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16 April 2024

Mrs Takara Raymond
4 Chester Court
PETRIE QLD 4502

Dear Takara & Travis

RE: Mrs Takara Raymond DOB: 18/6/1991 & Travis Harvey DOB: 19/4/1990
4 Chester Court, Petrie QLD 4502

Your recent Carrier Gene Screening has shown that you have no abnormal genes in common and so that certainly lessens the risk of you having a child with an abnormality (but it obviously does not exclude it completely).

I wish you all the best.

Kind regards

Yours sincerely

.....
Dr Scott Salisbury

EXTENDED CARRIER SCREEN
DUO CARRIER SCREEN

SPECIMEN TYPE: Blood

RESULT FOR FEMALE PARTNER:
- **Name: TAKARA RAYMOND**
- **Date of birth: 18/06/1991**
- VHSD lab ID: 24-1917336
- CFTR (cystic fibrosis): No variants detected
- SMN1 (spinal muscular atrophy): 2 or more gene copies detected
- HBB, HBA1, HBA2 (alpha/beta thalassemia): No variants detected
- FMR1 (fragile X syndrome): 23 and 29 CGG repeats detected
- X-linked genes: No variants detected
- All other tested genes: Please see Interpretation

RESULT FOR MALE PARTNER:
- **Name: TRAVIS HARVEY**
- **Date of birth: 19/04/1990**
- VHSD lab ID: 24-1917343
- CFTR (cystic fibrosis): No variants detected
- SMN1 (spinal muscular atrophy): 2 gene copies detected
- HBB, HBA1, HBA2 (alpha/beta thalassemia): No variants detected
- All other tested genes: Please see Interpretation

INTERPRETATION:
This couple is at LOW RISK of having offspring affected by the tested conditions. A small residual risk of having affected offspring remains. This is the lowest risk result a couple can receive from this screening test.

Unless otherwise specified, carrier states are only reported when both partners are carriers of the same condition, when the female partner is a carrier of an X-linked condition, or for selected conditions where a result has health implications for carriers themselves. The stated reproductive risk is therefore specific to this reproductive couple and the offspring they have together; if this coupling changes, repeat carrier screening with any new reproductive partners is recommended.

TEST INFORMATION:
Autosomal genes tested in both male and female partners (n=361): AAAS; ABCA12; ABCB4; ABCB11; ABCC6; ABCC8; ACAD9; ACADM; ACADVL; ACAT1; ACOX1; ACSF3; ADA; ADAMTS2; ADGRG1 (GPR56); AGA; AGL; AGPS; AGXT; AIRE; ALDH3A2; ALDH7A1; ALDOB; ALG6; ALMS1; ALPL; AMT; AP1S1; AQP2; ARG1; ARSA; ARSB; ASL; ASNS; ASPA; ASS1; ATM; ATP6V1B1; ATP7B; ATP8B1; BBS1; BBS2; BBS4; BBS9; BBS10; BBS12; BCKDHA; BCKDHB; BCS1L; BLM; BSND; BTD; CANT1; CAPN3; CASQ2; CBS; CC2D1A; CCN6 (WISP3); CDH23; CEP290; CERKL; CFTR; CHRNE; CHRNG; CIITA; CLN3; CLN5; CLN6; CLN8; CLRN1; CNGA3; CNGB3; COL4A3; COL4A4; COL7A1; COL11A2; CPS1; CPT1A; CPT2; CRB1; CTNS; CTSC; CTSD; CTSK; CYBA; CYP1B1; CYP11B1; CYP11B2; CYP17A1; CYP19A1; CYP21A2; CYP27A1; CYP27B1; DBT; DCLRE1C; DDB2; DHCR7; DHDDS; DNAH5; DNAI1; DNAI2; DNAL1; DLD; DOK7; DPYD; DYSF; EDAR; EIF2AK3; EIF2B5; ELP1 (IKBKAP); ERCC2; ERCC3; ERCC4; ERCC5; ERCC6; ERCC8; ESCO2; ETFA; ETFB; ETFDH; ETHE1; EVC; EVC2; EXOSC3; EYS; FAH; FAM161A; FANCA; FANCC; FANCG; FH; FKRP; FKTN; G6PC; GAA; GALC; GALE; GALK1; GALNS; GALNT3; GALT; GAMT; GBA1 (GBA); GBE1; GCDH; GCH1; GDF5; GFM1; GH1; GHRHR; GJB2; GJB6; GLB1; GLDC; GLE1; GNE; GNPTAB; GNPTG; GNS; GORAB; GUCY2D; GUSB; HADHA; HADHB; HAX1; HBA1; HBA2; HBB; HEXA; HEXB; HGSNAT; HJV (HFE2); HLCS; HMGCL; HMOX1; HPD; HPS1; HPS3; HPS4; HSD3B2; HSD17B3; HSD17B4; HYLS1; IDUA; IVD; KCNJ11; LAMA2; LAMA3; LAMB3; LAMC2; LCA5; LDLR; LDLRAP1; LHCGR; LIFR; LIPA; LOXHD1; LPL; LRPPRC; LYST; MAN2B1; MCOLN1; MED17; MESP2; MFSD8; MKKS; MKS1; MLC1; MLYCD; MMAA; MMAB; MMACHC; MMADHC; MMUT; MOCS1; MPI; MPL; MPV17; MRE11; MTHFR; MTRR; MTTP; MYO7A; MYO15A; NAGLU; NAGS; NBN; NDRG1; NDUFAF5; NDUFS4; NDUFS6; NEB; NEU1; NPC1; NPC2; NPHP1; NPHS1; NPHS2; NR2E3; NTRK1; OAT; OPA3; PAH; PANK2; PC; PCCA; PCCB; PCDH15; PDHB; PEPD; PET100; PEX1; PEX2; PEX6; PEX7; PEX10; PEX12; PFKM; PHGDH; PIGN; PKHD1; PLA2G6; PMM2; PNPO; POLG; POLH; POMGNT1; POR; PPT1; PREPL; PROP1; PSAP; PTS; PUS1; RAB23; RAG1; RAG2; RAPSN; RARS2; RDH12; RLBP1; RMRP; RNASEH2C; RPE65; RPGRIP1L; RTEL1; SACS; SAMD9; SAMHD1; SBDS; SEPSECS; SERPINA1; SGCA; SGCB; SGCD; SGCG; SGSH; SKIC3 (TTC37); SLC7A7; SLC12A3; SLC12A6; SLC17A5; SLC19A2; SLC22A5; SLC25A13; SLC25A15; SLC25A20; SLC26A2; SLC26A3; SLC26A4; SLC35A3; SLC37A4; SLC39A4; SLC45A2; SMARCAL1; SMN1; SMPD1; SRD5A2; ST3GAL5; STAR; SUCLA2; SUMF1; SURF1; TAT; TCIRG1; TECPR2; TFR2; TGM1; TH; TMC1; TMEM216; TPO; TPP1; TREX1; TRIM32; TRIM37; TRMU; TSEN54; TSFM; TSHB; TSHR; TTN; TTPA; TYMP; TYR; TYRP1; UGT1A1; USH1C; USH2A; VPS13A; VPS13B; VPS45; VPS53; VRK1; VSX2; VWF; WNT10A; WRN; XPA; XPC; ZFYVE26.

