

Genome puzzle

The Clinical Laboratory Geneticist Programme

2011

Genome puzzle

Netherlands Society for Clinical Genetic Laboratory Diagnostics

2011

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Preface

You hold in your hands the description of the Clinical Laboratory Geneticist Programme with the intriguing title "Genome puzzle". This description is the outcome of efforts exerted by the committee for "Restructuring the Clinical Laboratory Geneticist Programme". The committee was composed of representatives from all three disciplines within clinical genetic laboratory diagnostics. For clinical molecular genetic diagnostics: Dr. Ieke Ginjaar (LUMC, Leiden) and Dr. Hans Kristian Ploos van Amstel (UMCU, Utrecht); for clinical cytogenetics: Dr. Lia Knegt (AMC, Amsterdam) and Dr. Dominique Smeets (UMCN, Nijmegen); for trainee clinical laboratory geneticists: Dr. Rolf Pfundt (UMCN, Nijmegen); and for clinical biochemical genetics: Dr. Kees Schoonderwoerd (EUMCR, Rotterdam) and Dr. Ben Poorthuis (AMC, Amsterdam), also chair of the committee and representative of the Netherlands Society for Clinical Genetic Laboratory Diagnostics (VKGL) executive.

The laboratory specialism had three subspecialisations until now, namely clinical molecular geneticist, clinical cytogeneticist and the clinical biochemical geneticist. The rapid technological developments especially in molecular genetic diagnostics make the traditional distinction in the fields of interest of clinical molecular geneticist and clinical cytogeneticist less relevant. This fact has important consequences for the programme, which by definition must be oriented towards the future. Therefore, it was decided to integrate the separate programmes for clinical molecular geneticist and clinical cytogeneticist into a new Clinical Laboratory Geneticist Programme. The new programme is competence-oriented in imitation of the programmes for medical specialist and clinical chemical laboratory specialist.

The committee has expressly studied the possibility to integrate the programmes for all three subspecialisms into one programme. This is currently not possible for contextual and practical reasons. It is possible that future technical and social developments will be of such a nature to allow integration later. Therefore, the committee recommends considering this point again in five years' time. For now, a separate clinical biochemical geneticist programme will remain, which will strive to integrate as far as possible with the new clinical chemist programme with a specialisation in the diagnostics of hereditary metabolic diseases. The committee will at a later stage produce a separate proposal for the clinical biochemical geneticist programme.

This plan for the programme provides a firm foundation for a challenging and high-quality Clinical Laboratory Geneticist Programme. We assume that this will strengthen further the diagnostics of genetic diseases, both congenital and hereditary, in the near future.

The possibilities to detect and interpret genetic defects have grown tremendously: from microscope to array and from blot to 'personal genome'. There is still no end in sight: the genome puzzle is not complete and seems only to increase the number of its pieces. The new style of clinical laboratory geneticist has an important role to fill in solving the genome puzzle.

Amsterdam, February 2011,

Dr. Ben Poorthuis

Chair of committee for restructuring the Clinical Laboratory Geneticist Programme

Introduction

The clinical laboratory geneticist acts as clinical genetic laboratory expert, communicator, team player, manager, scientist, advocate and professional. These are the 7 general competences of the medical laboratory specialist. They demand from the clinical laboratory geneticist the necessary knowledge, skills and attitudinal aspects that s/he must acquire during the programme by carrying out practical clinical laboratory experience and by participating in formal educative activities. It is important for the clinical laboratory geneticist, like every medical laboratory specialist, to achieve a strong sense of autonomy and thus be able to make decisions when providing advice to professionals and when selecting diagnostic studies according to the clinical indication. S/he must have insight into the critical factors in the conduct and interpretation of the research.

This VKGL document specifies which changes have taken place within the Clinical Laboratory Geneticist Programme (formerly clinical cytogeneticist and clinical molecular geneticist). The most important changes are listed below:

- Explicit attention is paid to all essential competences of the clinical laboratory geneticist.
- The ALDIO (trainee laboratory diagnostics assistant) plays an active and central role in acquiring these competences.
- The intrinsic focus on the competences is translated not only into specific educational activities but also into the formal and informal examinations.
- Observing and recording observations about the functioning of the ALDIO in daily practice form an important source of information when giving explicit feedback.
- Giving explicit feedback regularly and in a structured format is essential for an optimal learning and working environment, which demands an active role from all members of the programme group.
- Progress in the programme can be made visible through a rise in the level of competence.
- The subject matter of the domain to be mastered can never be completely covered by the programme; this means that "life-long learning" will be a significant characteristic of the clinical laboratory geneticist.
- Both members of the programme team and the ALDIO should be trained in the new systematic of supervision, feedback and evaluation.

After a brief description of what competence-oriented training is and of its legal framework, in which the programme occurs, the revised programme is filled in step by step. First, the profile of genome diagnostics is described along with the position of this laboratory specialism within the health care sector. Then the 7 general competences, subdivided into several tasks, are described in general and in specific detail in relation to the clinical laboratory geneticist.

It was decided to describe the discipline in 5 themes. The description of operationalised competences and of the critical professional situations continues on from this. It also specifies which competency should be tested in which situation. Then the testing is considered in more depth. Next, there is an inventory of the programme methods, programme activities and programme materials. The quality policy and especially the professionalisation of the programme team members and the ALDIO are described in the following section. The portfolio is an essential component through the entire course of the programme and forms the basis for the planned progress reviews. This portfolio is described in a separate section. Finally, the design/structure of the programme in modules is explained along with the link between the themes and the modules.

Competence-oriented training / Legal framework

The general competences required of a clinical laboratory geneticist build on the general competences used in the medical specialist programmes and established as the 7 general competences formulated in the CanMEDS. In the 1990s these seven roles were formulated in Canada on the basis of wide-ranging research. They are the criteria that the medical specialist must fulfil and thus form the principal classification for the programmes' exit qualifications. These roles are now also standard in the Dutch medical specialists' programmes. The CZO has taken this on and uses the same seven roles for the arrangement of the exit qualifications, with a somewhat modified description of the hospital professional given below. The VKGL follows this system, bringing a new impulse to the evaluation of the ALDIO.

The clinical laboratory geneticist applies aspects of knowledge, skills and behaviour in an integrated manner in the exercise of his/her profession. The education and evaluation of the ALDIO should follow suit. This concept is translated in a structuring of all knowledge, skills and behavioural aspects according to certain "professional roles" the clinical laboratory geneticist normally fulfils. These professional roles are called competences. There are seven different areas of competence: laboratory technical methods, communication, knowledge & science, collaboration, organisation, social interaction and professionalism.

A further distinction is made in the different competences between general ones (transcending the specialism) and specialist ones.

Within this framework the competence-oriented training is mandatory. The ALDIO must be able to prove with a portfolio the level s/he has achieved for each competence. The Short Practice Evaluations (KPB; see below) should also be used to assess an ALDIO at least 10 times per year in practice.

After this report is approved by the members of the VKGL, this programme plan becomes legally valid. For the formal legal framework to which this programme belongs, refer to the programme regulations "Clinical Laboratory Geneticist Programme criteria".

