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Patient Name	Jane Doe	**Requesting Doctor**	Dr John Doe
Date of Birth	01/01/2002	**Provider Number**	123456AB
Sex	F	**Address**	1 John St
Address	1 Jane St	**Requested**	26 Jan 2025
Specimen Type	Saliva	**Reported**	30 Jan 2025
Collected	27 Nov 2025	**Episode ID**	EP-2048-251128
Clinical Indication	Preconception		

Familee Screen

Patient Summary

What We Did

We looked at your DNA to check for three things:

(1) If you are a carrier of any genetic conditions that could be passed on to your children
(2) If there are any genetic variants that could affect your own health
(3) If there are any genetic variants that could increase the chance of learning or developmental conditions in your children

What We Found

We didn't find any genetic variants that need follow-up right now.

What This Means

Your results are reassuring. They make it less likely that you or your children will be affected by the conditions we looked for.

It's important to remember that no test can find everything. This screen lowers the chance of having or passing on certain conditions, but it doesn't remove that chance completely.

It's especially important to consider your results alongside your personal and family medical history.

What to Do Next

Your doctor can help explain what these results mean for you and whether any next steps are needed.

Preciselee Pty Ltd | ABN 16 677 209 728 | SE301 116-118 Christie St, St Leonards NSW 2065 | contact@preciselee.co

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Test: Basic Carrier Screen

Results

- CFTR: No reportable variants were identified.
- SMN1: Two or more gene copies detected.
- FMR1: 30 CGG repeats.

Interpretation

- This patient was not found to be a carrier for cystic fibrosis, spinal muscular atrophy, or fragile X syndrome.
- In the absence of relevant family history, this result significantly reduces (but does not eliminate) the chance that the patient is a carrier of one or more of the screened conditions.

Recommendations

- Please interpret this result in the context of the patient's personal and family medical history.
- If there is a family history of any of the screened conditions, please provide this information so we can assess the potential for a false negative and determine whether further testing is warranted.

[***] = Clinically significant or actionable finding

Test Method

SMN1 copy number was determined using the P460 SMA MLPA kit. FMR1 CGG repeat number was determined using the AmplideX PCR/CE FMR1 kit. Fragment analysis was performed by the Australian Genome Reference Laboratory (AGRF). SMN1 testing does not detect sequence variants, 2+0 silent carriers, or the SMN1 duplication haplotype (c.*3+80T>G and c.*211_*212del). FMR1 AGG interruption analysis is not performed, and mosaic CGG repeat expansions may not be detected.

The CFTR gene was tested by exome sequencing, which was performed by the Australian Genome Reference Laboratory (AGRF) using the TWIST Alliance Clinical Research Exome Sequencing Kit. Variants were assessed in coding exons and up to 8 intronic bases from exon boundaries. Variant analysis was limited to the following: Variants classified as pathogenic or likely pathogenic in ClinVar with a review status of two stars or higher; premature termination variants (nonsense, frameshift, or canonical donor/acceptor splice site variants) that have not been classified as benign or likely benign in ClinVar (with a review status of two stars or higher). Missense variants that do not have an aggregate ClinVar classification of pathogenic or likely pathogenic have not been analysed. Reported variants may have been classified based on the aggregate ClinVar classification, or, where appropriate, according to ACMG/AMP guidelines (PMID: 25741868). Final decisions on variant reporting were made at the discretion of a pathologist, based on clinical relevance and the context of the test request. Variants have been classified based on current knowledge and this may change over time as new data become available. Variants of uncertain significance (VUS) have not been reported. This assay does not detect copy number variants, large insertions or deletions 10bp or more, or mosaic variants. Not all regions of CFTR known to harbour disease-causing variants have been sequenced.

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Test: Expanded Carrier Screen

Results

- No reportable variants were identified.
- Please refer to the 'Basic Carrier Screen' report for results in the CFTR (cystic fibrosis), SMN1 (spinal muscular atrophy), and FMR1 (fragile X syndrome) genes.

