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Full name:	Familial Hypercholesterolemia panel (9 genes) - ULG
Abbreviation:	ADRH panel
Description:	Autosomale dominant and recessive forms of familial Hypercholesterolemia. Investigation of : - coding sequence of APOB, APOE, LDLR (+5'UTR), PCSK9, LDLRAP1, LIPA, ABCG5, ABCG8 and STAP1 by NGS - deletion or duplication in LDLR using Multiplex ligation-dependent probe amplification (MLPA - MRC Holland) https://www.chuliege.be/jcms/c2_17345770/fr/genetique/formulaires-utiles (go to Panel NGS and Descriptif général des gènes du panel ADRH)
Type of panel:	Custom panel
Provider:	MASTR kit
Quality coverage:	99,91% at 30X for the whole panel (but 100% at 30X for core genes : LDLR - PCSK9 - APOE - APOB exons 26-29)
Laboratory:	Centre de Génétique Humaine - CHU Sart-Tilman
Created:	04 Jul 2019 - 14:21
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Related Diseases

- Cholesteryl ester storage disease
- Heterozygous familial hypercholesterolemia (NON RARE IN EUROPE)
- Homozygous familial hypercholesterolemia
- Sitosterolemia

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ABCG5	100.00	0	only for coding exons and intronic borders +/-14pb
ABCG8	100.00	0	only for coding exons and intronic borders +/-14pb
APOB	99.79	0	only for coding exons and intronic borders +/-14pb Exons 26 and 29 are entirely covered at 30x Exon 1 is not covered
APOE	100.00	0	only for coding exons and intronic borders +/-14pb
LDLR	100.00	1	only for 5'UTR + coding exons + intronic borders +/-14pb
LDLRAP1	98.69	0	only for coding exons and intronic borders +/-14pb exon 1 is not covered
LIPA	100.00	0	only for coding exons and intronic borders +/-14pb
PCSK9	100.00	0	only for coding exons and intronic borders +/-14pb
STAP1	100.00	0	only for coding exons and intronic borders +/-14pb