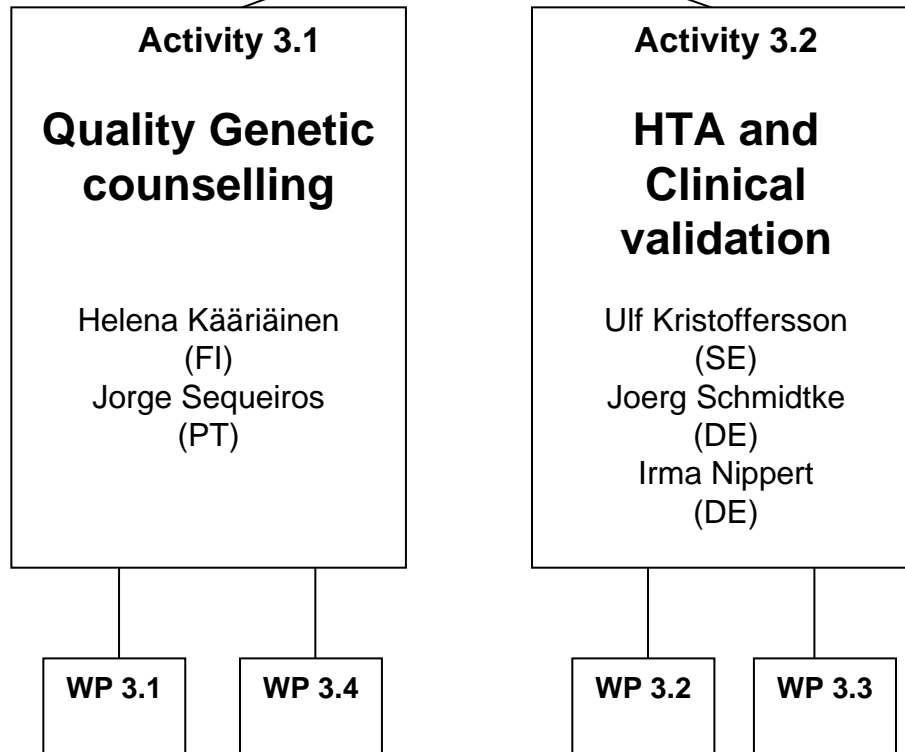


Unit 3

Clinical Genetics, Community Genetics and Public Health

Ulf Kristoffersson (SE) / Joerg Schmidtke (DE) / Helena Kääriäinen (FI) /
Jorge Sequeiros (PT) / Irma Nippert (DE)



Workpackages

- **WP3.1 Quality in Genetic Counselling**
(Helena Kääriäinen, FI)
- **WP3.2 Clinical Validity and Utility**
(Joerg Schmidtke, DE)
- **WP3.3 Evidence Based Utility**
(Irma Nippert, DE)
- **WP3.4 Definition of Genetic Testing**
(Jorge Sequeiros, PT)

Eurogentest Unit 3, WP 3.1

- The aim is to improve the quality of genetic counselling services associated with genetic testing across Europe

The network of WP 3.1

- Unit leaders:
 - Helena Kääriäinen
 - Jorge Sequeiros
 - Joerg Schmidtke
 - Ulf Kristoffersson
- Reseachers:
 - Elina Rantanen
 - Marja Hietala
 - Sirpa Soini
- Experts
 - Lauren Kerzin-Storarr
 - Heather Skirton
 - Aad Tibben
 - Andrew Faucett
 - Luis Pérez-Jurado
 - Elettra Ronchi
 - Gerry Evers-Kiebooms
 - Claire Julian-Reynier
 - Gerhard Wolff
 - Angus Clarke

What has been done:

- **Collection of National Regulation of Genetic Counselling** (Sirpa Soini, www.eurogentest.org)
- **Review of 56 Guidelines for Genetic Counselling** (Elina Rantanen, www.eurogentest.org, 1 paper in press, another submitted)
- **Guidelines of the minimal criteria for genetic counselling** (Helena Kääriäinen, www.eurogentest.org, comments collected from EuGT partners, ESHG membership and NHGSs, last step = endorsement by ESHG Board still missing), presented in ASHG meeting 2007. **More in panel discussion!**

What has been done:

- **“Regulations and practices related to genetic counselling in 38 European countries”** www.eurogentest.org
(Survey 1), presented in ESHG meeting 2007, paper submitted.

What is being done:

- **What the medical doctors tell about their counselling** (Survey 2), via laboratories to clinicians) Questionnaire sent out 2007 and last replies are expected by the end of the year, analysis and paper 2008.

What is being done:

- **How the patients have experienced counselling** (Survey 3).
- Steering Group's advice: No Survey, instead literature review (ongoing, some 100 studies from 1997-2007), will be finalized 2008.

How to **measure quality of genetic counselling?**

- A self assessment web tool at the level of the clinic has been drafted together with Expert Group and will be further discussed 13th December in an Expert Group meeting
 - About 40 criteria covering 11 topics, "traffic-light method"

Traffic light indicators

| | |
|--|---|
| | The goal was attained |
| | The goal was not attained, but the process was started |
| | The goal was not attained and the process was not started |

For example:

| 1) Guidelines | | | |
|---|--|--------------------|--|
| The clinical staff knows European and national professional guidelines for genetic counselling and follows them systematically. | | | |
| Criteria | Goal | Realisation | |
| Familiarity with the EuroGentest guidelines | Clinical staff knows the EuroGentest guidelines for genetic counselling. | | Guidelines have been handed to each member of the counselling team. |
| Familiarity with the national guidelines | Clinical staff knows the existing national guidelines for genetic counselling. | | There is only one national guideline concerning genetic counselling in prenatal diagnostic situations: this is well known in the clinic. |
| Implementation of knowing the guidelines | Clinical staff follows the guidelines that they know systematically. | | |

How to support quality development?

- "Peer-support" model
 - Presently the model is being developed by the expert group
- "Cook-book model"
 - Prof. Dr. Ursel Theile has written (in German) a book covering counselling in some hundreds (?) of genetic diseases; discussion on how to use this as a model is ongoing.

Possible other items for future?

- Harmonizing the consent process?
 - Usually under national regulation
- Training and curricula?
 - Collaboration with Unit 6
- Creating other ways than traditional genetic counselling for providing genetic information to individuals considering a genetic test?
 - WP 3.1 feels a lack of resources here

Implementation

- **Publishing** (2 reports in ESHG/ASHG meetings, www, 3 papers in press/submitted, more to come)
- **Investigating feed back** from the guideline of genetic counselling (Manchester group)
- Participation in **Road Show**
- **Web-tool(s)** being developed
- Collaboration with organizers of **training courses** (EGF, others)

What from WP3.1 will survive after EuGT?

- Publications
 - In addition to EuGT website, there will be at least 6 papers
- Guidelines of genetic counselling
 - Implementation (other than EuGT website) starts 2008
- Web-tools
 - Being developed, could be put also to ESHG website?