



# Update from EduCom

**Inga Prokopenko May 2025**

# Updates

The committee - 20 members

Committee focus areas:

- online courses,
- International relations,
- relationships with European Research Networks,
- Mentorship/observership programmes,
- online/digital genetic/omic education resources,
- Public education/Engagement
- Addressing disparities
- [DNADay essay and Video contest](#)



HOME

FOR GENETICS PROFESSIONALS

FOR UNIVERSITIES & STUDENTS

FOR PATIENTS & FAMILIES

NON-GENETICS SPECIALISTS & PRIMARY CARE

FOR SECONDARY SCHOOLS

FOR PRIMARY SCHOOLS

ETHICAL, LEGAL & SOCIAL IMPLICATIONS

# ESHG Genetic Educational Materials and Sources (EuroGEMS)

## For genetics professionals

This page provides information that is likely to be useful to professionals in clinical genetics/genomics e.g. information about individual clinical conditions (GeneReviews, OMIM and Orphanet), individual genetic variants (DECIPHER, LOVD, ClinVar, HGMD, VarSome, CanVar & VEP) and variant nomenclature (HGVS). Professionals are also likely to find sources that are listed on other pages here to be of relevance and interest, particularly the [Universities page](#) (including the genome browsers) and the [Ethical, Legal and Social Implications page](#). In addition, details and links to courses that are co-organised and/or supported by the ESHG can be found here.

On this page:

[COVID & CLINICAL INFO](#) | [GENOME BROWSERS](#) | [VARIANT ANALYSIS](#) | [HGVS-CHECK & GENERAL](#) | [LOCAL INITIATIVES](#)



# EuroGEMS ([EuroGEMS.org](http://EuroGEMS.org)) Ed Tobias

- The ESHG educational website has now been visited from 145 countries (still growing diversity).
- 4 languages
- Particularly noteworthy has been the growing number of visits to the professionally translated Spanish, Portuguese and French pages from numerous countries, within and outside Europe.
- Alongside the growing number of users, educators from many countries including Canada, Japan, Singapore, India, Egypt, South Africa, Argentina and Mexico have praised its usefulness.
- Web pages are now being used by the members of HUGO-International.



<https://www.genomicsapps.org> Ed Tobias

**associated free educational genomics terminology app**

[HOME](#)

## INHERITANCE MODE QUIZZES

## PRIVACY POLICY

## CONTACT

## GENOMICS IN BRIEF


MORE...

This page describes the Clinical Genomics Guide (glossary) App and its accompanying Quiz App.

[Click/tap here for the page regarding the Inheritance Mode Quiz Apps for students.](#)

### Clinical genomics guide


version 6.4.4



Copyright © 2019 Edward and Adam Tobias


### Circos genome plot

A circular diagram with chromosomes indicated around the outside. Many other data types can be added e.g. genes, repeats, coverage level, DNA variants, centromere positions (as shown here) and connections between pairs of locations, using lines stretching across to show gene fusions, as here. Plot created by Ed Tobias (see Image Source).

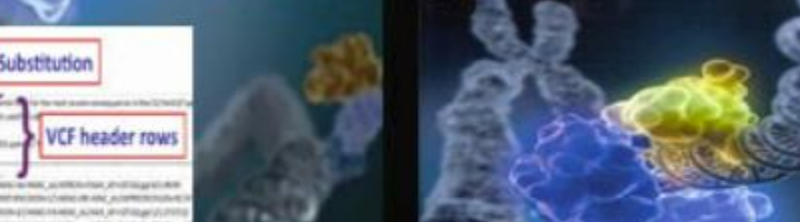


### Third generation sequencing

Also known as "long-read sequencing". Performed, by different methods, on long single DNA molecules rather than on many small segments of DNA (as in second generation sequencing i.e. NGS). Image shows DNA (white) entering an alpha-haemolysin "nanopore". See "long-read sequencing".



