3A Laboratories

Alphabetical List of Tests & Procedures

Molecular Biology
Molecular Genetics
Cytogenetics

2009-2010

13-18-21-X/Y CHROMOSOMES (FISH), AMNIOTIC FLUID
1p36 deletion FISH
21-Hydroxylase Deficiency (Congenital Adrenal Hyperplasia) -
21-HYDROXYLASE GENE, BLOOD
22q11 deletion (Di George Syndrome) -Karyotype FISH 7 mutations + deletions/duplications
A
ABCA4-Related Disorders - ABCA4 Sequencing
ABCA4-Related Disorders - ABCA4 Sequencing - KFM
ABCA4-Related Disorders - ABCA4 Sequencing - Prenatal
AcHE
Achondroplasia
Achondroplasia - 2 common mutations in FGFR3 (c.1138G>A + c.1138G>C)
ACHONDROPLASIA GENE, BLOOD
Acoustic Neuroma (NF2) - sequencing + deletions/duplications
Acylcarnitine/Carnitine Combination, Plasma
Adenosine Deaminase Deficiency, Amniotic Cell Culture
Adenosine Deaminase Deficiency, Cultured Chorionic Villus Cells
Adenosine Deaminase Deficiency, Red Blood Cells
Adenosine Deaminase Deficiency, Skin Fibroblast Culture
Adenosine Deaminase Deficiency, White Blood Cells
ad-PEO 2 (ANT1/SLC25A4) Sequencing
ad-PEO 2 (ANT1/SLC25A4) Sequencing - KFM
ad-PEO 2 (ANT1/SLC25A4) Sequencing - Prenatal
ad-PEO 3 (TWINKLE/C10orf2) Sequencing
ad-PEO 3 (TWINKLE/C10orf2) Sequencing - KFM
ad-PEO 3 (TWINKLE/C10orf2) Sequencing - Prenatal
ad-PEO with mtDNA Deletions-4 (PEOA4) - POLG2 Sequencing
ad-PEO with mtDNA Deletions-4 (PEOA4) - POLG2 Sequencing - KFM
Adrenal Hypoplasia Congenita FISH
Alagille Syndrome FISH
Alpha 1 Antitrypsin genotype, (PI*M, PI*S, PI*Z)
Alpha Fetoprotein on Amniotic fluid
Alpha Thalassaemia (alpha-globin sequencing)
Alpha Thalassaemia (alpha-globin sequencing) (multiplex PCR for common large deletions)
ALPHA-THALASSEMIA GENE, BLOOD
Alzheimer (Presenilin 1) Early onset dementia
AML1/ETO t(8;21) TRANSLOCATION, BLOOD
Amnio PCR only (Trisomy analysis QF-PCR)
Amniocentesis culture (karyotype)
AMT (11q22.3) DELETION, BONE MARROW/BLOOD
AMYLOGENIN GENE, BLOOD
Androgen Insensitivity (AR known mutation in relative)
Androgen Insensitivity Diagnosis (AR sequencing + deletions/duplications)
Androgen Insensitivity Syndrome - AR Sequencing
Androgen Insensitivity Syndrome - AR Sequencing - KFM
Androgen Insensitivity Syndrome - AR Sequencing - Prenatal
Aneuploidy FISH (Amnio or CVS)
Angelman Syndrome – karyotype/FISH
Angelman Syndrome – methylation PCR
Angelman Syndrome – UBE3A hotspot sequencing
Angelman Syndrome - UBE3A Sequencing
Angelman Syndrome - UBE3A Sequencing - KFM

Angelman Syndrome - UBE3A Sequencing - Prenatal
Angelman Syndrome – UPD 15 (parents and child)
Angelman Syndrome (Methylation)
Angelman Syndrome FISH
ANGELMAN SYNDROME, GENETIC STUDY
Aniridia Tier 1 – PAX6 deletions
Aniridia Tier 2 – PAX6 sequencing
Apo E genotyping by PCR (E2, E3, E4)
APOLIPOPROTEIN E GENOTYPE, BLOOD
Arginase Deficiency - ARG1 Sequencing
Arginase Deficiency - ARG1 Sequencing - KFM
Arginase Deficiency - ARG1 Sequencing - Prenatal
Argininemia, Liver
Argininemia, Red Blood Cells
Argininosuccinate Aciduria - ASL Sequencing
Argininosuccinate Aciduria - ASL Sequencing - KFM
Argininosuccinate Aciduria - ASL Sequencing - Prenatal
Argininosuccinate Lyase Deficiency, Amniotic Cell Culture
Argininosuccinate Lyase Deficiency, Cultured Chorionic Villus Cells
Argininosuccinate Lyase Deficiency, Liver
Argininosuccinate Lyase Deficiency, Red Blood Cells
Argininosuccinate Lyase Deficiency, Skin Fibroblast Culture
ARX Related Disorders
ARX Related Disorders - KFM
ARX Related Disorders - Prenatal
Ashkenazi Jewish Screen
Ashkenazic Disease Screen (ADS) (includes CF, Canavan, FD, and Tay-Sachs DNA)
Ashkenazic Disease Screen Plus (ADS+) (includes CF, Canavan, FD, Tay-Sachs DNA, and Tay-Sachs enzymatic)
Aspartylglucosaminuria, Skin Fibroblast Culture
ATPase Subunits Sequencing
ATPase Subunits Sequencing - KFM
Autoimmune Polyendocrinopathy-Candidiasis Ectodermal Dystrophy (APECED) - AIRE Sequencing - KFM
Autoimmune Polyendocrinopathy-Candidiasis Ectodermal Dystrophy (APECED) - AIRE Sequencing - Prenatal
Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED) - AIRE Sequencing
Autosomal Dominant Optic Atrophy 3 - OPA3 sequencing
Autosomal Dominant Optic Atrophy 3 - OPA3 sequencing - KFM
Autosomal Dominant PEO Panel (POLG1, ANT1, and TWINKLE)
AUTOSOMAL DOMINANT SPINOCEREBELLAR ATAXIA GENE, BLOOD
Azoospermia (Y deletion, karyotype, cystic fibrosis screen, AZFb)
B
Bannayan-Riley-Ruvalcaba Syndrome (BRRS) – PTEN sequencing
BCL1 GENE t(11;14)(q13;q32), BONE MARROW
BCL2 GENE t(14;18)(q32;q21), BONE MARROW
BCL6 GENE, BONE MARROW
BCR/ABL
BCR/ABL: t(9;22) [CML/ALL/AML] FISH Analysis
BCR-ABL GENE t(9;22)(q34;q11) QUANTIFICATION, BLOOD
BCR-ABL GENE t(9;22)(q34;q11), BLOOD
BCS1L Related Complex III Deficiency

