Summary of GENiaal, the Training Plan for Clinical Genetics

Dutch Clinical Genetics Association

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Introduction

In 2007-2008 a working group of the Dutch Clinical Genetics Association rewrote the training course for the clinical genetics specialism in accordance with the new framework decree and based on the final attainment levels in force. In 2009 that document, called GENiaal (ISBN-978-90-9024168-5), was officially agreed by the Central Board for Medical Specialisms.

GENiaal describes the profile of the specialism, the skills of the clinical geneticist, the 5 subjects of the specialism, putting the skills into operation both in critical professional situations and other instruction methods, the modular structure of the training course, national course teaching, test methods used, the requirements for the portfolio and the final attainment levels for clinical genetics.

The main new aspects concern the following points:

- Specification of all requisite skills for clinical geneticists on the basis of the CanMEDS model.
- The AIOS [Arts In Opleiding tot Specialist – doctor being trained as a specialist] himself plays an active and central role in the acquisition of these skills.
- Integration of the national course teaching, assessments and testing into the training plan.
- Assessment of defined practice situations forms the basis for structured periodic feedback.
- The whole group of trainers is to be schooled in and participate in new training.
- Progress in the training course can be made visible through growth in skill levels.

1. Profile of clinical genetics

Clinical genetics is a gateway specialism in the filed of second and third-line therapy. It is directed at:

1. The pre- and postnatal clinical genetic diagnosis of (possible) hereditary disorders, congenital abnormalities, dysmorphological syndromes and developmental disorders.
2. The preparation and provision of genetic counselling to individuals, families and relatives. This concerns both patients and healthy family members. Genetic counselling is a communication process between those seeking advice and professional advisers within the ethical and legal frameworks established for that purpose, and includes medical, genetic and socio-psychological aspects.
3. Participating in the management of patients with (possible) hereditary disorders, congenital abnormalities and developmental disorders.
4. Interpreting genetic laboratory results for other healthcare workers.

2. Skills of a clinical geneticist

1. Taking medical action

A clinical geneticist possesses broad up-to-date knowledge and a large arsenal of skills, so that he can draw up an appropriate plan of action within the boundaries of his discipline and expertise. That expertise specifically comprises clinical genetic diagnostics (prenatal, postnatal and presymptomatic), dysmorphology and genetic counselling. He applies the specialism's diagnostic and counselling arsenal correctly and where possible evidence-based across the whole spectrum of medicine. He provides effective and ethically responsible patient care and uses in principle a non-directive approach with scope for shared decision making. In drawing up the plan of action he takes into account the patient's gender, age, development level, stress capacity, ideology, family circumstances and cultural background.

2. Communication

Communication is essential for a clinical geneticist. Communicative skills are important when building up an effective counselling relationship with the patient/advice seeker and obtaining the necessary information about the matter on which help is being sought, previous medical history and the family anamnesis given in a three-generation genealogy. They are also essential for conveying the necessary information understandably and empathetically to the patient/advice seeker, written reporting, and recognising relevant psychological

aspects. He accompanies the patient/advice seeker when making choices between options for action and refers them if necessary. Also of the utmost importance is communicating effectively with the family (family dynamic) and with other care providers involved.

3. Cooperation

Since the working sphere of a clinical geneticist includes virtually the whole of medicine, intensive cooperation is required with other (para-) medical practitioners and laboratory specialists. There is also close cooperation with genetic consultants and the psychosocial carers in the same department, who often work under the supervision of a clinical geneticist. A clinical geneticist consults appropriately and refers effectively. He provides effective consultations among colleagues and contributes to effective interdisciplinary cooperation and chain care. He is able to handle effectively differences of opinion within multidisciplinary teams. He can interpret for other doctors the results of genetic laboratory tests.

4. Knowledge and learning

Throughout their professional lives, clinical geneticists strive to optimise their skills. They are in a position to contribute to scientific enquiry and research, both fundamental and clinical, and can translate its results into improved clinical genetic patient care. A clinical geneticist views medical information critically and in an evidence-based manner. He facilitates training of students, doctors' assistants, departmental colleagues, patients, advice seekers and other (para) medical staff. He develops and maintains a personal further training plan.

5. Social action

A clinical geneticist knows and recognises the determinants of illness and promotes the health of patients and of the community as a whole. A clinical geneticist acts in accordance with the relevant legal provisions and takes effective action in response to care incidents. Clinical geneticists are aware that their specialism is regularly the subject of public and political debate, and they know how to deal with this effectively. If required they support parent and patient organisations in the sphere of hereditary and congenital abnormalities. They contribute at (inter-)national level to the social debate about the applications of genetic knowledge.

6. Organisation

Clinical geneticists organise their own specialism, in which they take decisions on the deployment of resources and staff, the setting of objectives and the creation of a balance between work and private life. They do this in various spheres: individual patient care, practice/department organisation, regional knowledge networks, scientific research, training and professional associations. A clinical geneticist is at the forefront of applying information technology, and organises work from the perspective of the patient/advice seeker.

7. Professionalism

Clinical geneticists have specific skills which are directed at promoting the health and well-being of patients/advice seekers. Clinical geneticists strive for the highest possible level in clinical care and ethical behaviour and strive to perfect their specialist knowledge (permanent education).
Clinical geneticists apply the current professional, legal and ethical rules, as drawn up by (inter-)national professional organisations. They deal effectively with professional uncertainty and doubt, and are aware of the tension which can arise between the provision of care to the individual patient/advice seeker and to their family. A clinical geneticist is able to deal effectively with complaints/conflicts.

