorectal cancer ancestral tree** ***D. K. Shibata***
				18.15	**S12.4. Using microenvironmental microarrays to decipher the role of the niche signaling in breast progenitor cell fate** *M. LaBarge, R. Villadsen, O. Petersen,* ***M. J. Bissell***
18.45	*Bus departure to the Congress Party at the Jugendstiltheater in the Otto Wagner Spital*				

Time	D	E	F1
08.45 - 10.15	**S13 Epigenetics** Chair: T. Frébourg, O. Rittinger	**S14 Aging** Chair: P. Scambler, H. Schmidt	**S15 Development and pathogenesis** Chair: A. Wilkie, H.C. Duba
08.45	**S13.1. Reverse Phenotyping: Towards an integrated (epi)genomic approach to complex phenotypes and common disease** ***S. Beck***	**S14.1. The P53 pathway acts to delay in-vivo senescence and aging** ***J. van Deursen****, D. J. Baker, C. Perez-Terzic, F. Jin, N. J. Niederländer, K. Jeganathan, S. Yamada, R. Lois, A. Terzic*	**S15.1. Vangl genes and neural tube defects** *E. Torban, M. Gravel, A. Ilisescu, C. Horth, G. Andelfinger, D. J. Epstein Jr.,* ***P. Gros***
09.15	**S13.2. A novel genetic mechanism for Lynch syndrome resulting in heritable somatic methylation of MSH2** ***M. Ligtenberg****, R. Kuiper, T. L. Chan, M. Goossens, K. Hebeda, M. Voorendt, T. Lee, D. Bodmer, E. Hoenselaar, S. Hendriks-Cornelissen, W. Tsui, C. Kong, H. Brunner, A. Geurts van Kessel, S. Yuen, J. van Krieken, S. Y. Leung, N. Hoogerbrugge*	**S14.2. Aging and Cancer: Rival Demons?** ***J. Campisi***	**S15.2. The ciliopathies: a model for dissecting context-dependent pathogenesis** ***N. Katsanis***
09.45	**S13.3. Functional mechanism of genomic imprinting** ***A. Ferguson-Smith***	**S14.3. Insulin signalling, ageing and age-related disease** ***D. Withers***	**S15.3. Graded Sonic Hedgehog Signaling and the Control of Neuronal Subtype Identity in Vertebrate Embryos** ***J. Briscoe***
10.15 - 10.45	*Coffee / Poster Viewing / Exhibiton*		

Time	D	E	F1	F2	IK	LM
10.45 - 11.30	**C13 Cardiac Genetics** Chair: J. Burn, B. Streubel	**C14 Neurogenetics** Chair: G. Houge, C. Windpassinger	**C15 From genome to phenome** Chair: A. Metspalu, J. Neesen	**C16 Cytogenetics** Chair: A. Renieri, P.M. Kroisel	**C17 Complex genetics II** Chair: A. Read, A.Brandstätter	**C18 Genetic counseling and services** Chair: G. Evers-Kiebooms, J. Zschocke
10.45	**C13.1*. Clinical characteristics of distantly related families with idiopathic ventricular fibrillation linked to chromosome 7q36** ***I. Christiaans***, *M. Alders, T. T. Koopmann, P. G. Postema, K. P. Loh, K. Zeppenfeld, C. R. Bezzina, A. A. M. Wilde*	**C14.1*. The vertebrate inner ear microRNAs have unique spatial and temporal expression patterns and are crucial for inner ear development and survival** ***L. M. Friedman***, *A. A. Dror, E. Mor, T. Tenne, G. Toren, N. Shomron, D. M. Fekete, E. Hornstein, K. B. Avraham*	**C15.1*. Segmental copy number variation shapes tissue transcriptomes** ***E. A. C. Chaignat***, *C. N. Henrichsen, E. Aït Yahya-Graison, N. Vinckenbosch, S. Zollner, F. Schütz, J. Chrast, S. Pradervand, M. Ruedi, H. Kaessmann, A. Reymond*	**C16.1*. The Effect of Translocation-Induced Nuclear Re-organization on Gene Expression** ***L. Harewood***, *F. Schütz, S. Boyle, P. Perry, M. Delorenzi, W. A. Bickmore, A. Reymond*	**C17.1*. Genome-wide analysis in Parkinson's disease** ***J. Simón-Sánchez***, *C. Paisán-Ruiz, J. Bras, S. Scholz, R. Gibbs, PD Genetics Consortium, T. Gasser, A. B. Singleton*	**C18.1*. Links between psychological characteristics and Distress after genetic result announcment : emotional regulation and adjustment** ***C. Fantini-Hauwel***, *B. Dauvier, S. Lejeune-Dumoulin, J. Pedinielli, S. Manouvrier*
11.00	**C13.2. The yield of family screening in sudden unexplained death in the young** ***C. Van der Werf***, *N. Hofman, H. L. Tan, I. M. Van Langen, A. A. M. Wilde*	**C14.2. Molecular basis of alpha-dystroglycanopathies and phenotype-genotype correlations** ***P. Guicheney***, *S. Quijano-Roy, C. Bouchet, A. Yanagisawa, S. Vuillaumier-Barrot, P. Richard, N. Clarke, N. B. Romero, B. Estournet, N. Seta, French network on Congenital Muscular dystrophies*	**C15.2*. Methylation profiling in cases with uniparental disomy identifies novel imprinted genes on chromosome 15** ***A. J. Sharp***, *B. Steiner, Y. Dupre, M. R. Sailani, D. O. Robinson, H. Brunner, A. Baumer, A. Schinzel, S. E. Antonarakis*	**C16.2. 5q14.3 microdeletion encompassing MEF2C, a gene controlling excitatory synapse number, is associated with severe mental retardation, poor eye contact and seizures** ***A. Goldenberg***, *M. Holder-Espinasse, S. Jaillard, N. Le Meur, D. Bonneau, S. Joriot, A. Charollais, H. Journel, S. Auvin0, C. Boucher, J. Kerckaert, T. Frébourg, V. David, S. Manouvrier-Hanu, P. Saugier-Veber, C. Dubourg, J. Andrieux*	**C17.2*. A regional high risk isolate for schizophrenia reveals an enrichment of three large copy number variations overlapping developmental genes** ***O. P. H. Pietiläinen***, *T. Paunio, A. Tuulio-Henriksson, J. Suvisaari, J. Haukka, T. Varilo, K. Rehnström, E. Jakkula, J. Wedenoja, A. Loukola, J. Suokas, L. Häkkinen, S. Ripatti, S. Ala-Mello, M. Jussila, J. Lönnqvist, H. Stefansson, L. Peltonen,*	**C18.2*. Evaluation of risk prediction updates from commercial genome-wide scans** ***R. Mihaescu***, *M. van Hoek, E. J. G. Sijbrands, A. G. Uitterlinden, J. C. M. Witteman, A. Hofman, C. M. van Duijn, A. J. W. Janssens*
11.15	**C13.3. Mutations in the ANKRD1 gene encoding CARP are responsible for human dilated cardiomyopathy** *L. Duboscq-Bidot, P. Charron, V. Ruppert, L. Fauchier, A. Richter, L. Tavazzi, T. Wichter, B. Maish, M. Komajda, R. Isnard,* ***E. Villard***	**C14.3. Mutations of LRTOMT, a fusion gene with alternative reading frames, cause autosomal recessive nonsyndromic hearing impairment in DFNB63 families.** ***E. Kalay***, *S. Masmoudi, Z. M. Ahmed, I. A. Belyantseva, M. A. Mosrati, R. W. J. Collin, S. Riazuddin, M. Hmani-Aifa, H. Venselaar, M. N. Kawar, A. Tlili, B. van der Zwaag, S. Y. Khan, L. Ayadi, R. J. Morell, A. J. Griffith, I. Charfedine0, R. Caylan, J. Oostrik, A. Karaguzel, A. Ghorbel0, S. Riazuddin, T. B. Friedman, H. Ayadi, H. Kremer*	**C15.3. Closely spaced multiple mutations as potential signatures of transient hypermutability in human genes** ***J. M. Chen***, *C. Férec, D. N. Cooper*	**C16.3. 19q13.11 deletion syndrome: a novel clinically and biochemically recognizable genetic condition identified by array-CGH** ***V. Malan***, *C. Ottolenghi, O. Raoul, H. V. Firth, B. Chadefaux, G. Royer, C. Turleau, A. Bernheim, L. Willatt, A. Munnich, M. Vekemans, S. Lyonnet, V. Cormier-Daire, L. Colleaux*	**C17.3. Twenty-two Loci affecting haematological traits** ***N. Soranzo***, *C. Gieger, M. Mangino*	**C18.3*. Compliance to screening and prevention guidelines of women carrying a BRCA1/2 mutation** ***A. Contrain***, *L. Huiart, L. Rabayrol, S. Olschwang, V. Bourdon, T. Noguchi, E. Amar, G. Houvenaeghel, D. Margain, F. Eisinger, H. Sobol*

Time	D	E	F1	F2	IK	LM
11.30 - 12.15	**C13 Cardiac Genetics** Chair: J. Burn, B. Streubel	**C14 Neurogenetics** Chair: G. Houge, C. Windpassinger	**C15 From genome to phenome** Chair: A. Metspalu, J. Neesen	**C16 Cytogenetics** Chair: A. Renieri, P.M. Kroisel	**C17 Complex genetics II** Chair: A. Read, A.Brandstätter	**C18 Genetic counseling and services** Chair: G. Evers-Kiebooms, J. Zschocke
11.30	**C13.4. Clinical Features and Outcome of Hypertrophic Cardiomyopathy Associated with Triple Sarcomere Protein Gene Mutations** ***F. Girolami***, *I. Olivotto, C. Giuliani, A. Mariottini, I. Passerini, M. Ackerman, F. Cecchi, F. Torricelli*	**C14.4. Clinical and mutational spectrum of the Legius syndrome (or NF1-like syndrome)** ***L. M. Messiaen***, *S. Yao, H. Brems, T. Callens, E. Denayer, P. Arn, D. Babovic-Vuksanovic, C. Bay, L. Escobar, R. Greenstein, R. Hachen, M. Irons, E. Lemire0, K. Leppig, M. McDonald, V. Narayanan, L. R. Shapiro, D. Tegay, E. Zackai, K. Taniguchi, T. Ayada, A. Yoshimura, A. Parret, B. Korf, E. Legius*	**C15.4. The Human Phenotype Ontology** ***P. N. Robinson***, *S. Köhler, S. Bauer, M. H. Schulz, S. Dölken, G. V. Gkoutos, M. Ashburner, C. Mundlos, P. N. Schofield, S. Lewis, J. M. Hancock, D. Horn, C. Ott, S. Mundlos*	**C16.4. Parental origin and possible mechanisms of formation of de novo balanced reciprocal translocations** ***A. Spreiz***, *M. Höckner, S. Demuth, A. Dufke, V. Kalscheuer, M. Rieger, O. Rittinger, I. Rost, S. Singer, A. Tzschach, E. Wiedersberg, M. Erdel, C. Fauth, J. Zschocke, G. Utermann, D. Kotzot*	**C17.4. The association of Bardet-Biedl Syndrome and Hirschsprung Disease highlights the role of the primary cilium in ENS development** ***J. Amiel***, *L. De Pontual, S. Thomas, N. A. Zaghloul, D. M. McGaughey, E. E. Davis, H. Dollfus, C. Baumann, S. L. Bessling, S. Audollent, A. Pelet, P. Beales, A. Munnich, S. Lyonnet, H. C. Etchevers, M. Vekemans, T. Attié-Bitach, A. S. McCallion, N. Katsanis,*	**C18.4. Community views of population carrier screening for fragile X syndrome** ***S. A. Metcalfe***, *A. D. Archibald, Y. M. Bylstra, C. Hickerton, S. Wake, A. M. Jaques, J. Cohen*
11.45	**C13.5. Noncompaction Cardiomyopathy; mutation spectrum, distribution of disease genes and implications for diagnostic strategies** *Y. M. Hoedemaekers, K. Caliskan, M. van Tienhoven, M. Michels, D. F. Majoor - Krakauer,* ***D. Dooijes***	**C14.5. Three subgroups of neurodegeneration with brain iron accumulation (NBIA)** ***M. Hempel***, *H. Prokisch, T. Kmiec, E. Jurkiewicz, T. Meitinger, M. B. Hartig*	**C15.5. GEN2PHEN: An international effort to optimise and federate the databasing of gene-disease relationships** ***A. J. Brookes***, *& The GENPHEN Consortium*	**C16.5. 22q13.3 deletion syndrome is a multigenic disorder, with SHANK3 as the major pathogenic gene** ***M. Zollino***, *M. E. Grimaldi, L. Boccuto, C. Schwartz, D. Battaglia, E. Mercuri, F. Guzzetta, G. Marangi, D. Orteschi, D. Buccella, M. Lauri, P. Visconti, G. Gobbi, G. Neri, F. Gurrieri*	**C17.5*. The common and specific genetic backgrounds of rheumatoid arthritis and celiac disease** ***A. Zhernakova***, *M. J. H. Coenen, G. Trynka, S. Heskamp, B. Franke, C. C. van Diemen, M. van Leeuwen, D. A. van Heel, T. R. D. J. Radstake, P. L. C. M. van Riel, P. Barrera, C. Wijmenga*	**C18.5. Effect of Education, Knowledge and Experience on Acceptance of First Trimester Screening for Chromosomal Abnormalities** ***V. Stefansdottir***, *H. Skirton, K. Jónasson, H. Harðardóttir, J. J. Jónsson*
12.00	**C13.6. Common variants at ten loci modulate the QT interval duration in individuals of European ancestry: the QTSCD consortium** ***A. Pfeufer***, *S. Sanna, D. E. Arking, M. Müller, V. Gateva, C. Fuchsberger, C. Pattaro, M. F. Sinner, S. S. Najjar, W. Kao, T. W. Mühleisen0, S. Möhlenkamp, A. A. Hicks, B. Müller-Myhsok, P. P. Pramstaller, H. Wichmann, D. Schlessinger, E. Boerwinkle, M. Uda, S. Kääb, T. Meitinger, G. R. Abecasis, A. Chakravarti*	**C14.6. Congenital Disorders of Glycosylation (CDG) due to defects in N-glycan assembly in the endoplasmic reticulum: filling the gaps in the dolichol cycle** ***G. Matthijs***, *W. Vleugels, M. Haeuptle, F. Foulquier, V. Race, R. Barone, L. Keldermans, J. Michalski, A. Fiumara, T. Hennet*	**C15.6. Establishing a link between microRNAs, genes and hereditary diseases : the miRiFix database** ***A. Henrion Caude***, *C. Mugnier, S. Bandiera, M. Girard, M. Le Merrer, A. Munnich, S. Lyonnet*	**C16.6*. An International Standardized Cytogenomic Array (ISCA) Consortium approach to the design, implementation and reporting of constitutional oligo array-CGH** ***S. Huang***, *D. H. Ledbetter, C. L. Martin, S. Aradhya, S. J. L. Knight, K. Smith, K. Kok, J. R. Vermeesch, J. A. Crolla*	**C17.6. Genome-wide association scan reveals major susceptibility locus for non-syndromic cleft lip with or without cleft palate on chromosome 8q24** *S. Birnbaum, K. U. Ludwig, H. Reutter, S. Herms, M. Steffens, M. Rubini, C. Baluardo, J. Freudenberg, C. Lauster, B. Braumann, R. H. Reich, A. Hemprich, R. P. Steegers-Theunissen0, B. Pötzsch, S. Moebus, B. Horsthemke, F. Kramer, T. F. Wienker, P. A. Mossey, P. Propping, S. Cichon, P. Hoffmann, M. Knapp, M. M. Nöthen,* ***E. Mangold***	**C18.6. Genetic counselling and cardiological care in predictively tested hypertrophic cardiomyopathy mutation carriers: the patients' perspective** ***I. Christiaans***, *I. M. van Langen, E. Birnie, G. J. Bonsel, A. A. M. Wilde, E. M. A. Smets*
12.15 - 13.15	*Lunch / Poster Removal / Exhibiton*					

Presentations highlighted by a * and a gray background are from Young Investigator Award Finalists.

Time	
13.15 - 14.00	**Plenary Session P4 ESHG Award Lecture** Chair: D. Donnai, H.Brunner
13.15	**PL4.1 ESHG Award Lecture** ***K. Stefansson***
14.00 - 15.30	**Plenary Session P5** Chair: D. Donnai, H.Brunner
14.00	**Young Investigator Awards** ESHG Young Investigator Award for Outstanding Science Isabelle Oberlé Award Lodewijk Sandkuijl Award Poster Awards
14.45	**Special Lecture** **PL5.1 "I have seen the future, and it works"** ***J. Burn*** **Closing**

For the second time, the ESHG proposes the ESHG Poster award for the 7 best posters presented at the meeting. Finalists receive a complementary ESHG online membership for 1 year. The 2 winners will receive an iTouch.

An additional set of iTouchs will be drawn within the attendees of the final plenary session.

Workshops

Information on workshops can be found on the separate programme sheet in the conference bag.

Sunday, May 24, 2009, 13.15 - 14.45

WS1 Direct-to-consumer genetic tests: confronting the issues for Europe
Room D
Organisers: Pascal Borry and Heidi Howard

WS2 Dysmorphology I
Room E
Organisers: Dian Donnai and Jill Clayton-Smith

WS3 Quality control
Room F1
Organisers: Els Dequeker, Michael Morris

WS4 Genetic education
Room F2
Organisers: Domenico Coviello and Peter Farndon

WS5 Using arrays in a diagnostic setting
Room IK
Organisers: Nicole de Leeuw

WS6 Preimplantation and Prenatal diagnosis
Room LM
Organisers: Inge Liebaers and Milan Macek

Monday, May 25, 2009, 13.15 - 14.45

WS7 Community Genetics
Room D
Organisers: Martina Cornel and Ulf Kristofferson

WS8 Dysmorphology II
Room E
Organisers: Dian Donnai and Jill Clayton-Smith

WS9 Diagnostic Cytogenetics
Room F1
Organisers: Ros Hastings and Marta Rodrigues de Alba

WS10 Genetic counselling and predictive testing: a dynamic perspective
Room F2
Organisers: Aad Tibben and Heather Skirton

WS11 To GWAS, or not to GWAS. How to spend your money wisely
Room IK
Organisers: Cisca Wijmenga

WS12 Debate: Why test for moderate or low genetic risk of cancer anyway?
Room LM
Organisers: Jan Lubinski and Cecile Janssens

Official Satellite Meetings open to all participants

As per date of printing. Please check the pocket programme for possible updates.