BCS1L Related Complex III Deficiency - KFM
BCS1L Related Complex III Deficiency - Prenatal
Becker Muscular Dystrophy – deletions/duplications
Beckwith-Wiedemann Syndrome – methylation studies in KCNQ1OT1 (DMR2) + H19 (DMR1)
Beckwith-Wiedemann Syndrome (Familial) – CDKN1C sequencing
Beckwith-Wiedemann Syndrome FISH
Beta Thalassaemia (beta-globin sequencing)
BETA-THALASSEMIA GENE, BLOOD
Biotinidase Deficiency - BTD Sequencing
Biotinidase Deficiency - BTD Sequencing - KFM
Biotinidase, Serum
Bloom Syndrome – common c.2207-12 deletion
Bloom's Syndrome
Bloom's Syndrome - BLM Sequencing
Bloom's Syndrome - BLM Sequencing - KFM
Bloom's Syndrome - BLM Sequencing - Prenatal
BRCA1 GENE, BLOOD
BRCA2 GENE, BLOOD
C
CADASIL – NOTCH 3 Common mutations
Canavan Disease – 2 common Ashkenazi mutations p.Y231X + p.E285A
Canavan Disease Screen
Carbamoyl Phosphate Synthetase I Deficiency - CPS1 Sequencing
Carbamoyl Phosphate Synthetase I Deficiency - CPS1 Sequencing - KFM
Carbamoyl Phosphate Synthetase I Deficiency - CPS1 Sequencing - Prenatal
Carbamyl Phosphate Synthetase Deficiency, Liver
Carnitine Palmitoyltransferase 1 Deficiency - CPT1A Sequencing
Carnitine Palmitoyltransferase 1 Deficiency - CPT1A Sequencing - KFM
Carnitine Palmitoyltransferase 1 Deficiency - CPT1B Sequencing
Carnitine Palmitoyltransferase 1 Deficiency - CPT1B Sequencing - KFM
Carnitine Palmitoyltransferase II Deficiency - CPT2 Sequencing
Carnitine Palmitoyltransferase II Deficiency - CPT2 Sequencing - KFM
Carnitine, Free and Total, Plasma
Carnitine-Acylcarnitine Translocase Deficiency - SLC25A20 Sequencing
Carnitine-Acylcarnitine Translocase Deficiency - SLC25A20 Sequencing - KFM
Cartilage Hair Hypoplasia - RMRP Sequencing
Cartilage Hair Hypoplasia - RMRP Sequencing - KFM
Cartilage Hair Hypoplasia - RMRP Sequencing - Prenatal
CBFB: inv(16) [AML] FISH Analysis
CDKL5 Related Atypical Rett Syndrome Sequencing
CDKL5 Related Atypical Rett Syndrome Sequencing - KFM
CDKL5/STK9 Sequencing - Prenatal
Centromere FISH Panel
Charcot-Marie-Tooth Disease (CMT1A) FISH
CHARGE Syndrome
CHARGE Syndrome - CHD7 Sequencing
CHARGE Syndrome - CHD7 Sequencing - KFM
CHARGE Syndrome - CHD7 Sequencing - Prenatal
CHIC2: Deleted 4q [hypereosinophilic syndrome] FISH Analysis
CHOLIA I GENE, BLOOD
CHROMOSOMAL INSTABILITY, SYNDROME STUDY
Chromosomal Microarray 105K (CMA)

Chromosomal Microarray 105K (CMA) Prenatal - Amniotic Cell Culture
Chromosomal Microarray 105K (CMA) Prenatal - Cultured Chorionic Villus Cells
Chromosomal Microarray 105K (CMA) Prenatal - Tissue
Chromosomal Microarray Analysis (CMA) - SNP Array
CHROMOSOME 11 (FISH), BONE MARROW
CHROMOSOME 12 (FISH), BLOOD/BONE MARROW
CHROMOSOME 13 (FISH)
CHROMOSOME 15 (FISH)
CHROMOSOME 17 (FISH)
CHROMOSOME 18 (FISH)
CHROMOSOME 21 (FISH)
CHROMOSOME 22 (FISH), BLOOD
CHROMOSOME 7 (FISH), BLOOD
CHROMOSOME 8 (FISH), BLOOD/BONE MARROW
Chromosome Analysis - Blood
Chromosome Analysis - Bone Marrow
Chromosome Analysis - Oncology Blood
Chromosome Analysis (Amniocentesis) by culture
Chromosome Analysis (Amniocentesis) by PCR and culture
Chromosome Analysis (Chorionic Villus)
Chromosome Analysis (Chorionic Villus) – culture
Chromosome Analysis (CVS)
Chromosome Analysis (Products of Conception)
Chromosome Analysis (Slide for opinion)
Chromosome Analysis (Solid Tissue)
Chromosome Analysis + AFP
Chromosome Analysis, No AFP (AMNIO)
Chromosome Analysis+ AFP + AchE (AMNIO)
Chromosome Analysis+ AFP + AchE + FH (AMNIO)
CHROMOSOME INVERSION 16 Inv(16)(p13;q22), BLOOD/BONE MARROW
CHROMOSOME X/Y (FISH)
Chromosome Y Deletion – AZFa, AZFb, AZFc + SRY
CHROMOSOMES 13-18-20-21-22-X/Y, SEMEN
Chronic Granulomatous Disease Primary Screen – X-linked CYBB sequencing + NCF1 common GT deletion
Citrin Deficiency (Citrullinemia Type II) - SLC25A13 Sequencing
Citrin Deficiency (Citrullinemia Type II) - SLC25A13 Sequencing - KFM
Citrin Deficiency (Citrullinemia Type II) - SLC25A13 Sequencing - Prenatal
Citrullinemia Type I (CTLN1) - ASS Sequencing
Citrullinemia Type I (CTLN1) - ASS Sequencing - KFM
Citrullinemia Type I (CTLN1) - ASS Sequencing - Prenatal
Citrullinemia, Amniotic Cell Culture
Citrullinemia, Cultured Chorionic Villus Cells
Citrullinemia, Liver
Citrullinemia, Skin Fibroblast Culture
CLCN1 GENE SEQUENCING, BLOOD
Cleidocranial Dysplasia - RUNX2 Sequencing
Cleidocranial Dysplasia - RUNX2 Sequencing - KFM
Cleidocranial Dysplasia - RUNX2 Sequencing - Prenatal
CLL FISH Panel (6q, 13q14, 13q34, cen12, 17p13, 11q23)
COAGULATION FACTOR V GENE, BLOOD
COAGULATION FACTOR XII (GENETIC STUDY), BLOOD