3. Subjects within clinical genetics

In order to cover the clinical geneticist specialism in characteristic professional situations, five subjects have been chosen.

1. Diagnostics and genetic counselling in connection with the wish to have children.
2. Diagnostics and genetic counselling in connection with the patient's own health.
3. Postnatal syndrome diagnostics.
4. Prenatal diagnostics.
5. Family implications of genetic diagnostics.

The 5 subjects cover all activities within clinical genetic patient care, and 1 or more subjects can be recognised in all activities within the specialism.

4. Operationalised skills and critical professional situations by subject

By subject the skills are operationalised and then linked to 58 critical professional situations. These are case and work situations which are illustrative of day-to-day practice in clinical genetics. They occur frequently and are readily testable by means of short practice assessment. They have been divided up under the 5

subject headings and both general genetics and the various focal areas are dealt with, so that the final attainment levels for clinical genetics are largely covered. The numerical focus is on the skills Taking medical action, Communication and Knowledge and learning.

Skills	**Description**
Taking medical action	Effective ascertainment of symptoms further to the referral. Anamnesis, family anamnesis (at least 3 generation genealogy), physical examination and interpretation thereof. Differential diagnostics. Effective use of supplementary diagnostics, including genome analysis and interpretation of results. Genetic probability calculation and risk assessment. Knowledge of action options, including prenatal diagnostics. If necessary referral to other specialists, psychosocial carer and/or parent and patient organisations.
Communication	Understandable explanation of complex material. Basic conversational skills, imparting bad news, recognising psychological risk factors (dealing with bereavement, relational aspects, stress capacity, risk communication, helping to make comparative assessments. Effective reporting to advice seeker and referer.
Knowledge and learning	Using the latest information, including from the area between diagnostics and research. Having a knowledge of genetic heterogeneity, phenotype-genotype correlation, non-genetic phenocopies. Contributing to the increase of knowledge and learning. Using EBM [evidence-based medicine].
Cooperation	With the first, second and third line. With diagnostic laboratories. With multidisciplinary (para)medical colleagues involved.
Organisation	Patient-oriented organisation: short waiting times, efficient surgery hours, fast reporting, easily contactable.
Social action	Legal and ethical aspects of asking for patients' and third parties' medical data and of various action options, including pregnancy termination on medical grounds. Working in accordance with relevant legal provisions such as the WGBO [Wet op de geneeskundige behandelingsovereenkomst – Law on the medical treatment agreement].
Professionalism	Ability to deal with fundamental life questions. Complying with professional organisations' current ethical and professional standards (good clinical practice).

5. Structure of the training course

The four-year training course comprises a number of modules, some compulsory and some optional. These are made up of specific traineeships (such as the laboratory traineeships and the scientific traineeship), or of set periods of time during which experience is gained in a particular sphere, intensive supervision takes place and the trainee participates in specific (multidisciplinary) discussions/surgery hours. The following table shows the link between subjects and modules.

Link between subjects and modules

Subjects	**Wish to have children**	**Own health**	**Syndrome diagnostics**	**Prenatal diagnostics**	**Family aspects**
Modules					
Cyto laboratory traineeship			x	x	
DNA laboratory traineeship			x	x	
Discussion training	x	x		x	x
Course “counting with genes”	x	x	x	x	x
Optional (scientific) traineeship	x	x	x	x	x
General genetics	x	x	x		x
Prenatal diagnostics	x		x	x	
Clinical cytogenetics			x	x	x
Oncogenetics	x	x			x
Cardiogenetics	x	x			x
Dysmorphology/MCA	x	x	x		
Mental retardation	x		x	x	x
Neurogenetics	x	x	x	x	x
Genodermatoses	x	x	x	x	x
Metabolic disorders	x		x	x	x
Sensory abnormalities	x	x	x	x	x
Skeletal/connective tissue abnormalities	x	x	x	x	x
Nephrogenetics	x	x	x	x	x
Preconception	x			x	

6. National and regional course instruction

The course instruction's content is based on the 7 CanMEDS skills areas and the 5 clinical genetics subjects, and supplements learning on the job. The instruction programme is flexible and aimed at the teaching objectives of the AIOS. Work is undertaken according to the self-learning principle, whereby the AIOS has more responsibility for the learning process. Interactive forms of instruction are the starting point, together with a clear link with patient care and research. Both the lecturers and the AIOS must apply the techniques of new learning.

7. Testing, portfolio and assessment

Testing takes place by means of short practice assessments, multi-source feedback, CAT (critically appraised topic) assessments and portfolio development (increased skills, positively assessed modules).
The portfolio is the central training document and contains all available information about the AIOS, who is himself responsible for it. The portfolio provides the trainer with information about the AIOS's activities (courses, conferences, lectures, publications and the like), experiences (self reflection) and personal development plans, in response to which the trainer can give development-oriented feedback. The portfolio is used as the basis for discussing progress.
Assessment takes place by means of structured discussions based on the portfolio, every 3 months during the first year and every 6 months thereafter. After the first year and half way through the training course a formal aptitude assessment takes place, based on multi-source feedback and the expansion of the portfolio.

8. Quality policy

With the introduction of GENiaal, a quality policy will also be developed with regard to the training course. This will be aimed at products of the training course, appreciation of the AIOS, appreciation of the training team and appreciation of the working field. Various aspects will be developed to supplement the periodic training inspection by the MSRC [Medisch Specialisten Registratie Commissie – Medical Specialists Registration Board], and will form part of an integrated quality cycle.

9. Final attainment levels

Please refer to the original GENiaal document for the final attainment levels.