Profile of the clinical genetic laboratory diagnostic system

Clinical genetic laboratory diagnostics is the laboratory specialism of clinical cytogenetics and clinical molecular genetics focussing on:

1. detecting aberrations in the genome, both postnatally and prenatally, whether hereditary, sporadic or somatic.
2. evaluating, interpreting, recording and storing laboratory orders and laboratory results according to international directives like ISO15189, ISCN and HGVS.
3. reporting the results and risk estimates to the referring party/medical specialist of individuals and their families and suggestions for further investigation that is important for clarifying the prognosis and/or possible treatment or therapy.
4. participating in the management of patients with hereditary conditions (potential), congenital aberrations and development disorders.
5. carrying out management tasks (e.g. progress discussions, performance reviews, financial management) and the quality assurance of the laboratory work.

Clinical genetic laboratory diagnostics is part of the specialist and hospital care. There are links to other medical specialisms. Clinical laboratory geneticists are associated with a clinical genetics/human genetics department of a university medical centre.

Clarification

The clinical laboratory geneticist determines which tests are to be conducted, based on the clinical symptoms as ascertained by the clinician. Test results are reported to the clinician, and they always include an interpretation of the results and even a recommendation for further diagnostic testing. The expertise of the clinical laboratory geneticist extends to the technical aspects of the laboratory testing, the interpretation and the reporting of the results to the clinician. S/he also knows the symptoms and has insight into the consequences for the treatment of hereditary conditions. Finally, the laboratory specialist has extensive knowledge of factors that can affect the results of the tests.

The programme criteria for the Clinical Laboratory Geneticist Programme were set by the VKGL registration committee. The laboratory specialism currently has three subdisciplines, namely clinical biochemical genetics, clinical cytogenetics, and clinical molecular genetics. The new Clinical Laboratory Geneticist Programme integrates the clinical cytogeneticist and clinical molecular geneticist programmes.

The laboratory specialist must be in close contact with the involved clinical specialists (clinical geneticists, paediatricians, neurologists, etc.) and have access to relevant patient data like clinical and laboratory results and ultimately medication. An adequate interpretation of the genetic tests is only possible when the laboratory specialist has this information. The clinical laboratory geneticist participates in a network of professional experts. Via that network national and international experts can be consulted. There are multiple collaboration links between the clinical genetic laboratories and other laboratories both inside the UMCs (pathology, immunology, clinical chemistry) and outside them (FMLS).

At the European level (ESHG, Eurogentest) a start has been made to describe the required core competences for professionals in genetics (see

http://www.eurogentest.org/laboratories/documents/info/public/unit6/core_competences.xhtml).

Competences of the clinical laboratory geneticist

The general competences that a clinical laboratory geneticist must have build on the general competences used in the medical specialist programmes and established as the 7 general competences formulated in the CanMEDS. In table 1 these general competences are subdivided into partial competences. In most cases the clinical laboratory geneticist does not have a medical degree and no direct contact with patients. Although the competences for medical specialists are used as the basis, the general competences for a clinical laboratory geneticist differ in a number of essential points. The competences for a clinical laboratory geneticist have been elaborated in the sections on knowledge, skills and attitudinal aspects.

Table 1. General competences and partial competences of the clinical laboratory geneticist

Competences	Partial competences in clinical genetic laboratory diagnostics
Clinical genetic laboratory technical methods (LTH)	<ul style="list-style-type: none">- has adequate knowledge and skills reflecting the status of the specialism- applies the diagnostic process of the specialism well- provides effective and ethically responsible patient care- finds the required information quickly and applies it properly
Communication (C)	<ul style="list-style-type: none">- establishes effective relationships with clients and colleagues- listens well and obtains relevant patient information- discusses medical information properly with colleagues and clients- prepares adequate written and verbal reports about the patient's case
Collaboration (S)	<ul style="list-style-type: none">- discusses efficiently with colleagues and other caregivers- asks for advice as necessary- provides effective peer advice- contributes to an effective interdisciplinary collaboration and integrated care
Knowledge & Science (K & W)	<ul style="list-style-type: none">- examines medical information critically- promotes the broadening and development of scientific specialist knowledge- develops and maintains a personal in-service and refresher training plan- promotes the clinical genetic laboratory diagnostics expertise of ALDIOs, students, colleagues and others involved in health care
Social skills (M)	<ul style="list-style-type: none">- knows and recognises the determinants of diseases- promotes the patient's health care and the welfare of the community as a whole- acts in accordance with the relevant legal stipulations- responds adequately to mistakes in care
Organisation (O)	<ul style="list-style-type: none">- organises the work to effectively balance patient care and personal development- works effectively and efficiently in a health care organisation- responsibly commits the available means to patient care- uses information technology for optimal health care and for in-service and refresher training
Professionalism (P)	<ul style="list-style-type: none">- provides high-quality patient care in an upright, sincere and committed manner- behaves in an adequately personal and inter-personal professional manner- knows the limits of his/her own competence and confines his/her actions within them- conducts laboratory diagnostics in line with the standard ethical norms of the profession.

Themes in genome diagnostics

Here the specialism of genome diagnostics is described in themes with the aim to summarise it in typical professional situations. This is an essential step in the entire process, because the chosen description is a guiding principle for practically everything that comes after it. Classification into 5 themes which are characteristic for the specialism was ultimately selected, rather than one according to syndromes and/or organ systems, in which the specific clinical genetic features are less obviously recognisable.

Themes

Processes

decision points in the preanalytical and analytical phases of the prenatal and postnatal genome diagnostics

Analysis

practical experience and background knowledge of the analysis techniques and methods in genome diagnostics

Interpretation

structural aberrations/changes in the genome and the relation to gene product and the phenotype

Technology

implications & implementation

Quality care & Management

validation & guarantee

Operationalised competences and critical professional situations for each theme

The Critical Professional Situations (KBS) are a collection of case studies and work situations that are exemplary for the everyday practice in genome diagnostics. They occur frequently, can be generalised and are easily testable using the Short Practice Evaluation (KPB). They are classified according to the themes in genome diagnostics, and in the chosen KBS both general genetics and the different fields of interest are addressed: this mostly covers the exit qualifications for genome diagnostics.

To keep the testability of each KBS manageable, it was decided to score several competences in each KPB if that suited the practical case. The above overview reveals that the emphasis lies numerically speaking with the competences laboratory technical methods, communication and professionalism and knowledge & science. Both the chosen practice situations/case histories and the choice of competences to be tested in each KBS are arbitrary, and offer space for accents.

As about 40 KPBs are scheduled during the programme, it may be that not all KBSs can be tested. It therefore seems reasonable to agree at the start of the programme which part of the KBS must be tested at the end of the programme and record this choice in the portfolio.