Friday, May 22, 2009

The Human Variome Project Forum
Hilton Vienna Danube, Vienna

"Ensembl workshop"
Vienna University Computer Service, Vienna

2nd International Meeting on Rare Disorders of the RAS-MAPK pathway
Austria Trend Hotel Donauzentrum, Vienna

Saturday, May 23, 2009

2nd International Meeting on Rare Disorders of the RAS-MAPK pathway
Austria Trend Hotel Donauzentrum, Vienna

Human Genome Variation Society –
"Genome Wide Association Studies: the last mile problem"
Hilton Vienna Danube, Vienna

Monday, May 25, 2009

10.15 - 12.30 hrs **"Innovative Techniques in Genome Diagnostics" - EuroGentest**
ACV, Room IK

Applied Biosystems – Sunday 24 May 2009, 10.45 – 12.15 hrs **Room H - Blue Level** Stand # A 300

Advances in CNV and SNP Analysis: from Discovery to Validation

This satellite will explore discovery and validation tools for your genotyping workflow needs. With the expansion of the resequencing applications, more structural variants will be discovered. In our first talk, our AB research scientist will show how the SOLID™ System combines ultra high throughput capability and unmatched accuracy with broad application flexibility, and will detail how SOLID can be used for SNP and CNV discovery.

Following downstream in the workflow, our guest speaker will address how the new TaqMan® Copy Number Variation assays have been used to genotype the complement factor 4 gene cluster at chromosome 6p21.

Our third guest speaker will present another validation tool for SNP analysis, The Taqman® OpenArray® Genotyping System. This newly launched high sample throughput technology, allows analysis of over 90,000 genotypes a day using the recognized gold standard in genotyping, the Taqman® SNP Genotyping Assays.

Discovery of CNVs and SNPs by Human Whole Genome Resequencing using SOLiD™ System
Dr. Fiona Hyland, Senior Scientist Research and Development, Bioinformatics, Applied Biosystems USA

Genotyping Copy Number Variation in the Complement Factor 4 Gene Cluster at Chromosome 6p21
Dr. Andre Franke, Institute for Clinical Molecular Biology, Christian-Albrechts-University Kiel, Kiel, Germany

Simple and Flexible Genotyping using the TaqMan® Open Array System™
Invited Speaker

Roche Applied Science - Sunday, 24 May 2009, 13.15 - 14.45 hrs **Room H - Blue Level** Stand # B 532

Enabling Technologies for Genomics Discovery

qPCR-HRM on the LightCycler® 480 System
A New Screening Approach and its Applications in Oncogenetics
Dr. Etienne Rouleau, Centre Renè Huguenin, St. Cloud, France

Several techniques have been proposed to screen genes for deleterious mutations in dominant disease. Until now, two different techniques were required to screen for both point mutations and large rearrangement. The talk presents a new approach combining qPCR and high resolution melting (HRM) on the LightCycler® 480 Real-Time PCR System, enabling rapid and cost-effective testing of a broad series of samples. Applications for genes implied in cancer predisposition (MLH1, MSH2, BRCA1, BRCA2) will be discussed.

The Genome Sequencer FLX System from Roche
Comprehensive Detection of Genetic Variations needs Long Reads
Dr. Marcus Droege, Roche Applied Science, Penzberg, Germany

The Genome Sequencer FLX System (GS FLX), powered by 454 Sequencing, is a next-generation DNA sequencing technology featuring a unique mix of long reads, exceptional accuracy, and ultra-high throughput. It has been proven to be the most versatile of all currently available next-generation sequencing technologies, supporting many high profile studies in many applications categories. This presentation will focus on applications possible with the Genome Sequencer FLX system, with special emphasis on human re-sequencing.

Affymetrix - Sunday, 24 May 2009, 13.15 - 14.45 hrs **Room G - Blue Level** Stand # A 216

Faster, better, simpler Genomic Analysis: Next-generation Cytogenetics and Genotyping with Affymetrix Microarrays

Come and join us to hear about exciting research and new developments in the fields of cytogenetic and genomic analysis.

Chair	Nick Brain – Marketing Programmes Manager, Affymetrix Welcome and Introduction
Key speakers	*Dr Ingrid Simonic - Medical Genetics, Cambridge University Hospital, UK* The Diagnostic Usefulness of Human SNP Array 6.0 Microarray Analysis in Management of Individuals with Disability and High Risk Obstetric Patients with Normal Karyotypes
	Dr Joris Veltman - Radboud University Nijmegen Medical Centre,The Netherlands Validation of the New Affymetrix Cytogenetics Solution

Places are limited so please be prompt! Come and say hello to us at stand A-216. We look forward to meeting you!

Illumina - Sunday, 24 May 2009, 18.45 - 20.15 hrs **Room H - Blue Level** Stand # A 330

Illumina Technology Workshop

Complimentary wine and cheese will be served.

Come hear about the latest breakthroughs in Illumina's complete range of next-generation life science technologies from leading experts in the industry. Topics will cover the use of arrays to examine rare and common variants in human disease, and several now generation sequencing applications that allow disease to be studied at the DNA and RNA level. World class speakers will participate to cover these topics and a short 10-15 minute update on latest technology improvements will be given by an Illumina scientist.

Speakers: To be announced.

Abbott Molecular - Monday, 25 May 2009, 10.45 - 12.15 hrs **Room G - Blue Level** Stand # A 136

Breakthrough for a Comprehensive Molecular Diagnosis of Fragile X Syndrome

Chair Dr Elaine Lyon, University of Utah/ARUP Laboratories, Salt Lake City, USA

Talk 1 Evaluation of Abbott Molecular Fragile X PCR with the QIAxcel Capillary Electrophoresis System
Dr Damian Heine, Hospital Universitari Son Dureta, Palma, Spain

Talk 2 A New Era for Fragile X Testing
Dr Judy Yu, Abbott Molecular, Chicago, USA

Talk 3 Towards a Simple Screen for Fragile X Expanded Alleles
Dr Elaine Lyon, University of Utah/ARUP Laboratories, Salt Lake City, USA

Agilent Technologies - Monday, 25 May 2009, 10.45 - 12.15 hrs **Room H - Blue Level** Stand # A 310

Insights in Human Disease and Systems Biology enabled by New aCGH and Targeted Resequencing Tools

Lyndsey Connell, Oxford Genetics Laboratory, Oxford, UK
New Generation High Density Agilent Microarrays – Incorporating Technological Improvements in a Clinical Diagnostic Setting
This talk will focus on how we have incorporated the continual improvements offered by Agilent microarrays whilst maintaining a safe and cost effective arrayCGH diagnostic service.

Daniel Turner, Wellcome Trust Sanger Institute, Hinxton, UK
Targeted Resequencing on the Illumina Genome Analyzer using Agilent SureSelect ™ Genome Partitioning
As it is still not feasible to perform whole genome resequencing on large numbers of eukaryotic samples, it is desirable to be able to isolate and sequence specific genomic regions of interest. Here we present an evaluation of the Agilent SureSelect ™ Target Enrichment System in combination with the Illumina Genome Analyzer sequencing platform.

Vladimir Lazar, MD, Ph.D, Institut Gustave Roussy, Villejuif, France
An Integrated System Biology Approach to Power Research Capabilities
Understanding the complexity of diseases requires today an integrated view of biology. I will demonstrate that combining CGH, Gene expression and microRNA profiling together with identifying relevant mutations proves an extremely powerful approach. The crucial role of integrative bioinformatics analysis will be illustrated.

Applied Biosystems – Monday 25 May 2009, 13.15 – 14.45 hrs **Room H - Blue Level** Stand # A 300

Resequencing Today: from Discovery to Clinical Applications

By offering systems to address the expansion of resequencing applications and the evolving needs of today's research environment, Applied Biosystems continues to lead the way in sequencing applications. We will share our vision of limitless progression of scientific questions, from Discovery enabled by Next Generation Sequencing on the SOLiD™ System, to the field of validated clinical applications with continued developments in capillary electrophoresis sequencing.

The arena of genomic analysis is continuously expanding through new technology and scientific initiative. Invited speakers will present their progress in human health research and diagnostic applications to improve the human condition. You will learn about their projects with data generated using capillary electrophoresis or SOLiD System and the various applications supported by these platforms.

Advances in Sequencing Technologies Applied to Human Disease Research
Dr Shaf Yousaf, Division President Genomic Analysis, Applied Biosystems, USA

Nonsyndromal XLMR Diagnostic Panel- First Year Experience
Michael Friez, Director, Diagnostic Laboratories, Greenwood Genetic Center, USA

Development of a STR-based Assay for the Detection of Human Uniparental Disomy
Dr Emiliano Giardina, Facoltà di Medicina e Chirurgia, Sezione di Genetica, Università degli Studi di Roma "Tor Vergata", Rome, Italy

Towards Molecular MHC Haplotype Sequencing
Dr Margret Hoehe, Max Planck Institute for Molecular Genetics, Berlin, Germany

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Roche NimbleGen - Monday 25 May 2009, 13.15 - 14.45 hrs **Room G - Blue Level** Stand # B 532

High-Definition Microarrays for Genomic Exploration in Medical Research

High Resolution Array CGH in Chronic Myeloid Leukemia
Alistair Reid, PhD, Division of Investigative Science, Imperial College of London, UK

Developing an Imprinting Map of the Human Genome
Andrew J Sharp, PhD, Department of Genetic Medicine and Development, University of Geneva, Switzerland

Sequence Capture for Genetic Studies in Medical Research
Alexander Hoischen, PhD, Department of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands

As per date of printing. Please check the pocket programme for possible updates.

Unless otherwise mentioned, all meetings are „on invitation/registration only".

Friday, May 22, 2009

14.00 - 19.00 hrs	ESHG Executive Board Meeting I	Room P

Saturday, May 23, 2009

09.00 - 13.00 hrs	ESHG Board Meeting I	Room P
10.00-12.00	Hemoglobin Satellite – ITHANET (open)	Meeting Room 3
13.00 - 15.30 hrs	ESHG Education Committee Meeting	Meeting Room 1
14.00 - 16.00 hrs	IFHGS Executive Committee Meeting	Meeting Room 2
18.00-18.30 hrs	ESHG SPC Meeting I	Meeting Room 1

Sunday, May 24, 2009

10.15 - 12.15 hrs	ESHG Accreditation Committee for Clinical/Medical Geneticists Meeting	Meeting Room 3
10.15 - 12.15 hrs	Case studies on quality management - EuroGentest	ACV, Room P
10.30 - 13.00 hrs	ESHG PPPC Meeting	Meeting Room 1
19.00 - 20.00 hrs	ESHG Membership Meeting and General Assembly (open to all members)	Room F2

Monday, May 25, 2009

08.30 - 12.30 hrs	ESHG Genetics Services Quality Committee Meeting	Meeting Room 1
09.15 - 14.15 hrs	ISCGG Kick-off Meeting	Meeting Room 2
10.15 - 11.15 hrs	DYCERNE meeting	Meeting Room 3
11.15 - 13.15 hrs	4th Meeting of the Presidents of the National Human Genetics Societies (open to all representatives)	Lounge 7-8
12.00 - 13.00 hrs	EJHG Editorial Board meeting	Room P

Tuesday, May 26, 2009

08.00 - 8.45 hrs	ESHG Board Meeting II	Room P
12.15-13.15 hrs	ESHG SPC Meeting II	Meeting Room 1
16.00 - 18.00 hrs	ESHG Executive Board Meeting II	Room P

Technical Information

- **Posters** will be on display from **Sunday May 24 to Tuesday, May 26, 2009, (12.15 hrs)**.
- **Poster mounting** will be possible on: **Saturday May 23, 2009, from 14.00 - 16.00 hrs (strict) and Sunday, May 24, from 8.30 hrs.**
- **Removal** will be possible on: **Tuesday, May 26, 2009, from 12.15 hrs - 14.00 hrs (strict).**
  Please note that posters not removed until then, will be taken down by the staff of the conference center and will not be stored or sent to the authors after the meeting.
- You will find your **poster board number** in the author index at the end of the abstract book.

Presence at Posters

In order to enable discussion and interaction with other participants, we request you or one of your group to be at your poster board between:

- **11.15 and 12.15 hrs on Sunday, May 24** for **posters with odd numbers**
  (e.g. P04.001, P04.003 - this refers to your final poster board number - not the abstract control number!)
  or
- **11.15 and 12.15 hrs on Monday, May 25** for **posters with even numbers**
  (e.g. P07.002, P07.004 - this refers to your final poster board number not the abstract control number!)

If this is not possible, please leave a note on your poster board detailing the times when you will be present at the board.

Programme Changes

The organisers cannot assume any liability for changes in the programme due to external or unforeseen circumstances.

Spoken Presentations - Projection and Technical Setting

- All rooms will be equipped with data- and overhead projection (no slides).
- It is essential that you load and view your presentation in the media check/preview room (Level OE) preferably in the morning of the day your talk is scheduled, but not later than 2 hours in advance.
- The lecture rooms are exclusively equipped with Windows-PCs (no Macintosh machines). In case you absolutely need to use your own laptop or notebook, please contact the preview center well in advance of your talk.
- Please bring a USB-stick, CD-ROM, ZIP- or floppy disk all formatted for Windows® (PC). You may want to carry a second disk/CD as a back-up in case there is any insoluble technical problem.
- File Format: Microsoft® Power Point™ presentation formatted for Windows® (PC) only. (Operating system: Windows XP®)
- Preferred Resolution: XGA (1024 x 768 pixel)

ESHG Poster Awards

For the second time, the ESHG proposes the ESHG Poster award for the 7 best posters presented at the meeting. Finalists receive a complementary ESHG online membership for 1 year. The 2 winners will receive an iTouch.

The ESHG Scientific Programme Committee has selected a number of candidates for the ESHG Poster Award. Posters can be identified by a rosette on the board.

ESHG Poster Award Candidates

P01 Genetic counseling, including Psychosocial aspects, Genetics education, Genetic services, and Public policy
P01.03 The role of religious involvement, knowledge about and attitudes towards adult genetic testing in formation of intentions-to-test - a structural equation modeling approach, *A.* ***Botoseneanu*** *et al. Ann Arbor, MI*

P02 Clinical Genetics and Dysmorphology
P02.008 Delineation of a lethal autosomal recessive disorder characterized by alveolar capillary dysplasia and limb anomalies, *A.* ***Innes****, Calgary*
P02.055 A very early juvenile Huntington disease revealed by cerebellar ataxia in a 2 years old boy, *T.* ***Frébourg****, Rouen*
P02.142 Transcriptional hallmarks of Noonan syndrome in peripheral blood mononuclear cells, *G. B.* ***Ferrero****, Torino*
P02.146 Molecular analysis of the Noonan (-like) Syndromes: overview of 7 years of DNA diagnostics in the Netherlands, *H. G.* ***Yntema****, Nijmegen*
P02.155 Array-CGH analysis in a series of 54 index patients with limb malformation identified more than 10% anomalies, *S.* ***Manouvrier-Hanu****, Lille*
P02.168 Expression and functional analysis of EFNB1 mutations in craniofrontonasal syndrome, *R.* ***Makarov****, Magdeburg*
P02.185 Congenital variant of Rett syndrome due to the FOXG1 gene, *F.* ***Mari****, Siena*
P02.195 Developmental delay and a distinctive facial appearance in two families with Xq25 duplications, *A.* ***Philippe****, Paris*
P02.199 Towards understanding the pathogenetic mechanism of PQBP1 mutations in X-linked mental retardation, *V. M.* ***Kalscheuer****, Berlin*
P02.200 A recurrent copy number gain at Xq28 in four families with mental retardation reveals a dosage-dependent severity of the phenotype and suggests a novel recombination mechanism, *H.* ***Van Esch****, Leuven*

P03 Cytogenetics
P03.022 A disposable “microfluidic” chip for diagnostic FISH screening of genetic diseases, *R.* ***Carbone****, Milan*
P03.054 Evidence for significance of epigenetic inactivation of the cell-cycle checkpoints genes into etiology of chromosomal mosaicism during embryo development, *I. N.* ***Lebedev****, Tomsk*
P03.069 Independent patients with an identical 9q31.1q31.3 deletion showing similar clinical features: a new microdeletion syndrome?, *P.* ***Magini****, Bologna*
P03.080 De novo cryptic deletion at 2q14 in two female patients with Turner syndrome stigmata, *S.* ***Giglio****, Florence*
P03.099 Detection of low-level mosaicisms by array CGH, *I. Vanhevel, Leuven*
P03.145 De novo unbalanced translocations: how many of them have a post-zygotic origin?, *M. C.* ***Bonaglia****, Bosisio Parini (LC)*

P04 Reproductive genetics
P04.07 Superovulation in mice alters the methylation pattern of imprinted genes in the sperm of the offsprings, *A.* ***Paoloni-Giacobino,*** *Geneva*

P05 Prenatal and perinatal genetics
P05.46 Is nuclear transfer morally acceptable as a means to prevent mtDNA disorders if it cannot avoid residual health risks?, *A. L.* ***Bredenoord****, Maastricht*
P05.59 Large scale application of QF-PCR for rapid prenatal diagnosis of common chromosome aneuploidies, results of nine years clinical experience, *V.* ***Cirigliano****, Barcelona*

P08 Statistical genetics, includes Mapping, linkage and association methods
P08.10 Genome-wide association of pulse wave analysis phenotypes in two isolated populations, *C. S.* ***Franklin****, Edinburgh*
P08.19 Selection and migration as possible causes of failure of case-control study replications, *C. G. F.* ***de Kovel****, Utrecht*
P08.49 Genome-wide parametric linkage analysis of adult height, *T. I.* ***Axenovich****, Novosibirsk*

P09 Complex traits and polygenic disorders
P09.003 Genetic variation within adiponutrin is associated with lipoprotein metabolism and liver function, *B.* ***Kollerits****, Innsbruck*
P09.032 Novel insights into pathogenesis of coeliac disease: a second genome-wide association study, *G.* ***Trynka****, Groningen*
P09.048 A genome wide analysis identifies genetic variants in the RELN gene associated with otosclerosis, *I.* ***Schrauwen****, Wilrijk*
P09.108 Genetic variation modifies the effect of ACE-inhibitor treatment: a step towards personalized medication, *A.* ***Isaacs****, Rotterdam*

P10 Evolutionary and population genetics, and Genetic epidemiology
P10.31 Genetic differences between four European populations, *V.* ***Moskvina****, Cardiff*
P10.32 Conditional linkage and genome-wide association studies identify UGT1A1 as major gene for anti-atherogenic serum bilirubin levels - a Framingham Heart Study, *F.* ***Kronenberg****, Innsbruck*
P10.43 Coevolution of the repeated glutamine and proline codons in the mammalian Huntington disease gene, *D.* ***Savic-Pavicevic****, Belgrade*

P11 Genomics, Genomic technology including bioinformatics methods, gene structure and gene product function and Epigenetics
P11.016 The regulation of CDK5R1 gene expression by miRNAs may have a role in Alzheimer’s disease, *S.* ***Moncini****, Milan*
P11.020 Long range expression effects of copy number variation: insights from Smith-Magenis and Potocki-Lupski syndrome mouse models, *G.* ***Ricard****, Lausanne*
P11.021 Assessment of Copy Number Variation on a Large Population-Based Cohort, The Rotterdam Study, *K.* ***Estrada****, Rotterdam.*
P11.065 The Gen2Phen project: Collecting gene sequence variants and their phenotypic consequences in web-based LSDBs for Mendelian disorders, *I. F. A. C.* ***Fokkema****, Leiden.*
P11.072 Expression profiles of small RNAs from various tissues generated by SOLiD™ sequencing, *R.* ***Tanzi****, Foster City, CA*

P12 Molecular basis of Mendelian disorders
P12.020 Mutations of the EPHA2 receptor tyrosine kinase gene cause autosomal dominant congenital cataract, *X.* ***Zhang****, Beijing*

P14 Therapy for genetic disorders
P14.12 Systemic gene therapy for cardiomyopathy and muscular dystrophy of the BIO14.6 hamster, *I.* ***Rotundo****, Naples*

P17 Genetic analysis, linkage and association
P17.27 Large scale genome wide association study identifies new genetic loci determining homocysteine levels, *J. B. J.* ***van Meurs****, Rotterdam*

ESHG Award

The ESHG Award, formerly "Mauro Baschirotto Award", was founded in 1992 and is presented by the European Society of Human Genetics during its annual European Human Genetics Conference in recognition of individual achievement in human genetics. The laureate receives a cheque of EURO 1.500.- to cover the expenses of participating in the meeting.

