3B-VARIANT

Unique ID: [Unique ID]
3billion ID: [3billion ID]

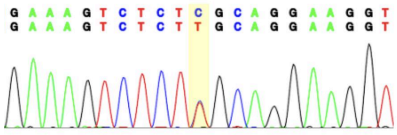
PATIENT INFORMATION

Unique ID	[Unique ID]	**Physician**	[Physician]	**Sample type**	DBS
3billion ID	[3billion ID]	**Department**	Pediatrics	**Collected on**	yyyy-mm-dd
DOB* / Sex	yyyy-mm-dd / Male	**Institution**	[Institution]	**Ordered on**	yyyy-mm-dd
Ethnicity	Latino/Admixed American			**Accessioned on**	yyyy-mm-dd
Relationship	Mother / [3billion ID]				

* (YYYY-MM-DD)

Variant 1: NC_000023.11:g.25013650NGC[23] (GRCh38) — Detected

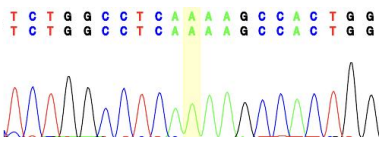
Results	Variant was observed
Gene	*ARX*
cDNA	NM_139058.3:c.298GCN[23] (p.Ala100[23])
Zygosity	Heterozygous
Disease [Inheritance Mode]	Developmental and epileptic encephalopathy 1 (OMIM: 308350)
Classification	Pathogenic



NM_139058.3:c.298GCN[23] (p.Ala100[23])
Chromatogram showing +/-10bp flanking region of the variant breakpoint (vertical line).

Variant 2: 2-210606931-A-G (GRCh38) — Not detected

Results	Variant was not observed
Gene	*CPS1*
cDNA	NM_001875.5:c.2182A>G (p.Lys728Glu)
Zygosity	Not applicable
Disease	Carbamoylphosphate synthetase I deficiency (OMIM:237300)
Classification	VUS



NM_001875.5:c.2182A>G (p.Lys728Glu)
Reverse chromatogram showing +/-10bp flanking region of the variant. The variant position is highlighted in yellow

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METHODS

Genomic DNA was extracted from dry blood spat specimen using standard protocol. PCR primers were designed using Primer3 (v. 0.4.0), (Whitehead institute; http://bioinfo.ut.ee/primer3-0.4.0/)[1,2] and NCBI GenBank reference sequence. PCR amplification and Sanger sequencing were performed following the standard protocol using PCR Master Mix Kit (ThermoFisher Scientific, Waltham, MA, USA) and SeqStudio Genetic Analyzer (Applied Biosystems, Foster City, CA, USA). The sequencing results were manually analyzed using Sequence Scanner Version 1.0 (Applied Biosystems, Foster City, CA, USA).

REFERENCES

1. Untergasser A, Cutcutache I, Koressaar T, Ye J, Faircloth BC, Remm M, Rozen SG (2012) Primer3 - new capabilities and interfaces. Nucleic Acids Research 40(15):e115
2. Koressaar T, Remm M (2007) Enhancements and modifications of primer design program Primer3 Bioinformatics 23(10):1289-91

DISCLAIMER

This test was developed by 3billion in the purpose of identifying single nucleotide variants, small insertions and deletions, and structural variant with breakpoint at a specific genomic position. This test is intended for clinical purposes and should not be regarded as investigational or for research. This laboratory is certified under the Clinical of American Pathologists (CAP#:8750906) and Clinical Laboratory Improvement Amendments (CLIA#: 99D2274041) as qualified to perform high complexity clinical laboratory testing. Limitations for this test include, but are not limited to, false positive findings due to co-amplification of homologous genomic regions and false negative findings from allelic dropout caused by unknown polymorphism(s) within the primer binding region, low-level mosaicism, preferential PCR amplification of the smaller amplicon and structural variants interfering with PCR amplification. This report may not be copied or reproduced, except in its totality.

ACCREDITATIONS AND CERTIFICATIONS

CAP License #	CLIA ID #
8750906, AU-ID# 2052626	99D2274041

This case has been comprehensively reviewed by our clinical team of physicians, geneticists and informaticists.
Report electronically signed by:

Go Hun Seo, M.D, Ph.D.
Chief Medical Officer & Laboratory Director