

## Documentation of Eurogentest Unit 3 workshop on prioritisation in genetic testing in MIL-gården (near Lund), Sweden on Sept. 2<sup>nd</sup>-4<sup>th</sup> 2008

### Participants

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### Programme

#### **Tuesday September 2**

18.30 Dinner

20.00 Welcome, house keeping issues (Ulf Kristoffersson), Introduction (Jörg Schmidtke)

20.30 "Prioritizing health services - Concepts and issues in their application to genetics services" (Wolf Rogowski, presentation and discussion)

22.00 end of day one

#### **Wednesday September 3**

09.00 Continuation of "Prioritizing health services - Concepts and issues in their application to genetics services" (Wolf Rogowski, Presentation and discussion)

10:30 "Resolving Health Care's all to difficult choices" (Per Carlsson, presentation and discussion)

12.30 Lunch

13.30 Continuation of discussion: Concepts, issues and pitfalls in developing a prioritisation system that can be universally used for prioritisation in clinical genetics services in Europe

19.00 End of workshop. Debriefing for those who do not have to leave.

19.30 Dinner

#### **Thursday September 4**

Breakfast and departure.

### Results

#### **Overall impression**

All EGT participants felt that the workshop was an excellent and intensive learning experience and that the collaboration with the Swedish PrioriteringsCentrum / Per Carlsson should be maintained and potentially extended.

#### **Clarification of concepts**

A number of concepts were discussed and clarified to guide the future work on prioritization in genetics services:

Prioritization is a term associated with the broader concept of allocation, i.e. the conscious choice about the distribution of scarce resources. Allocation is also used to refer to the results of the decisions, i.e. the existing distribution of resources. In its latter definition, allocation of resources to genetic services in Europe has been assessed in previous EGT Unit 3 work (Javaher et al.: Genetic Testing Services in Europe. DNA-based testing for heritable disorders. Background Document, Part I, Comm Genet 11: 75-120 (2008)). In our current approach, the *decision process is of interest*.

Allocation decisions have to be made on different levels in a health care system: on a **micro-level**, every service provider has to decide how to allocate his or her time and financial resources. On a **macro (policy) level**, decisions have to be made whether certain services are covered by health insurance or tax-funded health care systems. This work is restricted to the *macro-level of coverage decision making*.

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Allocation of scarce resources can be subdivided into three distinct but interrelated steps: **Rationalization** (increasing the outcomes with given resources or decreasing resource consumption with given outcomes), **prioritization** (placing in rank order to choose accordingly) and **rationing** (limiting the possibilities to optimally satisfy needs for health care services). Rationing can take different forms, e.g. exclusion (not addressing selected conditions), goal rationing (not fully addressing health care needs), time rationing (delaying treatment) or dilution (decreasing quality). In genetics services, the issue of rationalization is e.g. related to technical and quality improvements in laboratories which is addressed by other EUGT work packages. Rationing decisions strongly depend on the amount of resources available and the current reimbursement practice in a health care context. It is therefore difficult to generalize on a European level. Even if there is no sharp line between the three concepts, the *focus of this work is, therefore, on prioritization* in the narrow sense of placing in a rank order without explicit consideration of the rationing consequences that may follow from prioritization. A recent study showed that already today, there is a mismatch between medical need and available resources in genetics (Krawczak, M., et al., Genet Test, 2007 11(4): p. 417-9) which is likely to increase with the rising number of predictive genetic tests. The expected drop of DNA test cost is unlikely to resolve the problem of scarce resources due to the remaining and potentially increasing costs of counseling and follow-up testing. As implicit prioritization and rationing already takes place, explicit prioritization is a highly relevant issue in genetics.

Work on prioritization can be purely **positive**, investigating how prioritization decisions have been made or **normative**, drawing upon ethical concepts and providing guidance for the question how the objects under consideration should be prioritized. This study aims at providing *normative guidance for the prioritization of genetics services*. This should account for principles and criteria actually used in prioritization decision, yet also for the ethical reflection of these criteria. The discussion of principles of deontological, teleological and formal ethics during the workshop lead to the conclusion that different, potentially conflicting principles apply to prioritization in genetics services. Therefore, it is unlikely that ethical reflection will lead to one fixed criterion or ranking list but rather guide the decision process.

In the case of **vertical prioritization**, prioritization objects are to be ranked within a disease / professional group. **Horizontal prioritization** addresses allocation problems between diseases / professional groups. This work *focuses on vertical prioritization within genetics* because the core competence of EuroGentest is on genetics. In collaboration with representatives of other medical disciplines, future work should also address horizontal prioritization.

Coverage decisions may relate to different types of genetics services. **Diagnostic testing** targets individuals with symptoms of the disease tested for while **predictive testing** targets individuals without symptoms. This work addresses *predictive testing* because diagnostic genetic testing can hardly be separated from the treatment context in other medical disciplines and coverage of diagnostic testing may be embedded in reimbursement of these services.

