MedGenome Labs Ltd.
3rd Floor, Narayana Nethralaya Building, Narayana Health City,
#258/A, Bommasandra, Hosur Road, Bangalore - 560 099, India.
Tel : +91 (0)80 67154989 / 990, Web: www.medgenome.com

MEDGENOME

DNA TEST REPORT - MEDGENOME LABS

Full Name / Ref No:	**AATHMAJA D.**	Order ID/Sample ID:	**913408/8454341**
Gender:	Female	Sample Type:	Blood
Date of Birth / Age:	6 months	Date of Sample Collection:	16th April 2024
Referring Clinician:	Dr. Shyma M M., Sunrise Hospital, Ernakulam	Date of Sample Receipt:	17th April 2024
		Date of Order Booking:	18th April 2024
		Date of Report:	15th May 2024
Test Requested:	**Whole exome sequencing**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Baby *Aathmaja D*, presented with clinical indication of global developmental delay. MRI is suggestive of demyelination. She is suspected to be affected with Leukodystrophy and has been evaluated for pathogenic variants.

RESULTS

LIKELY PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

SNV(s)/INDELS

Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification$
DDX3X **(+)** (ENST00000644876.2)	Exon 15	c.1678_1680del **(p.Leu560del)**	Heterozygous	Snijders Blok type of X-linked syndromic intellectual developmental disorder (OMIM#300958)	X-linked dominant	Likely Pathogenic **(PM1,PM2,PM4,PP5)**
ATP11A **(+)** (ENST00000375645.8)	Exon 1	c.28G>A **(p.Val10Met)**	Heterozygous	Hypomyelinating leukodystrophy-24 (OMIM#619851)	Autosomal dominant	Uncertain Significance **(PM2)**

Parental testing is recommended, and classification of the variant(s) may change based on segregation analysis.

COPY NUMBER VARIANTS CNV(s)

No significant CNVs for the given clinical indications that warrants to be reported was detected.

VARIANT INTERPRETATION AND CLINICAL CORRELATION

VARIANT 1 (*DDX3X* gene):

CAP ACCREDITED