Critical professional situations theme 1: Processes

Decision points in the pre-analytical and analytical phases of the prenatal and postnatal genome diagnostics

<i>Processes: decision points in the pre-analytical and analytical phases of the prenatal and postnatal genome diagnostics</i>		<i>LTH</i>	<i>C</i>	<i>S</i>	<i>K&W</i>	<i>M</i>	<i>O</i>	<i>P</i>
1	Assessing orders in terms of missing data and contacting the client	X	X	X	X	X		
2	Assessing patient material for suitability for testing	X	X	X		X		
3	Choosing genome tests according to request	X	X	X	X			X
4	Choosing suitability of technology for detecting changes (point mutations, deletions/insertions/duplications, repeats, submicroscopic and chromosomal aberrations)	X	X	X	X		X	X
5	Assessing laboratory results in terms of quality parameters	X	X	X	X			
6	Assessing and applying line verifications	X	X		X			
7	Recognising purity of the patient material	X	X	X		X		
8	Prenatal diagnostics: taking maternal contamination into account	X	X	X				
9	Assessing unexpected results in relation to the request	X	X	X	X			X
10	Assessing suitability of request in relation to the phenotype		X	X	X	X		X
11	Decision about further testing (confirmation of finding, family testing, functional assessment)		X	X	X			X
12	Taking sensitivity of test into account	X	X		X			
13	Coping with switching of patient material in the laboratory	X	X	X		X	X	X
14	Coping with incorrect results	X	X	X		X	X	X
15	Coping with equipment failure		X	X			X	X
16	Coping with overrunning testing result deadline		X	X				
17	Data mining/bioinformatics referring to reported aberrations	X	X	X	X			X
18	Presymptomatic/carriership testing without index patient of the family	X	X	X				
19	Storage life, storage and archiving of material	X	X	X				

20 Archiving the registered data

X X X

21 Organising urgent requests

X X X

X X

Critical professional situations theme 2: Analysis

Practical experience and background knowledge of the analysis techniques and methods to be applied in genome diagnostics

		<i>LTH</i>	<i>C</i>	<i>S</i>	<i>K&W</i>	<i>M</i>	<i>O</i>	<i>P</i>
1	Cell culture (lymphocytes, fibroblasts, bone marrow, amniotic fluid and chorionic villi)	X						
2	DNA isolation [blood, tissue (fresh and fixed)]	X			X			
3	Karyotyping and specific staining techniques	X						
4	FISH (selecting probe, interphase, metaphase)	X			X			
5	PCR (selecting primer, conditions)	X			X			
6	Number of copy variations (PCR-based QF-PCR, MLPA)	X			X			
7	Gel electrophoresis (Agarose, Bioanalyzer)	X						
8	Sanger sequence analyse	X			X			
9	Massive parallel sequencing (next generation sequencing) (e.g. 454, Solid, Illumina)	X			X			
10	Array technology (CGH, SNP-based arrays)	X			X			
11	Southern Blotting technique	X						
12	Scanning analysis (HRMCA, SSCP, PTT, etc.)	X			X			
13	Fragment length analyses	X			X			
14	RNA examination (Northern blotting, RT-PCR)	X			X			
15	Technology (current/latest, situational)	X			X			

Critical professional situations theme 3: Interpretation

Structural aberrations in the genome and the relationship to the phenotype

Interpretation: structural aberrations in the genome and the relationship to the phenotype		LTH	C	S	K & W	M	O	P
1	Identify the structural genome variations/chromosome aberrations according to international guidelines (ISCN, HGVS)	X			X			X
2	Evaluation of the quality of the results and tests used in the final conclusion	X	X		X			X
3	Determining recurrence risks and genetic risk factors	X			X			X
4	Being aware of the possibility of non-paternity	X	X		X			X
5	Being aware of the possibility of mosaicism (somatic, gonadal)	X		X				X
6	Literature search and application of bioinformatics to assist interpretation of the pathogenic nature of the genome aberration (chromosomal, submicroscopic, extragenic, intragenic), taking into account copy number variations (CNVs), single nucleotide polymorphisms (SNPs), short tandem repeats (STRs), chromosome polymorphisms and fragile sites and unclassified variants (UVs)	X			X			X
7	Describing results in relation to the phenotype, treatment and prognosis		X	X	X			X
8	Determining clinical significance of the findings, possibly in relation to the course of treatment	X	X	X	X			X
9	Communication of negative findings for suspicion of chromosomal aberration, autosomal recessive, dominant and X-linked diseases, partner testing and genetic heterogeneous diseases in relation to test sensitivity and sensitivity		X	X	X			X
10	Probability calculation & risk estimation for familial aspects in autosomal recessive, dominant and X-linked diseases, genetic heterogeneous diseases, sporadic genetic diseases patients		X					X
11	Implication of influence of ethnicity on relevance of the aberration found				X			X
12	Reporting in clear and understandable language in relation to the request according to best practice guidelines for professional groups	X	X	X		X		X
13	Suggest options for prenatal diagnostics	X	X	X		X		X

Critical professional situations theme 4: Technology Evaluation & Implementation

<i>New technology: Evaluation & Implementation</i>		<i>LTH</i>	<i>C</i>	<i>S</i>	<i>K &W</i>	<i>M</i>	<i>O</i>	<i>P</i>
1	Draw attention to potential new methods, techniques, developments in bioinformatics, ICT possibilities and equipment	X	X	X	X			X
2	Imagining diagnostic possibilities for new methods, techniques, developments in bioinformatics, ICT possibilities and equipment	X	X	X	X	X		X
3	Estimating efficacy, quality, cost of new methods, techniques, developments in bioinformatics, ICT possibilities and equipment	X			X	X	X	X
4	Preparing implementation criteria for new methods, techniques, developments in bioinformatics, ICT possibilities and equipment	X	X		X		X	X
5	Validating new methods, techniques, developments in bioinformatics, ICT possibilities and equipment	X		X	X		X	X
6	Ensuring there is quality documentation for new methods, techniques, developments in bioinformatics, ICT possibilities and equipment	X	X	X	X		X	X

Critical professional situations theme 5: Quality care
Validation & Guarantee

Quality care: Validation & Guarantee		LTH	C	S	K & W	M	O	P
1	Validating new methods, techniques and equipment	X	X	X	X			
2	Knowledge and application of occupational health and safety and environmental legislation	X			X			
3	Primary care, specialist and intramural verifications	X	X		X		X	X
4	Holding quality audits	X	X	X				
5	Writing/updating quality documents (procedures, SOPs, handbook)	X	X		X			
6	Reporting and processing internal and external error statements and suggestions for improvement	X	X	X	X	X	X	X
7	Creating insight into efficacy of the testing (specificity and sensitivity)	X	X	X	X		X	X

Training methods

The Clinical Laboratory Geneticist programme plan is aimed at competence-oriented training. Competence-oriented training prepares the ALDIO for life-long learning. The diagnostics form the backbone of the competence-oriented programme plan.

Competence-oriented training should take place in a stimulating and safe learning and work environment. The ALDIO is expected to show considerable initiative and take responsibility for his/her learning process. The institute where the ALDIO is working should create possibilities to realise the set study objectives. It should be an experience-based learning process, in which the competence development continues to build on earlier experiences, leading to increasing autonomy and proactive action. The ALDIO must learn from his/her mistakes. This is incorporated in the personal development plan (POP). The proactive action is carried out in close consultation with the instructor and the entire programme group, according to the information from the portfolio. The instructor and the programme group take on the function more of a role model and coach. It is preferable if every ALDIO is assigned a personal mentor to discuss any problems (personal or programme-related) arising. There should be space for flexible individual adjustments of the POP on the basis of the portfolio. Options to continue studying should be available (self-study, courses/national training).

Programme activities

Courses, training and secondary activities

1. Courses:

Course (national or international) in genetic diagnostics (mandatory)

2. Education through individual courses:

Attending local courses (mandatory)

Attending national courses (mandatory)

National council/refresher courses:

3.

Attending the LOC/LOD (mandatory)

Presenting a case at the LOC/LOD (mandatory)

Presentation at scientific meeting of VKGN/LOG

Attending LOG/LOC/LOD (mandatory)

Attending the NVHG scientific meetings

Science:

4.

Scientific writing course

Writing article/presentation for scientific congress / symposium (mandatory)

Course on database searches

Other relevant courses

5. Management:

Management course (mandatory)

Other relevant courses (personal effectiveness and attitudinal aspects)

Active participation in local/national/international committees and societies

Assisting with management tasks in the department

Educational development:

6.