Interpretation

- This patient was not found to be a carrier for the screened conditions.
- In the absence of relevant family history, this result significantly reduces (but does not eliminate) the chance that the patient is a carrier of one or more of the screened conditions.

Recommendations

- Please interpret this result in the context of the patient's personal and family medical history.
- If there is a family history of any of the screened conditions, please provide this information so we can assess the potential for a false negative and determine whether further testing is warranted.

[***] = Clinically significant or actionable finding

Test Method

Exome sequencing was performed by the Australian Genome Reference Laboratory (AGRF) using the TWIST Alliance Clinical Research Exome Sequencing Kit. The gene list screened in this analysis is provided in the Appendix. This screen does not include genes associated with alpha thalassemia (HBA1, HBA2) or congenital adrenal hyperplasia (CYP21A2).

Variants were assessed in coding exons and up to 8 intronic bases from exon boundaries. Variant analysis was limited to the following: Variants classified as pathogenic or likely pathogenic in ClinVar with a review status of two stars or higher; premature termination variants (nonsense, frameshift, or canonical donor/acceptor splice site variants) that have not been classified as benign or likely benign in ClinVar (with a review status of two stars or higher). Missense variants that do not have an aggregate ClinVar classification of pathogenic or likely pathogenic have not been analysed. Variants in X-linked genes have only been analysed in females, except for EFNB1 and PCDH19, where variants in males have also been analysed.

Reported variants may have been classified based on the aggregate ClinVar classification, or, where appropriate, according to ACMG/AMP guidelines (PMID: 25741868) or gene-specific ACMG classification criteria. Final decisions on variant reporting were made at the discretion of a pathologist, based on clinical relevance and the context of the test request. Variants have been classified based on current knowledge and this may change over time as new data become available. Variants of uncertain significance (VUS) have not been reported. This assay does not detect copy number variants, oligonucleotide repeat expansions, large insertions or deletions 10bp or more, or mosaic variants. E.g., the F8 intron 22 inversion cannot be detected by this assay. Test sensitivity and specificity may be reduced in repetitive regions, low complexity regions, regions of homology elsewhere in the genome, or high GC content. E.g., sensitivity/specificity is reduced in the *GBA* gene.

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Test: Personal Genetic Screen

Results

- No reportable variants were identified.

Interpretation

- In the absence of relevant family history, this result significantly reduces (but does not eliminate) the likelihood of a clinically significant variant in the screened genes.
- This result does not rule out the possibility of developing health conditions in the future, including cancer or cardiac disease.

Recommendations

- Please interpret this result in the context of the patient's personal and family medical history.
- As this is a screening test, false negative results are possible. If there is strong clinical suspicion for a specific condition, consider further diagnostic testing.
- If a familial variant is known to exist in one of the screened gene, please provide this information so we can evaluate the risk of a false negative and the need for further testing.

[***] = Clinically significant or actionable finding

Test Method

Exome sequencing was performed by the Australian Genome Reference Laboratory (AGRF) using the TWIST Alliance Clinical Research Exome Sequencing Kit. The gene list screened in this analysis is provided in the Appendix.

Variants were assessed in coding exons and up to 8 intronic bases from exon boundaries. Variant analysis was limited to the following: Variants classified as pathogenic or likely pathogenic in ClinVar with a review status of two stars or higher; premature termination variants (nonsense, frameshift, or canonical donor/acceptor splice site variants) that have not been classified as benign or likely benign in ClinVar (with a review status of two stars or higher). Missense variants that do not have an aggregate ClinVar classification of pathogenic or likely pathogenic have not been analysed.

