



Belgian Society of Human Genetics absl-vzw

Guidelines for analysis of the CFTR gene

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
- Foetus 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 the Belgium Society of Human Genetics Work Group on “Good Clinical Practice”, modified by Geert Mortier, reviewed and approved on 13/07/2012 at the Meeting of the High Council for Anthropogenetics (now College for Medical Geneticists).