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Case Report

Cri du Chat Syndrome in Pediatric Patient: A Case from Jordan as Model Review Article. Genotype-Phenotype Correlations and Recommendations for Clinical Management

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Abstract

Rare syndrome caused by a deletion of the short arm of chromosome 5 (5p-). The main clinical features include a high-pitched cry, facial asymmetry, and microcephaly, round face at birth, epicanthic folds, hypotonic, delayed growth and development.

As a workout description, a new genomic region associated with microcephaly and cat-like cry and highlight the importance of precise delineation of 5p deletion breakpoints and detection of other CNVs in CdCS patients to improve genotype-phenotype correlation to perform a complete clinical and molecular diagnosis.

Keywords: *Cri du Chat Syndrome; Genotype-Phenotype Correlations; Chromosome 5 (5p-)*

Case Presentation

Case from Jordan

A 15 years young girl from Jordan was born in Madaba city in Jordan from a healthy woman and healthy Man.

Unfortunately, through the pregnancy period, the woman has been misdiagnosed with polyhydramnios and sever amniotic fluid through the last trimester and how the obstetrician failed to follow up the patient in the 3rd trimester and generally at the time of control pregnancy.

After birth, the signs and symptoms began to be clearer, at 2 months age, cryptic genomic alterations were uncovered, offering new insights into the genotype-phenotype relationship. In this study, we report the involvement of a previously unrecognized genomic region linked to microcephaly and the characteristic cat-like cry. Our results highlight the critical role of accurately mapping the 5p deletion boundaries and identifying coexisting for additional copy number variations (CNVs) in individuals with Cri-du-chat syndrome. Such comprehensive analysis enhances the precision of genotype-phenotype correlations and supports a more complete clinical and molecular assessment [1].

We report a female infant presenting with a partial deletion of the short arm (5p) and a partial duplication of the long arm (5q) of chromosome 5, resulting from a paternal per centric inversion.

Her karyotype was identified as 46, XY, rec (5) dup q, inv(5)(p15).

The case involving the 1q35.1 region of paternal origin is presented alongside the observed oral manifestations. The overall clinical presentation was primarily consistent with Cri-du-chat syndrome.

The patient exhibited delayed mental and motor development, along with microcephaly and congenital heart defects. Additional features included a distinct cry and facial characteristics typical of Cri-du-chat syndrome.

Minor perioral and intraoral findings included thin upper lip, down-turning corners of mouth, micrognathia, shallow palate, without cleft of soft palate.

Gene description

Humans normally have 46 chromosomes in each cell, divided into 23 pairs.

Chromosome 5 is present in two copies in each cell, with one copy inherited from the mother and the other from the father.

Covering approximately 181 million base pairs, chromosome 5 accounts for nearly 6% of the entire human genomic DNA content [2].

The discovery and characterization of genes located on each chromosome remains a dynamic and ongoing focus in genetic research.

Estimates of gene numbers on each chromosome can differ, as researchers employ various methodologies to make these predictions.

It is estimated that chromosome 5 harbors approximately hundreds genes responsible for coding proteins. These proteins perform a variety of different roles in the body.

Methods

Genomic array analysis is used in patients with CdCS to more accurately determine the breakpoints on 5p and to identify copy number variations (CNVs) that may influence the clinical features of the syndrome.

Pathophysiology and genetic embryology

A notable female predominance observed in the affected population. In just over 10% of families, parental chromosomal rearrangements such as inversions were identified, whereas less common abnormalities-such as isodicentric chromosomes, small supernumerary marker chromosomes, and chromosomal insertions-observed in fewer than 10% of cases.

The number of deciduous teeth and their eruption timing appeared normal, with no deviations observed.

In humans, each cell typically contains 46 chromosomes, organized into 23 pairs [3].

Conclusion

With advancements of molecular cytogenomic methods in the last two decades, it was possible to evidence cryptic alterations and improve the genotype-phenotype correlation.

As a workout description, a new genomic region associated with microcephaly and cat-like cry and highlight the importance of precise delineation of 5p deletion breakpoints and detection of other CNVs in CdCS patients to improve genotype-phenotype correlation to perform a complete clinical and molecular diagnosis [4,5].

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