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Research Article

A Rare Case Of Ring Chromosome 18

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ABSTRACT

Ring chromosome is an exceptionally rare condition wherein the ends of the chromosome break and result in abnormal fusion of the chromosome. This loss in genetic material results in abnormal growth and development in a child and can result in multi system involvement and dysfunction. Herein we report a case of a 12 year old female child who was brought with complaints of significant developmental delay in all domains when upon further investigation revealed a chromosomal abnormality: 46, XX, r(18)(11.3q 23), indicative of a ring chromosome 18 abnormality.

Keywords: Ring chromosome 18, developmental delay, chromosomal abnormality.

Introduction: -

A ring chromosome is an exceptionally uncommon chromosomal disorder, rarely encountered in clinical practice and is an uncommon chromosomal abnormality typically arising spontaneously (de novo). This anomaly arises from the deletion of both ends of a chromosome, leading to the formation of a circular or ring-shaped structure. The break in the chromosome results in the loss of genetic material from the terminal ends.The formation of a ring chromosome occurs through the fusion of both ends of a chromosome due to deletion. (1,9)

Ring chromosomes have been documented across all human chromosomes, with an estimated frequency ranging from 1 in 30,000 to 1 in 60,000. Notably, nearly 50 percent of all ring chromosomes derive from acrocentric chromosomes. Intriguingly, 90 percent of these cases stem from maternal origins, as ring chromosomes can induce abnormalities in spermatogenesis, leading to infertility in affected men.(2)

The clinical manifestations vary depending on the extent of chromosomal deletion. In the case of ring 18 syndrome, distinct features include severe mental and growth retardation, microcephaly, brain and ocular malformations, hypotonia, and other skeletal abnormalities.ends. The clinical manifestations of this disorder vary, contingent upon the amount of genetic material lost during the deletion.

Studies have shown that individuals with a ring chromosome 18 defect typically exhibit features such as intellectual disability, brain malformations, microcephaly, hypertelorism, and associated skeletal abnormalities, contributing to short stature and hypotonia (3,4). Additionally, some affected children may present with immunological abnormalities.

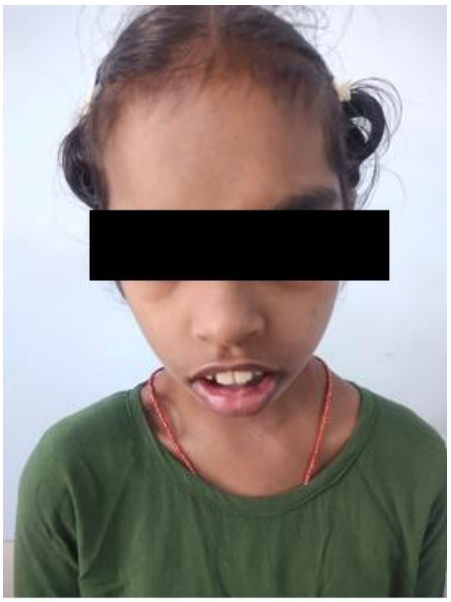
It is noteworthy that this condition is usually de novo, meaning it spontaneously arises and is not inherited from parents. The exact cause of this condition remains unidentified. Interestingly, most reported cases have traced the origin of the defect to the maternal side.

Case report: -

A 12-year-old female, the first child of a non-consanguineous marriage, presented with concerns about delayed development in all domains. The parents reported feeding difficulties since the age of 1 and noticed excessive limb flaccidity. The antenatal, natal, and postnatal periods were uneventful with no complications or neonatal intensive care unit (NICU) admission. The child's immunizations were up to date. There was postnatal history of delay in attainment of age appropriate milestones involving all domains (Gross motor , Fine motor, Language and Social). Development quotient was calculated and found to be 34%,43%,50% and 46% respectively.