Award Holders

1992 Lore **Zech**
1993 Pierre **Maroteaux**
1994 Mary **Lyon**
1995 Jean **Weissenbach**
1996 Malcolm **Ferguson-Smith**
1997 Leena **Peltonen**
1998 Jean-Louis **Mandel**
1999 Pat **Jacobs**
2000 Dirk **Bootsma**
2001 Robin **Winter**
2002 Albert **de la Chapelle**
2003 Peter S. **Harper**
2004 Bernhard **Horsthemke**
2005 Stylianos Antonarakis
2006 Veronica van Heyningen
2007 Andrea Ballabio
2008 Arnold Munnich
2009 Kari Stefansson

ESHG Young Investigator Awards

The ESHG Young Investigator Awards are granted for outstanding research by young scientists presented as a spoken contribution at the conference.

The Scientific Programme Committee has shortlisted the following for the **ESHG Young Investigator Award**. The committee will judge finalists' presentations during the conference. Winners will be announced, and awards made in the closing ceremony on Tuesday, May 26, 2009 at 14.00 hrs.

The **Isabel Oberlé Award** is awarded yearly since 2002 for best presentation by a young scientist on research concerning the genetics of mental retardation.

The **Lodewijk Sandkuijl Award** was instituted in 2004 to be awarded to the author of the best presentation at the ESHG conference within the field of complex disease genetics and statistical genetics.

Talks of finalists are highlighted by a * as well as a gray background in the detailed programme.

The profiles as well as a short interview of the finalists can be found on the next pages.

Mirna Assoum, Illkirch, France
Talk: C03.4 Search for genes implicated in new forms of recessive ataxia
Session: C03 Cerebellar disorders
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: Dec 15th,1979,Tripoli-Lebanon
What is your current position? PhD Student in the department of Neurobiology and Human Genetics, IGBMC, Illkirch, France
Why did you choose a career in genetics? Human Genetics is my favorite subject.I was always fascinated by the work of researchers in the field of human genetics. Now i am glad to be one of them.
What do you really like about this research? There are many rare human genetic diseases which are still not discovered. The study of these diseases allows us to define a clinical table and can be useful for therapeutic approaches.
What is your ultimate goal in life? To be always ambitious .I hope to improve the understanding of human diseases.

Bérénice A. Benayoun, Paris, France
Talk: C11.4 Positive and negative feedback regulates the transcription factor FOXL2 in response to cell stress: evidence for a regulatory imbalance induced by disease-causing mutations
Session: C11 Molecular Basis of Mendelian Disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: June 19th, 1984, Neuilly-sur-Seine, France
What is your current position? PhD candidate, Pr. Reiner A. Veitia's laboratory, Université Paris VII/Institut Jacques Monod, Paris, France
Why did you choose a career in genetics? After reading a vulgarization article about the genetic mechanisms of evolution as a teenager, I became most interested in working out how life and evolution worked and how genes contained the necessary information to build up a complete living organism. As I continued my formation, I realized I wanted to participate in the elucidation of how genetic dysfunctions could produce diseases and how genetic variations could regulate complex processes, such as ageing. With this path, I love knowing I'm going to learn something everyday, or at least try to do so.
What do you really like about this research? I like that I'm participating in the collective effort to decrypt and understand the molecular mechanisms/bases of ageing. Moreover, since the gene I'm focussing on is involved in a human syndrome, I really like that what I do and what I can find out through research could ultimately help patients.
What is your ultimate goal in life? Be useful, be happy and never stop learning.

Sarah Boissel, Paris, France
Talk: C11.5 Loss-of-function mutation in the dioxygenase-encoding, obesity-associated FTO gene causes severe growth retardation and multiple malformations
Session: C11 Molecular basis of Mendelian disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: March 27th, 1982, Paris, France.
What is your current position? I am a 3rd year PhD student in the Department of Human Genetics of Necker-Enfants Malades Hospital in Paris. I am looking for genes involved in autosomal recessive mental retardation.
Why did you choose a career in genetics? For me, working in human genetics is the prefect combination of biological and medical research. A better knowledge of the genetic basis of diseases allows scientists to better understand how these diseases develop and progress. Moreover, I hope that it opens new fields in molecular medicine development.
What do you really like about this research? I enjoy keeping my mind active everyday and I like the excitement and the ups and downs of research. Last but not least, it is important to me that my work could help the patients and their families.
What is your ultimate goal in life? To learn a lot of things everyday and to try to be useful at work. And of course, to be happy in my personal life!

Evelyne Chaignat, Lausanne, Switzerland
Talk: C15.1 Segmental copy number variation shapes tissue transcriptomes
Session: C15 From genome to phenome
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs
Date and city of birth: 21 May 1981

What is your current position? PhD student at University of Lausanne
Why did you choose a career in genetics? Aiming to understand what we are made of...
What do you really like about this research? It is a very challenging field where new techniques and concepts are continuously uncovered.
What is your ultimate goal in life? Become a Tea Master...

Randy Chandler, Bethesda, United States
Talk: C12.3 Rescue of a Lethal Murine Model of Methylmalonic Acidemia using rAAV8 Mediated Gene Therapy- One Year Post-Treatment
Session: C12 Pathophysiology and therapy for genetic disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: June 9, 1966 C<amden, New Jersey
What is your current position? PhD. candidate George Washington University, Washington, D.C. and biologist, National Human Genome Research Institute, National Institutes of Health, Bethesda Maryland

Why did you choose a career in genetics? I was trained as a biologist and work as an environmental scientist after graduating from college. After sometime, I realized that my real passion was in genetic research and changed careers.

What do you really like about this research? I truly enjoy being involved in translational research of metabolic disorders knowing that my research efforts may benefit individuals with this group devastating diseases.

Brigitte Chhin, Lyon, France
Talk: C12.6 Ciliary beating recovery in deficient human airway epithelial cells after lentivirus ex vivo gene therapy
Session: C12 Pathophysiology and therapy for genetic disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

D**ate and city of birth:** September 23th, 1979, in Venissieux, France

What is your current position? Post-doc in the team of Prof Bénédicte Durand at the Centre de Génétique Moléculaire et Cellulaire, Lyon.
Why did you choose a career in genetics? I was touched by the case of patients affected by rare diseases and here I am!
What do you really like about this research? We can work either on basic (what I am currently doing) or applied research (what I used to do) to contribute at the understanding and therapy of human genetic diseases.
What is your ultimate goal in life? To be happy and to clarify the maximum of scientific 'mysteries' before having Alzheimer's disease...

Imke Christiaans, Amsterdam, Netherlands
Talk: C13.1 Clinical characteristics of distantly related families with idiopathic ventricular fibrillation linked to chromosome 7q36
Session: C13 Cardiac Genetics
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs
and
Talk: C18.2 Genetic counselling and cardiological care in predictively tested hypertrophic cardiomyopathy mutation carriers: the patients' perspective
Session: C18 Genetic counseling and services
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: March 29th, 1979, Wijk bij Duurstede, the Netherlands
What is your current position? I'm working as a resident in Clinical Genetics at the Academic Medical Centre in Amsterdam, the Netherlands. Currently, I am also finishing my PhD project on hypertrophic cardiomyopathy.
Why did you choose a career in genetics? During my medical internships I became interested in the field of clinical genetics because it combines two elements of care that are important for me. Firstly, genetics is a very broad field that involves all organ systems but a the same time I also have to focus and do research for difficult patient cases. Secondly, I very much enjoy communication with patients; understanding their situation and explaining what the genetic condition could mean for them.
What do you really like about this research? My research allows me to look for answers on questions I come across in clinic. I especially enjoy it when my research is of direct use for patients.
What is your ultimate goal in life? To optimise my happiness in personal and professional life.

Sebastian Eck, Munich, Germany
Talk: C06.5 Detection of tumor-specific somatic mutations by transcriptome sequencing of a cytogenetically normal acute myeloid leukemia
Session: C06 Cancer genetics
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 11.07.1981, Munich
What is your current position? PHD student at the Helmholtz-Zentrum München, Institute of Human Genetics.
Why did you choose a career in genetics? Having studied Bioinformatics it was always a priority for me to not only work theoretically on algorithm design but rather employ these methods to real biological problems and research.
What do you really like about this research? To have an interesting and interdisciplinary field of work where I can work in a motivated team with scientists from different branches of study. Additionally, my positions allows me to work with state of the art technology.
What is your ultimate goal in life? To live happily ever after.

Carole Fantini-Hauwel, Aix en Provence, France
Talk: C18.1 Links between psychological characteristics and Distress after genetic result announcment : emotional regulation and adjustment
Session: C18 Genetic counseling and services
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Ilse Feenstra, Nijmegen, Netherlands
Talk: C02.4 Interstitial 18q21 microdeletions and a microduplication including the TCF4 gene causing Pitt Hopkins syndrome
Session: C02 Clinical Genetics I
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: October 12, 1975, Heemskerk, The Netherlands
What is your current position? I am working as a clinical geneticist in training in the department of Human Genetics in Nijmegen. Furthermore, I am doing a PhD project concerning genotype-phenotype correlations in patients with various unbalanced chromosome aberrations.
Why did you choose a career in genetics? There are several reasons why I choose for a profession within clinical genetics. For example the broad working field; as a clinical geneticist you see patients of all ages with very different sort of diseases. Also the continuing development of new diagnostic techniques and the implementation of that in daily genetic practice is something I like. The contact with patients and explaining sometimes complex matters is very valuable to me.
What do you really like about this research? My research is about rare unbalanced chromosome aberrations and this rarity makes it very difficult for parents and patients to get information about the disorder. By collecting as much data as possible on genotype-phenotype correlations, we can provide better information for patients and their family. This information can be crucial in the medical follow up of patients. Furthermore, when parents receive the message their child has a rare chromosome disorder, this can be a huge shock leading to many emotions and the desire to learn more about the disorder, also including non-medical issues.
What is your ultimate goal in life? To keep on growing, in various aspects of life.

Lilach Friedman, Tel Aviv, Israel
Talk: C14.1 The vertebrate inner ear microRNAs have unique spatial and temporal expression patterns and are crucial for inner ear development and survival
Session: C14 Neurogenetics
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: May 23, 1971 in Rishon Le-Zion, Israel.
What is your current position? I am a post-doctoral fellow in Prof. Karen Avraham's laboratory, studying genetics of the mammalian inner ear, at Tel Aviv University, Israel. I am studying microRNAs that may have a role in the inner ear.
Why did you choose a career in genetics? As of a young age, I was interested in biology of mammals in general and in medical sciences in particular. Most of all, I was intrigued by studies with a practical outcome. My Ph.D. is in immunology and aimed to develop better immunotherapies against cancer. During the last year of my Ph.D. studies, I was introduced to microRNAs, the roles of which have only begun to be revealed. I was fascinated by the possible applicatons hidden in these new tiny regulatory molecules. Therefore, I decided to do my post-doctoral training in genetics and study microRNAs.
What do you really like about this research? This is a whole new area in molecular biology, which brought about a revolution in our understanding of the regulation of gene expression. It is exciting to work in an area, where new findings may overturn our (molecular biology) world view. It is fascinating to participate in exposing the tip of the complex regulation of gene networks in mammalian cells, which are currently only superficially understood.
What is your ultimate goal in life? My professional goal is to conduct research with practical implications that will improve human health and quality of life. In addition, I hope to build a happy and warm family – and to successfully combine these two goals for a happy and satisfactory life.

Louise Harewood, Lausanne, Switzerland
Talk: C16.1 The Effect of Translocation-Induced Nuclear Re-organization on Gene Expression
Session: C16 Clinical Genetics II
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: 28th February, 1978 in Warrington, UK
What is your current position? Postdoc in the lab of Alexandre Reymond at the Centre for Integrative Genomics, Lausanne
Why did you choose a career in genetics? I'd always had an interest in genetics throughout school but then the reading of 'Perilous Knowledge' by Tom Wilkie made me realise that I wanted to be a part of the human genetics field. This was enforced by a thoroughly enjoyable degree course and the discovery of my love of human chromosomes. Since then, I've never looked back.
What do you really like about this research? I enjoy the puzzle of connecting genotypes to phenotypes and the unpredictability of human genetics in general. I also still, to this day, get excited at the appearance of a human G-banded karyotype!
What is your ultimate goal in life? The same as everybody else, I imagine. To be happy and content both personally and professionally and to feel that I've done something worthwhile with my life.

Iris Heid, Neuherberg, Germany
Talk: C10.6 Meta-analysis of genome-wide scans identifies three novel loci influencing central obesity including one with a women-specific association with waist-hip-ratio
Session: C10 Complex genetics I
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: 1968, 16th January, Munich, Germany
What is your current position? Post-Doc at the Helmholtz Zentrum München, Institute of Epidemiology
Why did you choose a career in genetics? The many open questions about biological pathways to dieases and the possibilities from new technologies to answer these important questions
What do you really like about this research? The interdisciplinary work, the puzzles to be solved, the (however small) contribution that a researcher can provide
What is your ultimate goal in life? to combine a fulfilled professional life with a balanced family life with my husband and my two children

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Alexander Hoischen, Nijmegen, Netherlands
Talk: P2.2 Massive parallel sequencing of ataxia genes after array-based enrichment
Session: P2 What's New?
Date: Saturday, May 23, 2009, 16.30 - 20.00 hrs

Date and city of birth: 11.07.1977, Hamm, Germany
What is your current position? Research post-doc in the Genomic Disorders Group, Department of Human Genetics, St. Radboud University Medical Center Nijmegen, The Netherlands.
Why did you choose a career in genetics? Isn't genetics the basic of (almost) everything (in life-sciences)? A research career in this field combines the possibility to do what you're curious about and do that in a network of creative people. It offers great freedom and the chance to discover something new.
What do you really like about this research? The combination of basic research with high clinical impact is of course very attractive. To know that you do work for patients, e.g. in deciphering the genetic causes of a disease, gives you certainly high motivation. On top, it is fascinating that the fast evolving technologies offer possibilities to answer questions researches haven't been able to answer for years.
What is your ultimate goal in life? To be surrounded by family and friends when aged at least 90 and look back on a fulfilled life (which always combines private and professional life), without regretting too much.

Shuwen Huang, Salisbury, United Kingdom
Talk: C16.6 An International Standardized Cytogenomic Array (ISCA) Consortium approach to the design, implementation and reporting of constitutional oligo array-CGH
Session: C16 Clinical Genetics II
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Eva Klopocki, Berlin, Germany
Talk: P2.4 Duplication of conserved non-coding sequence elements - a novel mechanism in the pathogenesis of congenital malformations
Session: P2 What's New?
Date: Saturday, May 23, 2009, 16.30 - 20.00 hrs

Date and city of birth: May 4th, 1975 in Wuppertal, Germany
What is your current position? Postdoctoral fellow and head of the array CGH lab at the Institute for Medical Genetics, Charité Universitätsmedizin Berlin, Germany
Why did you choose a career in genetics? The mechanisms underlying human development as well as human disease fascinated me already as a student. After my PhD in cancer research at a Berlin Biotech company I took the opportunity to join the Institute for Medical Genetics at the Charité to work at the exciting interface between basic and clinical research.
What do you really like about this research? Human Genetics is one of the fastest moving fields in Science - it is a great experience for me as a researcher to be part of this development. In my work I am currently investigating the impact of genetic changes in long-range regulators of gene expression on embryonic development and the relevance for congenital malformations which continues to yield interesting as unexpected results. Last but not least I enjoy the interdisciplinary work with other scientists and clinicians as well as the interaction with patients.
What is your ultimate goal in life? On and on 'cause the road is never ending ...

Zoltan Kutalik, Lausanne, Switzerland
Talk: C05.6 A Comparison of Methods for Testing Association Between Uncertain Genotypes and Quantitative Traits
Session: C05 Statistical genetics
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 30 December 1978, Budapest, Hungary
What is your current position? Post doc at the Department of Medical Genetics at the University of Lausanne, Switzerland.
Why did you choose a career in genetics? I found all branches of biology quite boring at high school, except for genetics. However, maths was my utmost favourite. It was not until my exchange year at the university when I met the first real application of mathematics... in biology. This made me realise that rather than pursuing a career in pure mathematics, I want to find an exciting field where maths is of quintessential use. That is how I stumble across genetics.
What do you really like about this research? This field is extremely interdisciplinary where so many skills are needed. It is like putting together a gigantic puzzle, where solving a part of it just triggers new questions. I like the freedom of research and the creativity it requires.
What is your ultimate goal in life? To discover all dimensions of happiness.

Iñigo Landa, Madrid, Spain
Talk: C06.4 Identification of Low Penetrance Genes associated to thyroid cancer susceptibility using a two-step case-control approach
Session: C06 Cancer genetics
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: November 18th, 1982, Pamplona (Spain)
What is your current position? PhD student, Hereditary Endocrine Cancer Group, Spanish National Cancer Research Centre

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Pirkka-Pekka Laurila, Helsinki, Finland
Talk: C10.2 Genome-wide association analysis and expression analysis from adipose tissue reveals coagulation factor XIII as a novel candidate gene for low HDL-cholesterol
Session: C10 Complex genetics I
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: March 3, 1982 Helsinki, Finland
What is your current position? I'm doing my thesis on cardiovascular genetics at the National Institute for Health and Welfare in Professor Leena Peltonen's lab as an MD/PhD student. I'm also finishing my medical studies at the University of Helsinki.
Why did you choose a career in genetics? I have always been fond of science, even if it was physics and chemistry rather than biology during secondary school. Starting my thesis work on human genetics has shown to me that biology itself is thrilling and genetics, being the fundamental basis for all molecular disciplines, is the most exciting thing in the world! Genetics is evolving at an extremely fast pace as a result of the complete sequence of the human genome, and major breakthroughs leading to a better understanding of human diseases emerge almost every week. It is a treat to work in the global team of geneticists having such a common spirit!
What do you really like about this research? This study successfully combines the powers of genome-wide association analysis and genome-wide expression analysis from adipose tissue. Many GWAs yield results that are unexpected, paving the way for new, previously unimaginable discoveries. Who would have thought that coagulation factors play a role in lipid metabolism? There is enormous potential in this hypothesis-free approach on human macro-molecules when used wisely. The old guys get brand new roles, and nothing can be more exciting for a scientist than taking 'the road less traveled'!
What is your ultimate goal in life? I want to set an example, whether it was in science, art, or personal life. I hope I can remain active and forward-looking in my life and be able to help other people get the best out of them.

