Immunoreactive Trypsin

General:

Cystic fibrosis (Mucoviscidosis) is an autosomal recessively inherited disease with a frequency of 1:2500, which predominantly affects the white population. The gene is located on chromosome 7, encoding the 'CysticFibrosis-Transmembrane-Conductance Regulator' (CFTR). Its physiological function is the intra- and extracellular regulation of chloride transport. Besides many known mutations, one common mutation is a deletion at position 508 in the CFTR gene. The mutations lead to less functional protein and electrolyte disturbances occur (positive sweat test) by transmembraneous transportation disturbances.

Typical symptoms are: formation of solid mucus in the lung, pancreas, liver and the small intestine, which affects the function of these organs. The first symptom is often an intestinal obstruction (meconium ileus) in infancy. Later, increased deterioration of the respiratory function is of major importance. Since a strong mucous obstruction blocks the way of premature sperms in the oviduct of the testes and epididymis, over 90% of the men are not fertile, many women are similarly infertile. Please note that one heterozygous mutation - without presenting symptoms of CF - can be sufficient to cause infertility (treatable!).

The diagnosis of CF/mucoviscidosis can be done by gene analysis or by pre-screening of immune reactive trypsin in native blood drops collected on a Guthrie filter card (as with newborn screen or metabolic screen).

The following tests are available:

- **Immunoreactive trypsin**

Indication:	Pre-screening for cystic fibrosis **(only valid up to the 21st day of life!)**
Material:	filter card
TAT:	5-7 days*
Method:	RIA
Units:	ng/ml
Ref.- range:	see report

- **Cystic fibrosis, genetic test**

Indication: meconium ileus, chronic or relapsing bronchitis, pneumonia, borderline or positive sweat test, pancreatic insufficiency, fertility disorder (suspicion of congenital bilateral aplasia of the vas deferens, CBAVD), analysis of the transmitter status in risk families.

Material: EDTA blood

TAT: Analysis of Del 508 mutation : up to 3 weeks

Screening for 50 mutations : 3- 4 weeks

Sequencing of the complete gene: 3-4 weeks

Method: PCR, sequence analysis

For more information please contact FML.

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit http://www.fml-dubai.com/parameter-listings/

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