Coeliac Disease (HLA DQ2/DQ8 markers)
Coenzyme Q10 Deficiency - COQ2 Sequencing
Coenzyme Q10 Deficiency - COQ2 Sequencing - KFM
Coenzyme Q10 Deficiency - COQ2 Sequencing - Prenatal
Coenzyme Q10 Deficiency - PDSS1 Sequencing
Coenzyme Q10 Deficiency - PDSS1 Sequencing - KFM
Coenzyme Q10 Deficiency - PDSS1 Sequencing - Prenatal
Coenzyme Q10 Deficiency - PDSS2 Sequencing
Coenzyme Q10 Deficiency - PDSS2 Sequencing - KFM
Coenzyme Q10 Deficiency - PDSS2 Sequencing - Prenatal
Complex 1 Deficiency - NDUFAF2 sequencing
Complex 1 Deficiency - NDUFAF2 sequencing - KFM
Complex I Deficiency - C6ORF66 Sequencing
Complex I Deficiency - C6ORF66 Sequencing - KFM
Complex I Deficiency - NDUFA1 Sequencing
Complex I Deficiency - NDUFA1 Sequencing - KFM
Complex I Deficiency - NDUFS3 sequencing
Complex I Deficiency - NDUFS3 sequencing - KFM
Complex I Deficiency - NDUFS4 sequencing - KFM
Complex I Deficiency - NDUFS6 sequencing
Complex I Deficiency - NDUFS6 sequencing - KFM
Complex I Deficiency - NDUFS7 sequencing
Complex I Deficiency - NDUFS7 sequencing - KFM
Complex I Deficiency - NDUFV1 sequencing
Complex I Deficiency - NDUFV1 sequencing - KFM
Complex I Deficiency- NDUFS4 sequencing
Complex I Subunits Sequencing
Complex I Subunits Sequencing - KFM
Complex IV (COX) Deficiency - COX10 Sequencing
Complex IV (COX) Deficiency - COX10 Sequencing - KFM
Complex IV (COX) Deficiency - COX10 Sequencing - Prenatal
Complex IV (COX) Deficiency - COX15 Sequencing
Complex IV (COX) Deficiency - COX15 Sequencing - KFM
Complex IV (COX) Deficiency - SCO1 Sequencing
Complex IV (COX) Deficiency - SCO1 Sequencing - KFM
Complex IV (COX) Deficiency - SCO1 Sequencing - Prenatal
Complex IV (COX) Deficiency - SCO2 Sequencing
Complex IV (COX) Deficiency - SCO2 Sequencing - KFM
Complex IV (COX) Deficiency - SCO2 Sequencing - Prenatal
Complex IV (COX) Deficiency - SURF1 Sequencing
Complex IV (COX) Deficiency - SURF1 Sequencing - KFM
Complex IV (COX) Deficiency - SURF1 Sequencing - Prenatal
Complex IV (COX) Deficiency - SURF1, SCO2, SCO1, & COX10 Sequencing
COMT (Val158Met MUTATION) GENE, BLOOD
Congenital Absence of Vas Deferens (karyotype, cystic fibrosis screen and polyT(5T) + Y deletions)
Congenital Adrenal Hyperplasia (21-Hydroxylase Deficiency) – 7 mutations + deletions/duplications
Connexin 26 Related Hereditary Hearing Loss - GJB2 Sequencing
Connexin 26 Related Hereditary Hearing Loss - GJB2 Sequencing - KFM
Connexin-26 Associated Deafness (sequencing + Connexin-30 common deletion)
CONSTITUTIONAL KARYOTYPE HIGH RESOLUTION, BLOOD
CONSTITUTIONAL KARYOTYPE, BLOOD

CONSTITUTIONAL KARYOTYPE, SKIN BIOPSY
Costello Syndrome - HRAS Sequencing
Costello Syndrome - HRAS Sequencing - KFM
Costello Syndrome - HRAS Sequencing - Prenatal
Cowden Syndrome – PTEN sequencing
Creatine Transporter (CRTR) Deficiency - CT1/SLC6A8 Sequencing
Creatine Transporter (CRTR) Deficiency - CT1/SLC6A8 Sequencing - KFM
Creatine/Guanidinoacetate Analysis, Plasma
Creatine/Guanidinoacetate Analysis, Urine
Cri du Chat Syndrome – karyotype/FISH
Cri-du-Chat FISH
CVS PCR and Culture
Cystic Fibrosis – 32 common mutations
Cystic Fibrosis – 7 Ashkenazi Jewish mutations
Cystic Fibrosis (CF) - CFTR Sequencing
Cystic Fibrosis (CF) - CFTR Sequencing - KFM
Cystic Fibrosis (CF) - CFTR Sequencing - Prenatal
Cystic Fibrosis (CF) - Prenatal
Cystic Fibrosis (CF) Mutation Analysis
Cystic Fibrosis (CFTR 5T) Variant Analysis
CYSTIC FIBROSIS GENE (CFTR) SPECIFIC MUTATION, BLOOD
CYSTIC FIBROSIS GENE (CFTR), BLOOD
Cystic Fibrosis Poly T (5T,7T,9T)
CYSTIC FIBROSIS-ASSOCIATED POLY T HAPLOTYPE, BLOOD
Cystine, White Blood Cells, Heparinized Blood
Cytochrome b Subunit
Cytochrome b Subunit - KFM
Cytochrome c Oxidase (COX) Subunits
Cytochrome c Oxidase (COX) Subunits - KFM
Cytochrome P450 microarray for 2D6 + 2C19 genotyping
Cytomegalovirus (CMV) DNA, Qualitative Real-Time PCR
D
D4Z4 (FACIO-SCAPULO-HUMERAL DYSTROPHY) GENE SEQUENCING, BLOOD
DAZ GENE (OLIGO/AZOOSPERMIA), BLOOD
Deafness – Connexin-26 sequencing + Connexin-30 common deletion
Deafness – Non-Syndromic Hearing Loss (10 mitochondrial mutations in MTRNR1 + MTTS1)
Deafness – Pendred Syndrome
Deafness-Dystonia-Optic Neuropathy Syndrome - TIMM8A Sequencing
Deafness-Dystonia-Optic Neuropathy Syndrome - TIMM8A Sequencing - KFM
Deletion 20q12: [MDS] FISH Analysis
Deletion 5: [MDS] FISH Analysis
Deletion 7: [MDS] FISH Analysis
Dentatorubral Pallidoluysian Atrophy (DRPLA)
Deoxyguanosine Kinase (DGUOK) - Prenatal
Deoxyguanosine Kinase (DGUOK) Sequencing
Deoxyguanosine Kinase (DGUOK) Sequencing - KFM
Di George Syndrome (22q deletion) – karyotype/FISH
Diamond-Blackfan Anemia - RPS19 Sequencing
Diamond-Blackfan Anemia - RPS19 Sequencing - KFM
Diamond-Blackfan Anemia - RPS19 Sequencing - Prenatal
DiGeorge Syndrome I (22q only) FISH
DiGeorge Syndrome II (10p only) FISH

Dihydropyrimidine Dehydrogenase deficiency (5-FU Toxicity) – common mutation IVS14+1G>A
Dihydropyrimidine Dehydrogenase deficiency (5-FU Toxicity) – rare mutations
DNA EXTRACTION
DNA Extraction & Storage – 3 years (longer upon request)
DNA fragmentation of semen
Duchenne Muscular Dystrophy –
Duchenne Muscular Dystrophy – deletions/duplications
DUCHENNE MUSCULAR DYSTROPHYMUSCULAR DYSTROPHY GENE, GENETIC STUDY
DVT/Pre-travel screen
E
Ectrodactyly-Ectodermal Dysplasia-Clefting
Ehlers-Danlos Syndrome - PLOD1 Sequencing
Ehlers-Danlos Syndrome - PLOD1 Sequencing - KFM
Ehlers-Danlos Syndrome - PLOD1 Sequencing - Prenatal
Emery Dreifuss Myotonic Dystrophy
Emery Dreifuss Myotonic Dystrophy, X-linked – Emerin sequencing
Encephalomyopathic mtDNA Depletion Syndrome - SUCLA2 Sequencing
Encephalomyopathic mtDNA Depletion Syndrome - SUCLA2 Sequencing - KFM
Erythrokeratodermia Variabilis – Connexin 31 sequencing
ETO/AML1: t(8;21) [AML] FISH Analysis
F
Fabry Disease
Fabry Disease - GLA Sequencing
Fabry Disease - GLA Sequencing - KFM
Fabry Disease - GLA Sequencing - Prenatal
Fabry Disease , Skin Fibroblast Culture
Fabry Disease , White Blood Cells
Facioscapulohumeral Muscular Dystropy (FSHD) – D4Z4 repeat deletion
Factor II Prothrombin – G20201A mutation
Factor V Leiden – G1691A mutation
Factor V Leiden Mutation
Factor VIII (Haemophilia A) – common 1/22 exon inversion
Factor VIII (Haemophilia A) – detection of known mutation in a relative
Factor VIII (Haemophilia A) – sequencing
Familial Adenomatous Polyposis (FAP) - APC Sequencing
Familial Adenomatous Polyposis (FAP) - APC Sequencing - KFM
Familial Adenomatous Polyposis (FAP) - APC Sequencing - Prenatal
Familial Adenomatous Polyposis (FAP) - Deletion/Duplication Assay
Familial Adenomatous Polyposis (FAP) - Deletion/Duplication Assay - KFM
FAMILIAL ADENOMATOUS POLYPOSIS GENE (APC), BLOOD
FAMILIAL AMYLOIDOSIS, BLOOD
Familial Cutaneous Malignant Melanoma – CDKN2A + CDK4 exon 2 sequencing
Familial Dysautonomia (FD)
Familial Dysautonomia (Reilly Day Syndrome) – 2 common Ashkenazi mutations p.R696P + c.2204+6T>C
Familial Hibernian Fever (TRAPS) – TNFRSF1A hotspot sequencing
Familial Hypercholesterolaemia – 20 common mutations in LDLR, APOB + PCSK9
Familial Hypercholesterolaemia - comprehensive LDLR sequencing + deletion/duplication analysis
Familial Hypercholesterolemia - LDLR Deletion/Duplication Analysis
Familial Hypercholesterolemia - LDLR Sequencing
Familial Hypercholesterolemia - LDLR Sequencing - KFM
Familial Mediterranean Fever – MEFV hotspot sequencing

