CASE REPORT

Case of a Floppy Infant: Contiguous Gene Syndrome

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ABSTRACT

Case of a 29-day-old male baby having hypotonia and respiratory distress, which on further evaluation came to be diagnosed as contiguous gene syndrome (15q11–13 gene deletion).

Key words: Hypotonia, malformations, genetic or metabolic disorders

INTRODUCTION

Hypotonia is characterized by unusually low muscle tone or diminished resistance to passive, quick movements. It can be caused by various factors affecting central neural function, such as brain injuries, malformations, genetic or metabolic disorders, trauma, anatomical issues, or unknown (idiopathic) reasons. Central hypotonia can be widespread, affecting the limbs, trunk, and neck, or it can be localized, where some areas of the body exhibit low muscle tone whereas others may have normal or increased tone.[1]

CASE REPORT

A 29-day-old male baby came with complaints of hypotonia and respiratory distress. Birth history – full term, cesarean section delivery (i/v/o uteroplacental insufficiency) with a birth weight of 1.8 kg, and required resuscitation at birth. Physical examination – stridor, weak cry, generalized hypotonia. Systemic examination – suprasternal, sub-xiphoid, subcostal retractions, and bilateral crepitations. On admission, taken on continuous positive airway pressure. Started on orogastric tube feeds, continued for 15 days. Special investigations were done, which were as follows: Total leucocyte count – 14,300/L, Sr. Calcium – 9.3 mg/dL, Sr. Sodium – 131 mmoL/L, Sr. Potassium – 4.6 MmoL/L. Electroencephalogram – right frontocentral and frontotemporal epileptiform activity whole exome sequencing-deletion, overlaps with 15q11.2–q13 microdeletion corresponding to Angelman syndrome.

RESULTS

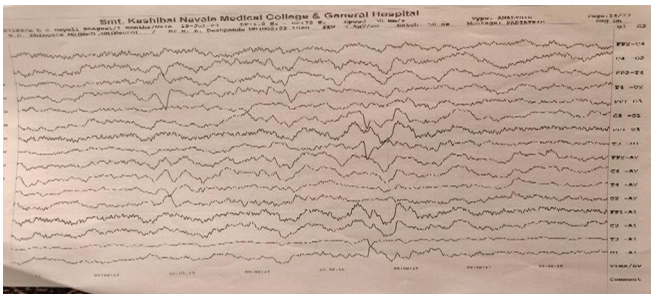
LIKELY PATHOGENIC COPY NUMBER VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

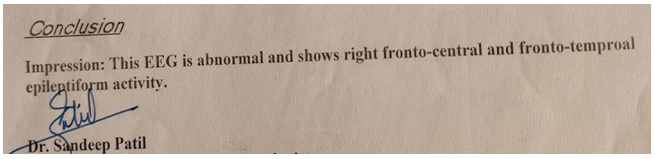
SNV(s)/INDELS

No significant SNV(s)/INDELS for the given clinical indications that warrants to be reported were detected.

Copy Number Variants CNV(s)

Variant	Zygosity	Size (KB)	Disease (OMIM)	Inheritance	Classification[5]
chr15:g.(?_23447098)_(28299516_?)del	Heterozygous	4852.42	-	-	Likely Pathogenic





The baby was discharged on oral feeds with weight gain and no respiratory support.

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DISCUSSION

Three distinct neurodevelopmental disorders are primarily associated with deletions or duplications at the 15q11–q13 locus: Prader–Willi syndrome, Angelman syndrome, and 15q11–q13 duplication syndrome.[2] Angelman syndrome is characterized by developmental delays, intellectual disability, lack of speech, seizures, an unsteady gait, a cheerful demeanor, and distinctive facial features. Management typically involves speech, occupational, and physical therapy, along with dietary and nutritional support, and ophthalmologic evaluations for conditions such as strabismus and hemianopia. Family counseling is recommended.

CONCLUSION

Contiguous gene syndrome (15q11–13 gene deletion) is a rare cause of hypotonia presenting early in life.

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