PATERNITY For Life
DNA TESTING SPECIALISTS

Focusing on
Personalised Medicine

PRECONCEPTION SCREENING

Genetic testing is performed on a buccal swab taken from the mouth using DNA, the genetic material which contains all the genes, extracted from the sample.

Our preconception screening test can be used for preconception testing of prospective parents to screen for potential overlapping genetic disorders as well as diagnosis of inherited conditions.

The preconception screening panel tests the coding exons of over 300 genes associated with more than 700 unique commonly inherited diseases and Mendelian diseases, including the most common forms of inherited deafness, blindness, heart disease, Parkinson's disease, immunodeficiency, and various ataxias, anemias, and treatable metabolic syndromes.

This panel has been developed in consultation with clinical molecular geneticists. Our technology uses a multiplexed approach using the latest technology. Multiplexing enables the testing of multiple genes simultaneously with high cost effectiveness compared to single gene testing.

Whilst individually each disease is rare, about 25% of people will carry at least one abnormal mutation. These conditions are not routinely detected using chorionic villous sampling or amniocentesis.

These disorders are usually autosomal recessive, which means that a baby must inherit a defective gene from each parent to have the disease. For autosomal recessive conditions, if a person is a carrier of the disease, this means they have one defective copy of the gene and one normal copy. Carriers don't usually have any symptoms of the disease.

If both you and your partner are carriers of an autosomal recessive disorder like cystic fibrosis your child will have a 1 in 4 chance of inheriting one defective gene from each of you and being born with the disease.

Preconception screening is not routinely offered and may only be discussed if there is a risk factor such as a family history or if a person belongs to a specific ethic group associated with an increased risk. However, most people don't know they may be at increased risk so there is no easy way of determining who is at an increased risk of being a carrier for any specific gene mutation.

The best approach is to have the option of preconception screening before you try to conceive. The inherited disease screening panel enables screening for a wide range of inherited diseases (expanded carrier screening), however, it does not cover every possible inherited disease. Specific diseases and genes covered are listed on the reverse. If you are discussing conception with a doctor then if preconception screening identifies you and your partner are both carriers for a condition, you will be able to consider a wider range of options. Genetic counselling may also be an option and they can provide more information about the condition and discuss reproductive choices.

How do I organise Testing?

