

S.No	Test Name	Gene list	Specimen Type	Methodology	TAT
1	Fragile X Syndrome		Peripheral Blood in Heparin (Green Top)	Karyotyping	15 days
2	Ataxia Telangiectasia(chromosome instability syndrome)		Peripheral Blood in EDTA (Purple Top)	Karyotyping	15 days
3	CSF IgG index			Colorimetric / Calculated	2 days
4	Anti - IgLON5 Antibody IgG		Serum	Immunofluorescence Assay (IFA)	4 days
5	VGKC (LG1 and CASPR2) Antibody (CSF and SERUM)		Cerebrospinal fluid (CSF),Serum	Immunofluorescence Assay (IFA)	24 Hours
6	NMO (Aquaporin-4) +MOG IgG -Serum		Serum	Immunofluorescence Assay (IFA)	24 Hours
7	NMO (Aquaporin 4+MOG) IgG – CSF		Cerebrospinal fluid (CSF)	Immunofluorescence Assay (IFA)	24 Hours
8	NMO Profile 2 IgG (CSF and Serum)		CSF & Serum	Immunofluorescence Assay (IFA)	24 Hours
9	Aquaporin 4 -CSF		Cerebrospinal fluid (CSF)	Enzyme Linked Immunosorbant Assay (ELISA)	4 days
10	Aquaporin 4 -Serum		Serum	Enzyme Linked Immunosorbant Assay (ELISA)	4 days
11	NMDA Receptor Antibody-NR-1		Serum,Plasma	Immunofluorescence Assay (IFA)	24 Hours
12	NEURONAL ANTIBODY PROFILE -3		Serum	Immunoblotting	4 days
13	Neuronal Antibody Profile 1		Serum	Immunofluorescence Assay (IFA)	24 Hours
14	MuSK Antibody		Serum	Enzyme Linked Immunosorbant Assay (ELISA)	2 days
15	Ganglioside IGM Antibody Panel (GM1, GM2, GM3,GD1a, GD1b, GT1b,GQ1b)		Serum	Immunoblotting	3 days
16	Ganglioside IgG Antibody Panel (GM1, GM2,GM3,GD1a,GD1b,GT1b,GQ1b)		Serum	Immunoblotting	3 days
17	Ganglioside (GM1) IgG and IgM Antibodies Serum		Serum	Enzyme Linked Immunosorbant Assay (ELISA)	7 days
18	GANGLIOSIDE IgG ANTIBODY SERUM		Plasma	Enzyme Linked Immunosorbant Assay (ELISA)	7 days
19	GANGLIOSIDE IgM ANTIBODY SERUM		Plasma	Enzyme Linked Immunosorbant Assay (ELISA)	7 days

20	DPYD (DEOXYPYRIDINOLINE), URINE		Urine	Lateral flow Chromatographic immunoassay	3 days
21	Spinal Muscular Atrophy (SMN1) gene analysis by NGS	SMN1	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	21 days
22	ExomeMAX reflex Whole Genome Sequencing (30x)		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	21 days
23	Neurosense		SALIVA,Peripheral Blood in EDTA (Purple Top)	Combo test	14 days
24	CARRIERGEN	CYP4F2,SLCO1B1, CYP2C19,HLA-A,VKORC1,CYP3A5,RYR1,CYP2B6,CACNA1S,G6PD,UGT1A1,CYP2D6,NUDT15,CFTR,ABCG2,DPYD,TPMT,CYP2C9,HLA-B,IFNL3	Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
25	Alzheimer's disease		SALIVA,Peripheral Blood in EDTA (Purple Top)	Whole Genome Genotyping Array	14 days
26	Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication analysis [Expedited TAT]		DNA (POC),Amniotic fluid (AF),Cord Tissue / Umbilical Cord,DIRECT DNA,Product of conception (POC),Cord blood in EDTA (Purple Top),Peripheral Blood in EDTA (Purple Top),Fetal DNA,Chorionic Villus Sample (CVS)	MLPA	8 days
27	Friedreich ataxia (FXN) - Repeat expansion analysis		DNA (POC),Fetal DNA,DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
28	Huntington's disease (HTT) - Repeat expansion analysis		DNA (POC),Fetal DNA,DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
29	PRKN gene deletion/duplication analysis		Product of conception (POC),DIRECT DNA,Cardiac Blood in EDTA (Purple Top),Placenta,Cord Tissue / Umbilical Cord,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top),Cord blood in EDTA (Purple Top),Fetal DNA,Chorionic Villus Sample (CVS)	MLPA	14 days

30	RSRC1 gene sequencing	RSRC1	DIRECT DNA,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top),Cord Tissue / Umbilical Cord,Fetal DNA,Chorionic Villus Sample (CVS),Product of conception (POC),DNA (POC),Cardiac Blood in EDTA (Purple Top),Placenta	Next Generation Sequencing (NGS)	19 days
31	CLP1 gene sequencing	CLP1	DNA (POC),Chorionic Villus Sample (CVS),Product of conception (POC),DIRECT DNA,Amniotic fluid (AF),Placenta,Cord Tissue / Umbilical Cord,Peripheral Blood in EDTA (Purple Top),Cord blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
32	GBE1 gene sequencing	GBE1	Chorionic Villus Sample (CVS),Fetal DNA,Cord Tissue / Umbilical Cord,Product of conception (POC),DNA (POC),Cardiac Blood in EDTA (Purple Top),Placenta,DIRECT DNA,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
33	NDUFS7 gene sequencing	NDUFS7	Cord Tissue / Umbilical Cord,Fetal DNA,Cord blood in EDTA (Purple Top),Product of conception (POC),DNA (POC),Chorionic Villus Sample (CVS),Placenta,DIRECT DNA,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
34	PRKRA gene sequencing	PRKRA	Chorionic Villus Sample (CVS),Product of conception (POC),Cord Tissue / Umbilical Cord,Cardiac Blood in EDTA (Purple Top),Placenta,DIRECT DNA,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top),Cord blood in EDTA (Purple Top),Fetal DNA	Next Generation Sequencing (NGS)	19 days
35	RANBP2 gene sequencing	RANBP2	DNA (POC),Cardiac Blood in EDTA (Purple Top),Placenta,DIRECT DNA,Chorionic Villus Sample (CVS),Peripheral Blood in EDTA (Purple Top),Cord blood in EDTA (Purple Top),Amniotic fluid (AF),Fetal DNA,Cord Tissue / Umbilical Cord,Product of conception (POC)	Next Generation Sequencing (NGS)	19 days