X-linked genes tested in female partner only (n=29): ABCD1; AR; ATP7A; ATRX; BTK; CHM; COL4A5; CYBB; DKC1; DMD; EDA; EMD; F8; F9; FMR1; GJB1; GLA; IDS; IL2RG; MECP2; MTM1; NR0B1; OCRL; OTC; PDHA1; PRPS1; RS1; SLC6A8; WAS.

FMR1 gene CGG repeat number is determined using the CarrierMax FMR1 Reagent Kit. All other genes are tested using the Ion AmpliSeq CarrierSeq ECS Panel, Ion GeneStudio S5 System, and Ion Reporter and Carrier Reporter software.

CarrierSeq target regions include all coding exons and flanking splice sites of the listed genes. Variants that can be detected by this assay include single nucleotide variants, small indels (<10bp), and large copy number variants. SMN1 (NM_000344.4) exon 7 copy number will be detected; the presence of c.*3+80T>G, and c.*211_*212del variants will be reported if copy number is 2. Variants unable to be detected by this test include (but are not limited to): single nucleotide variants in SMN1 and FMR1 (unless otherwise specified); 2+0 SMN1 carriers; complex structural variants (eg. intron inversion variants in F8 gene; recombinant alleles in GBA1 gene). Test sensitivity and specificity is reduced for: copy number variants involving one or a few coding exons; indels adjacent to homopolymer regions; single nucleotide variants in genes affected by pseudogenes (eg. GBA1; CYP21A2; HBA1; HBA2); mosaic variants. The CYP21A2 c.955C>T, p.(Gln319*) variant may only be detected when present with a whole gene duplication, as occurs in approximately 84% of carriers (PMID: 19773403).

Variants are classified using ACMG criteria (PMID: 25741868) and relevant ClinGen guidelines and reported using HGVS nomenclature (v20). Pathogenic and likely pathogenic variants are reported; variants of uncertain significance (VOUS), likely benign, and benign variants are not reported. Classifications are based on current knowledge in the literature. Variants that have solely been associated with mild and/or adult-onset phenotypes may not be reported. Unsolicited findings with potential implications for heterozygous carriers are only reported in the following autosomal genes: ATM, FH, LDLR, TTN (predicted premature termination variants only). The CFTR intron 9 polyT and TG region is not routinely analysed or reported. FMR1 repeat size thresholds applied: normal (5-44); intermediate (45-54); premutation (55-200); full mutation (>200). Female individuals with a single repeat allele are most likely homozygous for that allele.

This report has assumed that DNA extracted from the provided sample(s) represents the tested individuals constitutional genome and the laboratory has not been advised of any confounding factors (eg. bone marrow transplant, recent blood transfusion, or other known chimerism).

This test has been performed by Virtus Genetics (1800 837 284; info@virtusgenetics.com.au). Test Information v1.7.