MOLECULAR PATHOLOGY REPORT

CONSULTING PHYSICIAN: MUHAMMAD SOHAIL SALAT
REQUESTED BY : MUHAMMAD SOHAIL SALAT
CLINICAL INFORMATION / COMMENTS :

123-16-11
JUNAID,BABY GIRL OF SUMAIRA
Female 3M8D ACC # 81291189 (DIS)

LOC: D0 STATUS: IN
2008 : MB7418R - 16375928 [COMP]
COLL: 14/02/2008 -17:30 RECD: 14/02/2008 - 17:30

SOURCE : BLOOD
DELTA 508 MUTATION [Final Report]

DELTA F508 MUTATION FOR CYSTIC FIBROSIS:

Homozygous for Delta F508 mutation.

COMMENTS:
Cystic Fibrosis is an autosomal recessive disease characterised by the prescence of defects in cystic fibrosis transmembrane receptor gene, which is located on chromosome 7q31.2. The gene encodes a chloride channel transmembrane protein responsible for chloride electrolyte metabolism. If both parents are carrier for CFTR mutation (heterozygous i.e. one abnormal copy of the gene) then there is a 1 in 4 chance that the fetus will have a homozygous (i.e to carry both abnormal copies of the gene) genotype and will inherit cystic fibrosis.

ICN#216128

This is a computer generated report therefore does not require any signature. Date: 27/04/2013 18:31:35 {Dup}

276121300458909