Derek Lim, Birmingham, United Kingdom
Talk: C08.6 Germline mutation in NLRP2 (NALP2) in a familial imprinting disorder (Beckwith-Wiedemann Syndrome).
Session: C08 Molecular dysmorphology
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: September 27th 1976, Petaling Jaya, Malaysia
What is your current position? Clinical Research Fellow in the Department of Medical and Molecular Genetics at University of Birmingham and Honorary Specialist Registrar in Clinical Genetics at the West Midlands Regional Genetics Service.
Why did you choose a career in genetics? The close links between the science and clinical work in genetics is both exciting and rewarding.
What do you really like about this research? I have the opportunity to investigate epigenetic mechanisms in genomic imprinting and cancer, which are pet interests of mine,and also work under an inspirational supervisor.
What is your ultimate goal in life?
Making a difference.....and also starting a rock band of geneticist called The Polymorphisms! Any takers?

Lucia Micale, San Giovanni Rotondo, Italy
Talk: C06.3 TheTRIM8 gene is a novel player of p53 pathway
Session: C06 Cancer genetics
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: Febrary 6th, 1976, Modugno, Italy
What is your current position? Post-Doc at the Medical Genetics Unit, IRCCS, Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy.
Why did you choose a career in genetics? To understand the mysterious and molecular mechanisms underlying the human genetic diseases and the genetics events that lead to the tumors.
What do you really like about this research? I really like the several molecular strategies used in this study to understand the role of the TRIM8 gene in the pathogenesis of brain tumors. In a near future, I'd like that our results could improve somehow the life-expectancy of patients.
What is your ultimate goal in life? That all events of my life come from choices done with passion and serenity

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Raluca Mihaescu, Rotterdam, Netherlands
Talk: C18.3 Evaluation of risk prediction updates from commercial genome-wide scans
Session: C18 Genetic counseling and services
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: 6-09-1977, Iasi, Romania
What is your current position? PhD Student at the Department of Epidemiology, Erasmus University Medical Center, Rotterdam, The Netherlands
Why did you choose a career in genetics? Genetic profiling is expected to have a huge impact on the prediction of common diseases and both scientists and the general public show an increased interest in genetic testing. Therefore, I think that it is very important to accurately assess the existing methodology and to explore new methods for the evaluation of genomic profiling.
What do you really like about this research? My project focuses on the methods needed to translate genetic association discoveries into clinical and public health applications. I like that it integrates complex information and delivers simple practical messages, useful for the clinical and public health practice.
What is your ultimate goal in life? I would like to develop myself as a person and to be an inspiration for the others.

Sergey Nikolaev, Geneva, Switzerland
Talk: C01.3 Genomic variation detection by DNA selection and high throughput sequencing
Session: C01 Next Generation Sequencing
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 27.12.1978, Moscow
What is your current position? Research Associate, Department of Genetic Medicine and Development, University of Geneva Medical School
Why did you choose a career in genetics? I wanted to study how phenotypic and behavioral traits of an individual are encoded in the genome.
What do you really like about this research? I think that this is the field where the scientific achievements could be used in medical practics pretty soon.
What is your ultimate goal in life? I'd like to be involved in the development of the method that would allow high scale diagnostics of patients with genetic disorders

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Silke Nuber, Tuebingen, Germany
Talk: C12.2 Neuropathology of Alpha-Synuclein and Synphilin-1 transgenic Mouse Models of Parkinson's Disease
Session: C12 Pathophysiology and therapy for genetic disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Astrid Out, Leiden, The Netherlands
Talk: C01.5 Estimation of MUTYH variant frequencies in pooled DNA with massive parallel sequencing
Session: C01 Next Generation Sequencing
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 15 November 1980, Purmerend, The Netherlands
What is your current position? PhD student at the Center for Human and Clinical Genetics, Leiden University Medical Center, The Netherlands. I am working on genetics behind familial breast and colorectal cancer.
Why did you choose a career in genetics? Clinical and scientific internships in genetics during Medical School motivated me to continue in this direction. Both learning to understand our biology and to help patients are very rewarding to me. Furthermore, my monozygotic twin sister, who is genetically the same, but not identical to me as a person, has inspired me to start investigating how genetics work and maybe what more is there underlying our design.
What do you really like about this research? I really like to be involved in unraveling the functions of our DNA code that lies at the basis of our being in beneficial and disadvantageous ways. The fastly evolving technologies facilitating this may supply us with knowledge beyond our imagination. Implementing this knowledge will hopefully improve our quality of life.
What is your ultimate goal in life? To be creative and enjoy the full palette with the colors of life, in work, hobbies, nature and the people around me.

Olli Pietiläinen, Hinxton, Cambridge, United Kingdom
Talk: C17.2 A regional high risk isolate for schizophrenia reveals an enrichment of three large copy number variations overlapping developmental genes
Session: C17 Complex genetics II
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: January 10 1982, Helsinki, Finland
What is your current position? I'm a Ph.D student at Porf. Leena Peltonen's group in the Wellcome Trust Sanger Institute, Cambridge, UK
Why did you choose a career in genetics? I guess I have always liked science and especially been facinated by human biology. Already when I was a child I thought that I will be a researcher when I grow up. After graduating from high school I started studying chemistry at university of Helsinki. Then one summer I had the opportunity to work in Prof. Pelotnen's laboratory and soon I was swept away by genetics.
What do you really like about this research? It's difficult to say exactly what I like most in research. I like my work in general. One of the best things is the excitement when going through some new results waiting what you will find. It's like unwrapping a birthday present.
What is your ultimate goal in life? To live a happy and fulfilling life.

Sandra Puskaric, Heidelberg, Germany
Talk: C11.2 Shox2 mediates Tbx5 activity by regulating Bmp4 in the sinus venosus of the developing heart
Session: C11 Molecular basis of Mendelian disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: November 1, 1980, Mannheim, Germany
What is your current position? I am currently a PhD student at the Department of Human Molecular Genetics in Heidelberg, Germany
Why did you choose a career in genetics? I first became interested in molecular biology in high school. During this time my biology teacher Mr. Sommer convinced me to study biology, a decision which I have never regretted. My studies taught me that genetics is one of the most interesting and fascinating scientific fields.
What do you really like about this research? I like the combination of biological and medical research that genetics provides. Genetics also creates the exciting opportunity to be involved in elucidating the molecular mechanisms that underlie genetic disorders.
What is your ultimate goal in life? To stay a happy and content person in all aspects of life.

Jihane Romanos, Groningen, Netherlands
Talk: C09.1 Genetic risk model for coeliac disease helps identify high-risk individuals.
Session: C09 Evolutionary and population genetics and Genetic epidemiology
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: 23 January 1982, Beirut, Lebanon
What is your current position? 2nd year PhD student at the Department of Genetics, University Medical Center of Groningen, Gronigen, the Netherlands.
Why did you choose a career in genetics? Since high school, genetics was one of the themes that I loved to study and learn more about. When I started my undergrad in biology, genetics was one of the fields that were developing very fast with a promise that one day, all this knowledge could be used to predict, prevent or treat patients. Since this was what I would like to do in my career, I chose genetics and mainly human genetics.
What do you really like about this research? Human genetics is developing very fast and every day we are closer to use genetics in prediction, prevention and treatment of diseases. Being able to help in getting closer to this goal is my main satisfaction.
What is your ultimate goal in life? Try to combine a happy personal life and a successful professional life.

Serena Sanna, Monserrato, Italy
Talk: C10.4 Genome-wide association scan for bilirubin levels in a Sardinian population
Session: C10 Complex genetics I
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: November 15, 1980, in San Gavino M.,Sardinia, Italy
What is your current position? I'm currently a Post doctoral collaborator at the Institute of Neurogenetic and Neuropharmacology of the Italian National Council of Research, in Monserrato, Italy.
Why did you choose a career in genetics? After I graduated in Math I found a job in a molecular lab. I honestly hated biology at the high school, and I could not remember any notion. So, I had to get back on my books and I realized that "chromosomes", "nucleotides" and "mitochondrial DNA" are not monsters but actually very interesting parts of everyone's body, including mine! I discovered how much of the biology is regulated by math models, and that all the theory on statistics, analysis, programming is extremely helpful to understand the protein's structure, the etiology of diseases, the evolution of a population, and more. How fantastic is Science: one, big, unlimited world - and I want to discover it!
What do you really like about this research? I think what it's attracting and motivating me is that I'm studying my population: the Sardinian. Isn't great that a Sardinian scientist, studying the Sardinia population in a Sardinian lab, is able to discover something that may be useful for Sardinian's and world while people's health?? I also found that bilirubin is interesting for salsa lovers as me - listen La Bilirrubina by Juan Luis Guerra!
What is your ultimate goal in life? I would like to have two big families: my private one, with my husband and my children, and the business one, with a lot of students and colleagues working in my OWN lab. Very recently two students started to work with me, so I hope the "other family" will grow soon!

Arne Schäfer, Kiel, Germany
Talk: C10.5 Identification of a Shared Genetic Susceptibility Locus for Coronary Heart Disease and Periodontitis
Session: C10 Complex genetics I
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: August 10th 1972, Mannheim
What is your current position? Postdoctoral fellow
Why did you choose a career in genetics? I read the cv of Kary Mullis
What do you really like about this research? It brings insight into the secrets of life with a respectable resolution.
What is your ultimate goal in life? As anyone's, to optimise my life reproductive potential

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Thomas Schwarzbraun, Graz, Austria
Talk: P2.5 Mitosis updated - PICH and the anaphase threads
Session: P2 What's New?
Date: Saturday, May 23, 2009, 16.30 - 20.00 hrs
Date and city of birth: June 27th 1980, Graz, Austria

What is your current position? Postdoc in Michael Speicher's group at the Institute of Human Genetics in Graz
Why did you choose a career in genetics? Didn't work out as an astronaut...
What do you really like about this research? Gaining insights into the very fundamentals of life
What is your ultimate goal in life? To finally learn an instrument and start playing in the other candidate's rock band

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Andrew Sharp, Geneva, Switzerland
Talk: C15.2 Methylation profiling in cases with uniparental disomy identifies novel imprinted genes on chromosome 15
Session: C15 From genome to phenome
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: 9th June 1974, Brighton, England
What is your current position? Marie Curie Fellow, Department of Genetic Medicine and Development, University of Geneva Medical School, Switzerland
Why did you choose a career in genetics? My career in Genetics began more by blind chance than intention. In fact, during my undergraduate degree I disliked lab work so much I decided that I never wanted to work in a lab! How wrong I was, as now many years later, I would not want to be doing anything else. I think that having a job which allows freedom and creativity of thought in doing research which discovers new things about the genetic causes of human disease is a fantastic career. I would thoroughly recommend it to anyone!
What do you really like about this research? I have always had an interest in epigenetics since I first started working in research. This project is a very elegant idea that has been in the back of my mind for a while, but has been catalysed only recently by technological advances that now allow the epigenome to be profiled on a single array. Like all good experiments, at its heart is a simple idea that asked in the right way, allows powerful discoveries to be made. I believe that some of the great advances in genetics over the next few years will come from the field of epigenetics, and this work gives some preliminary insights into the phenomenon of how variation in the epigenome can influence human phenotypes.
What is your ultimate goal in life? To remain happy and contented.

Javier Simón-Sánchez, Amsterdam, Netherlands
Talk: C17.1 Genome-wide analysis in Parkinson's disease
Session: C17 Complex genetics II
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: June 20th 1980, Madrid
What is your current position? I'm a postdoc at the Department of Clinical Genetics of the Vrije Universiteit Medical Center under the supervision of Peter Heutink.
Why did you choose a career in genetics? While finishing my B.S. degree in the "Universidad Autónoma de Madrid", Adriano Jiménez-Escrig gave me the opportunity to work with him at the "Hosptal Ramon y Cajal" on the genetic basis of Alzheimer's disease. Since I found this research fascinating I decided to make a career on genetics of human traits such as disease.
What do you really like about this research? What I like about this research is that using a variety of methodology we are able to understand the genetic basis of devastating human disorders. In a near future, this information will be clinically meaningful with respect to risk profile, disease prognosis and response to a specific treatment.
What is your ultimate goal in life? Identifying genetic variability that causes or contributes to the etiology of different neurological diseases (such as Parkinson's disease or Frontotemporal lobar degeneration) in order to facilitate the understanding of the molecular pathways that underly these neurological disorders.

Sérgio Sousa, Coimbra, Portugal
Talk: C08.1 Nicolaides-Baraitser Syndrome - Delineation of the Phenotype
Session: C08 Molecular dysmorphology
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: 31th December 1977, Coimbra Portugal
What is your current position? Medical/Clinical Geneticist, Medical Genetics Department, Paediatric Hospital of Coimbra, starting this year a PhD research project on molecular dysmorphology.
Why did you choose a career in genetics? Since the first years of medical school, genetics fascinated me as it enabled me to understand so many biological phenomenons. This attraction for the search of the ultimate causes accompanied me during the clinical years and the area of Medical/Clinical Genetics strikingly brings together these two aspects: the contact with the patient and his family and the complex scientific thinking needed to help him; the chair of the consulting room and the lab table; the human organism and the DMA. It is a constant challenge that simply fulfils my mind every day.
What do you really like about this research? What really excited me in this project was the field work, evaluating in detail these patients and trying to bring some light in this rare and puzzling condition - the Nicolaides-Baraitser syndrome. It was specially rewarding to meet the families; to soften their daily problems with just some information about other affected patients and our effort in better understanding and describing this condition. Finally, it was quite amazing to join forces with so many colleagues in this collaborative study.
What is your ultimate goal in life? To give my best in every situation and to be a happy, passionate, fair and sincere person throughout my life.

Lisette Stolk, Rotterdam, Netherlands
Talk: C10.3 Loci on chromosome 19 and 20 are associated with age at natural menopause: a meta-analysis of 10,399 women
Session: C10 Complex genetics I
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: 25 January 1981, Dordrecht in the Netherlands
What is your current position? PhD-student in the Genetic Laboratory of the Department of Internal Medicine at Erasmus MC, Rotterdam
Why did you choose a career in genetics? During my first year master reseach period someone once told me "everything is genetic", and at the end of that research period I was convinced of that too. And after finishing my graduation thesis on imprinting in mice it was clear to me that I just had to start a career in genetics.
What do you really like about this research? You already know how someone's DNA look like, but how it influences your risk for diseases, the way you look, the way you behave, it is still a mystery for a large part. And I like this unknown part of it.
What is your ultimate goal in life?Being happy in my personal life and being successful in my professional life, without one effecting the other.

Bernard Thienpont, Leuven, Belgium
Talk: C11.1 An aCGH screening study in 150 patients identifies a novel dosage-sensitive gene, TAB2, which is disrupted in multiple patients with cardiac defects
Session: C11 Molecular basis of Mendelian disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: 22 05 1980, Wilrijk, Belgium
What is your current position? PhD Student at the Center for Human Genetics, Leuven
Why did you choose a career in genetics? Because it combines my interest in fundamental research with clinically relevance
What do you really like about this research? Our research starts from a very simple and elegant idea which I could focus on for several years. It moreover combines high-throughput generation and analysis of large amounts of data (an approach which I value greatly) with further more detailed investigations on particular interesting cases.
What is your ultimate goal in life? To be happy in life: satisfied from what I achieved and eager to continue.

Bregje van Bon, Nijmegen, Netherlands
Talk: C02.2 Further delineation of the 15q13.3 microdeletion and duplication syndromes: A clinical spectrum varying from non-pathogenic to a severe outcome
Session: C02 Clinical Genetics I
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 09-07-1981, Wamel, The Netherlands
What is your current position? I am combining my training to become a clinical geneticist with my PhD project on different forms of genomic imbalance in mental retardation.
Why did you choose a career in genetics? During my medical internships I realized I enjoyed communicating with patients. However, most outpatient clinics had 5-10 minute consultations, in which doctors hardly had enough time to really listen en talk to their patients. This in contrast to the genetic outpatient clinic; for patients genetic information is not always easy to understand. Therefore, we take the time needed to help people making their own decisions on difficult matters such as pre-symptomatic testing and prenatal diagnostics. It are these moments with patients and their families which I enjoy the most: Being able to translate scientific findings to understandable language and practical information. Besides, working in clinical genetics gives me the nice opportunity to combine both patient care and research.
What do you really like about this research? Chromosomal aberrations are a well-known cause of mental retardation. Several new microdeletion and duplication syndromes have been delineated, since the introduction of high resolution micro-array techniques. Some of these give rise to a clinically recognizable phenotype. Others, however, give rise to a less clear phenotype such as deletions in chromosome band 15q13.3. By careful phenotyping large patient cohorts with similar aberrations, this research may be of direct benefit in daily practise for clinicians counselling patients and their families.
What is your ultimate goal in life? To stay curious.

Ramprasath Venkatachalam, Nijmegen, Netherlands
Talk: C06.2 Identification of novel genes involved in colorectal cancer predisposition
Session: C06 Cancer genetics
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 1982, Chennai, India
What is your current position? 3rd year Ph.d student at the Dep. of Human Genetics, Univ. Medical Centre Nijmegen, Netherlands.
Why did you choose a career in genetics? Being from engineering background I have always been intrigued by application of sciences in catering to human needs. Likewise genetics has a broad spectrum of applications from diagnoses to treating diseases.
What do you really like about this research? There is a great scope for innovation and improvisation. Most of all the satisfaction that my findings could be applied in diagnosing and treating cancer.
What is your ultimate goal in life? To take life as it comes and to try and give my best in whatever I pursue.

Esmé Waanders, Nijmegen, Netherlands
Talk: C06.6 Ikaros is a frequently affected hematopoietic differentiation factor in pediatric relapse-prone precursor B-cell acute lymphoblastic leukemia
Session: C06 Cancer genetics
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 15th September 1978 Groenlo, the Netherlands
What is your current position? Post doc at the Department of Human Genetics, Radboud University Nijmegen Medical Center, Nijmegen, the Netherlands
Why did you choose a career in genetics? Genetics always fascinated me. I am intrigued by the way diseases can be traced back to our ancestors. Also, the multidiscipline nature of the field and the close relation between clinic and lab appealed to me.
What do you really like about this research? The big puzzle
What is your ultimate goal in life? Don't worry, be happy.

Verena Wally, Salzburg, Austria
Talk: C12.5 Trans-splicing gene therapy in autosomal dominant skin disease
Session: C12 Pathophysiology and therapy for genetic disorders
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Date and city of birth: 21.02.1979
What is your current position? Post-doc at the eb-house Austria, University Hospital Salzburg.
Why did you choose a career in genetics? I have no idea, but it was the right decision!
What do you really like about this research? We are working on Epidermolysis bullosa, a severe disease of the skin and I really hope to contribute to find a therapy for this disease! This is work that makes sense!
What is your ultimate goal in life? I want to have the feeling not to have missed anything and to have done everything right!