Reported variants may have been classified based on the aggregate ClinVar classification, or, where appropriate, according to ACMG/AMP guidelines (PMID: 25741868) or gene-specific ACMG classification criteria. Final decisions on variant reporting were made at the discretion of a pathologist, based on clinical relevance and the context of the test request. Variants have been classified based on current knowledge and this may change over time as new data become available. Variants of uncertain significance (VUS) have not been reported. This assay does not detect copy number variants, oligonucleotide repeat expansions, large insertions or deletions 10bp or more, or mosaic variants. Test sensitivity and specificity may be reduced in repetitive regions, low complexity regions, regions of homology elsewhere in the genome, or high GC content. E.g., sensitivity/specificity is reduced in PMS2 exons 12-15 (NM_000535.7).

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Test: Chromosome Analysis

Results

- No reportable copy number variants were identified.

Interpretation

- This result does not exclude the following possibilities:
    - Clinically significant copy number variants below the resolution of this assay
    - Chromosomal rearrangements not detectable by this method, such as balanced reciprocal or Robertsonian translocations
    - Risks of having offspring with neurodevelopmental conditions or chromosomal disorders, such as Down syndrome

Recommendations

- In the pre-pregnancy setting, additional chromosomal investigations may still be appropriate depending on the clinical context. Consider the following where indicated:
    - Conventional karyotyping in cases of infertility or recurrent pregnancy loss
    - Y chromosome microdeletion testing in males with oligozoospermia or azoospermia

[***] = Clinically significant or actionable finding

Test Method

Microarray analysis was performed by the Australian Genome Reference Laboratory (AGRF) using the Illumina Global Screening Array (GSA) version 3.0 BeadChip. The genome build used for alignment and analysis was GRCh37.

Copy number variant (CNV) analysis was conducted for the following: CNVs that overlap genes on the Basic and Expanded Carrier Screen, or the Personal Genetic Carrier Screen, where the relevant test has been requested and loss-of-function variants are known to be disease-causing; CNVs that correspond to targeted susceptibility CNVs (see Appendix); any other CNVs 1 Mb or larger that have been classified as pathogenic or likely pathogenic. The effective resolution of this assay is approximately 200 kb for targeted genes and regions, and approximately 1 Mb across the rest of the genome.

All CNVs considered for reporting have been classified using ACMG/ClinGen guidelines (PMID: 31690835). Variant classification has been based on current knowledge and may change over time as new data become available. Variants of uncertain significance (VUS) have not been reported. Regions of homozygosity have not been reported. This assay does not detect balanced chromosomal rearrangements, Y chromosome microdeletions, or mosaic variants.

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Appendix

Expanded Carrier Screen - Gene List (v1.2):

