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Duplication 5q Syndrome in an 11-days-old Male Infant from Dernah, Libya: Case Report

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Abstract **Case Report**

Duplication 5q syndrome (dup 5q) is a rare genetic disorder characterized by the duplication of a specific region on long arm of chromosome 5. This case report describes an 11-day-old male infant from Dernah, Libya, who was diagnosed with dup 5q syndrome based on clinical presentation and karyotype testing. The infant presented with dysmorphic features, low set ears, closed eyes, abnormal genitalia and a left foot deformity. Karyotype analysis revealed the diagnosis of dup 5q syndrome. The infant received supportive care and was referred to a multidisciplinary team for further assessment and planning. This case highlights the importance of early recognition and diagnosis of dup 5q syndrome, as well as the need for multidisciplinary care and support for infants with this disorder. Additionally, it emphasizes the importance of ongoing research to improve the understanding, diagnosis, treatment, and long-term outcomes for individuals with dup 5q syndrome.

Keywords: Duplication 5q syndrome, chromosome 5, genetic disorder, dysmorphic features, Dernah, Libya.

INTRODUCTION

Duplication 5 syndrome (dup5) is a rare genetic condition resulting from the duplication of a specific segment on chromosome 5, leading to a diverse array of physical, developmental, and cognitive difficulties. The global prevalence of dup5 syndrome is estimated at 1 in 20,000 individuals (De Gregori *et al*., 2007). However, there is a lack of comprehensive data regarding the occurrence of this disorder in the specific region of Derna, located in the eastern part of Libya within the historical area of Cyrenaica.

In 2006, Chen and his collogues present a clinical report on an 11-year-old girl with a direct duplication of chromosome 5q35.2-q35.3. The girl exhibited microcephaly, moderate mental retardation, short stature, strabismus, brachydactyly, and facial dysmorphism. This direct duplication of 5q35.2-q35.3 was paternally derived and correlated with the observed phenotype, providing insights into the gene dosage effect of the NSD1 gene causing a reversed phenotype of microcephaly and short stature. The study highlights the association of this duplication with specific clinical features and emphasizes the importance of understanding the genetic mechanisms underlying such chromosomal abnormalities (Chen *et al*., 2006).

The genetic basis of dup5 syndrome lies in the duplication of a specific region on chromosome 5, known as 5q13.3 (Tinoco *et al*., 2021). This region contains several genes, including MEF2C, which is thought to play a critical role in brain development (Nowakowska, 2017, Tinoco *et al*., 2021). The duplication of this region can disrupt the normal function of these genes, leading to the various features associated with dup5 syndrome (Tinoco *et al*., 2021).

Generally speaking, the clinical presentation of dup5 syndrome can vary significantly between individuals. Some common features include: developmental delays, intellectual disability, behavioral problems, physical abnormalities. Some individuals with dup5 syndrome may have physical abnormalities, such as cleft lip/palate, heart defects, face and skeletal malformations (Shaffer *et al*., 2007, Mefford *et al*., 2008).

The diagnosis of dup5 syndrome is typically made through genetic testing. This may involve

karyotyping, which can detect large duplications, or more specific tests such as fluorescence in situ hybridization (FISH) or chromosomal microarray analysis (CMA). These tests can identify the presence of the duplication on chromosome 5 and confirm the diagnosis (Cheung *et al*., 2005).

There is no cure for dup5 syndrome. However, there are a variety of interventions that can help manage the symptoms and improve the quality of life for individuals with this condition. These interventions may include; early intervention services, special education, behavioral therapy and some medications: for treating specific symptoms, such as hyperactivity or aggression (Shaffer *et al*., 2007, Mefford *et al*., 2008). Future research has the potential to lead to significant improvements in the diagnosis, treatment, and long-term outcomes for individuals with dup5 syndrome. This includes the development of targeted therapies, more accurate diagnostic tools, and improved treatment options. Additionally, ongoing research will help us better understand the long-term outcomes of dup5 syndrome and develop strategies to optimize the health and well-being of individuals with this condition (Tinoco *et al*., 2021).