Familial platelet disorder with associated myeloid malignancy - RUNX1 Sequencing
Familial platelet disorder with associated myeloid malignancy - RUNX1 Sequencing - KFM
Fanconi Anaemia – common Ashkenazi mutation IVS4+4A>T
Fatal Infantile Lactic Acidosis with mtDNA Depletion - SUCLG1 Sequencing
Fatal Infantile Lactic Acidosis with mtDNA Depletion - SUCLG1 Sequencing - KFM
Fatal Infantile Lactic Acidosis with mtDNA Depletion - SUCLG1 Sequencing - Prenatal
Female Specific Comprehensive Autism Panel
FETAL RhD STUDY, AMNIOTIC FLUID
FETAL RhD STUDY, PLASMA
FISH - STAT (13 or 18 or X&Y only)
FISH-Each Additional Probe
Focal Dermal Hypoplasia - PORCN Sequencing
Focal Dermal Hypoplasia - PORCN Sequencing - KFM
Focal Dermal Hypoplasia - PORCN Sequencing - Prenatal
Fragile X Syndrome
Fragile X Syndrome - Prenatal
Fragile X Syndrome Prenatal Diagnosis
Fragile X Syndrome screen
Friedreich Ataxia
Friedreich Ataxia – FXN repeat analysis
Friedreich Ataxia (FRDA) - FXN Sequencing
Friedreich Ataxia (FRDA) - FXN Sequencing - KFM
FRIEDREICH ATAXIA GENE (FRDA/X25), BLOOD
Fucosidosis, Skin Fibroblast Culture
Fucosidosis, White Blood Cells
G
Gain 8 chromosome FISH analysis
Galactosemia (UDPG Transferase), Red Blood Cells
Gaucher Disease – 5 common mutations
Gaucher Disease (GD)
Gaucher Disease, Skin Fibroblast Culture
Gaucher Disease, White Blood Cells
GENETIC REORGANIZATION OF IMMUNOGLOBULINS, BLOOD
Genetic Reproductive Profile (Male)
GILBERT SYNDROME, BLOOD
GIRDLE DYSTROPHY LGMD2A, BLOOD
GJB2 GENE (CONEXIN 26) SEQUENCING, BLOOD
GJB6-Related DFNB1 Hereditary Hearing Loss
Glycerol Kinase Deficiency FISH
Glycogen Storage Disease type 0 -GYS2 sequencing
Glycogen Storage Disease type 0 -GYS2 sequencing - KFM
Glycogen Storage Disease Type 1A (GSD1A) - G6PC Sequencing
Glycogen Storage Disease Type 1A (GSD1A) - G6PC Sequencing - KFM
GM1 Gangliosidosis (Morquio B, MPS IV B), Serum
GM1 Gangliosidosis (Morquio B, MPS IV B), Skin Fibroblast Culture
GM1 Gangliosidosis (Morquio B, MPS IV B), Skin Fibroblast Culture
GM1 Gangliosidosis (Morquio B, MPS IV B), White Blood Cells
Gorlin Syndrome (PTCH)
Guanidinoacetate Methyltransferase (GAMT) - GAMT Sequencing
Guanidinoacetate Methyltransferase (GAMT) - GAMT Sequencing - KFM
Guanidinoacetate Methyltransferase (GAMT) - GAMT Sequencing - Prenatal
H

Haemochromatosis – HFE common mutations C282Y + H63D
Haemophilia A (Factor VIII) – common 1/22 exon inversion
Haemophilia A (Factor VIII) – detection of known mutation in a relative
Haemophilia A (Factor VIII) – sequencing
Haemophilia B (Factor IX) – mutation screen
Hay-Wells (Ectrodactyly-Ectodermal Dysplasia-Clefting) – hotspot TP63 sequencing
HEMATOLOGIC KARYOTYPE, BONE MARROW
HEMATOLOGIC KARYOTYPE, LYMPH NODE
Heme/Onc Chip-44K Oligo Array
HEMOCHROMATOSIS GENE (C282Y,H63D), BLOOD
HEMOCHROMATOSIS GENE (S65C), BLOOD
HEMOPHILIA A GENE, AMNIOTIC FLUID
HEMOPHILIA A GENE, BLOOD
HEMOPHILIA B GENE A233T, BLOOD
Hepatocerebral mtDNA Depletion Panel (POLG1, DGUOK & MPV17)
Hereditary Angioedema – C1NH detection of known mutation in a relative
Hereditary Angioedema – C1NH sequencing
Hereditary Fructose Intolerance - ALDOB Sequencing
Hereditary Fructose Intolerance - ALDOB Sequencing - KFM
Hereditary Fructose Intolerance - ALDOB Sequencing - Prenatal
Hereditary Hemochromatosis
Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) / Inherited Peripheral Neuropathies FISH
Hereditary Non-Polyposis Colon Cancer (HNPCC) Panel (MLH1, MSH2, MSH6)
Hereditary Non-Polyposis Colon Cancer (HNPCC): MLH1 Del/Dup Assay - KFM
Hereditary Non-Polyposis Colon Cancer (HNPCC): MLH1 Deletion/Duplication Assay
Hereditary Non-Polyposis Colon Cancer (HNPCC): MLH1 Sequencing
Hereditary Non-Polyposis Colon Cancer (HNPCC): MLH1 Sequencing - KFM
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSH2 Del/Dup Assay - KFM
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSH2 Deletion/Duplication Assay
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSH2 Sequencing
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSH2 Sequencing - KFM
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSH6 Sequencing
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSH6 Sequencing - KFM
Hereditary Non-Polyposis Colon Cancer (HNPCC): MSI & IHC
Hereditary Pancreatitis – PRSS1 hotspot sequencing + deletions/duplications + SPINK1-N34S common mutation
Herpes Simplex Virus, Type 1 & 2 DNA, Real-Time PCR
HFE gene (Haemochromatosis) – common mutations C282Y + H63D
HLA Tissue Typing A
HLA Tissue Typing A/B/Cw/DRB1/DQB1 (Class I & II)
HLA Tissue Typing A/B/DRB1/DQB1
HLA Tissue Typing A+B
HLA Tissue Typing A+B+Cw (Class I)
HLA Tissue Typing B
HLA Tissue Typing B27
HLA Tissue Typing B51 only
HLA Tissue Typing B5701
HLA Tissue Typing Coeliac Disease (DQ2/DQ8)
HLA Tissue Typing Cw
HLA Tissue Typing DRB1*
HLA Tissue Typing DRB1*/DQB1* (Class II)
HLA Tissue Typing Narcolepsy – DRB1*15/DQB1*06

