3B-VARIANT

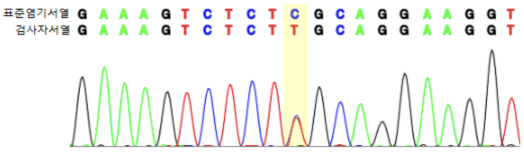
Clinical use

PATIENT INFORMATION

Unique ID	[Unique ID]	**Physician**	[Physician name]	**Sample type**	buccal swab
3billion ID	EPH23-XXXX	**Department**	Pediatrics	**Collected on**	2023-08-25
DOB / Sex	2016-08-08 / Male	**Institution**	[Institution name]	**Ordered on**	2023-08-25
Ethnicity	Latino/Admixed American			**Accessioned on**	2023-08-28

PRIMARY FINDING

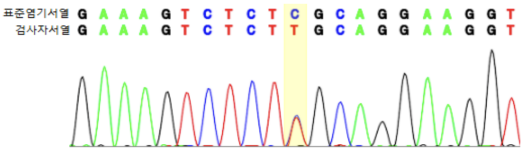
Variant 1: PRPH2(NM_000322.4):c.499G>A (p.Gly167Ser)	**Detected**
Results	Variant was observed
Gene	*HPS6*
cDNA (protein)	NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
Zygosity	Heterozygous
Disease [Inheritance Mode]	#619453 (OMIM ID) [Autosomal dominant]
Classification	Pathogenic



NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))

Forward chromatogram showing +/-10bp flanking region of the variant. The variant position is highlighted in yellow

Variant 1: PRPH2(NM_000322.4):c.499G>A (p.Gly167Ser)	
Results	Variant was observed
Gene	*HPS6*
cDNA (protein)	NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
Zygosity	Heterozygous
Disease [Inheritance Mode]	#619453 (OMIM ID) [Autosomal dominant]
Classification	Pathogenic



NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))

Reverse chromatogram showing +/-10bp flanking region of the variant. The variant position is highlighted in yellow

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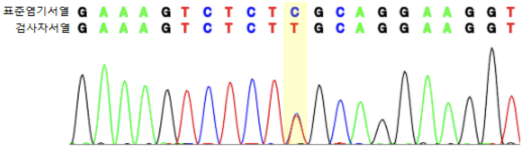
Variant 3: PRPH2(NM_000322.4):c.499G>A (p.Gly167Ser)	Not detected
Results	Variant was observed
Gene	*HPS6*
cDNA (protein)	NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
Zygosity	Heterozygous
Disease [Inheritance Mode]	#619453 (OMIM ID) [Autosomal dominant]
Classification	Pathogenic



NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))

Forward 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 buccal swab sample using AccuBuccal DNA Prep kit(AccuGene, Incheon, Korea).PCR primers were designed using Primer3 (v. 0.4.0, Whitehead institute; http://bioinfo.ut.ee/primer30.4.0/1,2 andNCBI GenBank reference sequence. PCR amplification and Sanger sequencing were performed following the standardprotocol using PCR Master Mix Kit ThermoFisher Scientific, Waltham, MA, USA and SeqStudio Genetic AnalyzerApplied Biosystems, Foster City, CA, USA. The sequencing results were manually analyzed using Sequence ScannerVersion 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 and small insertions and deletions at a specific genomic position. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical of American Pathologists as qualified to perform high complexity clinical laboratory testing (CAP#:8750906). 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