Stephen Williams, Richmond, United States
Talk: C02.1 Phenotypic and genomic evaluation of 52 subjects with a Smith-Magenis-like phenotype: identification of new syndromic regions associated with altered gene dosage
Session: C02 Clinical Genetics I
Date: Sunday, May 24, 2009, 15.00 - 16.30 hrs

Date and city of birth: 02/03/79 Leesburg, VA, USA
What is your current position? 4th year PhD student in the Elsea Lab. Department of Human and Molecular Genetics, Virginia Commonwealth University.
Why did you choose a career in genetics? More than any other science, genetics is on the forefront of identifying who we as people truly are and where we are going. I've always wanted to be a part of a culture that strives to help each other using these basic discoveries.
What do you really like about this research? I've been able to tie clinical work with molecular genetics. It helps me to not loose site of the human element that can sometimes be lost in science.
What is your ultimate goal in life? To be satisfied with my place in life whether it be in or outside of science.

Chi-Fan Yang, Taipei, Taiwan
Talk: C10.1 A genome-wide association study identified novel susceptibility loci for type 2 diabetes mellitus in Chinese population residing in Taiwan
Session: C10 Complex genetics I
Date: Monday, May 25, 2009, 15.00 - 16.30 hrs

Alexandra Zhernakova, Utrecht, Netherlands
Talk: C17.5 The common and specific genetic backgrounds of rheumatoid arthritis and celiac disease
Session: C17 Complex genetics II
Date: Tuesday, May 26, 2009, 10.45 - 12.15 hrs

Date and city of birth: 19th April 1971; St. Petersburg, Russia
What is your current position? After graduating from the Russian Medical Academy in 1994, I specialized in medical genetics and worked as a clinical geneticist in the Centre of Medical Genetics in St. Petersburg. After moving to the Netherlands, I started working on genetic research projects as a technician and I am now finishing my PhD in the Complex Genetics Group, University Medical Centre, Utrecht.
Why did you choose a career in genetics? It is challenging and exciting to work in this fastest developing field in science where fascinating discoveries are happening everyday.
What do you really like about this research? The co-morbidity of immune-related diseases was known from clinical studies. With recent genetic findings we can now understand why some diseases overlap. Understanding of shared pathways brings us closer to patient care - predicting a general immune-related profile rather than a disease-specific one opens up new possibilities for prevention and treatment of immune-related diseases

What is your ultimate goal in life? To be useful to people and society; and to enjoy the combination of exciting research, three kids and my various hobbies.

Conference venue
Austria Center Vienna
Bruno Kreisky Platz 1, 1220 Vienna, Austria
www.acv.at

Badges
Participants should collect name badges from the conference registration desks. As only registered participants will be permitted to attend the Scientific Sessions, the Exhibition and poster areas, you are kindly asked to wear your badge when entering the congress venue.
Accompanying persons and exhibitors will also receive badges to allow access to the appropriate areas.
Lost badges can be replaced at the registration desk. However, a handling fee of EURO 50.- will be charged.

Bank services - Money matters
Banks open from 8.30 – 12.30 and 13.30 - 15:00 hrs on Monday - Wednesday and Friday, from 8.30 – 12.30 and 13.30 - 17:30 hrs on Thursdays. There are multiple bank machines (ATMs) open 24 hours a day throughout the city that accept all major international bankcards. The official currency of Austria is the euro (€). Major credit cards are widely accepted, but please always check beforehand.

Cancellations and Refunds
Notice of cancellation had to be made in writing by registered letter or fax to the Congress Office.
The policy for refunding registration fees is as follows:
Written cancellation received:
- Before April 7, 2009: 75% refund
- Between April 7 and May 11, 2009: 25% refund
- After May 11, 2009: no refund

The date of the postmark or fax ID is the basis for considering refunds. Refunds will be made after the congress.

Car Parking
Participants may park their car in the parking lot of the Austria Center Vienna at cost.

Certificate of Attendance
Confirmations of attendance will be issued at the registration desk.

Climate
The weather in May is generally nice, evenings may be cooler and of course occasional rain showers have to be expected.

Cloakroom and Luggage
A cloakroom and luggage storage is available in the ACV.

CME credits
The ESHG has applied for credits at the European Accredication Council for Continuing Medical Education Institution of the UEMS. The result is currently still pending. The number of credits will figure on the respective confirmation of attendance, which will be sent after the meeting.
The rules of the EACCME state that participants are kindly asked to fill in the feedback form included in the conference bag. It can be returned to the registration desk.
Participants applying for credits will have to have their badge scanned daily at the CME desk in the registration area before entering the conference.

Coffee Breaks and Lunch
During the session breaks refreshments (coffee, tea, and water) will be served free of charge to participants wearing name badges.
Lunch tickets for lunch boxes for Sunday, Monday and Tuesday, had to be pre-ordered on the registration form - they cannot be purchased on site. Price per day: EUR 14.-.

Currency
The official currency of Austria is the Euro (€). 1 EUR = 1,32 USD = 0,88 GBP = 131 JPY = 1,50 CHF as per date of printing.

Drinking water
The tap water in Austria is of excellent quality and can be used without concern.

Electricity Supply
220V - 50Hz AC. Connectors can be obtained from your hotel reception or electronic stores.

Emergencies
A Medical Doctor is of duty in the conference center during meeting hours. Please contact the information desk of the ACV or the registration desk.
For emergencies outside of the conference center: Ambulance: 144, Police: 133, Fire Department: 122. European Emergency Call: 112 from a cell phone (even without inserted pin card).

Exhibition opening hours
Sunday, May 24, 2009:	08.30 – 18.00 hrs
Monday, May 25, 2009:	08.30 – 18.00 hrs
Tuesday, May 26, 2009:	08.30 – 13.30 hrs

GSM cell phone roaming
GSM cell/mobile phone roaming is available without any problems for all major international providers. It is advisable to inquire beforehand at your provider which roaming company in Austria offers the cheapest tariffs.

Insurance
In registering for the ESHG 2009 participants agree that neither the organising committee nor the congress office assume any liability whatsoever. Participants are requested to make their own arrangements for health and travel insurance.

Internet
An internet café as well as a WIFI access will be available in the exhibition area. You many also find small Internet cafés throughout the city.

Language
The official language of the congress will be English (no simultaneous translation). German is the official language in Austria.

Lunch and Refreshments
Lunch tickets for lunch boxes for Sunday, Monday and Tuesday, had to be pre-ordered on the registration form - they cannot be purchased on site. Price per day: EUR 14.-. Please note that lunch tickets are not refundable.

Message Centre
A Message Centre is available in the Registration Area.

Pharmacies
Pharmacies are open from 8.00 - 12.00 and 14.00 - 18.00 hrs on weekdays, from 8.00 - 12.00 hrs on Saturdays. Medicines that do not require a medical prescription may be purchased directly over the counter. If you need advice, do not hesitate to ask the pharmacist. If you usually take some medicine in particular, it is advisable to bring a sufficient amount with you for your trip. There are duty pharmacies open 24 hours a day in all city districts. Outside all the pharmacies there is a list of the nearest duty pharmacies.

Poster Removal
The organisers cannot assume any liability for loss or damage of posters displayed in the poster area. Posters that were not removed after the end of the meeting on Tuesday, May 26, 2009, 14.00 hrs, will be removed by the staff and **will not be kept or mailed to the author after the meeting.**

Preview Room/Media Ceck
Equipment for a final check of the sequence of your presentation is available in the media check/preview room on level OE. All presenters should bring their electronic presentation to the preview room preferably in the morning of the day of the talk, but not later than 2 hours before the start of the session.

Registration Desk opening hours
Saturday, May 23, 2009:	10.00 - 20.00 hrs
Sunday, May 24, 2009:	08.00 – 18.45 hrs
Monday, May 25, 2009:	08.00 – 18.45 hrs
Tuesday, May 26, 2009:	08.00 – 15.30 hrs

Safety - Crime
Visitor safety is generally adequate and Vienna can be considered a rather safe city. Like in any other major European city, you can walk everywhere using common sense. Pick pocketing in heavily visited tourist zones or in public transport lines might be a concern.
Unfortunately experience has shown that some basic precautionary measures should always be kept in mind in any city:
- Do not carry important items like flight tickets, passports etc. with you when visiting the conference or strolling through the city, leave them in the hotel safe during your stay. Rather carry a Xerox copy of your passport or an identity card with you.
- Try not to carry all documents, money, credit cards and other essential items and valuables in one bag. If it is lost or stolen, everything will be gone and might be difficult to replace on short notice, especially passports and visa to return to your country of residence.
- Take off your name badge when leaving the conference center.

Shops
The shops in Vienna open between 9.00 and 19.00 hrs and some close at lunchtime (12.00 - 14.00 hrs). The department stores and hypermarkets do not close for lunch. All retailers, including both small shops and department stores, open on Saturdays (generally 10.00 - 12.00 hrs, some, especially in the city center, until 18.00 hrs) and closed on Sundays, except for a few establishments with special permits.

Smoking Policy
The ESHG 2009 is a “No-smoking-Conference”. Please note the smoking is banned from all public buildings. Restaurants and bars may, depending on size, be declared either as “smoking” or “no-smoking” locations or have separate areas and ventilation systems.

Staff
If you should have any questions, the congress staff recognizable by a yellow badge and black polo shirts will be pleased to help you.

Taxis
A taxi is the most comfortable way of getting around the city but also the most expensive. Vienna's taxis do not have a special livery/colour. They are identifiable by their roof-sign which is lit when available. Prices must be displayed inside the cab and meters (which are compulsory in all cabs) will indicate the fare.

Tipping
Tips of 5-10% of the total bill are usually expected in restaurants and bars, although not mandatory. If part of a big group, you may of course ask the waiter to pay separately. Hotel and airport porters will accept tips, depending on the luggage.

Travelling - Accessibility
The fastest and cheapest way of moving around Vienna is using public transport. Tickets are available from machines at underground stations (maestro debit cards accepted),
at news agents', or at Vienna Transport Authority's ticket offices. Tickets bought in advance are cheaper and must be punched in a blue ticket cancelling machine on the tram or bus, or at the barrier before boarding the underground train.
Single trip tickets can be used for any single trip within Vienna. You may change lines (and switch between bus, tram, underground, or urban train), but you may not interrupt your journey. Ticket price: EUR 1.70 when bought in advance; EUR 2.20 when bought in the means of transport.
The 24-hour pass is valid throughout Vienna for exactly 24 hours from the time it is punched. Price: EUR 5.70
The 72-hour pass is valid throughout Vienna for exactly 72 hours from the time it is punched. Price: EUR 13.60
The 8-day ticket is valid for any eight days, not necessarily eight consecutive days. It is a rover ticket, which means you can travel all around Vienna. You can also use the ticket for several people travelling together. Simply punch one strip for each person in the group. Price: EUR 27.20
Week-pass valid from Monday to Monday, 09.00 hrs. Price: EUR 14.00
A taxi is the most comfortable way of getting around the city but also the most expensive. They are identifiable by their roof-sign which is lit when available. Prices must be displayed inside the cab and meters (which are compulsory in all cabs) will indicate the fare.

Travelling from and to the Vienna Airport (*Schwechat*)
Vienna International Airport is located 13 kilometres south east of Vienna.
City Airport Train (CAT)
The train service between the airport and the city centre (City Air Terminal – Station 'Wien Mitte') is every 30 minutes and takes 16 minutes. It is covered from 6:05 hrs to 00:05 hrs (Airport to City) and from 5:38 hrs to 23:38 hrs (City to Airport). Tickets may be purchased online, from ticket machines at the airport/station or on board.
Rapid Train
The S7 rapid transit line (blue train) goes to "Praterstern", where you can change to the metro line "U1" (red line) also going to the ACV.
Bus
The Vienna Airport Lines bus covers different routes downtown as well as to the conference venue*Airport transfer*
Airport transfers at a fixed rate of EUR 30.- can be pre-ordered by sending an email to: wolfgang.jechne@oracle.com
Taxis
Fares to the conference venue are about EUR 35.00 and take around 25-40 minutes to the ACV or the city center, depending on the time of the day. There is a taxi rank outside the airport terminal. There are surcharges for evening and night rides.

Travelling to the ACV by Public Transportation
To reach the ACV from the city centre (Stephansplatz) take the U1 underground line (red). Alight at Vienna International Centre/Kaisermühlen and take the exit marked Schüttaustraße. Travelling time: approx. eight minutes.

V.A.T.
A value-added tax (V.A.T.) of generally 20% is applied to most products and services (a 10% V.A.T. applies on e.g. books, drinks and a few other items). The VAT is included in the displayed price.

Vienna
Vienna, the capital of Austria, almost 2 million inhabitants, is situated on the banks of the Danube. Vienna is a dream city for anyone with an interest in history. Narrow, medieval alleyways and grand boulevards lead to historic sights such as the *Imperial Palace (Hofburg)*, *Belvedere Palace*, *Burgtheater*, the *Spanish Riding School*, *St. Stephen's Cathedral*, the *Opera House*, *Karlskirche*, or *Schönbrunn Palace*. Vienna has been synonymous with music for centuries, and was home to *Mozart*, *Beethoven*, *Schubert* and Johann Strauss. The *Vienna Philharmonic Orchestra* is one of the world's top orchestras; the *Vienna Boys' Choir* is triumphantly successful wherever it tours. Vienna possesses a lively and vast array of cultural attractions, from classical or experimental theatre, film or dance festivals, opera or operetta, to exhibitions and concerts. The *Museum of Fine Arts* is one of the world's largest and most distinguished museums, housing priceless works of art. Art accompanies you wherever you go in Vienna - even some of its underground stations are listed properties on account of their elegant, ornamental Jugendstil (Art Nouveau) style designed by *Otto Wagner*. For more information about Vienna, please see www.aboutvienna.org.

Registration Fees

Payment Received:	before March 31, 2009	after March 31, 2009
ESHG Members*	EUR 270.-	EUR 370.-
Non-Members	EUR 380.-	EUR 480.-
Students**	EUR 135.-	EUR 165.-
Day Ticket	-	EUR 135.-
Accompanying Persons	EUR 55.-	EUR 55.-
Conference Party (Members, Non-Members or Acc.Persons)	EUR 39.-	EUR 39.-
Conference Party (Students)	EUR 22.-	EUR 22.-

* Participants having applied for new ESHG membership and paid their contribution before the meeting may pay the Member's fee. Membership information is available at www.eshg.org. *Renewals* of existing memberships can be made on site. No refunds on registration fees can be made when applied or renewed on site.
** Please provide a copy of a Student's ID or a confirmation signed by the head of department at the moment of your registration by fax to +43 1 407 82 74 or together with the hardcopy of the registration form. Confirmations handed in at a later stage cannot be considered. Please note that the Student fee is not applicable for Post-docs.

What is covered by the registration fee?

Participants:

- Admission to all scientific sessions, exhibition and welcome reception
- Abstract Book & CDrom and Programme
- Coffee/Tea during breaks from Saturday, May 23 to Tuesday, May 26

Accompanying Persons:

- Admission to the Opening Ceremony
- Admission to the Welcome Reception
- Admission to the Exhibition Area
- Vienna City Tour on Sunday May 24, 2009, 10.00 hrs

Payment of Registration fees, may be made in Euro in cash or by credit card (Diners Club, Mastercard, VISA, American Express or Maestro Bank cards.

Please note

The reduced registration fee is only applicable, if it has been credited to the congress account before the deadline. Registering before March 31, 2009 without performing an actual payment is not sufficient to benefit from the reduction.

Cancellations and Refunds

Notice of cancellation had to be made in writing by registered letter or fax to the Congress Office.
The policy for refunding registration fees is as follows:
Written cancellation received:
- Before April 7, 2009: 75% refund
- Between April 7 and May 11, 2009: 25% refund
- After May 11, 2009: no refundThe date of the postmark or fax ID was the basis for considering refunds. Refunds will be made after the congress.

Social Events

Saturday, May 23, 2009, 20.00 hrs

Welcome Reception at the ACV (conference venue), Foyer D, level U2

Meet your colleagues at this event. Drinks and nibbles will be served in the ACV Congress Centre following the Opening of the Conference.

Monday, May 25, 2009, 20.00 hrs

Conference party: 20.00 hrs, Jugendstiltheater im Otto-Wagner-Spital

Join us for a congress party with "fingerfood", live music by "The Bad Powells" and dancing at the *Art-Nouveau Theatre* located within the Social Medical Center "Otto Wagner Hospital" in the Western outskirts of Vienna.

Bus departure from the conference venue at 19.00 hrs

Contribution towards expenses:

Participants, accompanying persons: EUR 39.-
Students: EUR 22.-

Note: Space is limited, please register early. Tickets will be checked at the entrance.

Exhibition Organiser

Name	**ROSE** INTERNATIONAL Exhibition Management & Congress Consultancy bv
Address	P.O. Box 93260 NL-2509 AG The Hague The Netherlands
Telephone	+31 70 383 89 01
Fax	+31 70 381 89 36
E-mail	eshg@rose-international.com
Telephone at Exhibition Service Desk in the ACV during the conference	Telephone + 43 (0)1 260 69 2701

Exhibition Dates & Opening Hours

Sunday, 24 May 2009	08.30 – 18.00 hrs
Monday, 25 May 2009	08.30 – 18.00 hrs
Tuesday, 26 May 2009	08.30 – 13.30 hrs

Location

Venue	Austria Centre Vienna
Exhibition Area	Hall X and XL
Address	Bruno-Kreisky-Platz 1, AT-1090 Vienna, Austria
Telephone	+43 (0)1 260 69-0 (general number)
Website	www.acv.at

Exhibition Floor Plan

You will find the floor plan of the exhibition in your conference bag, it is a separate document.

Lead Retrieval System used by Exhibitors

Again this year, a growing number of exhibitors will be using a so-called Lead Retrieval System on their stands.

Note the following please:

- exhibitors who rented this device will ask permission to scan the barcode on your badge
- this barcode gives the exhibitor access to your contact details as follows:
    - name and full postal address
    - e-mail address

Thank you for your understanding and cooperation.

