LAUNCESTON PATHOLOGY

Clare Pitt

LAB ID **413201707** DOB **20/12/1985 (37Y Female)**

Referring Doctor Dr Georgia Bavin

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Reproductive Carrier Screening

CONDITION	CARRIER RISK	RESULT
Cystic fibrosis (CF)	Low risk	No CFTR variants detected
Fragile X syndrome (FXS)	Low risk	Normal range allele(s) detected FMR1:c.-129CGG: 22,30
Spinal muscular atrophy (SMA)	Low risk	At least two copies of the SMN1 gene detected

INTERPRETATION

CF This individual is unlikely to be a carrier of CF. Residual risk varies with ethnicity and family history.

FXS This individual is unlikely to be a carrier of FXS. Residual risk varies with ethnicity and family history.

SMA This individual is unlikely to be a carrier of SMA. Residual risk varies with ethnicity and family history.

RECOMMENDATION

This individual is unlikely to be a carrier for CF, FXS or SMA. Carrier status for rare disease-causing variants in these genes has not been excluded. This report should be interpreted in conjunction with the reproductive partner carrier screen result.

TEST INFORMATION

CF carrier testing performed using the Elucigene CF-EU2v1 (YourGene) multiplex ARMS assay that detects more than 50 CFTR(LRG_663t1) variants including: c.54-5940_273+1025del21080, p.Glu60*, p.Pro67Leu, p.Gly85Glu, p.Leu88Ilefs*22, p.Ile105Serfs*2, p.Arg117Cys, p.Arg117His, p.Tyr122*, c.489+1G>T, c.579+1G>T, p.Leu206Trp, p.Phe316Leufs*12, p.Arg334Trp, p.Arg347Pro, p.Arg347His, p.Ala455Glu, p.Ile507del, p.Phe508del, p.Tyr515*, p.Val520Phe, c.1585-1G>A, p.Gly542*, p.Ser549Arg, p.Ser549Asn, p.Gly551Asp, p.Arg553*, p.Arg560Thr, c.1680-886A>G, c.1766+1G>A, p.Leu671*, p.Lys684Asnfs*38, p.Val739Tyrfs*16, p.Trp846*, c.2657+5G>A, p.Gln890*, c.2988+1G>A, c.3140-26A>G, p.Arg1066Cys, p.Tyr1092*, p.Met1101Lys, p.Asp1152His, p.Arg1158*, p.Arg1162*, p.Lys1177Serfs*15, c.3718-2477C>T, p.Ser1251Asn, p.Leu1258Phefs*7, p.Trp1282*, p.Asn1303Lys. Intron 9 polyT tract and adjacent TG repeat are not assessed unless the p.Arg117His variant detected. Targeted Sanger sequencing is undertaken in the presence of an atypical profile based on internal criteria. Variants of uncertain significance are not reported. Variants detected by this method include >30 most frequently encountered CFTR variants in the Australian population, that account for 95% of disease-causing variants in the CFTR2.org database (July 2020).

FXS carrier testing undertaken using the AmplideX FMR1 PCR (Asuragen) assay to detect normal to full expansion CGG repeat alleles in the 5'UTR of the FMR1 gene (LRG_762t1). Reference ranges: normal <45, intermediate 45-54, premutation 55-200, full mutation >200 CGG repeats. Single nucleotide variants and large deletions of the FMR1 gene that account for <1% of Fragile X syndrome cases will not be detected. Low-level mosaicism for an expanded allele may not be detected. A single repeat allele in a female is assumed to indicate homozygosity for that allele. The measurement uncertainty is +/- 2 repeats for alleles <, and +/- 3 repeats for alleles > 55 repeats, respectively.

SMA carrier testing performed using the AmplideX SMA Plus (Asuragen) kit to detect copy number of exon 7 of the SMN1 gene (LRG_676t1). This assay cannot detect rare sequence alterations (2% of SMN1 variants) or always identify SMA carriers who have two (or more) copies of SMN1 on one chromosome and zero copies on the other (4% of SMA carriers). Therefore, a two (or more) copy result does not exclude the possibility that an individual is a carrier of SMA. The kit also identifies the c.*3+80T>G and c.*211_*212del SMN1 variants that can be associated with a 2+0 carrier genotype; reported only when found together and in conjunction with two SMN1 copies. Accurate quantitation beyond two SMN1 copies is not possible; reflex testing using the MLPA P060-B2 SMA kit (MRC Holland) is undertaken in the presence of family history, detection of the 2+0 risk variants, partner carrier status, or when an accurate result cannot be obtained. Very rare polymorphisms in the primer binding sites for this assay can potentially lead to false-positive one SMN1 copy results. Post-test residual risk varies with ethnicity and family history; genetic counselling may be appropriate.

Genetic test results may have significant implications for both the individual and their relatives. Corroboration of this result by reference to other clinical or laboratory information or by repeat testing may be warranted.
Please note that from 1st May 2023, the three-gene reproductive carrier screen will be issued as a single combined report, replacing the previous four separate reports. Please contact the laboratory if there are any concerns.
This test was performed at Douglass Hanly Moir Pathology

Clinical notes:

Sample type: EDTA

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