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CASE REPORT

Phocomelia Syndrome - A Case Report

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Abstract:

Phocomelia is an extremely rare malformation in which babies are born with limbs that look like flippers on a seal. Although various factors can cause phocomelia, the prominent roots came from the drug use of thalidomide and from genetic inheritance. Phocomelia is transmitted as an autosomal recessive trait with variable expressivity and malformation is linked to chromosome 8.

Key Words: Phocomelia, meromelia, Thalidomide syndrome.

Case report:

A full term male child born of non consanguineous marriage with birth weight of 2.6 kg was delivered vaginally to 26 yrs old primi gravida at Civil Hospital attached to Dr. V. M. Govt. Medical College, Solapur. After birth, baby was referred to NICU for congenital limb anomaly. The mother did not have any antenatal history of drug intake or any other complications.

On examination, the neonate had underdeveloped upper limbs, thumbs were missing on both sides, with 4 fingers on right and 3 fingers on left side. On systemic examination, pansystolic murmur of grade 3 was present suggestive of ventricular septal defect, which was confirmed by 2-D-echo. On the basis of findings phocomelia syndrome was suspected.

Discussion:

Embryological basis of malformation of limbs show that an individual will have chromosome copies that do not connect at centromeres

making them unable to move to the equatorial plane of spindle in metaphase of cell division. As a result, the cell becomes incapable of division or slow in process, because of this the newly made cells contain an excessive or reduced amount of chromatin. In phocomelia, the cells ceased to develop or die, preventing proper development of limbs, eyes, brain, or other structures [1]. Abnormalities vary greatly, and they may be represented by partial [meromelia] or complete absence [Amelia] of one or more of the extremities. Sometimes rudimentary hands are attached to the trunk by small, irregularly shaped bones [phocomelia, a form of meromelia or 'seal limbs']. Sometimes all segments of the extremities are present but abnormally short [micromelia] [2]. Limb malformations occur in approximately 6 per 10,000 live births, with 3.4 per 10,000 affecting the upper limb [2].

Limb defects are caused by genetic factors, such as chromosomal abnormalities associated with trisomy 18, mutant genes, environmental factors, such as teratogens, a combination of genetic and environmental factors [multifactorial inheritance] vascular disruption and ischemia, as in limb reduction defects [3].

'Thalidomide syndrome' was caused by ingestion of drug thalidomide during the sensitive period of limb morphogesis. The limb malformations are included in major defects such as amelia, phocomelia as well as minor deformities such as hypoplasia of the thumb and syndactyly. Sometimes, it was accompanied by other defects such as anotia, duodenal stenosis and cardiac defects [4].

Typically the symptoms of phocomelia syndrome are undeveloped limbs, mental deciencies, and craniofacial anomalies. In severe cases encephalocele, hydrocephalus, bicornuate uterus, malformations in kidney and heart are found [1]. The diagnosis relies upon cytogenetic testing in peripheral blood of individuals with suggestive clinical findings [5]. This was a case of phocomelia syndrome with a difference that no associated craniofacial anomalies were found.

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