List correct as per date of printing - Exhibition floor plan in your conference bag (separate document)

Company name	Stand
5 PRIME	A-124
Abbott Molecular	A-136
Active Motif	A-120
Affymetrix	A-216
Agilent Technologies	A-310
AGOWA genomics	A-320
Applied Biosystems	A-300
Applied Maths	A-222
APPLIED SPECTRAL IMAGING	B-512
Asper Biotech	A-133
Atlas Biolabs	A-128
BaseClear	A-226
Bertin Technologies	A-118
Biocomputing Platforms	B-412
BIOKÉ	A-212
BIOLOGICAL INDUSTRIES	A-203
BioProducts, co-exhibitor of Biological Industries	A-203
Bio-Rad Laboratories	B-516
BioTrove	A-318
Biozym, co-exhibitor of Finnzymes	A-207
Cartagenia	B-422
CGC GENETICS	A-214
chemagen	B-602
CLC bio	A-332
Cogenics	B-514
CyberGene	A-316
Cytocell	B-638
Cytogen	B-611
deCODE genetics	B-430
DNA Genotek	B-615
ECARUCA - the Online Dosage Imbalance Network	B-536
Elchrom Scientific	B-410
Elsevier	B-634
Eppendorf	A-135
ESHG - European Society of Human Genetics	B-524
Eurofins MWG Operon	B-420
The Eurogene Project	B-534
EuroGentest	B-524
Exiqon	A-202
Expression Analysis	A-204
febit biomed	A-112
Finnzymes	A-207
Fluidigm	A-312
GATC Biotech	A-224
Gen-Probe (formerly TEPNEL)	A-137
Genial Genetic Solutions	B-616
Genizon BioSciences	A-206
GenoLogics	A-336
Genomed	B-604
GenVault	B-504

Company name	Stand
HAMILTON Robotics	A-127
HVD Life Science	A-116
Idaho Technology, co-exhibitor of BIOKÉ	A-212
Illumina	A-330
imaGenes	B-632
IMGM Laboratories	A-119
Innogenetics	B-700
Integrated DNA Technologies	A-208
Interactive Biosoftware	B-618
Invitrogen	A-110
JSI medical systems	B-500
KBiosciences	B-520
Kreatech	B-600
LGC Standards, co-exhibitor of AGOWA genomics	A-320
Macrogen	A-130
metabion	A-322
MetaSystems	B-610
The Microarray Facility Tübingen	A-314
MRC-Holland-MLPA	B-630
Multiplicon	A-114
NanoDrop Products, a part of Thermo Scientific	B-502
Nature Publishing Group	A-339
New England Biolabs	B-636
Nikon Instruments	A-125
ORPHANET	B-524
Oxford Gene Technology	B-613
PAA Laboratories	B-402
PASS Software	A-121
PerkinElmer	B-714
PhenoSystems	A-220
POSSUM, Murdoch Childrens Research Institute	A-228
PRESTAGEN	A-123
Progeny Software	B-400
Promega	B-612
Protalix Biotherapeutics	A-122
QIAGEN	B-706
RainDance Technologies	B-506
Roche Applied Science	B-532
Rosetta Biosoftware	B-712
Science / AAAS	A-200
Tecan	A-132
TEPNEL : see Gen-Probe	A-137
Thermo Scientific	B-510
TIB Molbiol Syntheselabor	B-530
TRANSGENOMIC	A-134
ViennaLab Diagnostics	B-710
Wiley - Blackwell	A-126
Wisepress Online Bookshop	B-526
Carl Zeiss	B-614

5 PRIME GmbH **Stand number: A-124**
Email: info@5prime.com
Website: www.5prime.com

5 PRIME GmbH has started in April 2007and had been able to establish a portfolio of over 210 products for Life Science researchers during the last year. All 5 PRIME products are produced under high quality standards and are designed for customers requiring high performance products at a competitive price. The competitive strength of 5 PRIME is based on the unique features of the products and has already lead to exciting feedback from our customers. As the one source for your everyday laboratory research, the 5 PRIME portfolio includes technologies and reagents for all common molecular biology applications.

Abbott Molecular **Stand number: A-136**
Email: See Abbott Molecular web-site for local contact details
Website: http://international.abbottmolecular.com/home_2.aspx

Among Abbott Molecular's leading portfolio of molecular diagnostic products for infectious diseases, oncology, transplantation and human genetics, are FISH probes for pre- and postnatal diagnosis of chromosomal abnormalities and leading products for detection of the commonest inherited genetic diseases, cystic fibrosis and fragile X syndrome.

Active Motif **Stand number: A-120**
Email: tlsales@activemotif.com
Website: www.timelogic.com

Active Motif makes specialised computer hardware for processing Next Generation sequencing and other bioinformatic data. Our SeqCruncher™ FPGA accelerator card can run supported algorithms - BLAST, HMMs, Smith-Waterman and GeneDetective – up to several hundred times faster than a CPU, freeing up existing computing resources for other uses and saving electricity. We also develop innovative cell biology-based research tools to help researchers elucidate the function, regulation and interactions of genes and their encoded proteins. This includes products for epigenetics, chromatin biology and transcriptional regulation.

Affymetrix **Stand number: A-216**
Email: Sales_Europe@affymetrix.com
Website: www.affymetrix.com

Affymetrix is at the forefront of a scientific revolution. The genetic information gathered by scientists today is changing the healthcare of tomorrow by informing both diagnostic and treatment processes. From whole genome scans to targeted genotyping, resequencing experiments to cytogenetic and copy number analysis, Affymetrix GeneChip® high-density arrays are speeding discovery and leading to better treatments in research labs and clinical practice. We cordially invite you to learn more at the Affymetrix workshop on Sunday 24th May from 13:15 – 14:45.

Agilent Technologies **Stand number: A-310**
Email: info_agilent@agilent.com
Website: www.agilent.com/chem

Agilent Technologies is a leading supplier of life science research systems that enable scientists to study complex biological processes and disease mechanisms. Engineered for sensitivity, reproducibility and workflow productivity, Agilent's solutions include instrumentation, software, consumables and services for genomics, proteomics and metabolomics applications. Agilent's key products are 60-mer Oligo catalog and custom microarrays for array-based Comparative Genomic Hybridization (aCGH), GeneExpression and ChIP-on-Chip, GeneSpring software platform, microarray scanner, Real-time PCR and 2100 Bioanalyzer lab-on-a-chip system. Agilent's proteomics and metabolomics product portfolio includes LC/MS and HPLC-Chip/MS systems, protein pre-fractionation and separation solutions.

AGOWA genomics **Stand number: A-320**
Email: service@agowa.de
Website: www.lgc.co.uk/genomics

AGOWA GmbH is an international-acting genomics specialist. The company was established in 1993 and has been part of LGC since December 2005 - under the name AGOWA genomics.
AGOWA genomics has long-standing experience in large-scale sequencing, gained in international and national genome projects. The portfolio of products and services comprises:
- Nucleic acid preparation – products and services
- DNA sequencing services
- Genomic services (incl. Next generation services)

LGC is an international science-based company and market leader in analytical, forensic and diagnostic services and reference standards (www.lgc.co.uk).

Applied Biosystems **Stand number: A-300**
Email: abdirect@eur.appliedbiosystems.com
Website: www.appliedbiosystems.com

Applied Biosystems is now a part of Life Technologies Corporation, a global biotechnology tools company dedicated to improving the human condition. Applied Biosystems systems, consumables and services enable researchers to accelerate scientific exploration, driving to discoveries and developments that make life even better. New full range of Capillary Electrophoresis Instruments, SOLID v3.0 Next Generation Genetic Analysis System and the Taqman Open Array Genotyping System will be the highlights of the AB exhibition at the conference. Life Technologies was created by the combination of Invitrogen Corporation and Applied Biosystems Inc.

Applied Maths NV **Stand number: A-222**
Email: info@applied-maths.com
Website: www.applied-maths.com

Applied Maths develops cutting-edge software for the biosciences. The software BioNumerics® is a software suite for integrated analysis and databasing of biological data in the broadest sense. When linked to Kodon®, a high level sequence analysis package or GeneMathsXT®, a highly sophisticated mathematical toolbox for the analysis of data from genechips and genearrays, it becomes a universal, powerful platform for bio-databasing and analysis. The software is licensed to more than 1.000 research sites worldwide and served for the preparation of >1.400 peer-reviewed papers. For more information, visit our website www.applied-maths.com.

APPLIED SPECTRAL IMAGING **Stand number: B-512**
Email: sales@spectral-imaging.com
Website: www.spectral-imaging.com

Applied Spectral Imaging (ASI) is a leading developer and manufacturer of comprehensive solutions for Cytogenetics and Pathology imaging and data management needs. With its superior quality image capture (acquisition) abilit ies, ASI provides state of the art diagnostic aids. All solutions are integrated into the Laboratory Information System (LIS), which incorporates Picture Archive Communication System (PACS). The company products are geared towards cancer, pre-, and post-natal diagnosis in clinical and research use. Products range from the basic manual operation mounted on existing laboratory microscopes all the way to fully automatic operation.

Asper Biotech **Stand number: A-133**
Email: info@asperbio.com
Website: www.asperbio.com

Asper Biotech is a genetic testing company with a 10-year experience in developing robust and cost-effective tests in the range of 50-600 SNPs/mutations. Asper offers testing service, custom test development service, interpretation of testing results as well as genotyping platform Genorama® QuattroImagerTM with the accompanying software, consumables and support services. Current range of tests includes BRCA, Ashkenazi Jewish diseases, Hereditary Hearing Loss and DNA repair in addition to world's largest portfolio of genetic eye disease tests (Usher syndrome, Bardet-Biedl syndrome, Leber's Congenital Amaurosis, Retinitis Pigmentosa, etc).

Atlas Biolabs GmbH **Stand number: A-128**
Email: customer-support@atlas-biolabs.de
Website: www.atlas-biolabs.de

ATLAS Biolabs GmbH is a leading provider of microarray based genomic services such as genome wide gene expression and SNP analysis, CGH analysis and diagnostic services. Customers include registered doctors, hospitals, and pharmaceutical, biotechnological as well as diagnostic enterprises in the European market.

BaseClear B.V. **Stand number: A-226**
Email: info@baseclear.com
Website: www.baseclear.com

BaseClear Group is an independent service providing laboratory based in Leiden, The Netherlands. BaseClear was set up in the strong belief and vision that providing high quality services is essential for life science research. As a service provider, BaseClear distinguishes itself by the dynamic and flexible manner in which it applies its extensive experience and expertise in all aspects of DNA sequencing and molecular biology techniques. For more than 15 years many international academic and company clients, already build and rely on the quick and high quality customised solutions, from the BaseClear Group.

Bertin Technologies **Stand number: A-118**
Email: precellys@bertin.fr
Website: www.precellys.com

Precellys is a complete range of products for DNA, RNA, proteins extractions. The Precellys24 is a tissue homogenizer using bead-beating for high yield and integrity results. With the Cryolys cooling option and the ready to use beads kits, Precellys24 improves your lab practices and productivity.

Biocomputing Platforms Ltd **Stand number: B-412**
Email: info@bcplatforms.com
Website: www.bcplatforms.com

Biocomputing Platforms Ltd offers powerful data management solutions for modern genetic studies. Our main products - BC/GENE and BC/SNPmax - are data management platforms which combine genotype data with phenotypes, maps and pedigrees, and create highly productive data analysis pipelines. They eliminate unnecessary data management work, increase productivity in data analysis and improve research team collaboration. Both BC/GENE and BC/SNPmax scale up to handle the output of modern high-throughput SNP genotyping technologies, BC/SNPmax being especially designed for large genome wide association studies. We also offer BC/SAMPLE for sample management.

BIOKÉ **Stand number: A-212**
Email: info@bioke.com
Website: www.bioke.com

BIOKÉ is a key provider of products and services in life sciences in the field of genomics, proteomics and molecular diagnostics. It is our aim to accelerate the progress in R&D and diagnostics of our customers in academic, hospital and other research institutions as well as pharmaceutical and biotechnology companies. Our booth focusses on the LightScanner system: the fastest, highest-quality Hi-Res Melting instrument with unparalleled performance in mutation scanning, genotyping using labeled or unlabeled probes, small amplicon genotyping, methylation analysis & real-time PCR.

BIOLOGICAL INDUSTRIES **Stand number: A-203**
Email: daniela@bioind.com
Website: www.bioind.com

Biological Industries Ltd., founded in 1981, is a manufacturer of animal cell culture products and molecular biology products. A team of 60 employees is dedicated to providing products of the highest quality via distributors in 30 countries world wide. The products undergo extensive quality control, and are certified under ISO 13485:2003 and ISO 9001:2000. Tissue Culture products also are registered with the CE Mark. Our product lines include: Liquid and powdered synthetic media, Sterile serum, Novel Serum-free media formulations, Products for Cytogenetics , Products for Mycoplasma detection and treatment, ECM-coated disposable plastic ware, Products for Molecular Biology and Custom Formulations.

BioProducts - Co-exhibitor of Biological Industries **Stand number: A-203**
Email: thomas.langmann@bioproducts.at.
Website: www.bioproducts.at

BioProducts, an Austrian based company, is focused on molecular diagnostics, life science and cytogenetics. We offer cytogenetic media from Biological Industries and FISH Products from Cytocell for the Austrian markets.

Bio-Rad Laboratories **Stand number: B-516**
Email: uk.lsg.marketing@bio-rad.com
Website: www.discover.bio-rad.com

From RNA purification to profiling and quantification, Bio-Rad provides Life Science researchers with superior tools, service, and support that produce consistently reliable and sensitive results for performing Gene Expression, Gene Silencing and DNA Analysis. Whether you are analysing thousands of genes or just one, Bio-Rad Laboratories has solutions to meet your needs. Come and visit us to discover our new generation of thermal cycling platforms and real-time PCR systems; the 1000-series and CFX96/384. Learn more about our SsoFastTM EvaGreen® Supermix qPCR Reagents for superior results in qRT-PCR and High Resolution Melting Analysis (HRM). www.bio-rad.com/pcr/

BioTrove, Inc. **Stand number: A-318**
Email: info@biotrove.com
Website: www.biotrove.com

BioTrove's OpenArray® provides a flexible and rapid solution for real-time qPCR. Our nanofluidics technology significantly accelerates project completion time and reduces project costs. Design custom SYBR® or TaqMan® arrays or choose from a library of bench-validated panels in several research areas, such as cancer, inflammation, signal transduction and cardiovascular disease.

Biozym Scientific GmbH - Co-exhibitor of Finnzymes **Stand number: A-207**
Email: support@biozym.com
Website: www.biozym.com

Biozym Scientific is a leading provider for the European Life Science Market. Our product portfolio consists of high performance instrumentation, superior biochemical and specialized plastic ware, used in applications like PCR, DNA-sequencing, identification and purification.

Cartagenia **Stand number: B-422**
Email: bert.coessens@cartagenia.com
Website: www.cartagenia.com

Cartagenia delivers a unique combination of highly specialized data management, integration, and mining applications with a strong focus on servicing cytogenetics laboratories introducing arrayCGH into clinical practice. Store+Bench is Cartagenia's flagship database solution for arrayCGH. This intelligent software platform supports diagnosis and research in constitutional cytogenetics, easing characterization and interpretation of the clinical impact of copy number variation in patient genotypes. Store+Bench is used throughout Europe by several genetics laboratories, and speeds up the diagnostic workflow significantly. It is compatible with a large number of commercial and in-house platforms, raw data analysis tools, external databases, and public information sources.

CGC GENETICS **Stand number: A-214**
Email: dcc@cgcgenetics.com
Website: www.cgcgenetics.com

CGC Genetics is a Medical Genetics Diagnostics company, with a Clinical Department, and 5 laboratories working strongly together: Biotechnology, Molecular Diagnostics, Cytogenetics, Anatomy Pathology and Prenatal Screening. With a rigorous quality policy and installed capability for the development of new tests, CGC Genetics has a group of highly skilled professionals, offering a large panel of genetic tests (>1500) including microarrays (Patent Pending), for prenatal diagnosis, prenatal screening, oncohematology, preventive medicine, pharmacogenetics and rare diseases. CGC Genetics has facilities in Portugal, Spain and USA.

chemagen Biopolymer-Technologie AG **Stand number: B-602**
Email: info@chemagen.com
Website: www.chemagen.com

chemagen Biopolymer-Technologie AG is a key player in the field of automated nucleic acid isolation. Our kits and optimized protocols in combination with our chemagic automation platforms facilitate the preparation of many different sample materials such as blood, serum/plasma, saliva, tissue or buccal swabs. Sample volumes up to 10 ml can be processed easily. Outstanding robustness, minimal hands on time and fast automated applications translate to the highest throughput and flexibility of your laboratory workflow. Find out more at www.chemagen.com

CLC bio **Stand number: A-332**
Email: info@clcbio.com
Website: www.clcbio.com

CLC bio is the world's leading bioinformatics solution provider. Next Generation Sequencing is a major focus area and CLC bio delivers the first and only comprehensive cross-platform analysis solution, which can analyze and visualize data from all major platforms, like Illumina's Genome Analyzer, SOLiD by Applied Biosystems, 454 by Roche, and HeliScope by Helicos. CLC bio offers everything from user-friendly desktop applications to full enterprise solutions, completely integrated with existing workflows.

Cogenics **Stand number: B-514**
Email: heike.naserke@epidauros.com
Website: www.cogenics.com

Cogenics provides a full range of genomics services for both research and regulated projects, including services to support pharmacogenomics discovery and validation and biomarker research and development. Integrated services include DNA and RNA extraction, genotyping, full sequencing services including next generation genomic sequencing, and gene expression. Services are further offered to support multiple applications of QPCR and services for cell bank characterization and biorepository, for both research and regulated environments.

CyberGene AB **Stand number: A-316**
Email: chromoquant@cybergene.se
Website: www.cybergene.se

ChromoQuant® QF-PCR kit will rapidly diagnose the most common chromosomal disorders in foetuses, e.g. Down, Edward, Patau and Klinefelter syndromes. ChromoQuant® is used by clinical laboratories worldwide. The kit technology is based upon traditional molecular biology, i.e. Quantitative Fluorescent PCR. The diagnosis is performed in 24-48 hours, using standard DNA sequencers and PCR instruments. ChromoQuant® is a CE marked IVD product for clinical use. Visualizer™ software, data base and decision support system for safe and objective interpretation of data, is supplied with ChromoQuant®. Visualizer™ software comes together with the ChromoQuant® QF-PCR kit. CyberGene holds an ISO 13485/9001 certificate.

Cytocell Ltd. **Stand number: B-638**
Email: probes@cytocell.com
Website: www.cytocell.com

Founded in 1991, Europe's largest FISH probe manufacturer is based in Cambridge, UK. Cytocell offers two FISH delivery systems for routine Cytogenetics and Oncology. Aquarius liquid probes offer new extensive range of Microdeletions and new increased range of Oncology/Haematology probes. Paints, Subtelomeres, Enumeration probes also available. Chromoprobe Multiprobe macroarrays offer a convenient screening tool for genetic abnormalities across multiple chromosomes. Key applications include diagnostic/prognostic tools in the analysis of leukaemia (CML, ALL, AML/MDS). Also detection of cryptic chromosomal rearrangements, chromosome translocations and SMC chromosomes (Subtelomeres, Paints, Enumeration).

Cytogen - Produkte für Medizin + Forschung GmbH **Stand number: B-611**
Email: cytogen@eurobiz.de
Website: www.cytogen.info

The Cell Culture Media Company in Human Genetic Diagnostics with it' s Highlights Amniogrow, Lymphogrow, Marrowgrow… the „Gold Standards" in Pre- & Postnatal and Tumor Cytogenetics.

deCODE genetics **Stand number: B-430**
Email: services@decode.com
Website: www.deCODEservices.com

deCODE genetics provides world class scientific services to the global marketplace. Leveraging our state-of the-art CLIA certified facility and expert technical staff, deCODE has the capacity to execute any scale of genetics study on demand. To assure the best possible quality control, all biological sample transactions are monitored by our system-wide LIMS at every operational step from sample intake through all workflows and data delivery. We offer a range of services for SNP & microsatellite genotyping, DNA sequencing, DNA extraction and amplification, as well as products for automated biobanking.