Contributing to supervision internships and workgroups (mandatory)

Contributing to educational activities / knowledge transfer to analysts, students, colleagues and other people involved in health care

7. Quality:

Course quality & auditor (mandatory)

Actively contribute to and further optimise the quality assurance system

Other

8.

Genetic risk calculation (mandatory)

Dealing with complaints

Coping with personal problems

Ethical aspects and legislation regarding clinical genetic laboratory diagnostics

NB. Exemption is possible for parts of the programme if the request is well supported. This is to be assessed by the registration committee, VKGL

Educational materials

During his/her programme, the genome diagnostics ALDIO should acquire sufficient knowledge to fulfil the exit qualifications applicable to the Clinical Laboratory Geneticist Programme. Theoretical knowledge can be obtained from the specialist literature (textbooks, reference works, Internet, as well as recent publications), guidelines, protocols and course folders. Also, in clinical genetic laboratory diagnostics, databases (OMIM, HMGD, NCHGR, etc.) are used extensively.

Design & Structure of the programme

The Clinical Laboratory Geneticist Programme takes 4 years, consists of two phases and is constructed of modules. Phase 1 involves two programme years strongly focussed on technology, and phase 2 a specialisation lasting two years, in which the knowledge and skills from the first phase are applied to themes in practice. During the first years, courses in cytogenetics and molecular genetics are taken that cover the entire current spectrum of technological possibilities. It is also possible to take a module in a laboratory of another programme institute.

Concerning the mandatory courses, in this phase a general (possibly international) course in genetic diagnostics is taken along with courses/training like quality, management and genetic risk calculation. During the 2-year specialisation phase, in years 3 and 4, the programme is divided into different modules of 3 and 6 months, each with a particular theme. The themes are groups of hereditary conditions like neurogenetics, oncogenetics, cardiogenetics. There is some freedom of choice, because the foci can be strongly determined by the local priorities. The two mandatory modules for everyone are mental retardation & development disorders and oncogenetics. These two are important areas of interest in the medical genetics centres and contain all aspects (such as technology, complexity of aberrations and interpretation, treatable/not treatable) of clinical genetic laboratory diagnostics.

The modules are defined in terms of functional/technical, can therefore vary from institute to institute in terms of content and extent, but in all centres are tailored to the programme and the exit qualifications. They conclude with an evaluation (portfolio). The local instructor can thus influence the local availability of the elective modules and their scope in excess of the minimum content. In a separate table the link between themes and modules is given.

<i>Clinical genetic laboratory diagnostics</i>		<i>LTH</i>	<i>C</i>	<i>K&W</i>	<i>S</i>	<i>M</i>	<i>O</i>	<i>P</i>
Years 1 and 2								
1 month	General introduction and orientation to laboratory diagnostics genome analysis including Laboratory practical in molecular genetics Laboratory practical in biochemical genetics Laboratory practical in molecular cytogenetics	X		X	X		X	X
2 months	Laboratory & Technology Modules Patient material: sampling, forwarding, receipt, evaluation of order and patient material, storage and preservation of patient material (postnatal and prenatal). Sample preparation, isolation, concentration, purifying and storage (DNA, RNA). Culturing various cell types (lymphocytes, fibroblasts). Archiving the registered data	X		X	X		X	X
2 months	Karyotyping and specific staining techniques. Chromosome staining methods including conventional staining, G-banding, QFQ-banding, (C-banding), NOR staining, Distamycin-DAPI staining. The relationship between chromosome structure and banding. FISH (Fluorescence in situ Hybridisation). Making human chromosome preparations, including synchronisation, the role of mitosis inhibitors, hypotonic treatment and fixatives	X		X	X		X	X
2 months	Array technology (CGH, SNP-based arrays) SNP- and CGH-array for the detection of segmental aneuploidies. Bio-informatics: use of software (analysis), databanks for finding and denoting genome variants	X		X	X		X	X
2 months	Copy-number of variations (PCR-based QF-PCR, MLPA) Detection of deletions and duplications (MLPA, QF-PCR)	X		X	X		X	X
2 months	Sanger sequencing, PCR (incl RT-PCR, LR-PCR and other applications)	X		X	X		X	X
2 months	Next generation sequencing/massive parallel sequencing	X		X	X		X	X

2 months	Special stipulations: methylation-sensitive testing, RNA study, fragment analysis, mutation screening (like DGGE, HR-MCA, PTT, TaqMan for example), Northern blot analysis, Southern blot analysis	X	X	X	X	X
2 months (not consecutive)	Courses: Quality course, probability calculation in terms of recurrence risk, management course, training, internships, course abroad	X	X	X	X	X
2 months	Prenatal diagnostics	X	X	X	X	X
Years 3 and 4						
6 months	Mental retardation & Development disorders	X	X	X	X	X
6 months	Oncogenetics/Malignancies	X	X	X	X	X
3 months	Elective block 1 (see table p.22)	X	X	X	X	X
3 months	Elective block 2 (see table p.22)	X	X	X	X	X
3 months	Elective block 3 (see table p.22)	X	X	X	X	X
3 months	Elective block 4 (see table p.22)	X	X	X	X	X

Laboratory & Technology Phase 1	Processes	Analysis	Interpretation	Technology	Quality care
Sampling, forwarding, receipt, evaluation, storage and preservation of patient material (postnatal and prenatal)	X	X	X	X	X
Preparation, storage and working with reagents	X	X	X	X	X
Bio-informatics: use of software (for analysis), databanks for finding and denoting genome variants	X	X	X	X	X
Chromosome staining methods including conventional staining, G-banding, QFQ-banding, (C-banding), NOR staining, Distamycin-DAPI staining.	X	X	X	X	X
The relationship between chromosome structure and banding	X	X	X	X	X
Detection of deletions and duplications (MLPA, Q-PCR)	X	X	X	X	X
DNA-fingerprinting	X	X	X	X	X
Electrophoresis and related techniques	X	X	X	X	X
FISH (Fluorescence <i>in situ</i> Hybridisation)	X	X	X	X	X
Fragment analysis	X	X	X	X	X
Making human chromosome preparations, including synchronisation, the role of mitosis inhibitors, hypotonic treatment and fixatives	X	X	X	X	X
Probability calculation for recurrence risk	X	X	X	X	X
Culturing and storage of different cell types (e.g. lymphocytes, fibroblasts)	X	X	X	X	X
Laboratory equipment (use and maintenance)	X	X	X	X	X
Methylation-sensitive tests	X	X	X	X	X
Sample preparation, isolation, concentration, purification and storage (DNA, RNA)	X	X	X	X	X
Mutation screening (like DGGE, HRMCA, PTT, TaqMan)	X	X	X	X	X
Next generation sequencing/massive parallel sequencing	X	X	X	X	X
Northern analysis	X	X	X	X	X
PCR (incl RT-PCR, LR-PCR and other applications)	X	X	X	X	X
Working with radioactivity	X	X	X	X	X
Sanger sequencing	X	X	X	X	X
SNP- and CGH-array for detecting segmental aneuploidies	X	X	X	X	X
Southern blot analysis	X	X	X	X	X
Sterile techniques	✓	✓	✓	✓	✓

Working with GMOs (genetically modified organism)