AAAS, AARS2, ABAT, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCD1, ABCD4, ABHD5, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1,* ACE, ACO2, ACOX1, ACTA1, ADA, ADA2, ADAMTS13, ADAMTS2, ADAMTSL2, ADAR, ADAT3, ADGRG1, ADGRV1, ADK, ADPRS, ADSL, AFF2, AGA, AGBL5, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGXT, AHCY, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG3, ALG6, ALG8,* ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALS2, AMH, AMHR2, AMN, AMPD2, AMT, ANKS6,* ANO10, ANO5,* ANTXR1, ANTXR2, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4M1, AP4S1, APTX, AQP2, AR, ARFGEF2, ARG1, ARHGEF9, ARL13B, ARL6, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX,* ASAH1, ASCC1, ASL, ASNS, ASPA, ASPM, ASS1, ATAD1, ATCAY, ATF6, ATM, ATOH7, ATP13A2, ATP6AP1, ATP6V0A2, ATP6V0A4, ATP6V1B1, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATR, ATRX, AUH, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT7, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCHE, BCKDHA, BCKDHB, BCKDK, BCS1L, BGN, BIN1, BLM, BLOC1S3, BLOC1S6, BLTP1, BMP1, BMPER, BMPR1B, BOLA3, BRAT1, BRF1, BRIP1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C19orf12, C1QA, C1QB, C1QC, C2CD3, C3, C5, CA2, CABP4, CAD, CANT1, CAPN3, CARD11, CARS2, CASK, CASP14, CASQ2, CASR, CAVIN1, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC39, CCDC40, CCDC8, CCDC88C, CCN6, CCNO, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD8A, CDAN1, CDC45, CDCA7, CDH11, CDH23,* CDH3, CDK10, CDK5RAP2, CDT1, CENPJ, CEP104, CEP120, CEP152, CEP290,* CEP41, CEP78, CERKL, CERS3, CFAP410, CFAP418, CFD, CFH,* CFI,* CFL2, CFP, CHAT, CHKB, CHM, CHMP1A, CHRNA1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHSY1,* CIB2, CIITA, CISD2, CIT, CKAP2L, CLCF1, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKB, CLDN1, CLDN10, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3,* CNNM4, CNTNAP1, CNTNAP2, COA8, COASY, COG6, COG7, COL11A1, COL11A2,* COL17A1, COL18A1, COL27A1, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COLEC11, COLQ, COQ2, COQ4, COQ6, COQ8A, COQ8B, COX10, COX15, COX20, COX6B1, CP, CPLANE1,* CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRLF1,* CRPPA, CRTAP, CRYL1, CSPP1, CSTB, CTC1, CTNS, CTPS1, CTSA, CTSC, CTSD, CTSF, CTSK, CUL4B, CUL7, CWC27,* CYB5R3, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP27A1, CYP27B1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DARS1, DARS2, DBT, DCAF17, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD2, DDR2, DDX11, DDX59, DENND5A, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHODH, DIS3L2, DKC1, DLAT, DLD, DLG3, DLL3, DMD, DNAAF1, DNAAF11, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC12, DNAJC19, DNAJC21, DNAJC6, DNAL1,* DNMT3B, DOCK2, DOCK6, DOCK8, DOK7, DOLK, DONSON, DPAGT1, DPH1, DPYD, DSP, DTNBP1, DUOX2, DUOXA2, DYM, DYNC2H1,* DYNC2I1, DYNC2I2, DYNC2LI1, DYSF, EARS2, ECEL1, ECHS1, EDA, EDAR, EFEMP2, EFNB1 (analysed in males only), EIF2AK3, EIF2AK4, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ELP1, ELP2, EMD, EML1, ENPP1,* EOGT, EPB42, EPCAM, EPG5, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2,* ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC,* EVC2, EXOSC3, EXOSC8, EXTL3, EYS, F11, F2, F5, F7, F8, F9, FA2H, FAH, FAM126A, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FARS2, FAT4, FBLN5, FBP1, FBXL4, FBXO7, FERMT1, FERMT3, FGA, FGB, FGD1, FGD4, FGG, FH, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLVCR1, FLVCR2, FMO3, FOLR1, FOXE3, FOXN1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRRS1L, FTCD, FTO, FTSJ1, FUCA1, FXN, FYCO1, G6PC1, G6PC3, GAA, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GAN, GAS8, GATM, GBA,* GBA2, GBE1, GCDH, GCH1,* GDAP1, GDF1, GDF5, GDI1, GFM1, GFPT1, GH1, GHR, GHRHR, GJA1, GJB1, GJB2, GJB6, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLIS3, GLYCTK, GM2A, GMPPA, GMPPB, GNAT2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNRHR, GNS, GORAB, GOSR2, GP1BA, GP9, GPAA1, GPC3, GPC6, GPHN, GPR143, GPR179, GPSM2, GPT2, GRHPR, GRIP1, GRM1, GSS, GTF2H5, GTPBP3, GUCY1A1, GUCY2C, GUCY2D, GUSB, GYS2, HACE1, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HCFC1, HELLS,* HEPACAM, HES7, HESX1, HEXA, HEXB,* HGD, HGSNAT,* HIBCH, HINT1, HJV, HK1, HLCS, HMGCL, HMGCS2, HMOX1, HOGA1, HOXA1, HPD, HPGD, HPRT1,* HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPD1, HSPG2, HTRA2, HUWE1, HYAL1, HYLS1, IARS1, IARS2, IBA57, ICOS, IDH3B, IDS, IDUA, IER3IP1, IFNGR1, IFNGR2, IFT122, IFT140, IFT172, IFT80, IGF1R, IGHMBP2, IGSF1, IKBKB, IL10RA, IL10RB, IL11RA, IL12RB1, IL17RA, IL1RAPL1, IL1RN, IL2RA, IL2RG, IL7R, INPP5E, INPP5K, INPPL1, INSR, INVS, IQCB1, IQSEC2, ISCA2, ITCH, ITGA2B, ITGA6, ITGB2, ITGB3, ITGB4, ITK, ITPA, ITPR1, IVD, IYD, JAGN1, JAK3, JAM3, JUP, KATNB1, KCNJ1, KCNJ10, KCNJ11, KCNQ1, KCNV2, KCTD7, KDM5C, KIAA0586,* KIF14, KIF1A, KIF1C, KIF7, KIFBP, KLHL40, KLHL41, KLHL7, KNL1,* KPTN, KRT10, KRT14, KRT5, KY, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LARGE1, LARP7, LARS1, LARS2, LAT, LBR, LCA5, LCAT, LCK, LDHA, LDLR, LDLRAP1, LEP, LGI4, LHCGR, LHX3, LIAS, LIFR, LIG4, LINS1, LIPA, LIPC, LIPN, LIPT1, LMAN1, LMBR1,* LMBRD1,* LMNA, LMOD3, LONP1, LOXHD1, LPAR6, LPIN1, LPIN2,

