

CYSTIC FIBROSIS, NEWBORN, SCREEN**(Fluoro Immuno Assay)**

ng/mL B

<70.00

Note: 1. Results should be clinically correlated as individual / biological variations can affect the test results

2. Test conducted on heel prick blood performed for IRT

3. Genetic counseling available with prior appointment at Department of Genetics,

National Reference Lab, New Delhi

Comments

Cystic fibrosis is one of the most common autosomal recessive diseases due to mutation in Cystic fibrosis transmembrane conductance regulator (CFTR) gene seen mainly in people of Northern European ancestry. It is a multisystem disorder affecting pulmonary, gastrointestinal and reproductive organs. The phenotypic expression of the disease is heterogenous ranging from meconium ileus to severe respiratory disease in infants. Immunoreactive trypsinogen (IRT) is used to screen newborns for increased risk of Cystic fibrosis. Screening for Cystic Fibrosis has helped in increasing survival age largely due to organ transplantation, improved nutrition, new drug therapies and shall continue to do so with a potential of successful gene therapy.

Increased IRT Levels - Cystic fibrosis – Delta F 508 heterozygotes (Commonest mutation), Hypoxic insult to pancreas, Renal insufficiency, Congenital heart disease, Spina bifida, Gastroschisis, Viral infections, Trisomy 13,18 & Galactosemia

Impression**Advised**