Case Report

The patient is an 11 days old male infant from Dernah, Libya. He was born at term via cesarean delivery to non-consanguineous parents. The pregnancy was uncomplicated, and the infant's birth weight and length were within normal limits. The mother has been pregnant a total of two times, has given birth to two normal children after 24 weeks, and she has not experienced any spontaneous or elective abortions.

At this birth, the infant was noted to have several dysmorphic features, including a low set ear, cloesed eyes, abnormal genitalia and left foot deformity (equinovalgus). He also had octopic kidney and dry skin.

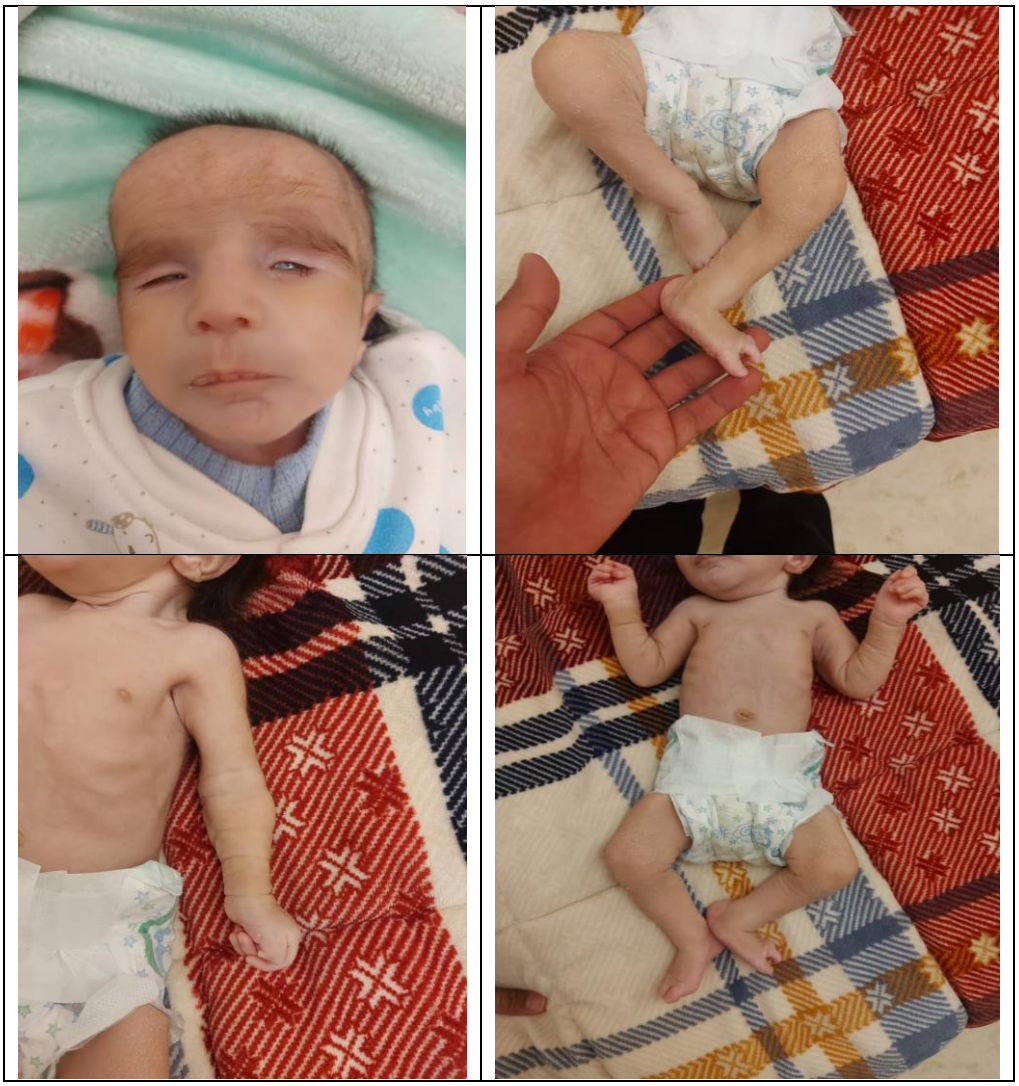

Figure 1: Case of dup5 syndrome

Genetic Testing:

Chromosomal karyotype was performed, which revealed a duplication of the 5q13.3 region. This finding needs to be confirmed by further investigations such chromosomal microarray analysis or FISH.

