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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	**Laboratory 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

- You are a carrier of Cystic Fibrosis, a condition that mainly affects the lungs and digestion.
- You are a carrier of Familial Hypercholesterolaemia, a condition that raises cholesterol levels and increases the risk of heart disease.
- You are a carrier of Duchenne/Becker Muscular Dystrophy, a condition that causes progressive muscle weakness, usually in boys.

What This Means

- Because you are a carrier of Cystic Fibrosis and Familial Hypercholesterolaemia, it is important to test your reproductive partner for the same conditions. If your partner is also a carrier for these conditions, you would be at high risk of having children affected by these conditions.
- For Duchenne/Becker Muscular Dystrophy, only mothers pass on this condition. If you are pregnant with a boy, there's a 1 in 2 (50%) chance that he may be affected. Testing your partner for this condition is not needed.
- The results for Familial Hypercholesterolaemia and Duchenne/Becker Muscular Dystrophy may also have health implications for you. You may be at higher risk of high cholesterol or changes to your heart.

It's important to remember that no test can find everything. This screen lowers the chance of having or passing on the other conditions we tested for, 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 your results in more detail and discuss whether further steps are needed.

They may recommend genetic counselling to help you understand your reproductive risks and options before pregnancy. They may also recommend a referral to a cardiologist to check your cholesterol levels and heart health, in order to manage any risks early.

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Test: Basic Carrier Screen

Results

- [***] A variant in the CFTR gene was identified. Heterozygous for NM_000492.3:c.1521_1523del, p.(Phe508del).
- SMN1: Two or more gene copies detected.
- FMR1: 30 CGG repeats.

Interpretation

- [***] This patient is a carrier of cystic fibrosis, a multisystem autosomal recessive disorder characterised by thickened airway secretions, chronic lung disease, pancreatic insufficiency, malabsorption, and recurrent respiratory infections.
- This patient was not found to be a carrier of 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 these conditions.

Recommendations

- [***] Carrier screening for cystic fibrosis is recommended in the patient's reproductive partner to assess the couple's risk of having a child with cystic fibrosis.
- 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

- [***] A variant in the LDLR gene was identified. Heterozygous for NM_000527.5:c.259T>G, (p.Trp87Gly).
- No other 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

- [***] Variants in the LDLR gene are associated with autosomal dominant and recessive familial hypercholesterolaemia (FH), conditions characterised by markedly elevated LDL cholesterol, tendinous xanthomas, corneal arcus, and increased risk of coronary artery disease. In heterozygotes, coronary artery disease usually manifests in the fourth or fifth decade. In those with biallelic variants (autosomal recessive FH), coronary artery disease manifests much earlier, often in childhood or adolescence.
- This patient was not found to be a carrier for the other 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 these other screened conditions.

Recommendations

- [***] Carrier screening for the LDLR gene is recommended in the patient's reproductive partner to assess the couple's risk of having a child with autosomal recessive familial hypercholesterolaemia.
- [***] Even if this patient's partner is not a carrier, each child has a 50% (1 in 2) chance of inheriting this variant and developing autosomal dominant hypercholesterolaemia.
- [***] This result also has implications for this patient's personal health. Please see the 'Personal Genetic Screen' report for further details.
- 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.

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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

- [***] A variant in the LDLR gene was identified. Heterozygous for NM_000527.5:c.259T>G, (p.Trp87Gly).
- No other reportable variants were identified.

Interpretation

- [***] Variants in the LDLR gene are associated with autosomal dominant and recessive familial hypercholesterolaemia (FH), conditions characterised by markedly elevated LDL cholesterol, tendinous xanthomas, corneal arcus, and increased risk of coronary artery disease. In heterozygotes, coronary artery disease usually manifests in the fourth or fifth decade. In those with biallelic variants (autosomal recessive FH), coronary artery disease manifests much earlier, often in childhood or adolescence.
- [***] This result would support a clinical diagnosis of LDLR-associated familial hypercholesterolaemia in this patient.
- In the absence of relevant family history, this result significantly reduces (but does not eliminate) the likelihood of a clinically significant variant in the other screened genes.
- This result does not rule out the possibility of developing health conditions in the future, including cancer or cardiac disease.

Recommendations

- [***] Correlation with the patient's clinical features and LDL cholesterol levels is recommended. Referral to a cardiologist should be considered for further evaluation and management of hypercholesterolaemia. Professional genetic counselling by a clinical geneticist or genetic counsellor is recommended.
- [***] This result also has implications for this patient's future reproductive planning. Please see the 'Expanded Carrier Screen' report for further details.
- 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

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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

- [***] A variant in the DMD gene was detected. Heterozygous for a deletion of exons 8 to 17 of the DMD gene (NM_004006.3).
- [***] ISCN 2020 nomenclature: arr[GRCh37] Xp21.1(32544026_32764130)x1
- No other reportable copy number variants were identified.

Interpretation

- [***] This patient is a carrier of a pathogenic DMD gene deletion associated with dystrophinopathies, a spectrum of muscle disorders that includes Duchenne and Becker muscular dystrophy. Male individuals who inherit a pathogenic DMD variant are typically affected, while female individuals are usually asymptomatic carriers.
- [***] This deletion is predicted to be out-of-frame. Out-of-frame deletions are typically, though not always, associated with the more severe Duchenne muscular dystrophy.
- [***] Each child has a 50% (1 in 2) chance of inheriting this variant. Male children who inherit the variant are typically affected, while female children who inherit the variant are usually carriers and may be asymptomatic or mildly affected.
- This result does not exclude the following possibilities:
    - Other 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

- [***] Professional genetic counselling by a clinical geneticist or genetic counsellor is recommended. Please consider prenatal and/or preimplantation genetic testing for future pregnancies.
- [***] Heterozygous female carriers of DMD gene variants are themselves at increased risk of dilated cardiomyopathy. Please consider referral to a cardiologist for further evaluation and management.
- 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.

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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	Laboratory 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	**Laboratory 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).