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Collected	27 Nov 2025	Episode ID	EP-2048-251128
Clinical Indication	Preconception		

LPL, LRAT, LRBA,* LRIG2, LRMDA, LRP2, LRP4, LRP5, LRPPRC, LRSAM1, LTBP3, LTBP4, LYRM7,* LYST, LZTFL1, MAK,* MALT1,* MAN1B1, MAN2B1, MANBA, MAOA, MAPKBP1, MARS1, MARS2, MASP1, MAT1A, MBOAT7, MBTPS2, MC2R, MCEE, MCFD2, MCOLN1, MCPH1, MECP2, MED12, MED17, MED23, MED25, MEFV, MEGF10, MEGF8, MERTK, MESP2, METTL23, MFN2, MFSD2A, MFSD8, MGAT2, MGME1, MGP,* MICU1,* MID1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMP2, MMP21, MMUT, MOCS1, MOCS2, MPDZ, MPI, MPL, MPLKIP, MPV17, MPZ, MRAP, MRE11, MTFMT, MTHFD1, MTHFR, MTM1, MTMR2, MTO1, MTR, MTRR, MTTP, MUSK, MVK, MYD88, MYMK, MYO15A, MYO5B, MYO7A, NAA10, NAGA, NAGLU, NAGS, NALCN, NANS, NARS2,* NAXE, NBAS, NBEAL2, NBN, NCF2, NCF4, NDE1, NDP, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFAF2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECTIN1, NEK1, NEK8, NEU1, NEXMIF, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHS, NIPAL4, NKX3-2, NKX6-2, NMNAT1, NNT, NONO, NPC1, NPC2, NPHP1,* NPHP3,* NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR2E3, NSDHL, NSUN2, NT5C2, NTRK1, NUBPL, NUP107, NUP93, NYX, OAT, OBSL1, OCRL, ODAD1, ODAD3, OFD1, OPA1, OPA3, OPHN1, ORAI1, ORC1, ORC6, OSGEP, OSTM1, OTC, OTOF, OTUD6B, P3H1, PAH, PAK3, PANK2, PAPSS2, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH15, PCDH19 (analysed in males only), PCNT, PCSK1, PCYT1A, PDE6A, PDE6C, PDHA1, PDHB, PDHX, PDP1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAP1,* PGAP2, PGAP3, PGK1, PGM1, PGM3, PHF6, PHF8, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIBF1, PIEZO2, PIGA, PIGG, PIGL, PIGN, PIGO, PIGT, PIGV, PIP5K1C, PJVK, PKHD1, PKLR, PLA2G6, PLAA, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLOD2,* PLP1, PLPBP, PMM2, PMPCA, PNKP, PNP, PNPLA1, PNPLA6, PNPO, POC1A, POLG, POLH, POLR1C, POLR3A, POLR3B, POMC, POMGNT1, POMGNT2, POMK, POMP, POMT1, POMT2, POP1, POR, POU1F1, POU3F4, PPA2,* PPIB, PPT1, PQBP1, PRCD, PRDM12,* PRDM5, PREPL, PRF1, PRG4, PRICKLE1, PRKDC, PRKRA, PROC, PROP1, PROS1,* PRPS1, PRUNE1, PRX, PSAP, PSAT1, PSMB8, PSPH, PTH1R, PTPN23, PTPRC,* PTS, PUS1, PUS7, PXDN, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, RAB18, RAB23, RAB27A, RAB33B, RAB39B, RAB3GAP1, RAB3GAP2,* RAD50, RAG1, RAG2, RAPSN, RARB, RARS1, RARS2, RAX, RBBP8, RBCK1, RBM10, RCBTB1, RD3, RDH12, RDH5, RECQL4, REEP6, REN, RETREG1, RFT1, RFX5, RFX6, RFXANK, RFXAP, RHAG, RIN2, RIPK4, RLBP1, RLIM, RMND1