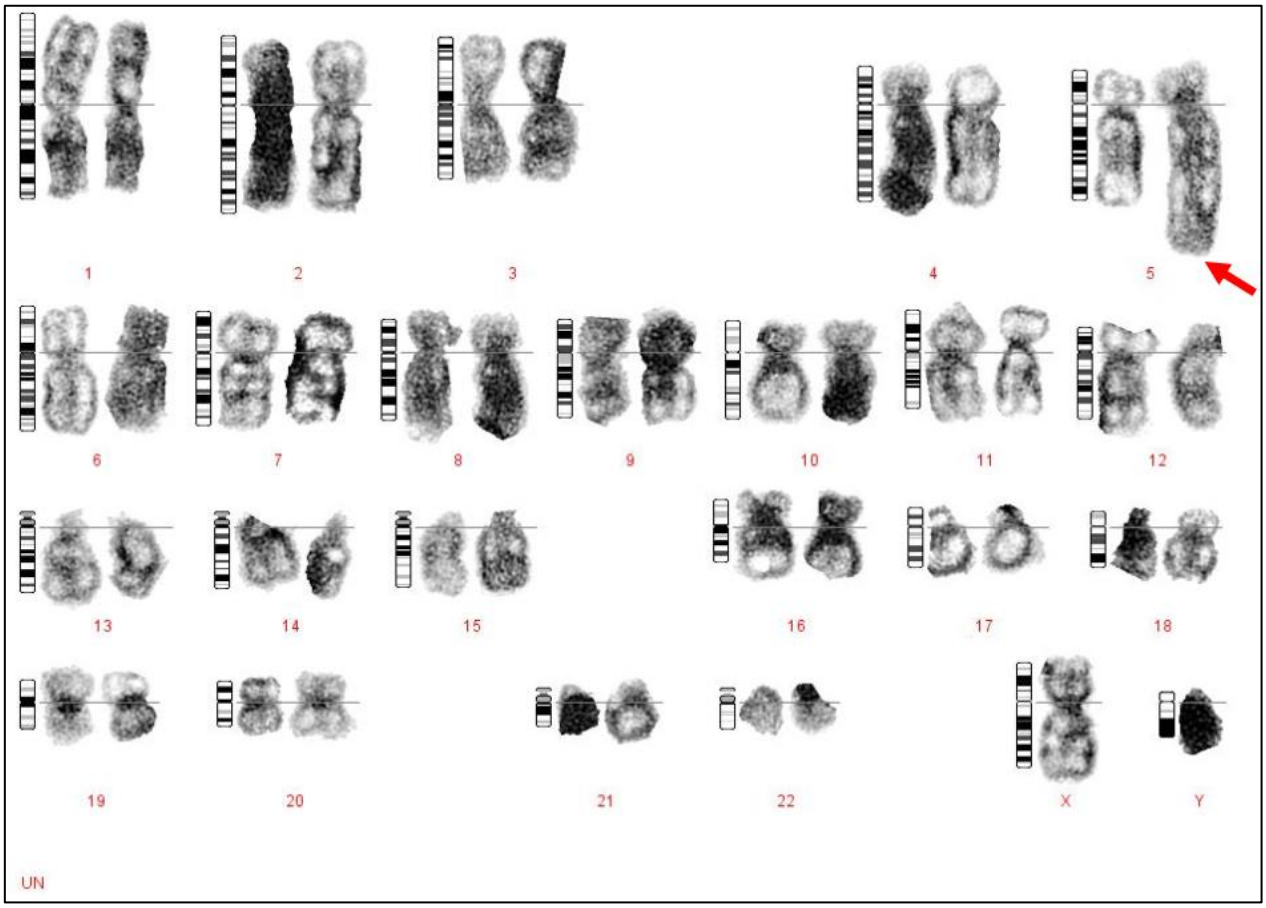

Figure 2: Karyotype result; red arrow indicates to the position of dup5

Initial Management:

The infant was admitted to the neonatal intensive care unit for further evaluation and management. He received supportive care, including nutritional support and respiratory support. Additionally, he was referred to a multidisciplinary team for further assessment and planning.