X

X

X

X

X

<i>Hereditary diseases (Examples) Phase 2</i>	<i>Processes</i>	<i>Analysis</i>	<i>Interpretation</i>	<i>Technology</i>	<i>Quality care</i>
Congenital aberrations and development disorders	X	X	X	X	X
Cardiovascular diseases	X	X	X	X	X
Deafness	X	X	X	X	X
Echoscopic aberrations of the fetus	X	X	X	X	X
Hereditary metabolic diseases	X	X	X	X	X
Haemoglobin and the haemoglobinopathies	X	X	X	X	X
Recurrent abortions/miscarriages	X	X	X	X	X
Skin diseases	X	X	X	X	X
Immune disorders	X	X	X	X	X
Cancer/Malignancy	X	X	X	X	X
Mental retardation	X	X	X	X	X
Possible genetic factors in commonly occurring diseases	X	X	X	X	X
Neurological conditions	X	X	X	X	X
Neuromuscular diseases	X	X	X	X	X
Renal diseases	X	X	X	X	X
Eye diseases	X	X	X	X	X
Orphan diseases (rare, difficult to place diseases)	X	X	X	X	X
Skeletal aberrations	X	X	X	X	X
Reproduction and fertility problems	X	X	X	X	X

Portfolio

The portfolio is a modified logbook, charting the individual planning and implementation of the ALDIO's programme. It is a collection of all available information about the ALDIO. S/he is responsible for keeping the portfolio updated. In practice the ALDIO is assessed at different timepoints in different situations in different ways by different people. The portfolio provides the instructor with information about the activities (courses, congresses, presentations, etc.), experiences (self-reflection) and the personal development plan (POP) of the ALDIO on which to base development-oriented feedback. The portfolio is used as a supporting document in the progress review. It along with other forms (see progress review) should be submitted a week in advance to allow the instructor enough time to examine all this information and request additional information.

The portfolio should contain the following components:

1. *Curriculum vitae*

Surname and initials:

First name:

Personal ID number (BSN):

Address:

Postal code and town/city:

E-mail:

Date of birth:

Preparatory programme:

Instructor(s):

Programme register number:

2. *Programme schedule+ evaluation documents*

Starting date of programme:

Termination date of programme:

Instructor(s):

Programme institution(s):

Internship(s):

Evaluation documents:

- short practice evaluations
- 360° evaluations and evaluations of other activities
- Agenda progress review: the ALDIO reports the points that must be discussed and the personal

development plan (competence development (see 3), what are the most important foci and objectives for the coming period and the reflection on the functioning and whether the objectives/foci of the previous period had been realised).

- 3 monthly/6 monthly/annual progress reviews
- The approved record of the progress review.
- Evaluation forms from registration committee.

3. Self-reflection competence development and the general theme

The ALDIO describes his/her development for the 7 competences supplemented with explicit examples and self-reflection on their own functioning.

4. Course-based education followed

The ALDIO records the national course-based education days and the locally organised course-based education with date carried out and topic.

5. Presentation and reports done and education given

The ALDIO records the presentations and reports done and education given with date carried out, location, organisation, setting and topic. Also, any evaluation of this is archived.

6. Publications

The ALDIO records any published articles with authors, title, journal and date of publication.

7. Scientific research

The ALDIO describes the scientific research s/he has carried out

8. Conferences, symposia, scientific meetings and courses attended

The ALDIO records the conferences, symposia, scientific meetings and courses attended with date, name, location and organisation and can produce proof (certificate/accreditation) for the mandatory programme components.

9. Activities carried out

The ALDIO records the number of laboratory activities/reporting done for each indication category and the

molecular-genetic or cytogenetic techniques applied. At the end of the programme, it must be apparent from this list that the ALDIO has carried out at least 1000 reports independently and under supervision, of sufficient quality and diversity. The reports should form a proportional representation of the different types of testing, in terms of indication, samples received and technique (postnatal, prenatal, tumour; sequencing, (QF)-PCR, MLPA, array, karyotyping, FISH, etc.). An evaluation of the results is also maintained.

10. Internship(s)

The ALDIO records the internship followed with name, period, supervisor and organisation and can produce proof in the form of a trial period form, checklist and/or a report.

11. Other activities during the programme

The ALDIO records any other activities during the programme and clarifies the contents. The ALDIO assists with preparing and writing protocols.

12. The instructor's declaration about the completeness and correctness of the portfolio

Progress review

In the progress review, on the basis of the collected material and the general and theme charts completed by the ALDIO and instructor, the objectives set in advance are examined for the internship/module followed. Also, attention is paid to the POP. This review provides information that can be incorporated in a new POP. The cycle starts again: POP and objectives for the new internship/module are compared, and new individual objectives set for the ALDIO. At each progress review the ALDIO is responsible for providing an updated portfolio.

The following topics are treated during the progress review:

- The ALDIO's experience with the learning environment in the department

- Self-reflection on competence development: the portfolio gives the ALDIO insight into his/her own development with regard to the themes and associated competences. The ALDIO runs through the 7 competences and briefly describes which proof can be furnished for development in these competences. Also, the quality of the self-reflection of the ALDIO and any discrepancies between the instructor's (or group's) evaluation and the ALDIO's one are discussed.

- Personal development plan (POP): the portfolio gives the ALDIO the opportunity with each progress review to examine his/her performance in the last period of 3, 6 or 12 months. Use is made of the testing timepoints and proof (courses followed, education/presentation given, etc.) assembled in the previous period. The ALDIO formulates on the basis of this a plan for the coming 3, 6 or 12 months: what are the most important foci and objectives.

- Notes: the ALDIO and instructor make notes of the progress reviews and record what should be discussed the next time and add them to the portfolio after approval (initials) by both parties.

- Strengths and weaknesses of the programme: evaluations based on the portfolio can highlight the weaknesses of the programme (instructor, instructor group and programme institute). If weaknesses are found, they are discussed during the mandatory three-monthly programme meetings, and a plan of action should be realised. Such weaknesses should also be indicated on the evaluation form of the programme that is completed each year by the ALDIO.

- Weaker ALDIOs are more likely to be spotted: the portfolio is primarily a means to stimulate the development and proactive action in the learning process of the ALDIO. Alternatively, it can point out the ALDIO's weakness. If insufficient growth is found during the progress review, an individual programme can be arranged. If despite the individual programme, insufficient growth/functioning persists, according to the prescribed rules, this evaluation can be used to terminate the programme (see addendum 1. summative evaluation).

- Declaration of competence: the growth and development in the themes and the associated competences, commitment and personal insight of the ALDIO are evaluated during the progress review of the preceding 3, 6 or 12 months. This evaluation is documented on the appropriate evaluation form by the Registration committee.

- An insufficient evaluation should be included in the dossier that the instructor receives from the ALDIO.

SHORT PRACTICE EVALUATION

The Short Practice Evaluation (KPB) concentrates on the ALDIO's competences. It is relatively easy to apply by staff members as part of the everyday routine and is suitable for the evaluation of the ALDIO in various programme years. The KPB is a short observation (10 minutes) of an action or an operation. The combinations of several KPBs provide insight into the ALDIO's activities.

The KPB can be constantly applied in the programme laboratory, during progress discussions, patient discussions, national or local schooling or case conferences. The staff members of the programme laboratory and internship placings can conduct assessments. The KPB is applied in a structured way by using a standard form. From each KPB a copy is given to the ALDIO and the original to the instructor. At least 10 KPBs should be conducted each year.

