



Sweat Test

Synonyms

Clinical Indication

In cystic fibrosis there is a defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7. This codes a transmembrane protein affecting chloride transport and giving rise to excessively viscous secretions. The disease shows an autosomal recessive pattern of inheritance, with a relatively high incidence of 1:2500 live births in white Northern European populations. Over 1000 mutations at this locus have been associated with cystic fibrosis, although some are very rare. The $\Delta F508$ mutation accounts for about 70% of UK cases.

The test is usually performed on young children with symptoms typical of cystic fibrosis, such as repeated respiratory infections, and/or exocrine pancreatic insufficiency. It is also undertaken if there is a family history of the disease, although if the genotype is known DNA studies may be appropriate.

Part of Profile / See Also

Request Form

Combined Pathology manual Blood form or ICE request

Availability / Frequency of Analysis

The test is performed in the Paediatric Department at Southend Hospital by appointment only.

Southend patients: Please telephone the POCT team on Ext 6612 for bookings.

Basildon patients: Please telephone the Biochemistry Laboratory, Basildon Hospital, where BMS staff can book in patients using the internal joint booking form. Alternatively please telephone POCT team at Southend on 01702 435555 ext 6612.

Email enquires can be sent to mse.poct.suhft@nhs.net

Patients/carers will be in the department for about an hour.

Turnaround Time

Chloride will be measured within 3 days of collection.

Patient Preparation

It is not normally possible to test children under two months old, because of the physical difficulty of sweat collection. In children up to six weeks old, a blood immuno-reactive trypsin assay may be used to screen for cystic fibrosis.

Sample Requirements

Specimen Type

Sweat

Volume

Minimum 20 μ l

Container

Micro cup

Reference Range & Units

Normal <40 mmol/L for children aged 6 months and older or <30mmol/L if less than 6 months of age.

Intermediate 40 – 60 mmol/L for children aged 6 months and older or 30 – 60 mmol/L if less than 6 months of age.

Elevated >60 mmol/L

Interferences

Sweat test should be deferred if patient on systemic corticosteroids.
 Sweat test should be deferred in subjects who are dehydrated, systematically unwell, oedematous, under Topiramate treatment or who have marked eczema covering potential stimulation sites.
 Chloride containing solutions cannot be used to clean the skin, nor local anaesthetic gel be applied.

Interpretation & Clinical Decision Value (if applicable)

The chloride levels in the sweat from affected individuals are high. Cystic fibrosis is unlikely with a sweat chloride level of <40 mmol/L.

A chloride level of >60 mmol/L is consistent with a clinical diagnosis of cystic fibrosis.

Abnormal results will be telephoned to the requesting Clinician.
 A confirmatory sweat test will be done if requested. A blood sample should be sent for DNA studies, and family studies may be required.

Equivocal results (sweat chloride 40 - 60 mmol/L) should have a further sweat test performed, and if this is also equivocal DNA studies should be considered. Equivocal results may be seen in unaffected children, or those with atypical cystic fibrosis - i.e. caused by gene defects other than homogenous $\Delta F508$ mutation.

References

If the results do not seem appropriate for the clinical condition of the patient, please discuss with a Clinical Biochemist.

Guidelines for the Performance of the Sweat Test for the Investigation of Cystic Fibrosis in the UK, An Evidence Based Guideline, Second Version, March 2014.

Test code

SWCL

Lab Handling

Sample processed within the lab.
 Sample stored at 2 - 8°C