Genetic counselling for family planning

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Who / When?

• **Before pregnancy:**
  – Young adults or couples who know a significant family history of a possibly hereditary condition
  – Exceptionally: young couples without any known risk

• **During pregnancy:**
  – Women who are identified because of family history taken in antenatal clinic as having a fetus at risk of a genetic condition
  – Women whose fetus is diagnosed as having abnormal findings in a screening test
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Example 1: microdeletion

- The child of the couple has been diagnosed as having a small chromosomal deletion in microarray examination.
- Risk for recurrence is very low
- The family may, however, want to discuss why the risk is low etc.
BAC array
Example 2: autosomal dominant disease in the parent

- One of the future parents has a genetic condition that is autosomally dominantly inherited.
- This means that in each pregnancy there is a 50% chance that the fetus inherits the mutation and will be affected, about similarly as the parent.
Achondroplasia as an example

- Genetic counsellor knows the chance of recurrence, possibility of prenatal diagnostics etc.
- The patient/family knows about the disease
Example 3: autosomal recessive disease in a previous child

- Healthy parents, usually without any family history, have a child affected with an autosomal recessive disease.
- The risk of recurrence in future pregnancies is 25% in each pregnancy.
Infantile neuronal ceroid lipofuscinosis as an example

- This progressive encephalopathy (common in Finland) starts during first year of life and quickly progresses to a situation, where the child is blind, unable to move or even swallow and appears to be in pain.
Prenatal diagnosis became available
Couples without any known risk

- Sometimes couples want genetic counselling without any known increased risk, simply to do everything possible to have a healthy child.
- These couples often ask for all possible genetic carrier tests.
- In genetic counselling (in the public health care) we ask their family history and explore the reason for the worry, but do not (usually) offer any carrier screening.
In the future?

- Possibly with new genetic methods, wide carrier screenings can be performed with reasonable price.
- Such tests can be offered direct to consumer.
- Genetic centres consider these possibilities complicated: the couples might get risk figures for diseases that they have never heard of, which would lead to difficult genetic counselling scenarios.
Principles for genetic counselling in the context of family planning

• Genetic counselling should be available for families with a possible increased risk for a rare disease in the offspring.
• It should be carried out by appropriately trained professionals.
• It should provide information and support
• The decisions should be based on this information and the values, wishes and cultural norms of the couple.