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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI: 10.21474/IJAR01/19694
DOI URL: http://dx.doi.org/10.21474/IJAR01/19694

RESEARCH ARTICLE

GENETIC INSIGHTS INTO 3-M SYNDROME: A NEONATAL CASE REPORT

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Manuscript Info

Manuscript History
Received: 19 August 2024
Final Accepted: 22 September 2024
Published: October 2024

Abstract

3-M syndrome is an uncommon autosomal recessive disorder. We present a case of a one-day-old infant with low birth weight, an enlarged head, and shortened limbs, initially suspected of skeletal dysplasia. Genetic testing confirmed the diagnosis of 3-M syndrome.

Introduction:-

3-M syndrome (OMIM 273750) is a rare genetic disorder inherited in an autosomal recessive pattern, characterized by short stature, distinct facial features, and skeletal anomalies [1]. The syndrome is named after three researchers—Miller, McKusick, and Malvaux—who were the first to describe it. Affected individuals typically experience intrauterine growth restriction (IUGR) and postnatal growth challenges, which can be exacerbated by early-life feeding difficulties. Mutations in the CUL7 gene are responsible for 3-M syndrome [2]. Since CUL7 is crucial for the growth and proliferation of chondrocytes, impaired cell division during early gestation may lead to growth retardation in affected individuals.

Case Presentation:

The patient, the child of a second-degree consanguineous marriage, had a history of a prior pregnancy that was terminated due to prenatal findings suggestive of skeletal dysplasia. At birth, the baby presented with a low birth weight, a disproportionally large head, shortened limbs, abdominal distension, and bluish discoloration of both lower limbs. Based on these clinical features and the family history, skeletal dysplasia was suspected. Genetic testing confirmed the presence of a CUL7 gene mutation, establishing the diagnosis of 3-M syndrome.

Discussion:-

In cases of 3-M syndrome, prenatal ultrasound may reveal growth retardation or other characteristic features. In our case, the diagnosis was made shortly after birth through a combination of clinical evaluation, which identified key physical findings such as low birth weight, short stature, and skeletal anomalies, and genetic testing confirming the diagnosis.

Conclusion:-

The presence of low birthweight, large head compared to body and small extremities, short stature and significant past history raised suspicion of skeletal dysplasia. Thus physician should keep high of suspicion.

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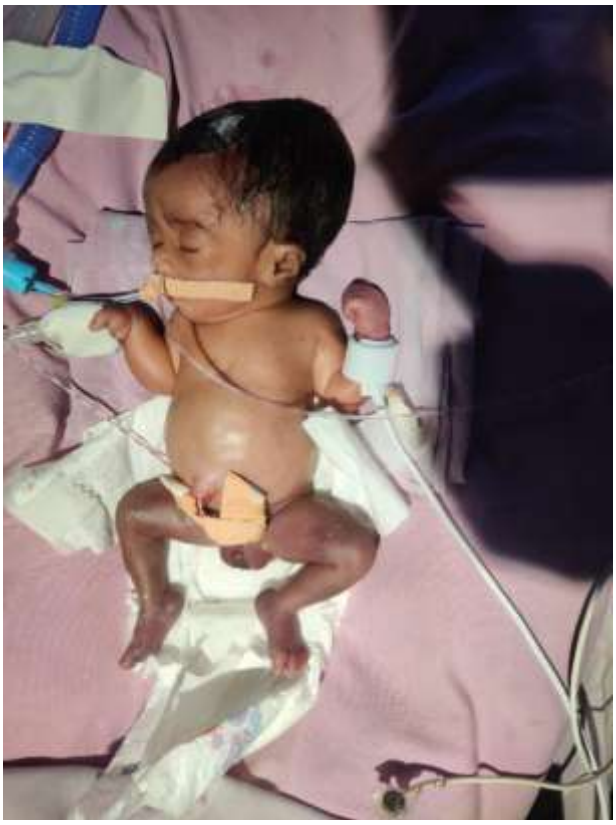

Figure 1:- Showing large head and short extremities.

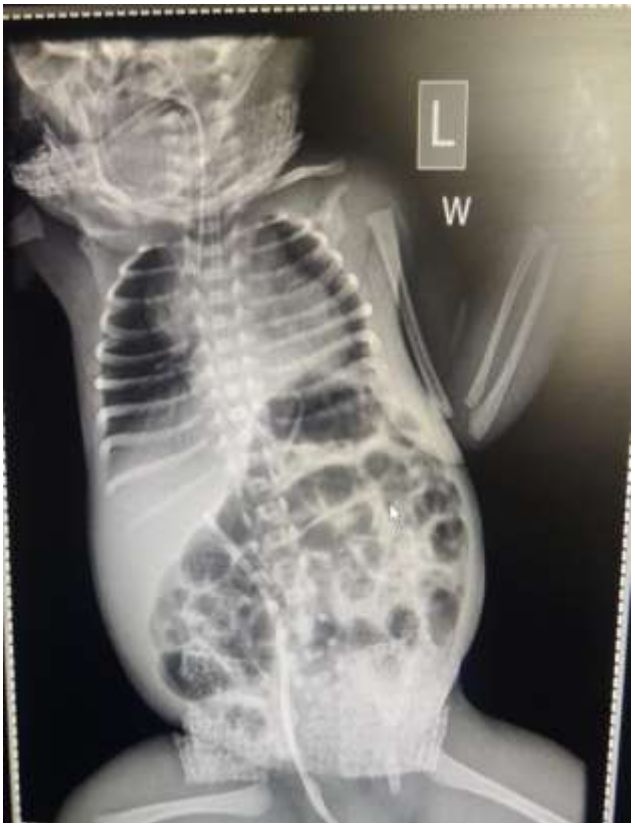

Figure 2:- Chest with abdomen x ray.

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