### UPF1 phosphorylation




### Understand the purpose of BWA & other bioinformatic software tools

BWA (BWA-MEM)

Bowtie2

IGV


IGV (Integrative Genomics Viewer) is a program for displaying e.g. BAM files, showing the sequence reads aligned to a reference sequence



### Understand bioinformatic software tools

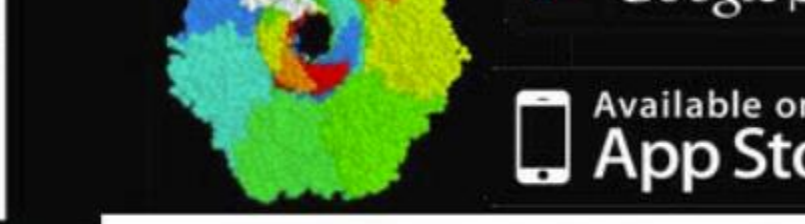
Available on the Google play

Available on the App Store



### Installation of the apps internationally

(in over 70 countries)



### Including GATK, GVCF and MNPs

Clinical genomics guide

Choose a topic below

- FASTQ file
- GATK
- Genome projects
- Genomics England
- Germline variant
- gnomAD database
- GVCF file
- IGV genome viewer
- Long-read sequencing
- MNPs
- NGS
- NMD
- Pathogenic variant
- PED file
- Phred score
- pLI score

### and Phred, pLI & Z scores

Clinical genomics guide

Choose a topic below

- NMD
- Pathogenic variant
- PED file
- Phred score
- pLI score
- Polygenic risk score
- SAMtools
- SGP
- SNV
- Somatic variant
- TAD
- Third gen. sequencing
- VCF file
- VUS
- WGS
- Z score


### Medical Genetics

ESSENTIALS

EDWARD S. TOBIAS, MICHAEL CONNOR, MALCOLM FERGUSON-SMITH

6TH EDITION

WILEY-BLACKWELL



### The Clinical Genomics Guide App



# **New free ESHG webinar series**

**In collaboration with President Bill Newman and SpC**

- **Episode 1 - Wednesday, February 26, 2025 (16:00 CET)**
- **The Dark Side of the Genome: Challenges and Opportunities in the Post-Genomic Era. Malte Spielmann, University Hospital Schleswig-Holstein Germany**
- **Episode 2 - Wednesday, March 26, 2025 (16:00 CET)**
- **The non-coding exome in rare disease: from diagnosis to therapies. Nicola Whiffin, University of Oxford, United Kingdom**
- **Episode 3 - Wednesday, April 30, 2025 (16:00 CEST)**
- **Making sense of genetic associations. Zoltan Kutalik, University of Lausanne, Switzerland**
- **Episodes 4 and 5 planned**

# New online pre-conference course

- "Presentation Skills Workshop for Conference Speakers"
- Organised by Julia Baptista, Hans Hennies, Sally Ann Lynch
- May 6 2025 - online live only, 08:30-12:15 BST (09:30-13:15 CET).
- received 30 applications, more than initially planned
- 26 attended
- The course focussed on presentation skills rather than scientific content.

# Pre-conference courses

## Half-day, CME credits

**Professional development workshop: Interactive approaches to genetic teaching**

**Course organizers :** *Bregje van Bon*, Radboud University Medical Center, NL; *Célia Azevedo Soares*, Universidade de Aveiro, PT

## Clinical NGS Data Interpretation Course

**Course instructors:** *Christian Gilissen*, bioinformatician, expert in WES/WGS analysis; *Rolph Pfundt*, clinical molecular geneticist, expert in cytogenetics of WES/WGS; *Erik-Jan Kamsteeg*, clinical molecular geneticist, expert in molecular genetics of WES/WGS; *Caroline Racine*, clinical geneticist, expert in molecular genetics of WES/WGS

### Learning goals:

- How to interpret SNVs/CNVs from WES data
- What practical tools/databases for WES interpretation are freely available
- How the interpretation of WGS data is different from WES data



# International Mentorship Scheme

Five successful candidates were selected in year 4 for 2025 :

Attend ESHG conference and week in mentor centre.

- . **Tea Mladenic** Croatia to Germany
- . **Daniela Oliveira** Portugal to Sweden
- . **Lein Dofash** Australia (Palestine) to UK
- . **Melda Erdogan** Turkey to Sweden
- . **Nesibe Saliha Bulut** Turkey to Austria

**Winners are invited to meet the committee – plan to give awards at the plenary on Tuesday**

# International Observer Scheme

Five successful candidates were selected in this 2nd year program.