For two reasons, this work *excludes pre-implantation and prenatal testing*. First, the bulk of new and currently upcoming tests that need to be prioritized can be expected to fall outside this area. The second reason is that in pre-implantation and prenatal testing a large number of very specific additional ethical problems arise which have been discussed elsewhere and, therefore, may not be of sufficient priority to be discussed by this NoE.

The experiences of Per Carlsson have shown that it is helpful to define **prioritization objects** for activities in question, based on various **combinations of health conditions and interventions**. *Appendix 1 contains a first preliminary list of prioritization objects* for the further work. This list contains a set of prioritization objects which should represent all different characteristics of prioritization objects as well as different characteristic values which may be of relevance for prioritization decisions (e.g. high & low severity; measurable and no measurable health benefit; high and low measurable health benefit; high and low evidence of medical benefit). It should focus on prioritization objects which are of importance to decision makers and should contain as little redundancy and as few health problems as possible to remain manageable and communicable. This preliminary is likely to be subject to further update.

## **Next steps**

As a subcontractor to EuroGentest through the Medizinische Hochschule Hannover, it is Wolf Rogowski's task to take the lead for the further work on this issue for the remaining time of the EuroGentest project (subject to acceptance of the acceptance of extending the funding until the end of 2009 by the EUGT General Assembly). He can draw upon the collaboration with the members of Unit 3 as well as support from its established network of experts. To receive support on ethical issues, the further work may involve research visits at adequate institutions, e.g. the Ethox Center at the University of Oxford. Wolf was asked to formulate a concept for the next steps (a draft of which can be found in Appendix 2 to this protocol).

**Appendix 1: first list of conditions**

Condition	Type of intervention
Periodontitis	Population predictive testing dentists
HNPCC	Diagnostic whole gene sequencing
HNPCC	Male FDR predictive testing
HNPCC	Female FDR predictive testing
Huntington	Passive cascade predictive testing
CF	Carrier testing
APOE4	Predictive testing
CF	Newborn screening
LQT (sudden cardiac death)	Cascade screening in at risk families
NF1 (neurofibromatosis)	Diagnostic testing
FVL	Pharmacogenetic test before c. pill
FVL	Predictive population screening
FVL	Passive cascade
HH	Diagnostic testing
HH	Passive cascade predictive testing
HH	Active cascade screening
HH	Caucasian male population screening
HH	Population predictive testing incl. femals and non-Europeans

The list should be complemented by additional tests which are of particular relevance because of the testing load in selected European populations or because they feature additional characteristics or combinations of characteristic values of genetic tests which may be important for priority setting. Redundant tests with similar characteristics should be excluded to keep it manageable and communicable also to experts and interested parties with little expertise in genetics.

## **Appendix 2: proposed next steps**

### ***Proposed overall aim***

- Establish (and potentially test) a model for prioritization of genetics services on a European level, based on generally acknowledged principles of ethics in resource allocation including their application in health economics.
- In case the test confirms that the model is feasible and helpful, it should serve as a basis for sustaining this prioritization process within the activities of the ESHG.

### ***Proposed deliverables and timelines***

- Background paper on the ethical and economic principles of resource allocation and their application to genetics services (laid down in current subcontract; timeline: Dec 15th, 2008) to determine important points to consider for an acceptable prioritization process and to inform its participants
- Concept of a model for prioritization in genetics services. A draft of this proposal of a procedure for a transparent and participative prioritization process is made available for discussion among all interested EGT and ESHG members by May 31st, 2009
- The procedures laid out in this report are tested in a first pilot prioritization study between June and October 2009 (given the limited time frame, only a fast-track process omitting selected elements may be possible). By the end of 2009, a paper documenting the process, its results as well as lessons to be learnt for future prioritization processes will be produced.

### ***Proposed further collaborations***

- To achieve a sustainable process, the model should be developed in collaboration with the other EGT units as well as interested parties within the ESHG. It would be desirable if the technical implementation (e.g. a platform for providing written comments) and official information (e.g. of subjects for discussion or results) was implemented within the EGT and / or the ESHG infrastructure (website).
- To draw upon existing experience in the field of prioritization, further collaboration with the PrioriteringsCentrum in Sweden (Per Carlsson) and potentially with individuals involved with decision processes in other institutions for prioritization like the English National Institute of Health and Clinical Excellence or others involved with the EU-funded EUnetHTA project would be desirable. WR could invite further collaborators.
- To ensure that the model fits with the currently acknowledged principles of ethics on the allocation of health care resources, collaboration with internationally leading ethicists who are experienced both in philosophy and in genetics or allocation of health resources would be desirable. Ideally, the first and second deliverable would be produced in conjunction with research visits by WR at their institutions.