# Guidelines for postnatal karyotyping and analysis of the FMR1 and CFTR genes

**Indications for standard postnatal karyotyping (chromosome analysis)**

- Clinical suspicion of a chromosomal disorder (e.g. Down syndrome, Turner syndrome, Klinefelter syndrome)
- Couples with 3 miscarriages at age $\leq$ 38yrs or with 2 miscarriages at age $\leq$ 33 yrs
- Couples entering an IVF program
- Male or female gamete donors
- Parents of a fetus with an abnormal karyotype
- Known chromosomal aberration in the family
- Suspicion of a balanced translocation
- Aberration detected with molecular karyotyping for which a standard karyotype could shed more light on the nature of the aberration (e.g. marker chromosome, unbalanced translocation)
- Suspicion of chromosomal mosaicism in an individual

**Indications for FMR1 analysis (diagnostic)**

- Individuals of either sex with developmental delay or unexplained intellectual disability
- Women who have reproductive or fertility problems
- Individuals with the suspected diagnosis of fragile X tremor/ataxia syndrome (FXTAS)

**Indications for FMR1 analysis (carrier testing)**

- Individuals at risk for being carrier because of a family history of either i) fragile X syndrome or possible X-linked mental retardation; or ii) fragile X tremor/ataxia syndrome (FXTAS); or iii) premature ovarian failure
- Women entering an IVF program
- Female gamete donors

**Indications for CFTR analysis (diagnostic)**

- Individuals with classical cystic fibrosis (CF)
- Individuals with atypical clinical CF and/or borderline sweat test
- Newborns with meconium ileus or elevated IRT
- Fetus with echogenic bowel and/or in parents
- Individuals with male infertility and CBAVD
- Individuals with other CFTR-related disorders (e.g. disseminated bronchiectasis, chronic pancreatitis, chronic rhinosinusitis, allergic bronchopulmonary aspergillosis)

**Indications for CFTR analysis (carrier testing)**

- Individuals with increased risk compared to the population risk
- Individuals with increased risk for their offspring because of consanguinity (in specific populations)
- Individuals whose partner is either carrier or affected by CF
- Males entering an IVF program
- Male or female gamete donors

*The above mentioned guidelines were prepared by Geert Mortier and reviewed and approved by the High Council for Antropogenetics at the meeting of 13/07/12.*