DISCUSSION

This case report sheds light on the real-world implications of this rare genetic disorder. The infant's presentation with dysmorphic features such as a low set ear, closed eyes, abnormal genitalia, left foot deformity (equinovalgus), ectopic kidney, and dry skin aligns with the characteristic phenotypic spectrum associated with dup5 syndrome. These physical anomalies are consistent with the diverse range of clinical features observed in individuals with chromosomal duplications involving chromosome 5, emphasizing the complexity and variability of the disorder. These infant's dysmorphic features prompted further investigation, leading to the identification of the genetic cause. Early diagnosis is crucial for ensuring appropriate management and support for the infant and family. The prevalence of dup5 syndrome generally is estimated to be 1 in 20,000 individuals globally, highlighting its rarity (De Gregori *et al*., 2007). However, there is a lack of data on the prevalence of this disorder in specific regions, such as Dernah, Libya, underscoring the need for further research and awareness.

The case report of an 11-day-old male infant from Dernah, Libya, sheds light on the real-world implications of this rare genetic disorder. The infant's presentation with dysmorphic features such as a low set ear, closed eyes, abnormal genitalia, left foot deformity (equinovalgus), ectopic kidney, and dry skin aligns with the characteristic phenotypic spectrum associated with dup5 syndrome. These physical anomalies are consistent with the diverse range of clinical features observed in individuals with chromosomal duplications involving chromosome 5, emphasizing the complexity and variability of the disorder. However, there is a lack of data on the prevalence of this disorder in specific regions, such as Dernah, Libya, underscoring the need for further research and awareness.

The genetic basis of dup5 syndrome, as highlighted in the literature, involves the duplication of

specific regions on chromosome 5, such as 5q13.3, which contains genes critical for brain development like MEF2C (Tinoco *et al*., 2021). The disruption of these genes due to the duplication leads to the observed features associated with dup5 syndrome. The case report's description of dysmorphic features and developmental anomalies underscores the importance of genetic testing and detailed phenotypic assessment in diagnosing and managing individuals with dup5 syndrome.

Moreover, the comparison of the infant's clinical presentation with previously reported cases of chromosomal duplications involving chromosome 5, as discussed in the literature, provides valuable context for understanding the variability and complexity of dup5 syndrome. The identification of dysmorphic features, developmental delays, and physical abnormalities in the infant aligns with the broader clinical spectrum associated with chromosomal duplications on chromosome 5, emphasizing the need for comprehensive genetic evaluation and multidisciplinary care for affected individuals.

Compared to many previous studies on chromosome 5 duplications, this study adds valuable clinical insights by reporting additional features not commonly associated with dup5 syndrome, such as low-set ears, closed eyes, abnormal genitalia, and a left foot deformity (equinovalgus) (Chen *et al*., 2006, Tinoco *et al*., 2021). These novel clinical characteristics provide a more complete picture of the diverse phenotypic spectrum associated with dup5 syndrome. The identification of these additional clinical features, in conjunction with the detailed genetic analysis, can contribute to a better understanding of the underlying mechanisms and the genotype-phenotype correlations in dup5 syndrome. This knowledge can ultimately lead to improved diagnostic approaches and more tailored management strategies for individuals affected by this rare genetic disorder.

Continued research on dup5 syndrome is essential to improve the understanding of the condition, develop new diagnostic tools, and identify potential treatment options. This research will ultimately lead to improved outcomes for individuals with dup5 syndrome.

CONCLUSION

In conclusion, the case report of the 11-day-old male infant from Dernah, Libya, exemplifies the challenges and complexities associated with dup5 syndrome. The infant's clinical presentation underscores the importance of early diagnosis, genetic testing, and tailored interventions to address the diverse clinical manifestations of this rare genetic disorder. By integrating clinical observations with genetic insights, healthcare providers can enhance the management and care of individuals with dup5 syndrome, ultimately striving to optimize their health and well-being.

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