

Complete *CFTR* gene mutation analysis in European patients with Cystic Fibrosis

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In cooperation with the ECFS Diagnostic Network Working Group (Nico Derichs)
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AIM

to provide a service for highly parallel sequencing of the complete *CFTR* gene (including intronic and promoter regions) in patients with confirmed CF (maximum 5 patients per individual site) in whom a disease-causing mutation was not found on both *CFTR* genes.

Criteria for inclusion:

- the local CF physician confirms the diagnosis of 'classic' CF according to the European CF diagnostic criteria (Thorax 2006): patient has symptoms compatible with CF OR a sibling with CF OR a positive test at newborn screening AND a sweat chloride value >60 mmol/L.
- routine *CFTR* mutation screening panels have not allowed identification of 2 CF-causing *CFTR* mutations.
- a signed written informed consent, according to the institute's ethical committee regulations and approvals, is signed by the patient and the local physician and archived locally. This consent must include that the mutation information and the clinical data of the patient will be listed anonymously in a central archive and that the results of the entire project will be published in a scientific journal.
- patient does not reside in UK, Spain or Czech Republic (separate program is available).
- 2 to 5 microgram of DNA is available.

How to proceed:

1. Send duly filled out request for *CFTR* gene analysis to els.aertgeerts@uzleuven.be
2. Approval /rejection of request will be mailed to the applicant physician within 2 weeks
3. For approved requests, send 2-5 microgram of DNA to: Martine Jaspers, Campus Gasthuisberg, O&N2, Exp. ORL, trilhaarlabo (7de verdiep), Herestraat 49, bus 721, 3000 Leuven, Belgium
4. Mail harry.cuppens@skynet.be that the DNA sample has been sent