Holocarboxylase Deficiency - HLCS sequencing
Holocarboxylase Deficiency - HLCS sequencing - KFM
Holoprosencephaly Panel – hotspot SHH+TGIF+ZIC2+SIX3 sequencing
Homocysteine, Plasma
HPN CLONALITY
HUNTINGTON CHOREA GENE (IT15), BLOOD
Huntington's Disease
Huntington's Disease - Prenatal
Hurler Disease (Hurler-Scheie, MPS I), Skin Fibroblast Culture
Hurler Disease (Hurler-Scheie, MPS I), White Blood Cells
Hyper-IgD Syndrome – MVK hotspot sequencing
HYPOACHONDROPLASIA GENE, BLOOD
I
IGH/BCL2: t(14;18) [follicular lymphoma] FISH Analysis
IGH/CCND1: t(14;11) [mantle cell lymphoma] FISH Analysis
Incontinentia Pigmenti – NEMO common mutation
Incontinentia Pigmenti (IP)
Incontinentia Pigmenti (IP) - Prenatal
Infantile Hepatic mtDNA Depletion - MPV17 Sequencing
Infantile Hepatic mtDNA Depletion - MPV17 Sequencing - KFM
Infantile Hepatic mtDNA Depletion - MPV17 Sequencing - Prenatal
Infertility Profile 2 (Male)
Iron overload Profile
Isolated Lissencephaly FISH
J
Jak 2 – V617F mutation
JAK2 GENE (V617F MUTATION), BLOOD
K
Kallman Syndrome FISH
Kallmann Syndrome – karyotype/FISH
KALLMANN SYNDROME, GENETIC STUDY
KARYOTYPE HEMOPATHIES, BLOOD
KARYOTYPE, FETAL TISSUE
Kennedy Disease
Kennedy Disease (Spinal Bulbar Muscular Atrophy) – AR repeat expansion
Krabbe Deficiency - GALC Sequencing
Krabbe Deficiency - GALC Sequencing - KFM
Krabbe Deficiency - GALC Sequencing - Prenatal
Krabbe Disease, Skin Fibroblast Culture
Krabbe Disease, White Blood Cells
L
Langer Giedion Syndrome (includes Ext 1 & TRPS 1) FISH Panel
L-Arginine:Glycine Amidinotransferase Deficiency - GATM sequencing
L-Arginine:Glycine Amidinotransferase Deficiency - GATM sequencing - KFM
L-Arginine:Glycine Amidinotransferase Deficiency - GATM sequencing - Prenatal
Lebers Hereditary Optic Neuropathy – LHON
Leber's Optic Neuropathy (LON) Point Mutations
Leber's Optic Neuropathy (LON) Point Mutations - KFM
LEIGH SYNDROME, GENETIC STUDY
Lesch-Nyhan - HPRT1 Sequencing
Lesch-Nyhan - HPRT1 Sequencing - KFM
Lesch-Nyhan - HPRT1 Sequencing - Prenatal

Lesch-Nyhan Disease, Amniotic Cell Culture
Lesch-Nyhan Disease, Cultured Chorionic Villus Cells
Lesch-Nyhan Disease, Red Blood Cells
Lesch-Nyhan Disease, Skin Fibroblast Culture
Leukoencephalopathy w/ Vanishing White Matter (VWN) - EIF2B5 Sequencing
Leukoencephalopathy w/ Vanishing White Matter (VWN) - EIF2B5 Sequencing - KFM
Leukoencephalopathy w/ Vanishing White Matter (VWN) - EIF2B5 Sequencing - Prenatal
LHON SYNDROME, GENETIC STUDY
Li-Fraumeni syndrome (LFS) - P53 Gene Mutation Analysis
Li-Fraumeni syndrome (LFS) - P53 Gene Mutation Analysis - KFM
Loeys-Dietz (Marfan-like) Syndrome – TGFBRI + TGFBR2 sequencing+ deletions/duplications
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) - Common Mutation Analysis (1528G>C & 1132>T)
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) - KFM
Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADL)
Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADL) - KFM
Long QT Syndrome Panel – KCNQ1+HERG+KCNE1+SCN5A+KCNE2 (sequencing of 20 hotspot exons)
Lowe (Oculocerebrorenal) Syndrome – OCRL sequencing+ large deletions
Lowe Syndrome - OCRL Sequencing
Lowe Syndrome - OCRL Sequencing - KFM
Lowe Syndrome - OCRL Sequencing - Prenatal
Lowe Syndrome, Amniotic Cell Culture
Lowe Syndrome, Cultured Chorionic Villus Cells
Lowe Syndrome, Skin Fibroblast Culture
M
Male Genetic Reproductive Profile
Male Specific Comprehensive Autism Panel
Mannosidosis - Serum
Mannosidosis - Skin Fibroblast Culture
Mannosidosis - White Blood Cells
Maple Syrup Urine Disease Type 3 - DLD Sequencing
Maple Syrup Urine Disease Type 3 - DLD Sequencing - KFM
Marfan Syndrome – FBN1 sequencing+ deletions/duplications
Marfan-like (Loeys-Dietz) Syndrome – TGFBRI + TGFBR2 sequencing+ deletions/duplications
MARIE CHARCOT TOOTH (CMT1/CMT2) GENE (PMP22), BLOOD
Maroteaux-Lamy (MPS VI), Skin Fibroblast Culture
Maternal Cell Contamination (performed on all prenatals)
Maternally Inherited Sensorineural Hearing Loss (MISNHL)
Maternally Inherited Sensorineural Hearing Loss (MISNHL) - KFM
Maturity-Onset Diabetes of the Young Type 2 (MODY2) – glucokinase sequencing
McCune-Albright Syndrome – 2 common mutations in GNAS1
MEDITERRANEAN FEVER (E148Q AND EXON 10 MUTATION), BLOOD
MEDITERRANEAN FEVER (E148Q MUTATION), BLOOD
MEDITERRANEAN FEVER (EXON 10)(MEFV), BLOOD
Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD)
Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD) - KFM
Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD) - One Mutation Analysis (K304E)
MEN1 GENE (11q13), BLOOD
Mental Retardation microdeletions/duplications
Metachromatic Leukodystrophy (MLD) - ARSA Sequencing
Metachromatic Leukodystrophy (MLD) - ARSA Sequencing - KFM

