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CONCOR

National registry and DNA-bank of adults with congenital heart disease

www.CONCOR.net

Number of participating hospitals: 103
Number of included patients: 10,613

Steering committee
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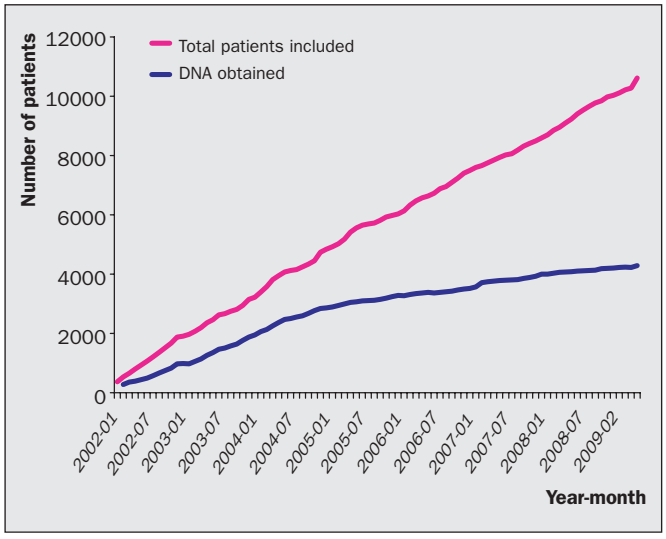

Figure 1. Progress of inclusion.

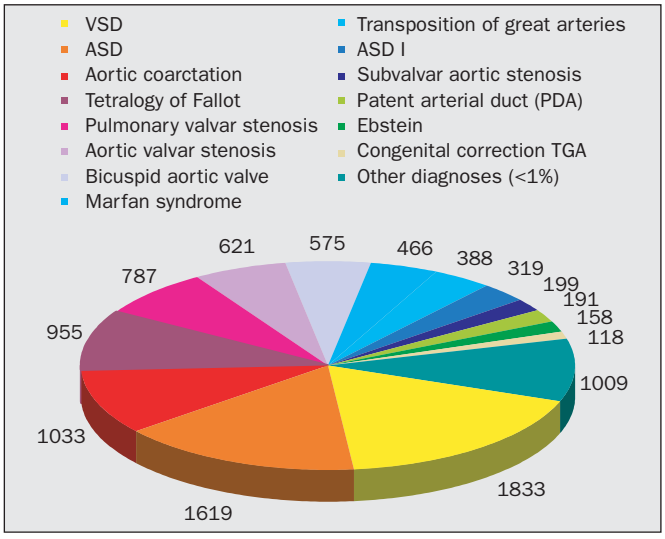

Figure 2. Most frequent main diagnoses.

22q11 deletion was present in 10% of tetralogy of Fallot patients in CONCOR. In 65% of the identified patients the 22q11 deletion was as yet unknown! As a cardiologist, you can request 22q11 deletion diagnostics. See the CONCOR website (under 'for cardiologists').

Table 1. Symptoms of 22q11 deletion.

Symptoms of 22q11 deletion include:
- Congenital heart disease, particularly conotruncal malformations
- (Submucosal) cleft palate or hypernasal speech
- Learning disabilities/mental retardation
- Psychiatric problems
- Characteristic facial features
- Immune deficiency
- Hypocalcaemia
- Thrombocytopenia

The clinical features are highly variable between patients. Offspring of an affected patient have a 50% chance of inheriting the disorder

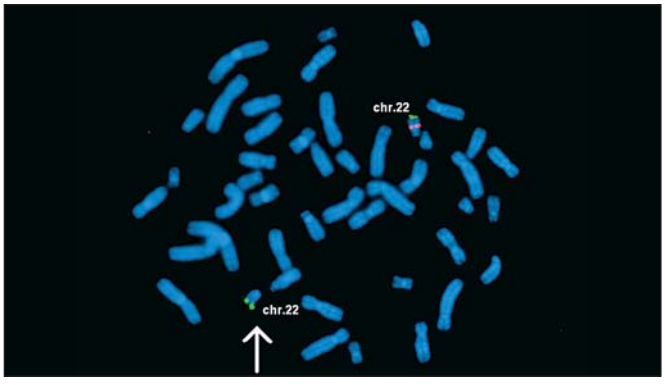

Figure 3. FISH 22q11 picture. The green dots (probes) mark the two chromosomes 22. The red probe marks the region 22q11. The red probe is missing on one of the chromosomes 22, confirming the deletion (see arrow).