Exploring the boundaries of life sciences research, Wednesday, January 17th

From monogenetic screening to screening of multifactorial diseases

8.45-9.30 (Dirk Stemerding)
Introduction to the group.
Discussion: what ideas for the future of public health genetics/genomics are present in contemporary life sciences?

Coffee break

9.45-10.30 (Dirk Stemerding)
Lecture: The path from lab to population screening involves more than just solving technical problems. Political and economic aspects, as well as ethical and social considerations shape the way genetic screening is implemented. Screening for haemoglobinopathies will serve an example of the kinds of processes and problems that are involved in realizing a genetic screening programme.

Coffee break

11.00-12.45 (Erwin van Rijswoud) Case study:
Screening for hereditary haemochromatosis: between science and practice
Hereditary or primary haemochromatosis (PH, iron overload) is a genetic disorder that causes the serum ferritin levels to rise to toxic levels. This results in many health problems, such as damage to the liver and chronic fatigue. A genetic test for PH is available, but as yet, no official screening programme was launched. Some pilot studies, however, have provided data on the feasibility of population screening. In fact, a Nijmegen pilot is about to be concluded (the HEFAS: HEmochromatosis FAmily Study).

Before an official population-screening programme for PH can be launched in the Netherlands, the Dutch Health Council committee has to see if the proposed screening programme complies with the regulation in place.

Based on the lectures, the reading and an introduction to the case study, the students will act as a Health Council committee which advises the Dutch government on population screening for iron overload. In groups, the following questions are discussed:
- Is screening for haemochromatosis scientifically & societally acceptable from the perspective current regulation?
- If screening is acceptable, what kind of model is most appropriate and will cause least societal and ethical concern?
- What kinds of questions remain to be answered by a) politics, b) researchers.

More information on this will be given in the introduction.

In preparation to the case study, the students are expected to have read the following papers.
- Delatycki et al., *Use of community genetic screening to prevent HFE related hereditary haemochromatosis*, Lancet vol 366, july 23rd 2005, 314-316 (2 p)