Metachromatic Leukodystrophy (MLD) - ARSA Sequencing - Prenatal
Metachromatic Leukodystrophy, Skin Fibroblast Culture
Metachromatic Leukodystrophy, White Blood Cells
Methylmalonic Acid, Plasma
Methylmalonic Aciduria and Homocysteinuria, cblC Type - MMACHC Sequencing - KFM
Methylmalonic Aciduria and Homocystinuria, cblC Type - MMACHC Sequencing
Miller-Dieker Syndrome – karyotype/FISH
Miller-Dieker Syndrome (Isolated Lissencephaly) - FISH
Mitochondrial DNA Deletion Analysis
Mitochondrial DNA Screen Panel Quantification
Mitochondrial DNA Screening Panel (Point Mutations and Deletions)
Mitochondrial Myopathy – 13 common mutations + DNA rearrangements
Mitochondrial phosphate carrier deficiency - SLC25A3 Sequencing
Mitochondrial phosphate carrier deficiency - SLC25A3 Sequencing - KFM
MitoMetSM Mitochondrial/Metabolic oligonucleotide array CGH analysis
MitoMetSM Mitochondrial/Metabolic oligonucleotide array CGH analysis-CNV
MLL (11q23), BLOOD/BONE MARROW
MLL: 11q23 FISH Analysis
MLS/MIDAS - FISH
MMAA-Related Methylmalonic Aciduria - MMAA Sequencing
MMAA-Related Methylmalonic Aciduria -MMAA Sequencing - KFM
MMAB-Related Methylmalonic Aciduria - MMAB Sequencing
MMAB-Related Methylmalonic Aciduria -MMAB Sequencing - KFM
MNGIE-Thymidine Phosphorylase Sequencing
MNGIE-Thymidine Phosphorylase Sequencing - KFM
MNGIE-Thymidine Phosphorylase Sequencing - Prenatal
MOLECULAR ANEUPLOIDY STUDY, AMNIOTIC FLUID
MOLECULAR ANEUPLOIDY STUDY, CHORIAL VILLOSITY
Mowat-Wilson Syndrome – ZEB2 mutation scanning + deletions/duplications
mtDNA content in Liver
mtDNA content in Muscle
mtDNA Depletion and Multiple Deletions (POLG1, TK2, SUCLA2, & DGUOK)
MTHFR – common C677T mutation
MTHFR GENE (C677T MUTATION), BLOOD
MTHFR Variant Analysis
Mucopolysaccharidosis I (MPS1) - IDUA Sequencing
Mucopolysaccharidosis I (MPS1) - IDUA Sequencing - KFM
Mucopolysaccharidosis I (MPS1) - IDUA Sequencing - Prenatal
Mucopolysaccharidosis Type II (MPS II) - IDS Sequencing
Mucopolysaccharidosis Type II (MPS II) - IDS Sequencing - KFM
Mucopolysaccharidosis Type II (MPS II) - IDS Sequencing - Prenatal
Mucopolysaccharidosis Type IVA (MPS4A) - GALNS Sequencing
Mucopolysaccharidosis Type IVA (MPS4A) - GALNS Sequencing - KFM
Mucopolysaccharidosis Type IVA (MPS4A) - GALNS Sequencing - Prenatal
Multiple Endocrine Nepoplasia Type 2B – RET hotspot sequencing
Multiple Epiphyseal Dysplasia – COMP hotspot sequencing
Multiple Exostoses 1 FISH
Multiple Exostoses 2 FISH (Potocki/Shaffer)
Multiple Exostoses FISH Panel (includes Ext 1 & Ext 2)
Multiple Myeloma FISH Panel (13q14, 17p, IgH)
MUT-Related Methylmalonic Aciduria - MUT Sequencing
MUT-Related Methylmalonic Aciduria -MUT Sequencing - KFM

MYC translocation FISH Analysis
MYELOID PROGENITOR CELLS, BLOOD
MYELOID PROGENITOR CELLS, BONE MARROW
MYH Associated Polyposis (MYH) - 2 Mutation Panel
MYH Associated Polyposis (MYH) - KFM
MYH Associated Polyposis Sequencing
Myopathic mtDNA Depletion Syndrome - RRM2B Sequencing
Myopathic mtDNA Depletion Syndrome - RRM2B Sequencing - KFM
Myopathic mtDNA Depletion Syndrome - RRM2B Sequencing - Prenatal
Myopathic mtDNA Depletion Syndrome (Thymidine Kinase) TK2 Sequencing
Myopathic mtDNA Depletion Syndrome (Thymidine Kinase) TK2 Sequencing - KFM
Myopathic mtDNA Depletion Syndrome (Thymidine Kinase) TK2 Sequencing - Prenatal
Myotonic Dystrophy
Myotonic Dystrophy - Prenatal
Myotonic Dystrophy Type 1 – DMPK repeat analysis
Myotonic Dystrophy Type 2 – ZNF9 repeat analysis
N
N-Acetylglutamate Synthase Deficiency - NAGS Sequencing
N-Acetylglutamate Synthase Deficiency - NAGS Sequencing - KFM
Nail-Patella Syndrome - LMX1B Sequencing
Nail-Patella Syndrome - LMX1B Sequencing - KFM
Nail-Patella Syndrome - LMX1B Sequencing - Prenatal
Narcolepsy (HLA DRB1*15/DQB1*06)
Neurofibromatosis 1 FISH
Neurofibromatosis Type 1 – NF1 sequencing + deletions/duplications
NEUROFIBROMATOSIS TYPE 1 GENE (NF1), BLOOD
Neurofibromatosis Type 2 (Bilateral Acoustic) –NF2 sequencing + deletions/duplications
NEUROFIBROMATOSIS TYPE II, BLOOD
Niemann-Pick Disease (Type A)
Niemann-Pick Disease Type C - NPC1 Sequencing
Niemann-Pick Disease Type C - NPC1 Sequencing - KFM
Niemann-Pick Disease Type C - NPC1 Sequencing - Prenatal
Niemann-Pick Disease Type C - NPC2 Sequencing
Niemann-Pick Disease Type C - NPC2 Sequencing - KFM
Niemann-Pick Disease Type C - NPC2 Sequencing - Prenatal
Niemann-Pick Disease, Skin Fibroblast Culture
Niemann-Pick Disease, White Blood Cells
Noonan Syndrome - KRAS Sequencing
Noonan Syndrome - KRAS Sequencing - KFM
Noonan Syndrome - KRAS Sequencing - Prenatal
Noonan Syndrome - PTPN11 Sequencing
Noonan Syndrome - PTPN11 Sequencing - KFM
Noonan Syndrome - PTPN11 Sequencing - Prenatal
Noonan Syndrome - RAF1 Sequencing
Noonan Syndrome - RAF1 Sequencing - KFM
Noonan Syndrome - RAF1 Sequencing - Prenatal
Noonan Syndrome - SOS1 Sequencing
Noonan Syndrome - SOS1 Sequencing - KFM
Noonan Syndrome - SOS1 Sequencing - Prenatal
Noonan Syndrome Tier 1 – PTPN2 sequencing
Noonan Syndrome Tier 2 – SOS1 hotspots + RAF1 sequencing
Noonan Syndrome Tier 3 – remaining SOS1 + KRAS sequencing