360° feedback

With this multi-source feedback, different parties from different perspectives make a valuable contribution to the evaluation. The concerned individuals in the ALDIO's surroundings provide the content, like genome diagnosticians, clinical geneticists, fellow ALDIOS, chief analysts, clients. Involving a person to evaluate only makes sense if there is sufficient opportunity to observe the ALDIO and enough aspects of his/her activities can be assessed.

The instructor and the ALDIO discuss the results of the 360° feedback and how the ALDIO deals with this feedback. This involves contrasting self-evaluation and the evaluation of others. If the ALDIO evidences signs of over- or underestimating him/herself, the instructor should assist them to a more correct self-image. A 360° feedback should be held at least twice during the programme.

Testing via internet and/or national/international broadcasts (already conducted UKNEQAS or EUROAGENTEST testing)

Each year there is a test conducted via internet which is mandatory for all ALDIOS in the 2nd, 3rd, or 4th year of their programme. A number of relevant case histories is presented, and the assignment is to analyse them in a complete and adequate report, using all the means available. The instructor has an important role as supervisor of the process.

This internet testing is prepared and evaluated by the programme committee with input from all of the centres.

Testing

Examination methods:

- Short Practice Evaluation (KPB) 10x/year
- 360° feedback 2x/programme
- Digital testing (internet) 3x/programme
- Portfolio throughout programme
- Presentations throughout programme
- Publications/protocols throughout programme
- Profession-oriented national activities (workgroup, management) throughout programme

Examination methods

LTH C K & W S O M P

360° feedback	X	X	X	X	X	X	X
Short Practice Evaluation	X	X		X			X
Portfolio	X	X	X	X	X	X	X
Chair / member of workgroup, administrative activities VKGL, LOD, LOC, etc.	X	X	X	X	X		
Presentations	X	X	X				X
Other activities, e.g. writing an article, protocol, active in a parents'/patients' association, etc.	X		X	X	X	X	X

Time of evaluation (see above also):

Evaluation interviews:

1st year of programme 4x/year

2nd - 4th year 2x/year

Two summative evaluations: end of 1st year and halfway through the programme. They are linked to a 360° evaluation.

What is assessed?

In principle, testing is done at the workplace. In addition, depending on how the course-based education develops, a form of testing is also done during the national meetings of the course-based programme.

Local employment/use of:

- Programme committee
- Mentor

Quality assurance for the programme

With the introduction of the revised programme plan for clinical genetic laboratory diagnostics, the VKGL has set objectives that must be achieved within a certain deadline. The Clinical Laboratory Geneticist Programme is not complete with the implementation of this new programme plan. Continuous adjustments to the programme will always be needed, along with improvements based on changing circumstances, and a flexible response is needed to questions from outside and within the specialism. This demands quality assurance of the curriculum in the form of a systematic evaluation of the programme. On the basis of the evaluation results, further actions can be undertaken and/or objectives adjusted.

Testing of the quality of the programme must involve objectifiable indicators like: the ALDIO's performance level, the realisation of the particular educational concept, information about the learning environment, the assessment by the ALDIO, the assessment by the instructors, the assessment by the other professional societies and by other health care workers.

It is important for the success of the proposed innovation that the key factors are unambiguous. The following points belong to the key factors of the clinical genetic laboratory diagnostics programme.

- Obtaining broad support for the new programme (its elaboration)
- ALDIO takes responsibility for his/her own learning process
- Being able to reflect on one's own learning process (ALDIO)
- Use of the portfolio by ALDIO and instructors
- Training of instructors' group and ALDIO
- Direct coaching of ALDIO with associated feedback by the instructor/supervisor
- Use and functionality of short practice evaluation and 360° feedback as testing methods
- Modular structure of the programme
- Course-based education and testing

Good information can be obtained about these key factors and/or whether they are successful through interviews. The evaluations of the ALDIO and staff members provide helpful sources. The most important instruments for this are: external reviews by programme institutes, external reviews by the trainee clinical laboratory geneticist, continuous evaluation of the programme process through annual evaluations and professionalisation of the instructors.

The outcomes can be used to:

- Provide those responsible for the programme with the necessary data
- Provide review committees with the necessary data
- Adjust objectives and procedures and formulate new objectives within a set deadline

Data of the quality cycle can serve as an indicator of the success of the implementation.

To be able to implement the entire programme plan properly, a large number of steps is essential. One of the important steps is training the instructors, members of the programme teams and the ALDIO. The content of this training must focus on aspects that are significant for instructors, members of the programme team and the ALDIO. Topics include:

- What is known in the literature about education in the workplace and testing (state of the art)?
- Giving feedback
- Being able to observe
- Conducting a progress review
- Preparing a suitability evaluation
- Putting together a portfolio
- Discussing the portfolio
- Quality care at the workplace
- Arranging the education at the workplace

Each programme institution should prepare a plan for the professionalisation of the instructors, members of the programme team and the ALDIO. Exchanging information among each other is desirable and necessary. A number of training activities could be organised in combination with other specialisms e.g. by the programme committee. Further elaboration of the skills enhancement/professionalisation will take place during the implementation process and will depend on the courses on offer in the country.

Exit qualifications

Clinical Genetic Laboratory Diagnostics

Lemmas (L)

L.1 Knowledge

- L.1.1.Genetics
- L.1.2.Laboratory procedure
- L.1.3.Quality assurance
- L.1.4.Ethics

L.2. Skills

- L.2.1.Research
- L.2.2.Practice
- L.2.3.Management
- L.2.4.Academic skills
- L.2.5.Education
- L.2.6.Organisation

L.3. Attitudinal aspects

- L.3.1.Professionality
- L.3.2.Communication
- L.3.3.Education
- L.3.4.Support
- L.3.5.Research
- L.3.5.Academic profile
- L.3.6.Direction & Management

L.1 Knowledge

L.1.1 Genetics

The basis principles of human & medical genetics

The assistant has insight into and knowledge of the basic principles of inheritance

- The molecular basis of inheritance
- Chromosome structure
- Cell division (DNA replication, mitosis and meiosis)
- Gene structure and gene expression (DNA, gene, genetic code, transcription, translation and regulation)
- Genomics
- Evolution (molecular evolution, natural selection)
- The history of genetics in medicine

The assistant has insight into and knowledge of the basic principles of hereditary diseases

- The nature and frequency of genetic diseases (chromosomal, monogenetic, multifactorial)
- Mendelian inheritance (autosomal, sex-linked, dominant, recessive)
- Mutations and diseases: nature and repercussion (genetic variation, neutral, disease causing, de novo, somatic, penetrance, anticipation)
- Polygenic and multifactorial diseases
- Non-classical inheritance (pseudo-autosomal, multifactorial, mitochondrial, imprinting, uniparental disomy, mosaicism)
- Identification and characterisation of disease genes and structural genome variations/chromosome aberrations
- Genetic pathogenesis (genotype-phenotype relationship)
- Population genetics (Hardy-Weinberg, consanguinity, genetic drift, inbreeding, selection, founder, fitness)
- Genetic epidemiology (genetic contribution to complex diseases)
- Epigenetics
- Linkage studies
- Probability calculations (family-tree analysis, risk determination, *Bayes' theorem*, penetrance, age of onset, linkage, mosaicism)
- Genetics of complex diseases (IBD, QTL, LD, association, sib-pair analysis)

Genetics in medicine

The assistant has insight into and knowledge of genome variations in specific areas of interest in medicine such as

