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Case Report:

Emanuel syndrome: a rare case report

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ABSTRACT:

Emanuel syndrome (ES), also known as Derivative 11;22 syndrome; is a rare chromosomal disorder that is characterized by multiple congenital anomalies and developmental disabilities. It is an unbalanced translocation syndrome usually arises through a 3:1 meiosis 1 malsegregation during gametogenesis in a balanced translocation phenotypically normal carrier. Here we are reporting a case of a neonate having typical features of Emanuel syndrome. On examination child had facial features of high arched palate, micrognathia, pre-auricular skin tags, microcephaly, sacral dimple, hypotonia and cardiac anomalies. The detection of this syndrome as early as possible helps in improving quality of life and perinatal diagnosis in future pregnancy by antenatal cytogenetic testing.

Keywords: Emanuel syndrome , chromosomal disorder

INTRODUCTION:

Emanuel syndrome (ES) is an unbalanced translocation syndrome, usually arising through a 3:1 meiosis I malsegregation during gametogenesis in a phenotypically balanced translocation normal carrier. While the true mortality rate in Emanuel syndrome is unknown, long-term survival is possible.(1) Emanuel syndrome is also referred to as derivative 22 syndrome, derivative 11;22 syndrome, partial trisomy 11;22, or supernumerary der(22)t(11;22) syndrome (2). In this partial duplication, 11(q23-qter) and 22(pter-q11) complex, congenital diaphragmatic hernia has been observed (3). There is growth retardation, mental retardation (severe), cardiovascular malformation, craniofacial anomalies (including pre auricular tags or sinuses, micrognathia, ear anomalies, cleft or high-arched palate), microcephaly, kidney abnormalities and genital abnormalities in males.(1-4)

CASE REPORT:

A 7 day old male neonate presented to us with complain of not feeding well. He was the first child of non consaguinous marriage born out of late pre term vaginal delivery with birth weight 2000 gm, 50 cm length and 33 cm head circumference. After brief clinical examination patient had features of high arched palate, micrognathia, pre-auricular skin tags(fig.1) , microcephaly, sacral dimple(fig.2),hypotonia. Child investigated for other features of anomalies. 2D-Ehco was done suggestive of ostium secundum atrial septal defect of about 6mm with left to right shunt. Ultrasonography of abdomen and neurosonogram did not reveal any abnormality.

After counselling the parents, karyotyping was done, which revealed the following chromosomal abnormality: 47XY, +der (22) (q23.3; q11.2)(fig3). To ascertain the origin and trait of this supernumerary marker chromosome (der 922) t (11;

22)(q23.3;q11.2)] karyotyping of his parents was performed . The mother was found to be a balanced carrier: 46XX t (11; 22)(q23.3;q11.2) (fig.4)The patient is thus diagnosed as EMANUEL SYNDROME.

DISCUSSION:

The exact incidence is unknown. This is a rare syndrome with reported cases of around 100. Highest mortality is in the first few months of life.

List of clinical features observed in Emanuel syndrome: (5-7)

Growth and development : Pre and postnatal growth retardation, delayed speech, and language development (more commonly).

Craniofacial : brachycephaly, prominent forehead, epicanthal folds, down slanting palpebral fissures, broad and flat nasal bridge, long pronounced philtrum, abnormal auricles, preauricular ear pits and/or tags (76%), deafness, and otitis media.

CNS : Microcephaly present most commonly, seizures, and delayed pschomotor development

Cardiac : 60% individuals with congenital heart defects like atrial septal defect, ventricular septal defect, Tetralogy of Fallot, and patent ductus arteriosus

Genitointestinal : Diaphragmatic hernia, anal atresia, inguinal hernias, biliary atresia, small penis 64%, and cryptorchidism 46%

Musculoskeletal : Centrally based hypotonia most commonly, congenital hip dislocation, arachnodactyly, club foot and joint, syndactyly of the toes delayed bone age, and hyperextensibility of joints.

Oral findings : Cleft palate (50%), micrognathia (60%), angular mouth pits, bifid uvula, and facial asymmetry.

Immunological : Congenital immunological deficiency.

Renal : Renal defects (36%)

Diagnosis: Investigations needed to find out associated anomalies

Include 2-D Echo, Renal ultrasound, ophthalmologic evaluation, hearing evaluation, . Developmental assessment .

Management: It involves multidisciplinary team approach involving pedodontist, pediatrician, plastic surgeon, geneticist, gastrologist, speech therapist, urologist, cardiologist, ENT surgeon, and ophthalmologist. Patients with cleft palate have feeding difficulties, which requires feeding plate and surgical closure of cleft palate. The long-term prognosis is directly related to the associated congenital malformations. Highest mortality is in the first few months of life.

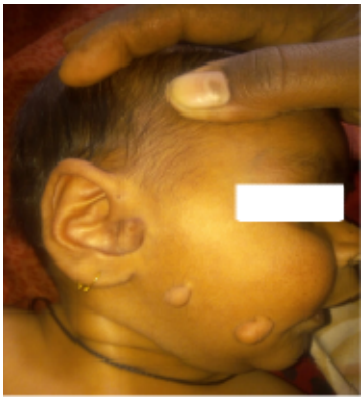

Fig 1

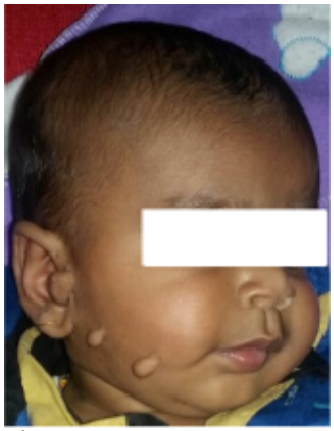

Fig 2

Fig 3

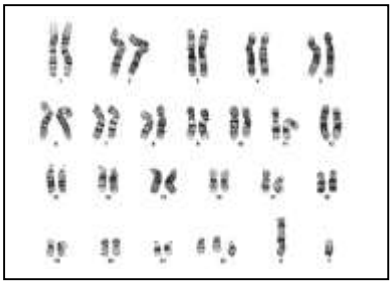

Fig.3:Karyotype of patient der(22)t(11;22)

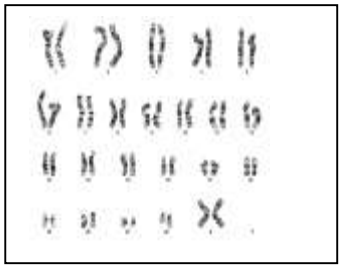

Fig.4:Karyotype of the mother [t(11;22)(q23.3;q11.2)].

Conclusion:

It is necessary to suspect this syndrome if a neonate presents with foresaid facial dysmorphism and congenital anomalies so that early diagnosis and interventions can be done. For such families,genetic counselling is very important as future pregnancies are at an increased risk for either ES, balanced t(11;22), or another meiotic malsegregation.Thus, prenatal cytogenetic testing should be offered and carrier testing of the unaffected siblings should normally be offered.

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For any images presented appropriate consent has been obtained from the subjects: NA
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