Norrie Disease – NPD sequencing + deletions
Nuclear Complex 1 Panel (NDUFS3, NDUFS4, NDUFS7, NDUFA1, NDUFAF2, C6ORF66, NDUFS6, & NDUFV1)
O
Oculopharyngeal Muscular Dystrophy – PABPN1 repeat analysis
Optic Atrophy Type I - OPA1 sequencing
Optic Atrophy Type I - OPA1 sequencing - KFM
Optic Atrophy Type I - OPA1 sequencing - Prenatal
Organic Acid Screen, Urine
Ornithine Transcarbamylase Deficiency, Liver
Ornithine Transcarbamylase Sequencing
Ornithine Transcarbamylase Sequencing - KFM
Ornithine Transcarbamylase Sequencing - Prenatal
Orotic Acid/Orotidine Analysis, Urine
Osteogenesis Imperfecta (OI), Autosomal Recessive - CRTAP Sequencing
Osteogenesis Imperfecta (OI), Autosomal Recessive - CRTAP Sequencing - KFM
Osteogenesis Imperfecta (OI), Autosomal Recessive - CRTAP Sequencing - Prenatal
Osteogenesis Imperfecta (OI), Autosomal Recessive - LEPRE1 Sequencing
Osteogenesis Imperfecta, Autosomal Recessive - LEPRE1 Sequencing - KFM
Osteogenesis Imperfecta, Autosomal Recessive - LEPRE1 Sequencing - Prenatal
p16 PROTEIN, EXUDATE
PAI-1 GENE, BLOOD
Pancreatitis (Hereditary) – PRSS1 hotspot -sequencing + deletions/duplications-+ SPINK1 N34S common mutation
Parvovirus (PARVO) B19 DNA, Qualitative Real-Time PCR
Paternity Testing (postnatal and prenatal) – sample required from each person being tested (3 people)
Pelizaeus-Merzbacher Disease - PLP1 Sequencing
Pelizaeus-Merzbacher Disease - PLP1 Sequencing - KFM
Pelizaeus-Merzbacher Disease - PLP1 Sequencing - Prenatal
Pelizaeus-Merzbacher Disease/Spastic Paraplegia 2 FISH
Peutz-Jeghers Syndrome – STK11 sequencing
Phenylalanine/Tyrosine - Serum or Plasma
Phenylketonuria (PKU) - PAH Sequencing
Phenylketonuria (PKU) - PAH Sequencing - KFM
PHEO and PGL-Associated Syndromes Panel (SDHB, SDHC, and SDHD)
PLASMA CELL CYCLE AND CELL PLOIDY STUDY, BONE MARROW
PML/RARA: t(15;17) [AML] FISH Analysis
PML-RARa GENE t(15;17)(q22; q11.2-21), BONE MARROW
PMS2-Related Hereditary Non-polyposis Colorectal Cancer (HNPCC) - PMS2 Sequencing
PMS2-Related Hereditary Non-polyposis Colorectal Cancer (HNPCC) - PMS2 Sequencing - KFM
POLG1 Related Disorders - POLG1 Sequencing
POLG1 Related Disorders - POLG1 Sequencing - KFM
POLG1 Related Disorders - Prenatal
Poly T (5T,7T, 9T) – cystic fibrosis gene
Polyols, Urinary
Pompe Disease, Muscle
Pompe Disease, Skin Fibroblast Culture
P-Q
PRADER WILLI GENE (SNRPN), BLOOD
Prader-Willi Syndrome – karyotype/FISH
Prader-Willi Syndrome – methylation-PCR

Prader-Willi Syndrome – UPD15 (parents and child)
Prader-Willi Syndrome (Methylation)
Prader-Willi Syndrome FISH
PRENATAL KARYOTYPE, AMNIOTIC FLUID
PRENATAL KARYOTYPE, CHORIAL VILLOSITY
PRENATAL KARYOTYPE, UMBILICAL CORD BLOOD
PRENATAL STEINERT MYOTONIC DYSTROPHY, AMNIOTIC FLUID; BLOOD
Progressive Familial Intrahepatic Cholestasis 1 (PFIC1) - ATP8B1 Sequencing
Progressive Familial Intrahepatic Cholestasis 1 (PFIC1) - ATP8B1 Sequencing - KFM
Progressive Familial Intrahepatic Cholestasis 2 (PFIC2) - ABCB11 Sequencing
Progressive Familial Intrahepatic Cholestasis 2 (PFIC2) - ABCB11 Sequencing - KFM
Progressive Familial Intrahepatic Cholestasis 3 (PFIC3) - ABCB4/MDR3 Sequencing
Progressive Familial Intrahepatic Cholestasis 3 (PFIC3) - ABCB4/MDR3 Sequencing - KFM
Proteus Syndrome – PTEN sequencing
Prothrombin (Factor II)
PROTHROMBIN GENE (20210 MUTATION), BLOOD
PROXIMAL SPINAL MUSCULAR ATROPHY GENE, BLOOD
Pseudoachondroplasia – COMP hotspot sequencing
PTEN-Related Disorders - PTEN Sequencing
PTEN-Related Disorders - PTEN Sequencing - KFM
PTEN-Related Disorders - PTEN Sequencing - Prenatal
Purine Nucleoside Phosphorylase Deficiency, Amniotic Cell Culture
Purine Nucleoside Phosphorylase Deficiency, Cultured Chorionic Villus Cells
Purine Nucleoside Phosphorylase Deficiency, Red Blood Cells
Purine Nucleoside Phosphorylase Deficiency, Skin Fibroblast Culture
Purine Nucleoside Phosphorylase Deficiency, White Blood Cells
Purine Profile, Urine
Pyruvate Dehydrogenase Deficiency - PDHA1 Sequencing
Pyruvate Dehydrogenase Deficiency - PDHA1 Sequencing - KFM
Pyruvate Dehydrogenase Deficiency - PDHA1 Sequencing - Prenatal
QF-PCR Trisomy Analysis
R
Recurrent Miscarriage Profile (female)
Reilly Day Syndrome (Familial Dysautonomia) – 2 common Ashkenazi mutations (p.R696P + c.2204+6T>C)
RENAL POLYCYSTOSIS AD GENE (PKD1,PKD2), BLOOD
Respiratory Chain (Electron Transport Chain) Enzymes - Skeletal Muscle
Respiratory Chain (Electron Transport Chain) Enzymes - Skin Fibroblasts
RET ONCOGENE (HIRSPRUNG DISEASE), BLOOD
RET ONCOGENE MEN2A (10q11,2), BLOOD
RET ONCOGENE MEN2B (T918C), BLOOD
RETINOBLASTOMA MOLECULAR ANALYSIS, BLOOD
Rett Sequencing of Exon 1 only
Rett Syndrome – MECP2 sequencing
Rett Syndrome Deletion/Duplication Analysis
Rett Syndrome Deletion/Duplication Analysis - Prenatal
Rett Syndrome MECP2 Sequencing
Rett Syndrome MECP2 Sequencing - KFM
Rett Syndrome MECP2 Sequencing - Prenatal
RETT SYNDROME, GENETIC STUDY
RhD Molecular Typing - Prenatal
RhD Molecular Typing (Trio)