Email: enquiries@paternityforlife.co.uk | Web: www.paternityforlife.co.uk

DISEASE	GENE SYMBOL
Agammaglobulinemia, X-Linked, Type 1	*BTK*
Alagille Syndrome	*JAG1*
Alopecia Universalis Congenita (ALUNC)	*HR*
Alpers Syndrome	*POLG*
Alpha-1-Antitrypsin Deficiency	*SERPINA1*
Alpha-Thalassemia - Southeast Asia	*HBA2*
Alport Syndrome	*COL4A5*
Amyotrophic Lateral Sclerosis (Lou Gehrig's Disease)	*SOD1*
Androgen Insensitivity Syndrome	*AKR1B1*
Angioedema, Hereditary, Types I and II	*SERPING1*
Aniridia	*PAX6*
APC-Associated Polyposis Conditions	*APC*
Argininosuccinate Lyase Deficiency	*ASL*
Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	*DSC2,DSG2,DSP,JUP,PKP2,RYR2,TMEM43*
Arylsulfatase A Deficiency	*ARSA*
Ataxia with Oculomotor Apraxia Type 2	*APTX*
Ataxia with Vitamin E Deficiency	*TTPA*
Ataxia-Telangiectasia	*ATM*
Atrial Septal Defect	*GATA4*
Autoimmune Polyendocrine Syndrome	*AIRE*
Beta-Hydroxyisobutyryl CoA Deacylase Deficiency (HIBCH Deficiency)	*HIBCH*
Biotinidase Deficiency	*BTD*
Blepharophimosis-Ptosis-Epicanthus Inversus	*FOXL2*
Brachydactyly	*GDF5*
Brachydactyly, Type B1	*ROR2*
Branchiootorenal Spectrum Disorders	*EYA1,SIX1,SIX5*
Brugada Syndrome	*CACNA1C,CACNB2,GPD1L,HCN4,KCNE3, SCN1B,SCN3B,SCN5A*
Campomelic Dysplasia	*SOX9*
Canavan	*ASPA*
Cardiomyopathy (Dilated)	*STARD3, TAZ*
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	*CASQ2*
Cerebrotendinous Xanthomatosis	*CYP27A1*
Ceroid Lipofuscinoses (Batten Disease)	*PPT1*
Charcot-Marie-Tooth Disease Type 2B	*DNM2*
Charcot-Marie-Tooth Neuropathy Type 1A	*PMP22*
Charcot-Marie-Tooth Neuropathy Type 1B	*MPZ*
Charcot-Marie-Tooth Neuropathy Type 2A	*MFN2*
Charge Syndrome	*CHD7*
Cherubism	*SH3BP2*
Choroideremia	*CHM*
Citrin Deficiency	*SLC25A13*
Coffin-Lowry Syndrome	*RPS6KA3*
Congenital Cataracts, Facial Dysmorphism, and Neuropathy	*CTDP1*
Congenital Disorder of Glycosylation Type 1a	*PMM2*
Congenital Myasthenic Syndromes	*CHRNA1, CHRNB1,CHRND,CHRNE,DOK7,RAPSN,CHAT*
Cornelia de Lange Syndrome	*NIPBL*
Familial Hypertrophic Cardiomyopathy	*ACTC1,CALR3, CAV3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK, MYOZ2, PRKAG2, RPS7, SLC25A4, TNNT2, TPM1, VCL*
Cystinosis	*CTNS*
Darier Disease	*ATP2A2*
Diamond-Blackfan Anemia	*RPL11, RPL35A, RPS10, RPS19, RPS24, RPS26*
Dilated Cardiomyopathy	*DES, LAMP2, LDB3, SGCD, TNNC1, TNNI3*
Dilated Cardiomyopathy 1AA	*ACTN2*
Dilated Cardiomyopathy 1HH	*BAG3*
Dilated Cardiomyopathy 1O	*ABCC9*
Dilated Cardiomyopathy 1P	*PLN*
Double Cortex Syndrome	*DCX*
Duane Syndrome - Autosomal Dominant	*SALL4*
Duchenne/Becker Muscular Dystrophy	*DMD*
Dysferlinopathy	*DYSF*
Dyskeratosis Congenita	*DKC1*
Early-Onset Familial Alzheimer Disease	*APP, PSEN1, PSEN2*
Ehlers-Danlos Syndrome	*COL3A1, COL5A1, COL5A2*
Ehlers-Danlos Syndrome, Hypermobility Type	*TNXB, PLOD1, EMD*
Epidermolysis Bullosa Simplex	*COL7A1, ITGB4, KRT14, KRT5, LAMB3, PLEC*
Exostoses, Multiple, Type 1	*EXT1*
Fabry Disease	*GLA*
Facioscapulohumeral Muscular Dystrophy	*FRG1*
Familial Dysautonomia (HSAN III)	*IKBKAP*
Familial Transthyretin Amyloidosis	*TTR*
Fanconi Anemia	*FANCA, FANCC, FANCF, FANCG*
FGFR-Related Craniosynostosis Syndromes	*FGFR1*
Friedreich Ataxia	*FXN*
FRMD7-Related Infantile Nystagmus	*FRMD7*
Fryns Syndrome	*MED12*
Galactosemia	*GALT*
Gaucher Disease	*GBA*
Glycine Encephalopathy	*AMT, GCSH, GLDC*
Glycogen Storage Disease Type VI	*GBE1*
Hemophilia A	*F8*
Hemophilia B	*F9*
Hereditary Hemorrhagic Telangiectasia	*ENG*
Hexosaminidase A Deficiency	*HEXA*
HFE-Associated Hereditary Hemochromatosis	*HFE*
Holoprosencephaly-7 & Basal Cell Nevus Syndrome	*PTCH1*
Holt-Oram Syndrome	*TBX5*
Hunter Syndrome (MPSII)	*IDS, IDUA*

