

Cystic Fibrosis

“Woe to the child that when kissed on the forehead tastes salty. He is bewitched and shall soon die.” – Old Northern European quote on Cystic fibrosis

Cystic fibrosis is an autosomal recessive disease that affects the transport of electrolytes in the body. It is predominantly expressed in epithelial tissue and impacts quite heavily on Cl^- and Na^+ levels.

1 in 2500 live births is a cystic fibrosis (CF) sufferer. 1 in 20 people carry a single defected copy of the gene that causes the disease. 50 years ago, it was rare for a CF patient to survive beyond their first birthday. Today, patients have a life span of ~45 years, due mainly to the medication that can be used to offset most of the symptoms.

CF affects almost all the parts of the body. Some of the main areas are:

- Airways
 - Prone to clogging and infections
- Liver
 - Blockages of small bile ducts and problems with liver function in 5% of patients
- Pancreas
 - Blockage of the ducts prevents the secretion of digestive enzymes in 65% of patients
- Small Intestine
 - Obstruction due to thick content in 10% of newborns – will require removal very soon after birth
- Reproductive Tract
 - Absence of vas deferens in 95% of males makes them infertile. A small number of women are also affected.

Infants with CF will not gain weight in the first weeks after birth due to pancreas blockage – cannot digest milk. This is one of the first signs of CF that medics can look for.