RhD ZYGOSITY STUDY (PATERNAL), BLOOD
Rickets-Alopecia Syndrome - VDR Sequencing
Rickets-Alopecia Syndrome - VDR Sequencing - KFM
RNA TYROSINASE (CIRCULATING CELLS), BLOOD
Rothmund Thomson Syndrome - RECQL4 Sequencing
Rothmund Thomson Syndrome - RECQL4 Sequencing - KFM
Rothmund Thomson Syndrome - RECQL4 Sequencing - Prenatal
Rubenstein-Taybi Syndrome – CREBBP mutation scanning
Rubinstein-Taybi Syndrome FISH
S
Sanfilippo B Mucopolysaccaridosis (MPS III), Serum
Sanfilippo B Mucopolysaccaridosis (MPS III), Skin Fibroblast Culture
Schmid Metaphyseal Chondrodysplasia (SMCD) - COL10A1 Sequencing
Schmid Metaphyseal Chondrodysplasia (SMCD) - COL10A1 Sequencing - KFM
Schmid Metaphyseal Chondrodysplasia (SMCD) - COL10A1 Sequencing - Prenatal
Short Stature SHOX – mutation screen
Sialidosis (Neuraminidase Deficiency), Skin Fibroblast Culture
Sickle Cell Disease
Smith-Magenis Syndrome – karyotype/FISH
Smith-Magenis Syndrome FISH
SMITH-MAGENIS SYNDROME, GENETIC STUDY
Sotos Syndrome (Cerebral Gigantism) – NSD1 sequencing + deletions/duplications
SOTOS Syndrome FISH
Sperm DNA Fragmentation
Sperm DNA Fragmentation and Aneupliody
SPG4 GENE (SPASTIN) SEQUENCING, BLOOD
Spinal Bulbar Muscular Atrophy (Kennedy Disease) – AR repeat analysis
Spinal Muscular Atrophy – SMN1 deletions/duplications
Spinal Muscular Atrophy (SMA) Diagnostic Test
SPINAL MUSCULAR ATROPHY GENE (SMN), BLOOD
SPINAL MUSCULAR ATROPHY GENE (SMN1) (CARRIERS), BLOOD
Spinocerebellar Ataxia – multiplex (SCA1+2+3+6+7+17)
SPINOCEREBELLAR ATAXIA (SCA-1) GENE, BLOOD
Spinocerebellar Ataxia Type 1 (SCA1)
Spinocerebellar Ataxia Type 1 (SCA1) - Prenatal
Spinocerebellar Ataxia Type 10 (SCA10)
Spinocerebellar Ataxia Type 10 (SCA10) - Prenatal
SRY (Sex-determining Region Y)
SRY Analysis FISH
SRY GENE, BLOOD
STEINERT MYOTONIC DYSTROPHY GENE, BLOOD
Steroid Sulfatase Deficiency, Amniotic Cell Culture
Steroid Sulfatase Deficiency, Cultured Chorionic Villus Cell Culture
Steroid Sulfatase Deficiency, Skin Fibroblast Culture
Steroid Sulfatase Deficiency, White Blood Cells
SUBTELOMERIC RESTRUCTURING, AMNIOTIC FLUID
Succinate Dehydrogenase Complex Subunit A - SDHA Sequencing
Succinate Dehydrogenase Complex Subunit A - SDHA Sequencing - KFM
Succinate Dehydrogenase Complex Subunit B - SDHB Sequencing
Succinate Dehydrogenase Complex Subunit B - SDHB Sequencing - KFM
Succinate Dehydrogenase Complex Subunit C - SDHC Sequencing
Succinate Dehydrogenase Complex Subunit C - SDHC Sequencing - KFM

Succinate Dehydrogenase Complex Subunit D (SDHD) Sequencing
Succinate Dehydrogenase Complex Subunit D (SDHD) Sequencing - KFM
Succinylacetone, Urine
SUPPRESSOR GENE P16 MICROSATELLITE INSTABILITY, BIOL. MATERIAL
Systemic Carnitine Deficiency - SLC22A5 Sequencing
Systemic Carnitine Deficiency - SLC22A5 Sequencing - KFM
T
T CELL RECEPTOR (TCR) GENETIC REORGANIZATION, BLOOD
t(11;14) TRANSLOCATION, BLOOD/BONE MARROW
t(15;17)(PML/RARa) TRANSLOCATION, BLOOD/BONE MARROW
t(9;22)(bcr/abl) TRANSLOCATION, BLOOD/BONE MARROW
Tay Sachs Screen – 3 common Ashkenazi mutations c.1421+G>C + c.1277-1278insTATC + p.G269S
Tay-Sachs Carrier Testing
Tay-Sachs Disease Screen
Tay-Sachs Disease, Serum
TEL/AML1 t(12;21)(p13;q22) TRANSLOCATION, BLOOD/BONE MARROW
TEL/AML1: t(12;21) [ALL] FISH Analysis
Telomeres FISH
Telomeric screen – deletions/duplications
Thrombophilia Panel (Factor V, Prothrombin & MTHFR)
Thrombotic Risk/Miscarriage Profile
Thymidine, Plasma
TNF BETA GENE, BLOOD
TOMM20 Sequencing
TOMM20 Sequencing - KFM
Torsion Dystonia (DYT1) – TOR1A common mutation c.904-906delGAG
TORSION DYSTONIA GENE (DYT1), BLOOD
Toxoplasma (TOXO) gondii DNA, Qualitative Real-Time PCR
TPMT (THIOPURINE METHYLTRANSFERASE) GENE, BLOOD
Trichorhinophalangeal Syndrome FISH
Trifunctional Protein Deficiency - HADHB Sequencing
Trifunctional Protein Deficiency - HADHB Sequencing - KFM
Trisomy Analysis (Amniotic Fluid) – PCR only
TUMOR SUPPRESSOR p53 GENE, BLOOD
Tyrosinemia, Type 1 - FAH Sequencing
Tyrosinemia, Type 1 - FAH Sequencing- KFM
U
Uni parental Disomy (UPD) – parents and child
UNIPARENTAL DYSSOMIA STUDY, BLOOD
V
Velocardiofacial Syndrome (22 & 10p) FISH Panel
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) - ACADVL Sequencing
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) - ACADVL Sequencing - KFM
VHL (VON HIPPEL LINDAU) GENE, BLOOD
Vohwinkel Syndrome – Connexin 26 sequencing
Von Hippel-Lindau Syndrome
W
Waardenburg Type 1 – PAX3 deletions/duplications
Waardenburg Type 1 – PAX3 sequencing
WAGR (Aniridia) FISH Panel
Werdnig-Hoffman Disease (Spinal Muscular Atrophy) – SMN1 deletions/duplications

Whole Genome Sequencing
Whole Genome Sequencing - KFM
Williams Syndrome – karyotype/FISH
Wilms Tumor (WT1) FISH
WILMS TUMOR GENE, BLOOD
Wilms Tumour (Nephroblastoma) – WT1 sequencing
Wilson Disease Tier 1 – ATP7B hotspot sequencing
Wilson Disease Tier 2 – ATP7B remaining sequencing
Wolf-Hirschhorn Syndrome – karyotype/FISH
WOLF-HIRSCHHORN SYNDROME, GENETIC STUDY
Wolman Disease - LIPA Sequencing
Wolman Disease - LIPA Sequencing - KFM
Wolman Disease - LIPA Sequencing - Prenatal
Wolman Disease (Cholesterol Ester Storage), Amniotic Cell Culture
Wolman Disease (Cholesterol Ester Storage), Cultured Chorionic Villus Cells
Wolman Disease (Cholesterol Ester Storage), Liver
Wolman Disease (Cholesterol Ester Storage), Skin Fibroblast Culture
Wolman Disease (Cholesterol Ester Storage), White Blood Cells
X
X-FRAGILE CHROMOSOME (CYTOGENETIC), BLOOD
X-FRAGILE CHROMOSOME (MOLECULAR), AMNIOTIC FLUID
X-FRAGILE CHROMOSOME (MOLECULAR), BLOOD
X-linked Angelman-like Syndrome - SLC9A6 Sequencing
X-linked Angelman-like Syndrome - SLC9A6 Sequencing - KFM
X-linked Angelman-like Syndrome - SLC9A6 Sequencing - Prenatal
X-linked Hydrocephalus/MASA Syndrome – L1-CAM sequencing
X-linked Hypohidrotic Ectodermal Dysplasia – ED1 sequencing
X-linked Ichthyosis/ Steroid Sulfatase Deficiency FISH
X-LINKED ICHTYOSIS (STS GENE), BLOOD
X-linked Ocular Albinism - GPR143 Sequencing
X-linked Ocular Albinism - GPR143 Sequencing - KFM
X-linked Ocular Albinism - GPR143 Sequencing - Prenatal

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