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CASE REPORT

FRASER SYNDROME IN A FEMALE BABY AGED 2MONTHS - A CASE REPORT

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ABSTRACT: Fraser Syndrome is an autosomal recessive congenital disorder first described by Canadian geneticist C.R. Fraser in the year 1962. The incidence of Fraser Syndrome is 0.043 per 10,000 live born infants and 1.1 in 10,000 still births making it a rare syndrome.
KEYWORDS: Fraser Syndrome, baby, rare.

INTRODUCTION: Fraser Syndrome or Meyer–Schwickerath's Syndrome or Fraser –Francois Syndrome is an autosomal recessive congenital disorder first described by Canadian geneticist C.R. Fraser in the year 1962. It is usually associated with acrofacial and urogenital malformations with or without cryptophthalmos, positive family history, umbilical hernia and bilobed lungs.[1]

CASE REPORT: Two month old female baby presented with:

- Bilateral cryptophthalmos(where the eyelids fail to separate in each eye)
- Syndactyly in both hands(fused fingers)
- Anomaly of the auricle of both ears
- Umbilical hernia
- Enlarged clitoris
- Deep breathing

DIAGNOSIS: Diagnosis of this Syndrome can be made on clinical examination. Diagnosis based on at least two major and one minor criterion or one major and four minor criteria [2].

Major criteria are:

i) cryptophthalmos
ii) syndactyly
iii) abnormal genitalia
iv) positive family history.

Minor criteria are:

i) congenital malformation of the nose, ears or larynx.
ii) cleft lip or palate
iii) skeletal defects
iv) umbilical hernia
v) renal agenesis
vi) mental retardation

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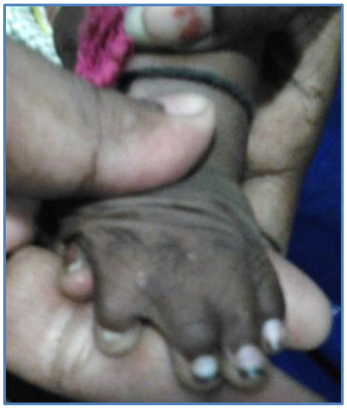
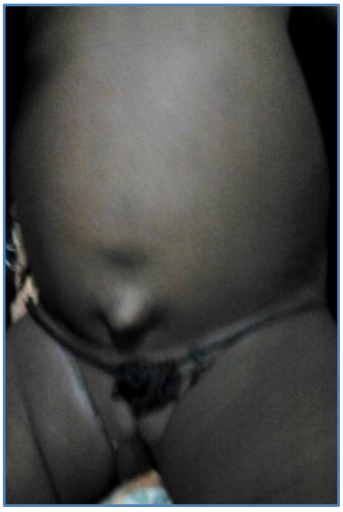
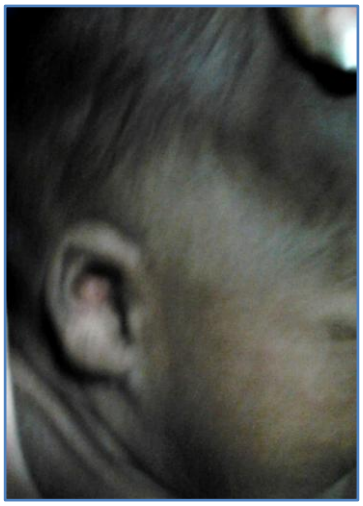
The female baby fulfils 3 Major criteria and 2 Minor criteria. So the diagnosis of Fraser Syndrome is confirmed.







Genetics: linked to a gene called FRAS1 which seems to be involved in skin epithelial morphogenesis during early development.[3]

Chromosomal abnormality: both the female baby and phenotypically normal father had a chromosomal anomaly: inv (9) (p11q21).

Epidemiology: The incidence of Fraser Syndrome is 0.043 per 10, 000 live born infants and 1.1 in 10,000 still births making it a rare syndrome.

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