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU4ATAC, ROBO3, ROGDI, ROR2, RORC, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L,* RPL10, RPS6KA3, RRM2B, RS1, RSPH1, RSPH4A, RSPH9, RTEL1, RTN4IP1, RTTN,* RXYLT1, RYR1,* SACS, SAG, SAMD9, SAMHD1, SAR1B, SARS2, SBDS, SBF2, SC5D, SCARB2, SCARF2, SCN9A, SCNN1A, SCNN1B, SCO1, SCO2, SCYL1, SDCCAG8, SDHAF1, SDR9C7, SEC23B, SELENON,* SEPSECS, SERAC1, SERPINF1, SERPINH1, SETX, SFTPB, SGCA, SGCB, SGCD, SGCG, SGPL1, SGSH, SH2D1A, SH3PXD2B, SH3TC2, SIL1, SKIV2L, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC24A5, SLC25A1, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A38, SLC25A46, SLC26A2, SLC26A3, SLC26A4, SLC27A4, SLC29A3, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC34A3, SLC35A3, SLC35D1,* SLC37A4, SLC38A8, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC45A2, SLC46A1, SLC4A1, SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A5, SLC5A7, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A7, SLC7A9, SLC9A3, SLC9A6, SMARCAL1, SMPD1, SMS, SNAP29, SNX10, SNX14, SOST, SP110, SPAG1, SPART, SPATA5, SPATA7, SPEG, SPG11, SPG21, SPG7, SPINK5, SPINT2, SPR, SQSTM1, SRD5A2, SRD5A3, SSR4, ST3GAL5, STAMBP, STAR, STAT1, STIL, STIM1, STK4, STRA6, STRADA, STUB1, STX11, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNE4, SYP, TAFAZZIN, TALDO1, TANGO2, TAP1, TAT, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBX19, TCAP, TCIRG1, TCN2, TCTN1,* TCTN2, TCTN3, TDRD7, TECPR2, TELO2, TERT, TF, TFR2, TG, TGM1, TH, THOC2, TIMM8A, TJP2, TK2, TMC1, TMCO1, TMEM107, TMEM126A, TMEM138, TMEM165, TMEM216, TMEM237, TMEM38B, TMEM67, TMEM70, TMPRSS3, TMTC3, TNFRSF11A,* TNFRSF11B, TNFSF11, TNNT1, TOE1, TPI1, TPK1, TPM3, TPO, TPP1, TRAPPC11, TRAPPC9, TRDN,* TREX1, TRHR, TRIM32, TRIM37, TRIP11, TRIT1, TRMT10A, TRMU, TRNT1,* TRPM6, TSEN2, TSEN34, TSEN54, TSFM,* TSHB, TSHR, TTC19, TTC21B, TTC37, TTC7A, TTC8, TTI2, TTN,* TTPA, TUBGCP4, TUBGCP6, TUFM, TULP1, TUSC3, TWNK, TXNL4A, TYK2, TYMP, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3B, UBR1,* UFM1,* UGT1A1, UMPS, UNC13D, UNC80, UPF3B, UROS, USB1, USH1C, USH1G, USH2A, USP9X, VAC14, VARS1, VARS2, VDR, VIPAS39, VKORC1, VLDLR, VMA21, VPS11, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WARS2, WAS, WDR19, WDR35,* WDR45B, WDR62, WDR73, WDR81, WFS1, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT7A, WRAP53, WRN, WWOX, XIAP, XPA, XPC, XRCC4, XYLT1,* XYLT2, YARS2, ZAP70, ZBTB24, ZC4H2, ZDHHC9, ZFYVE26, ZIC3, ZMPSTE24, ZMYND10, ZNF335, ZNF469, ZNF711, ZNHIT3.

