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DEPARTMENT OF GENOMICS AND MOLECULAR DIAGNOSTICS

Patient Name	Dr. PRAPHUL DIPANKAR	Visit ID	0482312020174	Registration Date	03-12-2023 12:29 PM
Barcode	25057177	Sample Type	WHOLE BLOOD EDTA	Reported Date	22-12-2023 02:02 PM

Targeted Sanger Sequencing

Referral Reason

Familial genetic testing for the targeted *AFF2* gene variant reported in the Index baby.

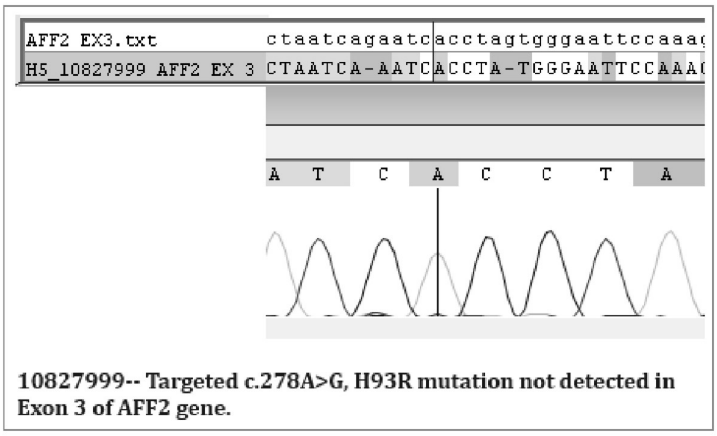
Test Result

Targeted *AFF2* gene variant is not detected.

Gene	Location	Variant	Zygosity	Inheritance
AFF2 ENST00000370460.7	Exon 3	c.278A>G,H93R	**Not Detected**	X-Linked Recessive

Interpretation

The Sanger analysis identified,normal (wild type) for the tested targeted AFF2 gene mutation in this individual.



10827999-- Targeted c.278A>G, H93R mutation not detected in Exon 3 of AFF2 gene.

Recommendations

- Genetic counseling is recommended.

Analyzed by

Kanaka Durga Devi.Y

Approved by

Kanaka Durga Devi.Y
Technical Lead -Molecular Diagnostics.

A. Radhika
Co-Head Genomics

Dr. NAUSHAD
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Targeted Sanger Sequencing

Methodology

PCR followed by bi-directional Sanger Sequencing was performed from the isolated DNA to identify the targeted variant in the AFF2 gene.

Clinical Information

Intellectual Developmental Disorder, X-Linked 109, also known as mental retardation, x-linked, fraxe type, is related to fraxe intellectual disability and fragile x syndrome, and has symptoms including agitation An important gene associated with Intellectual Developmental Disorder, X-Linked 109 is AFF2 (ALF Transcription Elongation Factor 2). Related phenotypes are intellectual disability and delayed speech and language development.

Disclaimer

- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- Although all precautions are taken while conducting these tests, there is a standard error rate of approximate 1% in all genetic tests and this should be taken into consideration before any clinical decision [1]
- It is presumed that the specimen used to perform the test belongs to the patient specified above, such verification having been carried out at the collection level of sample.

References

1. Katharina J Hoff (2009): The effect of sequencing errors on metagenomic gene prediction. BMCGenomics, 10:520

***** End Of Report *****

Suggested clinical correlation & follow up

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