DNA Genotek **Stand number: B-615**
Email: sales@dnagenotek.com
Website: www.dnagenotek.com

DNA Genotek is focused on improving nucleic acid sample collection. The company's Oragene® product line offers researchers a non-invasive, all-in-one system for the collection, stabilization, transportation and purification of high quality DNA or RNA from saliva. Oragenes' reliability and ease-of-use have resulted in rapid worldwide adoption by top-tier health institutions.

ECARUCA : the Online Dosage Imbalance Network **Stand number: B-536**
Email: info@ECARUCA.net
Website: www.ECARUCA.net

ECARUCA (the Online Dosage Imbalance Network) is an interactive online database collecting and providing clinical, cytogenetic and molecular information on rare chromosomal disorders, including microdeletions and microduplications. The ECARUCA database contains over 5000 cases and is easily and freely accessible for all participants. ECARUCA facilitates information exchange as well as exchange of technical knowledge between genetic centres throughout Europe, thereby improving patient care and collaboration in the field of clinical cytogenetics and molecular genetics.

Elchrom Scientific AG **Stand number: B-410**
Email: mleu@elchrom.com
Website: www.elchrom.com

Elchrom Scientific is specialised in providing excellent electrophoresis systems combined with ready-to-use gels of a novel material for the separation and analysis of DNA & RNA. Our mission is to provide unsurpassed levels of standardisation for various applications. In addition, Elchrom's products stand for outstanding performance and resolution in DNA & RNA analysis and purification.

Elsevier **Stand number: B-634**
Email: directenquiries@elsevier.com
Website: www.elsevierdirect.com

Elsevier is a world-leading publisher of scientific, technical, and medical information products and services. Working in partnership with the global science and health communities, Elsevier's 7,000 employees in over 70 offices worldwide publish more than 2,000 journals and 1,900 new books per year, in addition to offering a suite of innovative electronic products, such as Science Direct, MD Consult, Scopus, and online reference works. We have an outstanding range of books and journals in genetics, pharmacology, cardiology, and neuroscience which are available to purchase or subscribe to on our stand.

Eppendorf **Stand number: A-135**
Email: office@eppendorf.at
Website: www.eppendorf.com

Eppendorf is a biotech company which develops, produces and distributes systems for use in life-science research laboratories worldwide. Its product range includes pipettes, dispensers and centrifuges as well as consumables such as micro test tubes and pipette tips. In addition, Eppendorf provides instruments and systems for cell manipulation, automated devices for liquid handling, complete equipment for DNA amplification and biochips. Eppendorf products are aimed at academic and commercial research institutes as well as industrial companies in the field of biotechnology or in other sectors using biotech research processes.

ESHG - European Society of Human Genetics **Stand number: B-524**
Email: office@eshg.org
Website: www.eshg.org

The European Society of Human Genetics is a thriving international society offering high quality science through its European Human Genetics Conference and European Journal of Human Genetics. The several committees such as the Public and Professional Policy Committee and the Education Committee are making active contributions to the Society and to Genetics as a whole. When policy statements are finalised they are posted on our website. We are keeping in touch with our members through a regular newsletter.

Eurofins MWG Operon **Stand number: B-420**
Phone: +49 8092 8289-77
Website: www.eurofinsdna.com

Eurofins MWG Operon is an international provider of genomic services established around the core business lines DNA sequencing, oligonucleotide, siRNA and gene synthesis. The company's main mission is focussed on customer convenience and high quality services in industrial scale for the life science industries and academic research institutions around the world.

The Eurogene Project **Stand number: B-534**
Email: michele.zadra@eurogene.eu
Website: www.eurogene.eu

Eurogene is a European funded project which final aim is to establish a European reference portal for GENETIC MEDICINE that will bring together many multimedia resources into a single organised site. Through Eurogene authors and educators will be able to find, organise and develop multimedia educational materials. To achieve this goal EUROGENE will create a unique web portal for making digital educational contents in the field of GENETIC MEDICINE available and retrievable for end users. Eurogene is willing to find new educational material providers. For further info please contact michele.zadra@eurogene.org or visit www.eurogene.eu.

EuroGentest **Stand number: B-524**
Email: iris.rens@med.kuleuven.be
Website: www.eurogentest.org

EuroGentest is an EU-funded Network of Excellence (NoE) with 5 Units looking at all aspects of genetic testing - Quality Management, Information Databases, Public Health, New Technologies and Education. Through a series of initiatives EuroGentest encourages the harmonization of standards and practice in all these areas throughout the EU and beyond.

Exiqon A/S **Stand number: A-202**
Phone: +45 45 66 08 88
Website: www.exiqon.com/ls

Exiqon Life Sciences, inventors of the miRCURY LNA™ tools for microRNA Research. Exiqon's proprietary Locked Nucleic Acid (LNA™) technology provides unparalleled specificity in microRNA quantitation, expression profiling, detection and knockdown. The miRCURY LNA™ product line includes:
miRCURY RNA Isolation Kit
miRCURY LNA™ microRNA Power Labeling Kits
miRCURY LNA™ microRNA Microarray
miRCURY LNA™ microRNA PCR System
miRCURY LNA™ microRNA Detection Probes for Northern Blotting and in situ hybridization
miRCURY LNA™ microRNA Knockdown Libraries and Probes
We also provide microRNA Array and PCR Services to researchers, world-wide. Visit us at Booth A202 and view our technology.

Expression Analysis **Stand number: A-204**
Email: kmichailo@expressionanalysis.com
Website: www.ExpressionAnalysis.com

Providing whole genome to focused set gene expression and genotyping assays, along with sequencing services using Illumina, Affymetrix and Applied Biosystems. A leading provider of genomic services in clinical trials and research, offering solutions for challenging specimens such as whole blood and FFPE tissues, as well as nucleic acid isolation and bioinformatic services. Our CLIA-registered lab supports GLP compliance.

febit biomed gmbh **Stand number: A-112**
Email: info@febit.de
Website: www.febit.com

febit develops, produces and markets flexible automated solutions for enabling biochip applications in Life Sciences. The product portfolio spans various instruments, assay protocols and bioanalytical services and is complemented by bioinformatics software and consulting.

Finnzymes **Stand number: A-207**
Email: fz@finnzymes.fi
Website: www.finnzymes.com

Finnzymes, founded in 1986, is a leader in the development of PCR, quantitative real-time PCR, transposon and RNA technologies. Our aim is to develop products which make the difference, products that deliver results not achievable otherwise. Finnzymes' core strength is its long experience and extensive know-how in PCR and qPCR technology. Our PCR product line includes all components required for PCR: DNA polymerases, PCR instruments and reaction vessels. Our PCR and qPCR products are thoroughly validated with all the major systems on the market.

Fluidigm Europe B.V. **Stand number: A-312**
Email: biomarkeurope@fluidigm.com
Website: www.fluidigm.com

Fluidigm Corporation develops and markets life-science systems based on integrated fluidic circuit (IFC) technology. IFCs called dynamic arrays facilitate studies requiring high throughput gene expression and genotyping analyses. In addition, digital array IFCs perform digital PCR for copy number variation and for sample quantification with next generation sequencing. Fluidigm's IFC systems, consisting of instrumentation, software and single-use IFCs, increase throughput, decrease costs and enhance sensitivity compared to conventional laboratory systems. Fluidigm products have not been cleared or approved by the Food and Drug Administration for diagnostic purposes and are only available for research use.

GATC Biotech AG **Stand number: A-224**
Email: customerservice@gatc-biotech.com
Website: www.gatc-biotech.com

With 19 years' experience, GATC Biotech is a leading provider of DNA sequencing services and bioinformatics software for industry and academia worldwide. GATC Biotech offers complete sequencing solutions, from sample prep to high throughput genome sequencing and bioinformatics and uses all leading high throughput sequencing technologies, the ABI 3730XL™, Illumina Genome Analyzer™ and Roche GS FLX™. The company has subsidiaries in France, the UK and Sweden.

Genial Genetic Solutions Ltd **Stand number: B-616**
Email: info@genialgenetics.com
Website: www.genialgenetics.com

Genial Genetics, leaders in genetic automation & software:
- MultiPrep Genie In-Situ Cytogenetic Culture Harvester
- MultiPrep CellSprint Suspension Culture Harvester
- iGene Genetic Lab Management System & Clinical Genetics System - providing tools for patient, sample & test management; workflow configuration including bar-coding, culturing, worksheets & batch processing. Clinical tools for family management, appointment scheduling, diagnosis & follow-ups.
- mWare middleware engine facilitating connectivity to instrumentation/ information systems via extensibile plugin architecture.
- iPassport Quality Management System facilitating accreditation management (ISO, CAP, CPA). Full document control, auditing, incident tracking, equipment & training management.
- Procell Cytogenetic Reagents

Genizon BioSciences **Stand number: A-206**
Email: services@genizon.com
Website: www.genizon.com

Genizon offers GLP compliant genetic and Pharmacogenomics services - high throughput SNP genotyping, and statistical genetics - to provide industry leading statistical power.
Genotyping/DNA methylation: Genizon operates eight Illumina Readers with a capacity of 1 billion genotypes/week.
Quality: Genizon is a certified service provider of Illumina. Performance data from 52,000 samples show 99.9% accuracy and 99.7% reproducibility for call rates of 99%.
Statistical Genetics: Genizon offers statistical genetics services using automated quality-controlled genetic analysis computing pipeline. Genizon performs case-control or family trio analysis using individual SNPs and/or multi-marker haplotypes.

GenoLogics **Stand number: A-336**
Email: info@genologics.com
Website: www.genologics.com

GenoLogics provides discovery and biomedical solutions that can be implemented across multiple labs and support translational medicine and systems biology initiatives. Our vision is to catalyze life sciences research with a collaborative data management software platform, advancing the early detection, prevention, and treatment of disease.

Genomed Ltd. **Stand number: B-604**
Email: info@genomed-biotech.com
Website: www.genomed-biotech.com

Genomed: a British company involved in worldwide distribution, manufacturing, product development, turnkey projects, and joint ventures of biotechnology products and services. www.genomed-biotech.com
AneufastTM is a CE/IVD Quantitative Fluorescent PCR (QF-PCR) test for rapid prenatal diagnosis of chromosomes 21, 18, 13, X and Y aneuploidies with 30,000 published results.
F-HPVTM is a CE/IVD Type Specific F-Multiplex-HPV Typing test for detection of Human Papilloma Virus. The test is rapid, simple and very sensitive and can be used for automated genotyping of 15 HPV-types. Fluorescent-PCR allows multiplex amplification in the same tube.

Gen-Probe (formerly TEPNEL) **Stand number: A-137**
Email: info@tepnel.co.uk
Website: www.tepnel.com

Tepnel is pleased to announce that it is now part of Gen-Probe, a world class provider of nucleic acid tests. Tepnel Pharmaceutical Services offers a comprehensive range of single-plex and multi-plex genotyping services. A variety of platforms and techniques support a wide portfolio of SNP-based investigations from whole genome screens through to candidate gene studies. The Elucigene range of kits and reagents are specially designed for the simple and rapid analysis of human genetic disorders including cystic fibrosis, cardiovascular disease and chromosome aneuploidy such as Down Syndrome.

GenVault Corporation **Stand number: B-504**
Email: info@genvault.com
Website: www.genvault.com

GenVault is the leading global provider of next-generation technologies for room temperature storage and management of biosamples for genomic research. GenVault's products enable extraction, recovery and preservation of DNA and RNA. With newly launched GenTegraTM DNA, and scalable archiving solutions, GenVault is pioneering new standards for biosample management. Visit: www.genvault.com.

HAMILTON Robotics GmbH **Stand number: A-127**
Email: infoservice@hamiltonrobotics.com
Website: www.hamiltonrobotics.com

From Clark Hamilton's Microliter® Syringe to Leading Laboratory Automation Technology, Hamilton's expertise in liquid handling goes back to the development of the revolutionary Microliter® Syringe in 1947 by the company founder Clark Hamilton. Thousands of Hamilton liquid handling workstations are serving customers in laboratories all over the world, some of the instruments with a track record of over 15 years.

HVD Life Science Vertriebs G.m.b.H **Stand number: A-116**
Email: office1@hvdgmbh.com
Website: www.hvdlifesicences.com

HVD is a marketing and distribution company which specialises in the Life Science Area and acts as a representative for some of the biggest names in the industry. We offer a wide range of products and support services covering training, guarantees, warranties and running service contracts. With offices in Vienna, Athens, Moscow, Hong Kong, Riyadh, Sharjah and Cairo as well as an extensive network of over 40 partner companies, we are ideally positioned to provide you with fast and top quality products and services that you can rely on.

Idaho Technology, Inc. - Co-exhibitor of BIOKÉ **Stand number: A-212**
Email: cameron_gundry@idahotech.com
Website: www.idahotech.com

Idaho Technology now offers two platforms that utilize their high speed, Hi-Res Melting™ technology for mutation scanning and genotyping– the LightScanner® 96-384 well plate system and the new LightScanner 32, which also offers real-time PCR capabilities. This system offers versatility without sacrificing performance. Rapidly generate high quality gene expression data, accurately discriminate DNA mutations and affordably genotype samples with the same specificity as TaqMan® genotyping at a fraction of the cost. Both systems provide superb sensitivity and specificity and come pre-loaded with intuitive analysis software that provides automatic results.

Illumina, Inc. **Stand number: A-330**
Email: info@illumina.com
Website: www.illumina.com

Illumina develops, manufactures, and markets a complete range of next-generation life science technologies, allowing researchers to comprehensively study both human and non-human species for genetic variation and biological function.

imaGenes GmbH **Stand number: B-632**
Phone: +49 30 9489 2444
Website: www.imagenes-bio.de

imaGenes GmbH, Berlin, is a premier provider of genome research services in Europe. imaGenes manages one of the most comprehensive collections of clones of mammalian and vertebrate genes in the world offered via its clone search engine GenomeCube®, linked to international databases. The imaGenes research services feature a broad range of microarray applications on Affymetrix, Agilent, and NimbleGen platforms as well as next generation sequencing services on an Illumina Genome Analyzer System. imaGenes is fully compliant with ISO 9001:2000 quality standards.

IMGM Laboratories GmbH **Stand number: A-119**
Email: info@imgm.com
Website: www.imgm.com

IMGM Laboratories – your partner for genomic services. IMGM is located on the biotech campus Martinsried and we offer advanced genomic services tailored to the needs of customers coming from pharma, academia and biotech. We are accredited according to DIN EN ISO/IEC 17025 and are an Agilent Certified Service Provider (CSP) for gene expression microarrays. Our service portfolio covers the areas of RNA services, DNA services, bioinformatics and consulting. At IMGM Laboratories, we combine state-of-the-art technology with complex data analysis to provide you with meaningful results in an easy-to-understand format (www.imgm.com).

Innogenetics N.V. **Stand number: B-700**
Email: info@innogenetics.com
Website: www.innogenetics.com

Innogenetics NV develops, manufactures and markets a wide range of diagnostic assays focussing on molecular diagnostics and multiparameter testing. The Company is a global leader in the manufacturing and marketing of LIA and LiPA based products. Innogenetics is also positioned to become the first company to commercialize a micro-array designed for clinical diagnostics, its 4-MAT™ system. The company's products are sold in over 90 countries through its 6 subsidiaries and a large number of distributors. Innogenetics' diagnostics business focuses on five areas: infectious diseases, genetic testing, transplantation, neurodegeneration and oncology.

Integrated DNA Technologies **Stand number: A-208**
Email: eutechsupport@idtdna.com
Website: www.idtdna.com

Integrated DNA Technologies (IDT) is the largest supplier of custom-synthesized DNA and RNA in the world, with two manufacturing plants in the U.S. and one in Europe, serving the areas of academic research, biotechnology, and pharmaceutical development. IDT products support applications including DNA sequencing, DNA amplification, expression profiling, microarray analysis, SNP detection, gene quantification, and functional genomics. IDT is unique in its ability to accommodate even the largest orders without compromising quality.

Interactive Biosoftware **Stand number: B-618**
Email: contact@interactive-biosoftware.com
Website: www.interactive-biosoftware.com

Our flagship product, Alamut, is a clinical decision support system focused on the interpretation of genomic variants. Alamut is designed to help quickly and reliably interpret mutations by synthesizing relevant molecular data and prediction methods within a consistent and user-friendly graphical environment. Alamut is used on a daily basis in numerous molecular diagnostic laboratories throughout the world.

Invitrogen part of Life Technologies **Stand number: A-110**
Email: ian.sanders@invitrogen.com
Website: www.invitrogen.com

Invitrogen provides products and services that support academic and government research institutions and pharmaceutical and biotech companies worldwide in their efforts to improve the human condition. Invitrogen provides essential technologies for disease research, drug discovery, and commercial bio-production. Invitrogen's products and services accelerate discovery by providing optimized, ready-to-use tools and processes.

JSI medical systems GmbH **Stand number: B-500**
Email: mail@jsi-medisys.de
Website: www.jsi-medisys.de

JSI medical systems is located in the Black Forest area in southwest Germany. Our software Sequence Pilot is one of the leading products for the analysis of conventional sequencing, next generation sequencing, Affymetrix chip resequencing, sequencing based typing and MLPA data. For detailed information see www.jsi-medisys.de.

KBiosciences Ltd. **Stand number: B-520**
Email: info@kbioscience.co.uk
Website: www.kbioscience.co.uk

KBiosciences is a leading SNP Genotyping company generating >500.000 datapoints/day, having >100.000 validated assays in stock and processed >150 million genotypes so far. KBiosciences is offering both Genotyping services and its own range of SNP genotyping chemistry and polymerases, as well as novel fully automatable instrumentation, as there are laser plate sealing systems, heat sealing systems and High Throughput Thermocyclers. As a partner and co-developer of Covaris Inc. KBiosciences also sells the ultra-sound based gold standard instrumentation for DNA-shearing applications in Next-Gen-Sequencing: the Covaris Adaptive Focused Acoustics (AFA) DNA-shearing technology.

Kreatech **Stand number: B-600**
Phone: +31 (0)20 691 91 81
Website: www.kreatech.com.

KREATECH Diagnostics is focused on the development, manufacturing and commercialization of innovative labeling and detection products. The company's main focus is the molecular cytogenetic market, in which Kreatech sells a broad range of FISH DNA probes for oncology and prenatal testing under the brand name Poseidon. These probes are unique through the design with Repeat-Free™ technology, are CE-marked and registered as in vitro diagnostic devices. Kreatech owns a unique labelling technology; the Universal Linkage System (ULS™) that allows non-enzymatic, direct labelling and detection of DNA, RNA and proteins for applications in the life sciences and healthcare industry.