* Some regions in these genes are known to have incomplete sequencing coverage. As a result, sensitivity for variant detection in these regions may be reduced. Additional gene-specific testing should be considered if clinically indicated.

preciselee

Patient Name	Jane Doe	**Requesting Doctor**	Dr John Doe
Date of Birth	01/01/2002	**Provider Number**	123456AB
Sex	F	**Address**	1 John St
Address	1 Jane St	**Requested**	26 Jan 2025
Specimen Type	Saliva	**Reported**	30 Jan 2025
Collected	27 Nov 2025	**Episode ID**	EP-2048-251128
Clinical Indication	Preconception		

Personal Genetic Screen - Gene List (v1.2):

ABCD1, ACTA2, ACTC1, ACVRL1, APC, APOB, ATP7B, BAG3, BMPR1A, BRCA1, BRCA2, BTD, CACNA1S, CALM1, CALM2, CALM3, CASQ2, COL3A1, CYP27A1, DES, DSC2, DSG2, DSP, ENG, FBN1, FLNC, GAA, GLA, HFE (C282Y homozygotes only), HNF1A, KCNH2, KCNQ1, LDLR, LMNA, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYL2, MYL3, NF2, OTC, PALB2, PCSK9, PKP2,* PLN, PMS2,* PRKAG2,* PTEN, RB1,* RBM20, RET, RPE65, RYR1,* RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SMAD3, SMAD4, STK11, TGFBR1,* TGFBR2, TMEM127, TMEM43, TNNC1, TNNI3, TNNT2, TP53, TPM1, TRDN, TSC1, TSC2, TTN (truncating variants only),* TTR, VHL, WT1.

* Some regions in the genes marked with an asterisk are known to have incomplete sequencing coverage. As a result, sensitivity for variant detection in these regions may be reduced. Additional gene-specific testing should be considered if clinically indicated.

Chromosome Analysis - Targeted Susceptibility Copy Number Variants (v1.0):

1q21.1 deletion (ISCA-37421), 2p16.3 deletion (includes NRXN1), 3q29 deletion (ISCA-37443), 6q24 duplication (ISCA-37442), 15q11.2q13 duplication (BP1-BP3, BP2-BP3; ISCA-37404, ISCA-37478), 15q13.3 deletion (ISCA-37411), 16p11.2 deletion (proximal, distal; ISCA-37400, ISCA-37486), 17p12 deletion/duplication (ISCA-37436), 17q12 duplication (ISCA-37432), 22q11.2 duplication (proximal A-B, proximal A-D; ISCA-37433, ISCA-37446).