3B-VARIANT

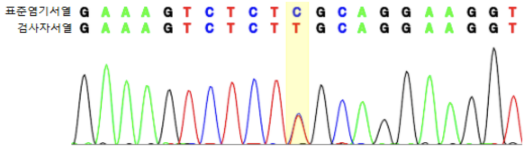
환자 기본 정보 (PATIENT INFORMATION)

Unique ID	[Unique ID]	**담당의**	[당담의]	**샘플 타입**	EDTA tube
3billion ID	EPH23-XXXX	**분과**	Pediatrics	**샘플채취일**	2023-08-25
생년월일 / 성별	2016-08-08 / Male	**의뢰기관**	[의뢰기관]	**검사등록일**	2023-08-25
인종	Latino/Admixed American			**샘플접수일**	2023-08-28

결과 요약 (RESULT SUMMARY)

Variant 1: *HPS6*(NM_000322.4):c.499G>A (p.Gly167Ser) **Detected**

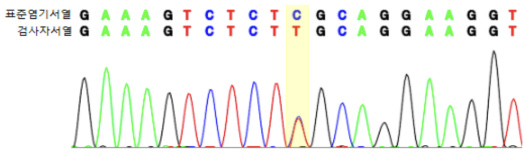
Results	Variant was observed
Gene	*HPS6*
cDNA (protein)	NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
Zygosity	Heterozygous
Disease [Inheritance Mode]	#619453 (OMIM ID) [Autosomal dominant]
Classification	Pathogenic



NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
변이 주변 +/-10bp 생어 크로마토그램 입니다. 변이 위치는 노란색으로 표시되어 있습니다.

Variant 2: *HPS6*(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))
변이 주변 +/-10bp 생어 역상보 크로마토그램 입니다. 변이 위치는 노란색으로 표시되어 있습니다.

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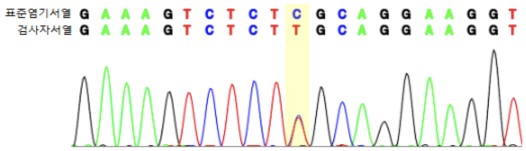
Variant 3: *HPS6*(NM_000322.4):c.499G>A (p.Gly167Ser)	Not detected
Results	Variant was observed
Gene	*HPS6*
cDNA (protein)	NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
Zygosity	Heterozygous
Disease [Inheritance Mode]	#619453 (OMIM ID) [Autosomal dominant]
Classification	Pathogenic



NM_XXXX:c.XXXREF>ALT (NP_XXXX:p.(AAAXXXAAA))
변이 주변 +/-10bp 생어 크로마토그램 입니다. 변이 위치는 노란색으로 표시되어 있습니다.

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검사방법 (METHODS)

표준작업지침서에 따라 EDTA tube를 사용하여 채취된 전혈 검체에서 genomic DNA를 추출했습니다. Primer3 (v. 0.4.0), (Whitehead institute; http://bioinfo.ut.ee/primer3-0.4.0/) [1,2] 및 NCBI GenBank 참조 서열을 사용하여 PCR primer를 디자인 하였습니다. 표준작업지침서에 따라 PCR Master Mix Kit (ThermoFisher Scientific, Waltham, MA, USA), ExoSAP-IT™ PCR Product Cleanup Reagent (ThermoFisher Scientific, Waltham, MA, USA), BigDye™ Terminator v3.1 Cycle Sequencing Kit (ThermoFisher Scientific, Waltham, MA, USA)와 SeqStudio Genetic Analyzer (Applied Biosystems, Foster City, CA)를 사용하여 PCR amplification과 Sanger sequencing를 수행하였습니다. 시퀀싱 결과는 Sequence Scanner Version 2.0 (Applied Biosystems, Foster City, CA, USA)를 이용하여 분석하였습니다.

참고문헌 (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

면책조항 (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

이 레포트는 의사, 유전학자, 정보학자로 구성된 임상팀이 종합적으로 검토했습니다.
Report electronically signed by:

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