

MOLECULAR GENETICS

For further information and clarification of fee please contact SCI on (02) 9855 5369 or 1800 010 447

Alpha Thalassaemia

SEA Gene Assay - Alpha Thalassaemia, Alpha Globulin Gene Test, A-Thalassaemia Gene Test

Asthma - Beta 2 Adrenoreceptor Gene

B-2 Adenoreceptor Mutation (Asthma)

C1 Esterase Inhibitor Gene Assay

Subcutaneous Oedema, C1 Inhibitor Deficiency Gene Assay, Serping-1 Gene Assay, Serpin Gene Test, Type 1,2 HA Gene Test (C1 Esterase Gene Test)

Coeliac Tissue Typing

HLA DR/DQ

Cystic Fibrosis dF508

CF delta F508 (Cystic Fibrosis), CF delta 5018 (Cystic Fibrosis), DF508 (Cystic Fibrosis delta F508 mutation), Delta F508 (Cystic Fibrosis)

Cystic Fibrosis PCR

32 common mutations tested: deltaF508, S549N, S549R, R553X, G551D, V520F, deltaI507, 3876delA, 1717-1G>A, G542X, R560T, 3120+1G>A, A455E, R117H, 394delTT, 2183AA>G, 2184delA, 2789+5G>A, 1898+1G>A, 621+1G>T, 711+1G>T, G85E, R347P, R347H, I148T, W1282X, R334W, 1078delT, 3849+10kbC>T, R1162X, N1303K, 3659delC,3905insT

DAZ GENE

AZF PCR (DAZ Gene) , Y Micro Deletion PCR

DNA Relationship & Paternity Testing

Familial Mediterranean Fever Gene Test

FMF (Familial Mediterranean Fever), Familial Fever (Familial Mediterranean Fever), Mediterranean Fever (Familial Mediterranean Fever)

Familial Hibernian Fever (TRAPs)

TRAPS (Familial Hibernian Fever), FHF (Familial Hibernian Fever)

Fragile X

Fragile X PCR, DNA Probe Fragile X, FRXDP (Fragile X DNA Probe), FRX (Fragile X), FMR1 Premutation (Fragile X)

JAK2 Gene Test

Tyrosine Kinase Gene Test, Janus Kinase 2, Polycythaemia Gene Testing, Myelofibrosis (Familial), Myeloproliferative Disease, Polycythaemia Rubra Vera, PRV Gene Test (Polycythaemia Rubra Vera), V617F (JAK2 Gene Test)

Hereditary Angioedema Factor XII HAE Test

Type III Angioedema, Factor XII Mutation - Hereditary Angioedema, Factor XII HAE Test (Type III Hereditary Angioedema), Type III HA (Hereditary Angioedema), Type III Hereditary Angioedema Test

HLA B27

Human Leucocyte Antigen B27, Seronegative spondyloarthropathies

HLA B1502

Human Leucocyte Antigen B1502, ADR-Carbamazepine (Adverse Drug Reaction),

HLA B5701

ADR-Abacavir (Adverse Drug Reaction) (HIV patients), Human Leucocyte Antigen B5701

Narcolepsy Tissue Typing

Sjogren's Tissue Typing, HLA DW3 (Tissue Typing), Tissue Typing (Narcolepsy/Sjogren's/Sleep Apnoea)

PRE NATAL TESTING

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First Trimester Screen (FTS)

PAPPA (Pregnancy Associated Plasma Protein A), FTS, BHCG Free, Free BHCG, uE3, Pregnancy Associated Plasma Protein A, Downs Syndrome Screen

Second Trimester Screen (STS)

STS, DS Screen (Downs Syndrome), Triple Screen, Biochem Aneuploidy, Downs Syndrome Screen, Oestriol

Alpha Feto Protein (AFP)

Neural Tube Defect, NTD, Alpha Fetoprotein - Maternal Serum, AFP - Pregnant, AFP, Maternal AFP, MS AFP