- Biochemical genetics
- Cardiovascular diseases
- Chromosomal diseases
- Deafness
- Echoscopic aberrations of the fetus
- Hereditary metabolic diseases
- Pharmacogenetics
- Genetics of congenital aberrations and development disorders
- Genetic factors in commonly occurring diseases
- Gynaecology-Obstetrics
- Haemoglobin and the haemoglobinopathies
- Recurrent abortions/miscarriages
- Skin diseases
- Immunology
- Neurology & Neuropsychiatry
- Neuromuscular diseases
- Kidney diseases
- Oncology
- Eye diseases
- *Orphan diseases*
- Skeletal aberrations
- Reproduction and infertility

Genome diagnostics, clinical genetics and genetic counselling

The assistant has insight in and knowledge of the following when applying genetics for the diagnosis, treatment and advising of patients and family members with a genetic disease in the family,

- Cytogenetic and molecular genetic nomenclature
- Inheritance and incidence/prevalence of monogenetic and multifactorial diseases
- Origin, incidence and inheritance of chromosomal aberrations
- Chromosome breakage syndromes
- Carriership determination and presymptomatic testing
- Prenatal diagnostics of genetic diseases (indications, possibilities and limitations)
- Preimplantation diagnostics
- Risk calculations
- Population screening and community genetics
- The human genome project, treatment of genetic diseases and gene therapy
- Ethical issues in medical genetics (informed consent, privacy, METC)
- Inheritance advice/counselling

L.1.2 Laboratory procedures

General clinical genome diagnostics

The assistant has knowledge of and is able to describe and explain

- The principles of the techniques below including the background, functions of the most important reagents, differences between the various methods and platforms.
- Which types of conditions are suited to analysis by laboratory molecular and/or cytogenetic techniques and can describe the limitations and added value of each technique.
- Relevant image processing and analysis software
- The relationship of the test result to the question and is thus able to establish and describe links between the genotype and the phenotype

Techniques

- Preparation, storage and working with reagents
- Sample preparation, isolation, concentration, purification and storage (DNA, RNA)
- Sampling, sending, receipt, evaluation, storage and preservation of patient material (postnatal and prenatal)
- Laboratory equipment (use and maintenance)
- Sterile technique
- Culturing different cell types (lymphocytes, fibroblasts, etc.)
- Working with radioactivity
- Working with GMOs (genetically modified organisms)
- Electrophoresis and related techniques
- PCR (incl RT-PCR, LR-PCR and other applications)
- Methylation-sensitive tests
- Detection of deletions, duplications, aneuploidies (MLPA, Q-PCR)
- Southern analysis
- Northern analysis
- Fragment analysis
- DNA-fingerprinting
- Sanger sequencing
- Next generation sequencing/massive parallel sequencing
- Mutation screening (DGGE, HRMCA, PTT, TaqMan, etc.)
- Making human chromosome preparations, including synchronisation, the role of mitosis inhibitors, hypotonic treatment and fixatives
- Chromosome staining including conventional staining, G-banding, QFQ-banding, (C-banding), NOR staining, Distamycin-DAPI staining.
- The relationship between chromosome structure and banding
- FISH (Fluorescence *in situ* Hybridisation)
- SNP- and CGH-array for the detection of segmental aneuploidies
- Bio-informatics: use of software (for analysis), databanks for finding and denoting genome variants/mutations

I 1.3 Quality care including occupational health and safety and environmental problems

- Theoretical knowledge of a laboratory quality system according to the norm NEN-ISO-15189
- *Quality care systems, CCKL (ISO), Accreditation Board*

L.1.4 Ethics

- Ethics in diagnostics and research (See add. C Norm ISO 15189)
- Use of informed consent
- Medical Ethics (METC, CCMO)

L.2 Skills

L.2.1 Research skills

The assistant can assess which diagnostic methods to choose (which level, which technique) for suspected structural aberrations of the genome or the possible genetic cause of monogenetic and multifactorial diseases, for example

- Congenital aberrations and development disorders
- Cardiovascular diseases
- Chromosomal diseases
- Deafness
- Echoscopic aberrations of the fetus
- Hereditary metabolic diseases
- Haemoglobin and the haemoglobinopathies
- Recurrent abortions/miscarriages
- Skin diseases
- Immune disorders
- Malignancies
- Possible genetic factors in commonly occurring diseases
- Neurological conditions
- Neuromuscular diseases
- Kidney diseases
- Eye diseases
- *Orphan diseases*
- Skeletal aberrations
- Reproduction and fertility problems

L.2.2 Practical skills

L.2.2.1. Conducting genome diagnostics.

Investigation of structural aberrations in the genome.

The assistant has experience with conducting methods and understands their principles and how they work (including limitations). S/he is capable of assessing which tests need to be done to answer the diagnostic question and can assess whether the material available for genome testing meets the minimal quality criteria.

- Preparation, storage and working with reagents
- Sample preparation, isolation, concentration, purification and storage (prenatal (maternal contamination), postnatal, DNA, RNA)
- Storage and preservation of patient material
- Laboratory equipment
- Spectrophotometry
- Working with radioactivity
- Electrophoresis and related separation techniques
- PCR
- Methylation-sensitive tests
- Detection of (large) deletions, duplications and aneuploidies (MLPA, Q-PCR)
- Southern, Northern analysis
- Mutation screening (DGGE, HR-MCA, PTT, etc.)
- Sanger sequencing
- Fragment analysis
- Profiling/DNA-fingerprinting
- UPD testing
- Next generation sequencing
- Genome-wide association studies (GWAS)
- SNP and CGH array
- Microscopy (fluorescence)
- Microphotography and/or image-processing methods
- FISH
- Karyotyping/analysis of chromosome preparations

L.2.2.2. The interpretation and statistical analysis of the data obtained and reporting

The assistant has experience with carrying out and understanding the principles and working (including limitations) of

Results

- Assessing the quality of the laboratory results and tests used
 - Denoting genetic variation: molecular pathology, copy number variations (CNVs), single nucleotide polymorphisms (SNPs), short tandem repeats (STRs), **unclassified variants (UVs)**, chromosome polymorphisms and fragile sites
 - Bio-informatics: use of software, databanks and literature data
 - Identification and characterisation of structural genome variations/chromosome aberrations
 - Denoting structural genome variations/chromosome aberrations according to international guidelines.
 - Determining clinical meaning of the findings.
 - Calculation: linkage analysis (LOD score), genetic association analysis, linkage disequilibrium
- Determining recurrence risks and risk factors (weak and dominant)

Advising: Determining whether follow-up is necessary.