Month in a different centre

- Juliana Miranda Cerqueira Finland (Brazil) to UK
- Vanessa Sousa Portugal to Belgium
- Purvi Majethia India to UK
- Luiza Lorena Pires Ramos Belgium to Sweden
- Sílvia Pires Portugal to Germany

Winners are invited to meet the committee on Monday – plan to give awards at the plenary on Tuesday

# ESHG podcasts (Rita Barbosa-Matos)

- Podcasts - good feedback- 3 series total 16 episodes; 4<sup>th</sup> series starting
- Range topics and speakers engagement across all continents
  - multiple platforms, including Buzzsprout, Apple, and Spotify
- Increased exposure after “Live Event” at ESHG 2022
  - New “Live Event” at ESHG 2025
  - “The diary of a journal editor. Voices, Values, and the Vocabulary of Science”



# ESHG Xpeer Channel – featured on ESHG website



## CME accredited medical education

**Paediatric Neurogenetics.** Sponsored through Unrestricted Educational Grant by Illumina. ~€30k

Course content led by Nicola Brunetti Pieri (with Nadia Buisson and Kathleen Gorman)

- Collaboration with European Pediatric Neurology Society
- **Updated classification of developmental and epileptic encephalopathies and brain malformations -**  
<https://web.xpeer.app/en/courses/340>
- **Past and present diagnostic approaches of genetic syndromes and inherited metabolic disorders -**  
<https://web.xpeer.app/en/courses/339>
- **Movement disorders, neurodegenerative diseases and neurocutaneous syndromes -**  
<https://web.xpeer.app/en/courses/341>
- 12 x 15 minute modules
- Total: 156 tests taken, and 115 diplomas obtained for the three modules
- Double from last year



CHANNEL BY NUMBERS

# New App Based course Xpeer

- **Fundamentals in Genetic Testing**
- Focus: educating clinicians in basics of human genetics and testing
- **Leads:** James O'Byrne, Ed Tobias
- More topics are discussed for development
- Meeting with Illumina to discuss the new course

# Courses

## New courses approved

- Pharmacogenomics Course, Lead: Vita Dolžan, University of Ljubljana (June, 4 - 6, 2025, Slovenia)
- Leena Peltonen School of Human Genomics, in collaboration with Wellcome Conferences, Wellcome Genome Campus, Hixton, UK, Tutors – 20 geneticists, great attention and many applications
- Translational Epigenetics in Precision Medicine, Lead: Eka Kvaratskhelia (October 2025, Georgia)

## Proposed – in development

- Master Class in skeletal dysplasia (Valérie Cormier-Daire) - December 8-10, 2025, Imagine Institute Paris



# Courses

- Delivered
- In French language - Dakar (Senegal)
- ESHG-S2GH Workshop:
- Introduction à l'analyse de données massives génétiques, épigénomiques et transcriptomiques: initiation aux principes théoriques et ateliers pratiques
  - Took place before the National Meeting of the Senegalese Society of Human Genetics.
  - leader: N Bouatia-Naji, local organiser: Khalid Sidki (President of Moroccan association of human genomics and genetics: SM2GH)
  - Number of attendees: 20 selected from more applications
  - French-speaking countries Senegal, Rwanda, Tanzania, and Côte d'Ivoire

# Recent Courses

2<sup>nd</sup> ESHG Precision Genomic Medicine Workshop: A Focus on Clinical Utility.

- Lead: James O'Byrne, Dublin, Ireland, April 9-11,2025

CME accredited.

- scholars coming from 13 countries and 5 continents.
- Hybrid workshop
- Partnership with the CCMG & HGSA
- one of the first times all 3 societies have worked together to deliver an educational event.
- 25 world-class educators at an iterative workshop
- Over 70 attendees

# Conference courses/ Educational Sessions

## Proposal

- Better collaboration with SpC
- Opportunity for Ad hoc review of proposed Conference courses/ Educational Sessions
  - Feedback from EduComm provided by two EduComm SpC members (Julia/Inga)
  - This will enable better interaction with ESHG-Y, SpC, and other committees.



# International Education Links

**co-led by Edith Coonen, Liz Loehrer, Carmen Navas and Ed Tobias**

- Focus on promoting genetic education
- we expanded our connections, engaging with
  - Indian, Irish, Georgian, and Romanian national societies.
  - genomics educators providing clinical genetic training in Tanzania.
- We are developing a summary on the future needs for genomic education in Europe.