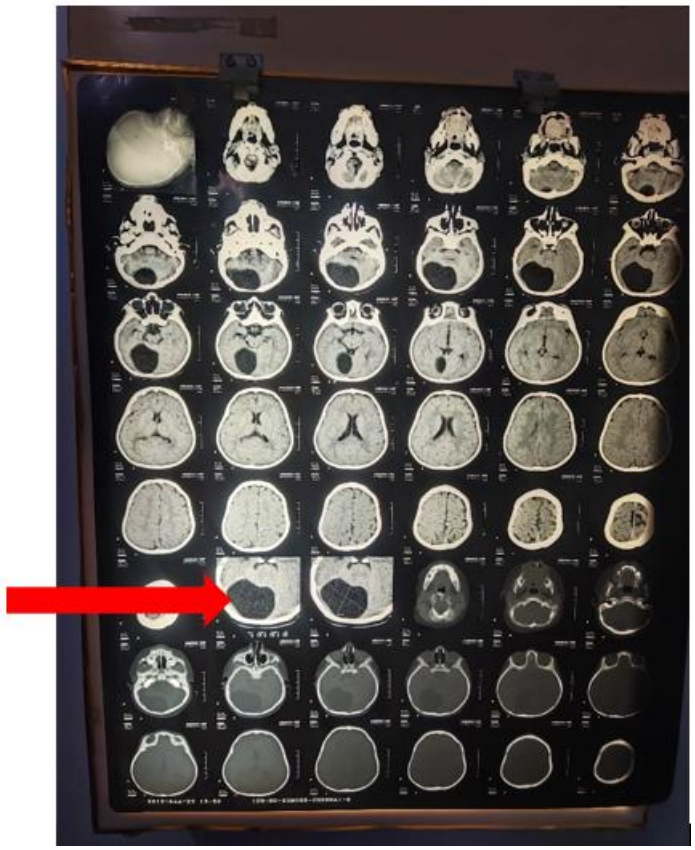
Upon examination, anthropometric measurements indicated height, weight, and height-for-weight ratios below the 3rd percentile, suggestive of short stature and undernutrition. The child exhibited dysmorphic facial features, including microcephaly (Figure 1), mild to severe mental retardation, hypertelorism, epicanthic folds (Figure 2), micrognathia, and small hands (Figure 3). Hypotonia and foot deformation were also observed.
CT brain which was done revealed a large arachnoid cyst in the left posterior cranial fossa measuring 5X3.5cm in size (Figure 4) with no complications of hydrocephalus or meningitis.
Cardiac workup done which included Echocardiography and electrocardiography were found to be normal.
Karyotyping was considered as a possibility of genetic cause was considered from the above clinical manifestations and dysmorphic features and as initial blood investigation reports were found to be inconclusive.
Karyotyping conducted on a venous blood sample revealed a chromosomal abnormality: 46, XX, r(18)(11.3q 23), indicative of a ring chromosome 18 abnormality(Figure 5). Karyotypes were assigned according to the International System of Human Cytogenetic Nomenclature (ISCN) 2005.Further genetic analysis showed the mother had a normal karyotype with an increased length of the satellite short arm of chromosome 22 (Figure 6). In contrast, the father's genetic analysis was normal. In the family history it was disclosed that the mother was mute and now deceased , with no other significant history or findings.(5,6)

(Figure 1)



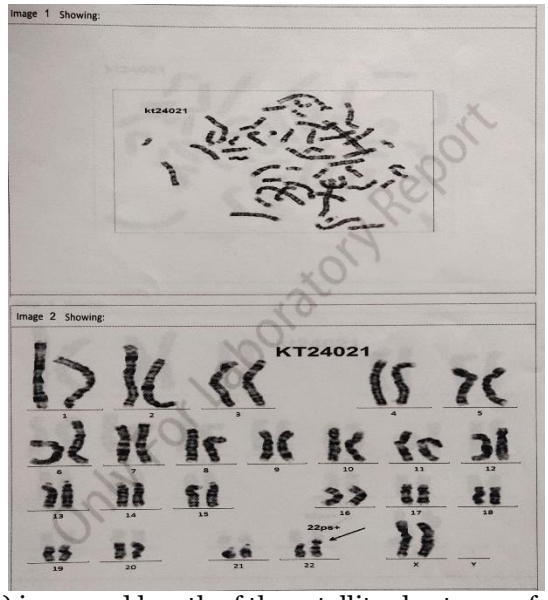
(Figure 2)

(Figure 3)
Short 5 th metacaral



(Figure 4)
Arachnoid cyst in the left posterior fossa measuring 5x3.5cm

(Figure 5) karyotyping -46, XX, r(18)(11.3q 23) ring chromosome 18



(Figure 6) increased length of the satellite short arm of chromosome 22

Discussion: -

In ring chromosome 18, both ends of the chromosome undergo a break, followed by fusion at these ends. (7)This process leads to the loss of genetic material from the broken ends, resulting in the clinical manifestations mentioned earlier. The karyotyping conducted on our patient revealed a chromosomal abnormality, specifically 46, XX, r(18)(11.3q 23), indicative of ring chromosome 18.(8,)

Literature reports include a case of mosaic ring chromosome 18 documented in Iran. Most of the cases reported worldwide are those reported among adults and less among the pediatric population. Until 2011, a total of 70 cases of ring chromosomes had been reported.

Shermineh Heydari et al. reported a case involving a 2.5-year-old male child with cleft lip, club foot, mild mental retardation, and undescended testicles (cryptorchidism), all associated with ring chromosome 18.(9) Chromosomal analysis was instrumental in identifying the chromosomal abnormality in this case as well.

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