Reporting: preparing report of results

- Can produce a complete and adequate description of the tests
- Can almost always independently interpret the results of the test in the context of the diagnostic question and formulate a conclusion, if necessary with the help of additional techniques or after consulting the literature
- Knows the most important implications of the diagnoses
- Is aware of the possibilities and limitations of the applied techniques and of their causes
- Understands the positive or negative predictive value of a test
- Integrates multidisciplinary data in a good way
- Integrates the national and international developments in the molecular and cytogenetic areas in the diagnostics
- Is aware that with more difficult case histories, internal or external consultation can often be essential
- Produces comprehensible reports in conformance with the internationally recognised standards
- Plays an active role in initiating new diagnostic possibilities

L.2.2.3 Experience with clinical patient care

Attending patient discussions in relevant specialisms

L.2.3 Quality care

- Quality care and accreditation according to accepted norms like CCKL and ISO15189, ISO 17025, GLP (Good Laboratory Practice)
- Standardisation, calibration, validation and evaluation of analysis techniques
- Line and ring studies

L.2.4 Management skills

- Management skills needed for the development and use of money and means in the laboratory like budget monitoring, strategic planning, writing a project proposal
- Experience with the everyday practice in the laboratory, service provision, safety, occupational health and safety and environmental matters
- Personnel matters like recruiting and selection, training, conflict management
- Knowledge of the organisation of the local, regional and national health care
- Demonstrates cost-consciousness

L.2.5 Academic skills

- Critical evaluation of medical information, scientific results and laboratory results in the literature
- Understanding of scientific methods to interpret experimental results
- Writing articles
- Can make adequate use of valid standard books, evidence-based literature, regulations, guidelines and internet sites
- Demonstrably uses 'evidence-based medicine', for example in the form of structured internet search actions
- Displays a critical approach to sources of information
- Follows generally accepted guidelines
- Can give a good oral presentation
- Shows active participation in the conduct of scientific research

- Has had at least one scientific article in the field of medical genetics published in a peer-reviewed journal
- Has given at least one scientific presentation or presented a poster during a scientific congress or symposium

L.2.6 Educational skills

- Educational skill for university and polytechnic students
- Development of educational material

L.2.7 Organisational skills

- Can carry out departmental tasks like services, educational programmes and meetings
- Can run a genetic diagnostics laboratory
- Knows the organisational aspects of a genome diagnostics laboratory as a whole and feels involved
- Takes appropriate measures to ensure the efficiency of health care
- Ensures balance between the question arising from care trajectories and the consequences for the laboratory
- Can assign priority in the diagnostic process
- Ensures a good handing over for holidays, absence and disease of oneself and others
- Copes well with peaks and troughs in work pressure and looks for help quickly if necessary
- Plays an active role in the implementation of guidelines on the workforce
- Plays an active role in the development and implementation of quality policy in the department
- Makes adequate use of the available ICT means
- Keeps a personal development plan up to date (portfolio)

L.2.8 Research skills

- Preparing a scientific research proposal
- Organisation and presentation of data
- IT skills

L.3 Attitudinal aspects

L.3.1 Professional

- Has insight into the knowledge levels of patients, clients and fellow technicians
- Can actively contribute to multidisciplinary teams
- Knows his/her limitations and when to consult others
- Recognises his/her own mistakes and those of others and acts appropriately
- Can deal with other points of view and interpretations, assuming progressive insight
- Takes a professional attitude towards the clients
- Poses ethical questions and works within the ethical norms of the profession
- Shows commitment to the professional conduct
- Is a good colleague and forthright towards fellow technicians
- Feels responsible for an optimal finishing off or handing over of his/her diagnostic tasks and smoothly takes over tasks from others as necessary
- Participates effectively in meetings
- Acts within set legal frameworks and can explain them if necessary
- Works according to the valid quality policy in health care
- Actively recognises faults in care

L.3.2 Communication

- Can communicate effectively and adequately approach paramedics, doctors, researchers, management, etc.
- Establishes effective relations with fellow technicians, analysts and clients
- Is capable when necessary of obtaining relevant background information to make a diagnosis
- Discusses the molecular diagnostic information clearly with colleagues and clients
- Can communicate adequately (both orally and in writing) with client(s), also about uncertain diagnoses
- Reports patients' case histories adequately both orally and in writing
- Can empathise with the problems of the client and caregiver
- Produces relevant diagnoses and information for the clinical practice
- Respects the patient's privacy and autonomy
- Shares the clinical information, whether written or spoken, only with relevant people

L.3.3 Education

Prepared to follow 'permanent education' and transfer knowledge to others

L.3.4 Support

- Understanding and positive attitude towards supervision
- Recognition of stress in yourself and others and being able to cope with it

L.3.5 Academic profile/research

- Innovative regarding scientific problems
- Supportive and active in research
- Collaborating attitude towards fellow technicians

L.3.6 Management/Personal effectiveness

- Interest in the organisation of departmental activities
- Understands the local management
- Constructive attitude towards decision-making
- Accepts joint responsibility
- Capable of responding effectively to complaints

Reading list (suggestions)

Emery's Elements of Medical Genetics: With Student CONSULT Online Access (Paperback)
by Peter Turnpenny MD (Author), Sian Ellard MD (Author)
Paperback: 436 pages
Publisher: Churchill Livingstone; 13 edition (July 9, 2007)
Language: English
ISBN-10: 0702029173
ISBN-13: 978-0702029172

Human Molecular Genetics, Third Edition
Human Molecular Genetics, Third Edition (Hardcover)
by Tom Strachan (Author), Andrew Read
Hardcover: 696 pages
Publisher: Garland Science/Taylor & Francis Group; 3 edition (November 21, 2003)
Language: English
ISBN-10: 0815341822
ISBN-13: 978-0815341826

Chromosome Abnormalities and Genetic Counseling (Oxford Monographs on Medical Genetics, No 46)
(Hardcover)
by R J McKinlay Gardner (Author), Grant R Sutherland (Author)
Hardcover: 604 pages
Publisher: Oxford University Press, USA; 3 edition (August 28, 2003)
Language: English
ISBN-10: 0195149602
ISBN-13: 978-0195149609

Thompson & Thompson Genetics in Medicine: With STUDENT CONSULT Online Access (Paperback)
by Robert MD Nussbaum MD (Author), Roderick R McInnes MD PhD FRS(C) (Author), Huntington F Willard
PhD (Author)
Paperback: 600 pages
Publisher: Saunders; 7 edition (June 8, 2007)
Language: English
ISBN-10: 1416030808
ISBN-13: 978-1416030805

Human Genetics and Genomics (HUMAN GENETICS: A PROBLEM-BASED APPROACH (KORF)) (Paperback)
by Bruce R Korf MD PhD (Author)
Paperback: 288 pages
Publisher: Wiley-Blackwell; 3 edition (December 22, 2006)
Language: English
ISBN-10: 0632046562

Oxford Desk Reference Clinical Genetics (Oxford Desk Reference Series) (Hardcover)
by Jane A Hurst (Author), Helen V Firth (Editor), Judith G Hall (Editor)
Hardcover: 752 pages
Publisher: Oxford University Press, USA; 1 edition (September 1, 2005)
Language: English
ISBN-10: 0192628968
ISBN-13: 978-0192628961

Abbreviations

ALDIO Trainee laboratory diagnostics assistant
CanMEDS Canadian Medical Education Derivatives for Specialists
CCKL Committee Certifying Clinical Laboratories
CCMO Central Committee on Research Involving Human Subjects
CZO Netherlands Board of Hospital Education
ESHG European Society of Human Genetics
FMLS Federation of Medical Laboratory Specialisms
HGVS Human Genome Variation Society
HGMD Human Gene Mutation Database
ISCN International System of Cytogenetic Nomenclature
KBS Critical Professional Situations
KPB Short Practice Evaluation
LOC Dutch National Board for Cytogenetics
LOD Dutch National Board for DNA diagnostics
LOG Dutch National Board for Clinical Genetics
METC Medical ethics committee
NCHGR National Centre for Human Genome Research
OMIM Online Mendelian Inheritance in Man
POP Personal development plan
UMC University medical centre
VKGL Society for Clinical Genetic Laboratory Diagnostics