Emanuel Syndrome : A Case Report

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Abstract : Emanuel Syndrome or Supernumerary derivative (22) syndrome is one of the rare genomic syndromes. It is characterised by severe mental retardation, microcephaly, failure to thrive, ear anomalies, pre-auricular tags or sinus, cleft palate or high arch palate, micrognathia, renal anomalies, congenital cardiac defects and genital abnormalities in males. In 99 percent of the cases, one of the parents is a balanced carrier of a translocation between chromosome 11 and chromosome 22. We report the first known case, a male neonate, of supernumerary derivative (22) syndrome. **Keywords :** Emanuel syndrome, supernumerary derivative (22) syndrome, t(11;22), trisomy 11, trisomy 22.

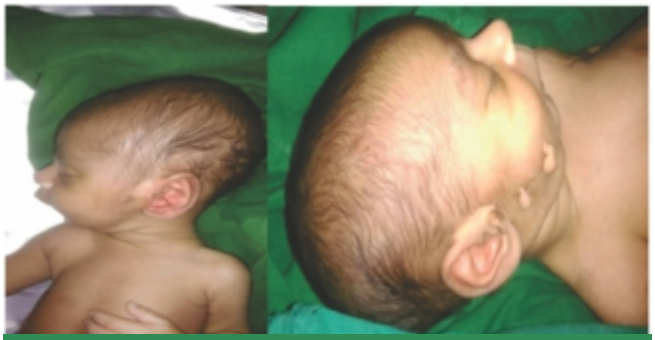
Introduction : Emanuel Syndrome is the constitutional translocation between chromosomes 11 and 22 and is the most common non-Robertsonian translocation in humans. Clustered breakpoints involving q23 of chromosome 11 and q11 of chromosome 22 have been reported in numerous unrelated families. Balanced translocation carriers are clinically normal, and are often identified after the birth of an offspring with supernumerary der (22) t (11;22) syndrome.[1]

This genomic syndrome was named as Emanuel syndrome in 2004 (OMIM # 609029). Patients with Emanuel syndrome has a distinctive phenotype, which consists of severe mental retardation, microcephaly, prominent forehead, epicanthal folds, down-slanting palpebral fissures, broad and flat nasal bridge, long and pronounced philtrum, micrognathia, cleft palate, abnormal auricles ranging from microtia to large ears often associated with a pre-auricular ear pit and/or skin tags, in developmental milestone, renal anomalies, congenital cardiac defects, and genital anomalies in boys.[2] This unbalanced translocation syndrome usually arises through a 3:1 meiosis I mal segregation during gametogenesis in a balanced translocation carrier.[3]

Case Report : A young mother aged 22 years was reported with a 12 days male neonate. The marriage of the infant's parents was consanguineous. The antenatal period of the infant was uneventful. The infant was delivered preterm by vaginal delivery. On examination, he was small for gestational age as the birth weight was 2 kg (<third percentile), length was 45 cm (<third percentile), and head circumference was 32 cm (<third percentile).

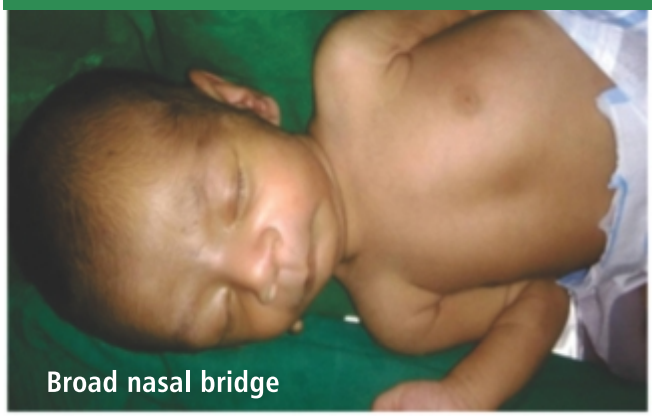
He had a remarkable facial appearance which included, Microcephaly, prominent forehead with dilated veins, broad nasal bridge, prominent philtrum, unilateral right sided microtia and large pre-auricular skin tags and pit. He was also having a small penis (1.4 cm), but both testes were completely descended.

Oral findings observed were high arched palate and micrognathia.



Micrognathia

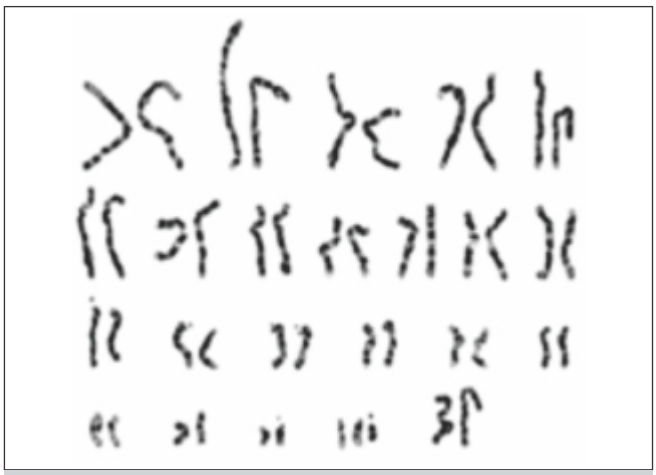
Pre-auricular tags & pit, Microcephaly, Microtia



Broad nasal bridge

His cardiovascular assessment revealed a moderately large ventricular septal defect and a small patent ductus arteriosus. Renal ultrasonography, ophthalmological assessment and the hearing assessment did not reveal any abnormality.

During the follow-up examination, he was found to have hypotonia, developmental delays, and all growth parameters remained below the third percentile.



The patient's karyotype shows an extra supernumerary chromosome.

His mother's karyotype shows a balanced non-Robertsonian translocation between chromosome 11 and chromosome 22.

Discussion : Emanuel syndrome is named after Dr. Beverly Emanuel, a cytogenetist in Philadelphia, USA. Emanuel syndrome is a chromosomal disorder that is characterized by learning problems and stunted growth and development[4]. The signs and symptoms are varied and may include hypotonia, developmental delay, microcephaly, epicanthal folds, microtia, distinctive facial features, micrognathia, cleft palate, heart defects, renal anomalies.Seizures and behavioral problems are very common in emanuel syndrome patients. Emanuel syndrome is caused by additional genetic material of chromosome 11 and 22 in each cell. This condition is usually inherited from a parent who has a balanced translocation between chromosomes 11 and 22[5,6]. This unbalanced translocation syndrome usually arises through a 3:1 meiosis I mal segregation during gametogenesis in a balanced translocation carrier.

Life expectancy in patients with emanuel syndrome is around 30 years. Management of child with emanuel syndrome is mainly supportive in the form of routine check up to find out any complications. Counselling is given to parents who are having one baby affected with emanuel syndrome to go for antenatal screening for emanuel syndrome.

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