DISEASE	GENE SYMBOL
Hydroxymethylbilane Synthase (HMBS) Deficiency	*HMBS*
Hypochondroplasia	*FGFR3*
Hypophosphatasia	*ALPL*
Inclusion Body Myopathy 2	*GNE*
Inherited Deafness	*COLL11A2, KCNQ4*
Inherited Deafness, Top Genes	*GJB2, GJB3, GJB6*
Juvenile Polyposis Syndrome	*BMPR1A, SMAD4*
Leber Congenital Amaurosis	*AIPL1, CEP290. CRB1, GUCY2D, IMPDH1, RDH12, RPE65, RPGRIP1*
Li-Fraumeni Syndrome	*CHEK2, TP53*
Limb-Girdle Muscular Dystrophy Type 2A - Calpainopathy	*CAPN3*
Limb-Girdle Muscular Dystrophy, Type 1B	*LMNA*
Lissencephaly 1	*PAFAH1B1*
Long QT Syndrome, Autosomal Dominant	*AKAP9, KCNE1, KCNE2, KCNH2, KCNQ1, SCN4B,SNTA1, ANK2*
Lowe Syndrome	*OCRL*
Malignant Hyperthermia Susceptibility	*RYR1*
Maple Syrup Urine Disease	*BCKDHA, BCKDHB, DBT,DLD*
Marfan Syndrome	*FBN1*
MECP2-Rett Syndrome	*MECP2*
Menkes/ATP7A-Related Copper Transport Disease	*ATP7A*
Methylmalonic Acidemia	*MMAA, MMAB,MMACHC,MUT*
Mucolipidosis II	*GNPTAB*
Multiple Endocrine Neoplasia Type 1	*MEN1*
Multiple Endocrine Neoplasia Type 2	*RET*
Myotonia Congenita	*CLCN1*
Nemaline Myopathy	*TNNT1*
Neonatal Adrenoleucodystrophy	*PEX5*
Neurofibromatosis Type 1	*NF1*
Neurofibromatosis Type 2	*NF2*
Niemann-Pick Disease Type C1	*NPC1*
Niemann-Pick Disease Type C2	*NPC2*
Noonan Syndrome	*KRAS,NRAS,PTPN11, RAF1, SOS1*
Ocular Albinism, X-Linked	*GPR143*
Oculocutaneous Albinism Type 1	*TYR*
Oculocutaneous Albinism Type 2	*MCR1, OCA2*
Oculopharyngeal Muscular Dystrophy	*PABPN1*
Ornithine Transcarbamylase Deficiency	*OTC*
Osteogenesis Imperfecta	*COL1A1, COL1A2*
Parkinson Disease	*FBX07,LRRK2,PINK1,SNCA*
Parkinson-Dementia Syndrome	*MAPT*
Pendred Syndrome/Syndromic Deafness	*SLC26A4*
Peroxisome Biogenesis, Zellweger	*PEX10, PEX13, PEX14, PEX19, PEX26, PEX3*
Phenylketonuria (PKU)	*PAH*
Polycystic Kidney Disease, Autosomal Dominant	*PKD1, PKD2, PKHD1*
Pompe Disease -GSD II	*GAA*
Primary Ciliary Dyskinesia	*CCDC39,CCDC40, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, RSPH4A, RSPH9, TXNDC3*
Retinitis Pigmentosa	*ABCA4, ARL6, BEST1, CA4, CERKL, CNGB1, CRX, EYS, FSCN2, KLHL7,LRAT, MAPRE2, MERTK, NR2E3, NUNDT19, PRCD, PROM1, PRPF31, PRPF8, PRPH2, RHO, RP9, RPGR,SEMA4A, SNRNP200, TOPORS, TULP1*
Retinoblastoma0	*RB1*
Saethre-Chotzen Syndrome	*TWIST1*
SCN9A-Related Inherited Erythromelalgia	*SCN9A*
Severe Combined Immunodeficiency	*ADA, RAG1, RAG2*
Short QT Syndrome	*KCNJ2*
Sickle Cell Disease Beta-Thalassemia	*HBB*
Smith-Lemli-Opitz Syndrome	*DHCR7*
Smith-Magenis Syndrome	*RAI1*
Sotos Syndrome	*NSD1*
Spastic Paraplegia 7	*SPG7*
Spastic Paraplegia 8	*KIAA0196*
Spastic Paraplegia Type 1 - L1 Syndrome	*L1CAM*
Spastic Paraplegia-3A	*ATL1*
Spinocerebellar Ataxia 1	*ATXN1, ATXN2, ATXN7*
Stickler Syndrome	*COL9A1*
Stickler Syndrome, AD	*COL11A1, COL2A1*
Supravalvular Aortic Stenosis	*ELN*
Tetralogy of Fallot	*NKX2-5*
Thoracic Aortic Aneurysms and Aortic Dissections	*ACTA2, COL4A1, MYH11, SMAD3, TGFBR1, TGFBR2*
Treacher Collins Syndrome	*TCOF1*
Trimethylaminuria	*FMO3*
Tuberous Sclerosis Complex	*TSC1, TSC2*
Turcot Syndrome	*MLH1, MSH2*
Usher Syndrome Type 1	*CDH23, MYO7A, PCDH15, USH1C*
Usher Syndrome Type 2	*USH2A*
Very Long Chain Acyl-Coenzyme A Dehydrogenase Deficiency	*ACADVL*
von Hippel-Lindau Syndrome	*VHL*
Waardenburg Syndrome, Type 1	*PAX3*
Werner Syndrome	*WRN*
Wilms Tumor, Classic	*WT1, GPC3*
Wilson Disease	*ATP7B*
Wiskott-Aldrich Syndrome	*WAS*
X-Linked Adrenoleukodystrophy	*ABCD1*
X-Linked Dystonia-Parkinsonism	*TAF1*
X-Linked Juvenile Retinoschisis	*RS1*
X-Linked Myotubular Myopathy	*MTM1*
X-Linked SCIDS	*IL2RG*
Zellweger Syndrome	*PEX1*