LGC Standards GmbH - Co-exhibitor of AGOWA genomics **Stand number: A-320**
Email: de@lgcstandards.com
Website: www.lgcstandards.com

LGC Standards (trading as LGC Promochem until November 2007) is Europe's most comprehensive source of reference materials and certified reference materials. LGC Standards supplies over 20,000 reference materials, pharmaceutical impurity reference standards, proficiency testing and training in analytical quality. As the exclusive European distributor for ATCC cultures and bioproducts, LGC Standards is also committed to providing the latest research tools to life science researchers across Europe.

Macrogen Inc. **Stand number: A-130**
Email: info@macrogen.com
Website: http://dna.macrogen.com/eng

Macrogen Inc. (www.macrogen.com) is an internationally operating, Korea-based company providing total genomic solutions. We have served 9,000 bio researchers over the world since 1997. From regular sequencing of a single reaction to large scale projects involving the sequencing of an entire microorganism, Macrogen provides optimized genomic solutions equipped with cutting edge technology in DNA sequencing to meet the exact standards of its clients.

metabion international AG **Stand number: A-322**
Email: info@metabion.com
Website: www.mymetabion.com

Founded in 1997 in Martinsried, metaBIOn has managed to become one of the leading biotechnology custom synthesis services, offering different biomolecules designed for the scientist's specific application. For a consistently high quality standard, metaBIOn's oligo production is certified according to DIN EN ISO 9001:2000 regulations. Products:
- DNA oligos (with / without modifications in tubes or plates)
- RNA oligos (with / without modifications)
- Real-time PCR probes (incl. LightCycler™ probes)
- Peptides
- Polyclonal/Monoclonal antibodies
- DNA purification kits
- dNTP
- DNA marker
- Polymerases
- Restriction enzymes

MetaSystems **Stand number: B-610**
Email: cschunck@metasystems.de
Website: www.metasystems.de

MetaSystems is a leading manufacturer of imaging automation systems for cytogenetics. With its ultra-fast slide scanning platform Metafer the highest degree of automation can be achieved for many applications, including metaphase finding, karyotyping, FISH image analysis, multicolor FISH, and multicolor chromosome banding. The MetaSystems portfolio also includes high quality DNA probe kits for chromosome painting, mFISH, and mBAND.

The Microarray Facility Tübingen **Stand number: A-314**
Email: info@microarray-facility.com
Website: www.microarray-facility.com

The Microarray Facility offers innovative and reliable services in the field of microarray applications and applied genomics to researchers in all fields, commercial and academic. As authorized Affymetrix Service Provider, The Microarray Facility is able to bring GeneChip experiments within reach of every researcher as well as all Illumina tools for large-scale analysis of genetic variation and function, including genome wide methylation and miRNA expression profiling. As the latest milestone, we offer custom made capture arrays for enrichment of specific sequences.

MRC-Holland-MLPA **Stand number: B-630**
Email: info@mlpa.com
Website: www.mlpa.com

MRC-Holland is a fast-growing biotechnology company based in Amsterdam, The Netherlands. MRC-Holland is the inventor of the Multiplex Ligation-dependent Probe Amplification (MLPA®) technique, a sensitive method to detect copy number changes in genes. MLPA® is used in numerous applications, from identifying hereditary diseases to investigating genetic alterations in tumors. We have over 300 SALSA MLPA kits available, such as kits for DMD, SMA, subtelomeres, BRCA1/2, RETT, CAH, APC, Williams, Prader-Willi/Angelman, DiGeorge, Cri du Chat, Pelizaeus-Merzbacher, CMT, HNPP. Our MLPA® products are used in more than 600 labs and hospitals worldwide.

Multiplicon **Stand number: A-114**
Email: geoffrey.henno@vib.be
Website: www.multiplicon.com

Multiplicon is active in the field of Applied Genomics and focuses on the development of innovative analysis-tools for genomic research. PCR Multiplexing is our central technique, enabling us to produce simple PCR tools and protocols for molecular diagnostics and next-generation sequencing purposes. We produce and market assays for prenatal and general genetic analysis, tools for DNA verification and CNV detection and we also offer custom assay development. Visit multiplicon.com for more details.

NanoDrop Products, a part of Thermo Scientific **Stand number: B-502**
Email: nanodrop@thermofisher.com
Website: www.nanodrop.com

Thermo Scientific NanoDrop products are specifically designed for 1ul samples, employing a patented retention system that eliminates the need for dilutions of sample. Using this novel system, the Thermo Scientific NanoDrop™ 2000 and 2000c spectrophotometers and the NanoDrop™ 8000 eight sample spectrophotometer enable full UV-Vis absorbance analysis while the NanoDrop™ 3300 Fluorospectrometer provides fluorescence spectra analysis.

NATURE PUBLISHING GROUP **Stand number: A-339**
Phone: +44 (0) 207 833 4000
Website: www.nature.com

Nature Publishing Group (NPG) brings leading scientific and medical research to your desk top. The NPG portfolio combines the continued excellence of Nature, NPG's flagship journal and its associated research and review journals, over 45 leading academic and society journals - Including European Society of Human Genetics. NPG also provides news content through Nature News and scientific career information through Naturejobs. Visit the NPG stand to pick up your free sample copies, subscribe at the conference rate, or enquire about advertising opportunities in any NPG title.

New England Biolabs GmbH **Stand number: B-636**
Email: info@de.neb.com
Website: www.neb-online.de

Established in the mid-1970s, New England Biolabs is a world leader in the production and supply of reagents for the life science industry. NEB offers the largest selection of enzymes for genomic research and continues to expand its product offerings into areas related to proteomics, drug discovery and cell imaging.
NEB GmbH is the service hub for Germany, Austria & Eastern Europe and also distributor for Cell Signaling Technology Inc., a world leader in the production of the highest quality activation-state antibodies utilized to expand knowledge of cell signaling pathways implicated in various diseases including cancer, diabetes and neurodegenerative diseases.

Nikon Instruments S.p.A **Stand number: A-125**
Email: cristiana.ricci@nikon.it
Website:

Software for Karyotyping and FISH analysis.

ORPHANET **Stand number: B-524**
Email: orphanet@orpha.net
Website: www.orpha.net

ORPHANET is a free access European database dedicated to information on rare diseases and orphan drugs. ORPHANET offers services adapted to the needs of patients and their families, health professionals and researchers, support groups and industry. ORPHANET includes an encyclopaedia and a directory of services in Europe : specialised outpatient clinics, clinical laboratories, research activities and support groups.

Oxford Gene Technology **Stand number: B-613**
Email: simon.walker@ogt.co.uk
Website: www.ogt.co.uk

The key focus areas of OGT include:
1. High Throughput Microarray Services. With a processing capacity of over 1,000 samples per week, applications available include aCGH, CNV, methylation studies and miRNA.
2. Cytogenetics products for high resolution detection of chromosomal abnormalities. With a range of high resolution oligonucleotide arrays, labelling kits and analysis software that together provide a unique, comprehensive solution for cytogenetics.
3. Single Cell analysis. OGT's innovative, patent-protected technology development programme is aimed at analysing genomic events at the single cell level for major applications such as stem cell and cancer biology.

PAA Laboratories GmbH **Stand number: B-402**
Email: s.deckers@paa.com
Website: www.paa.com

PAA Laboratories offers cell culture products for research and diagnostic applications, including liquid and powder media, sera, reagents and buffer solutions. Furthermore, PAA developed a new product standard for raw material used in the manufacture of Advanced Therapy Medicinal Products (ATMP) and was granted a GMP licence for the production of "in vivo products for tissue cultures". For specific applications such as neuronal stem cell culture and cytogenetics we also provide the appropriate products. With production facilities and sales subsidiaries on three continents and more than 50 distributors PAA acts globally.

PASS Software **Stand number: A-121**
Phone: +31 6 38931555
Website: www.pass-software.com

PASS Software offers solutions for clinical genetic research, consultancy and screening programmes for clinical genetic facilities and institutes. Pass Clinical ® is designed to support various genetic screening programmes, like the Dutch national screening programme for Familial Hypercholesterolemia. It offers a unique combination of functionalities: automatic pedigree drawing, flexible data collection and workflow management.

PerkinElmer Inc. **Stand number: B-714**
Phone: +358 2 2678 111
Website: www.perkinelmer.com

PerkinElmer, Inc. is a global company focused on improving the health and safety of people and their environment. PerkinElmer provides versatile complete aCGH solutions and is about to launch its new bead based high-throughput molecular karyotyping solution. PerkinElmer's molecular karyotyping solutions provide the speed, throughput and resolution required for today's cytogenetic laboratories.

PhenoSystems SA **Stand number: A-220**
Email: david@phenosystems.com
Website: www.phenosystems.com

PhenoSystems develops software for molecular genetics and presents its latest version of Gensearch, DNA sequence analysis software for mutation detection and interpretation. Its major features are high specificity and sensitivity as well as advanced tools to support interpretation of mutations (frameshifts, splice prediction, connection to certain locus specific databases). Gensearch has been developed together with leading diagnostics laboratories in Europe, packing powerful tools in an extremely user friendly interface. Customisation of the software can be done as well. PhenoSystems is partner of EuroGentest (FP6), Gen2Phen (FP7) and NMDchip (FP7).

POSSUM - Murdoch Childrens Research Institute **Stand number: A-228**
Email: possum@mcri.edu.au
Website: www.possum.net.au/

POSSUM-web is a web-based software tool that can help with diagnosis of syndromes in patients and teaching about syndromes. By providing a flexible search facility, POSSUM-web saves time and effort in researching and cross-referencing syndrome information. The POSSUM database contains information on nearly 4000 syndromes, including skeletal dysplasias and chromosomal disorders. There are over 30,000 images including photos, Xrays, diagrams, and histology, with an extensive trait dictionary and atlas. POSSUM-web is available by annual subscription, with monthly data updates, and images delivered by DVD and updated annually.

PRESTAGEN **Stand number: A-123**
Email: prestagen@prestagen.com
Website: www.prestagen.com

Prestagen is a company developing one step multiplex PCR kits designed to detect quantitative aberrations of any gene or chromosomal region. These kits are based on a new proprietary technology called PlexAmp, which allows the simultaneous quantitative PCR amplification of several DNA fragments in a quick one step procedure. The quantitative analysis of DNA fragments is then performed on a DNA sequencer using a specific fragment analysis software.

Progeny Software LLC **Stand number: B-400**
Email: progenyeurope@progenygenetics.com
Website: www.progenygenetics.com

Progeny is a premier provider of Clinical and Genetic data management systems. At ESHG we will be presenting our suite of products Progeny CLINICAL, LAB and LIMS and will demonstrate how an integrated solution can help you effectively organise your data to gain maximum value from it. Progeny aids in the management of clinical, phenotype, pedigree and case-control data , SNP/STR genotypes and all associated document information. We will present our latest WEB versions of Progeny CLINICAL and Progeny LIMS designed with collaboration in mind. VISIT US FOR A DEMO.

Promega **Stand number: B-612**
Email: katja.krauth@promega.com
Website: www.promega.com

Promega is a leader in providing innovative solutions and technical support to the life sciences industry. 2,000 products enable scientists to advance their knowledge in life science research, particularly in genomics, proteomics, and cellular analysis. The products are also used to support molecular diagnostics and human identification (e.g. PowerPlex®) and a complete portfolio of luminescent and fluorescent cell-based and biochemical assays that are amenable to drug-discovery screening and other high-throughput investigations (e.g. Dual-Luciferase, Glomax product line and HaloTag™). Maxwell® 16 System is part of the integrated solution portfolio and achieves consistent yield and purity in nucleic acid isolation.

Protalix Biotherapeutics Inc. **Stand number: A-122**
Email: shomrats@protalix.com
Website: www.protalix.com

Protalix is a biopharmaceutical company focused on the development and commercialization of recombinant therapeutic proteins based on our proprietary ProCellExtm - plant cell protein expression system. Our initial commercial focus is on complex therapeutic proteins, including proteins for the treatment of genetic disorders, such as Gaucher disease and Fabry disease. Our lead product candidate is prGCD, a glucocerebrosidase enzyme for the treatment of Gaucher disease. Protalix is performing a pivotal Phase III clinical trial under SPA agreement. The enrollment of all the required patients has been completed.

QIAGEN **Stand number: B-706**
Phone: +49 (0) 2103 29 00000
Website: www.qiagen.com

QIAGEN is the leading provider of innovative technologies and products for sample preparation, assay detection and molecular diagnostics solutions. We have developed a portfolio of consumable products and automated solutions for sample collection, nucleic acid and protein purification and detection. We also supply diagnostic tests, and assays for human and veterinary molecular diagnostics. Take advantage of QIAGEN's research and development capabilities for genotyping and genetics research. Over 320 researchers on 3 continents in 5 R&D centres of excellence provide outstanding expertise in preanalytical sample processing, assay development, detection and automation.

RainDance Technologies, Inc. **Stand number: B-506**
Email: info@raindancetech.com
Website: www.raindancetech.com

RainDance Technologies Inc. is a provider of innovative microdroplet-based solutions for human health and disease research. The speed and simplicity of the company's exciting new technology platform enable researchers to design experiments in ways that were previously unaffordable or unimaginable. The company's RainStorm™ technology produces picoliter-volume droplets at a rate of 10 million per hour. Each droplet is the functional equivalent of an individual test tube and can contain a single molecule, reaction, or cell. This versatile technology can adapt proven assays for high-speed workflows with minimized process-induced error.

Roche Applied Science
Stand number: B-532
Email: Friedhelm.Huebner@roche.com
Website: www.Roche-Applied-Sience.com

Roche Applied Science provides advanced systems, instruments, and reagents for life science research. Products include innovative instrument platforms such as the LightCycler® System for quantitative PCR and real-time, online detection of nucleic acids. The plate-based LightCycler® 480 Instrument was developed for medium- and high-throughput real-time PCR. The MagNA Pure Systems are innovative components for high-quality nucleic acid purification. The Genome Sequencer FLX System provides the flexibility to address a broad range of applications in various fields. LIGHTCYCLER, GS FLX and MAGNA PURE are trademarks of Roche.

Rosetta Biosoftware **Stand number: B-712**
Phone: + 1 206 926 1200
Website: www.rosettabio.com

Rosetta Biosoftware provides life science informatics solutions that enable breakthroughs in basic biological research, drug discovery and development, and translational research. Through superior integration and analysis of data and content from diverse sources, our products accelerate research and improve safety and therapeutic decision making. We continue to innovate leading technology that converts data into understanding in support of the promise of personalized medicine.

Science/AAAS International **Stand number: A-200**
Email: subs@science-int.co.uk
Website: www.sciencemag.org

Since 1848, AAAS and its members have worked together to advance science and serve society. As part of these efforts, AAAS publishes Science, a multidisciplinary peer-reviewed journal, and offers programs focused on science policy, international cooperation, science education, diversity, and career development for scientists. Learn more at aaas.org and sciencemag.org.

Tecan **Stand number: A-132**
Email: info.de@tecan.com
Website: www.tecan.com

Tecan is a leading global supplier of solutions for the biopharma, forensic and diagnostic industries. The company is a leader in the development, production and distribution of advanced automation and detection solutions for the world's leading life science laboratories. Through its REMP subsidiary, Tecan is the premier supplier of large-scale automated laboratory storage and logistics systems.
Founded in Switzerland in 1980, the company has over 1100 employees, owns production, research and development sites in both North America and Europe and maintains a sales and service network in 52 countries.

TEPNEL : see Gen-Probe **Stand number: A-137**

Thermo Scientific **Stand number: B-510**
Email: abgene.info@thermofisher.com
Website: www.thermo.com

Thermo Scientific offers customers a complete range of high-end analytical instruments as well as laboratory equipment, software, services, consumables and reagents to enable integrated laboratory workflow solutions. Thermo Scientific microplate instruments are used for cell biology, molecular biology and immunology applications in the field of cancer research, drug development, proteomics and genomics. ABgene – now sold as Thermo Scientific - manufactures and develops high-performance QPCR reagents and amplification plastics that facilitate reproducible, sensitive and accurate quantization of all nucleic acid targets. Through constant customer feedback and an innovative approach to product development, Thermo Scientific has developed a portfolio of industry-leading products.

TIB Molbiol Syntheselabor GmbH **Stand number: B-530**
Email: dna@tib-molbiol.de
Website: www.tib-molbiol.com/

TIB MOLBIOL is a manufacturer of oligonucleotide primers and Real-Time-PCR probes (Scorpion primers, TaqMan, LightCycler® FRET probes). TIB offers a wide spectrum of fluorophores and modifications, including Locked-Nucleic Acids (LNA). The R&D group provides a non-charged assay design service and the (charged) development of tests. The LightMix® Kits (under license from Roche Diagnostics) cover different genotyping applications for pharmacogenetics, cancer mutation search, clinical chemistry and hemachromatosis (licensed from Bio-Rad) as well as quantification tests for haematology or pathogen detection. In 2008 we introduced our LightMix® Methylation Kits (collaboration with Epigenomics).

TRANSGENOMIC **Stand number: A-134**
Email: sales@transgenomic.com
Website: www.transgenomic.com

TRANSGENOMIC is a global biotechnology company that provides unique systems, products (like SURVEYOR® Mutation Detection kits), discovery and laboratory testing services to the academic and medical research, clinical and pharmaceutical markets for automated high sensitivity genetic variation and mutation analysis in the fields of pharmacogenomics and personalized medicine. Use of Transgenomic's WAVE® System instrument platform has been cited in more than 1,700 research publications. Transgenomic also provides technology for: automated cytogenetic analysis with HANABI Metaphase Harvesters and Chromosome Spreaders; imaging and offline analysis with the BIOVIEW cell imaging platform.

ViennaLab Diagnostics GmbH **Stand number: B-710**
Email: info@viennalab.co.at
Website: www.viennalab.com

ViennaLab offers a range of innovative diagnostic tests based on reverse hybridisation. The user-friendly StripAssays with up to 48 positions allow the detection of DNA variants including mutations, deletions, methylation status and more. With the StripAsssays proven reliability, the diagnosis of genetic diseases and predispositions can be adressed with highest accuracy. The current application fields are cardiovascular dieseases, haemochromatosis, hemoglobinopathies and FMF as well as pharmacogenetics. The new KRAS StripAssay and more cancer-related tests currently under development are to deepen ViennaLab´s focus on cancer.

Wiley-Blackwell **Stand number: A-126**
Email: customer@wiley.co.uk
Website: www.wiley.com

Wiley-Blackwell was formed in 2007 as a result of the merger between Wiley's Scientific, Technical and Medical business and Blackwell Publishing. Together we publish more than 1,400 journals, including Annals of Human Genetics, Clinical Genetics and also Human Mutation. Our extensive and expanding books program includes classic textbooks, current protocols and professional references from leading authors in the field. Visit the Wiley-Blackwell exhibition booth at ESHG to view our extensive collection of print and online publications and take advantage of a 20% discount on all our books.

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Carl Zeiss GmbH **Stand number: B-614**
Email: a.wagner@zeiss.at
Website: www.zeiss.at

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Dr. Brian Gregory
Postdoctoral Fellow
The Salk Institute for Biological Studies

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