36	TCF4 gene sequencing	TCF4	Cord blood in EDTA (Purple Top),Product of conception (POC),DNA (POC),Cardiac Blood in EDTA (Purple Top),Placenta,DIRECT DNA,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top),Cord Tissue / Umbilical Cord,Fetal DNA	Next Generation Sequencing (NGS)	19 days
37	HINT1 gene sequencing	HINT1	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
38	PSEN1 gene sequencing	PSEN1	Cord Tissue / Umbilical Cord,Amniotic fluid (AF),Fetal DNA,Cord blood in EDTA (Purple Top),Product of conception (POC),DNA (POC),Chorionic Villus Sample (CVS),Placenta,DIRECT DNA,Cardiac Blood in EDTA (Purple Top),Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
39	CAPN3 gene sequencing	CAPN3	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
40	PIEZO2 gene sequencing	PIEZO2	Cord Tissue / Umbilical Cord,Peripheral Blood in EDTA (Purple Top),Cord blood in EDTA (Purple Top),DNA (POC),Chorionic Villus Sample (CVS),Product of conception (POC),DIRECT DNA,Amniotic fluid (AF),Placenta	Next Generation Sequencing (NGS)	19 days
41	GCH1 gene sequencing	GCH1	DNA (POC),Cardiac Blood in EDTA (Purple Top),Product of conception (POC),DIRECT DNA,Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top),Cord Tissue / Umbilical Cord,Fetal DNA,Chorionic Villus Sample (CVS),Placenta	Next Generation Sequencing (NGS)	19 days
42	ADSSL1 gene analysis	ADSSL1	Amniotic fluid (AF),Placenta,Cord blood in EDTA (Purple Top),Peripheral Blood in EDTA (Purple Top),Cord Tissue / Umbilical Cord,DIRECT DNA,Chorionic Villus Sample (CVS),Product of conception (POC),DNA (POC)	Next Generation Sequencing (NGS)	19 days
43	SMN and NAIP deletion/duplication analysis			MLPA	14 days
44	SPTBN2 gene sequencing	SPTBN2	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days

45	CSTB gene sequencing	CSTB	Fetal DNA, Chorionic Villus Sample (CVS), Product of conception (POC), Cord Tissue / Umbilical Cord, Amniotic fluid (AF), Placenta, Cord blood in EDTA (Purple Top), Peripheral Blood in EDTA (Purple Top), DNA (POC)	Next Generation Sequencing (NGS)	19 days
46	Familial temporal lobe epilepsy-1 (LGI1) gene sequencing	LGI1	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
47	Hereditary essential tremor (FUS and TENM4) gene sequencing	FUS, TENM4	Peripheral Blood in EDTA (Purple Top), Cardiac Blood in EDTA (Purple Top), DIRECT DNA, Amniotic fluid (AF), Placenta, Cord Tissue / Umbilical Cord, Chorionic Villus Sample (CVS), Product of conception (POC), DNA (POC), Fetal DNA	Next Generation Sequencing (NGS)	19 days
48	Neurofibromatosis type 1 (NF1) gene analysis	NF1	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
49	Sandhoff disease (HEXB) gene sequencing	HEXB	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
50	ATP1A2 and ATP1A3 gene sequencing	ATP1A3, ATP1A2	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
51	GAN and DCAF8 gene sequencing	DCAF8, GAN	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
52	Neurodegeneration due to cerebral folate transport deficiency (FOLR1) gene sequencing	FOLR1	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
53	MYH3 gene sequencing	MYH3	Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
54	DYSF gene sequencing [Expedited TAT]	DYSF	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	15 days
55	DYSF gene sequencing	DYSF	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
56	KCNT1 gene sequencing	KCNT1	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
57	PLP1 gene sequencing	PLP1	Placenta, DNA (POC), Chorionic Villus Sample (CVS), Product of conception (POC), DIRECT DNA, Cardiac Blood in EDTA (Purple Top), Peripheral Blood in EDTA (Purple Top), Cord Tissue / Umbilical Cord, Amniotic fluid (AF), Fetal DNA, Cord blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
58	PANK2 gene sequencing	PANK2	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days

59	Gordon Holmes syndrome (RNF216 and OTUD4) gene sequencing	OTUD4,RNF216	Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
60	NKX2-1 gene sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
61	IGHMBP2 gene sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
62	Fragile X Syndrome		Peripheral Blood in Heparin (Green Top)	Karyotyping	15 days
63	Ataxia Telangiectasia(chromosome instability syndrome)		Peripheral Blood in EDTA (Purple Top)	Karyotyping	15 days
64	Dystonia-28 (KMT2B) gene sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
65	ADGRG1 gene sequencing	ADGRG1	DIRECT DNA,Amniotic fluid (AF),Product of conception (POC),Cord blood in EDTA (Purple Top),Peripheral Blood in EDTA (Purple Top),Cord Tissue / Umbilical Cord,DNA (POC),Chorionic Villus Sample (CVS),Placenta	Next Generation Sequencing (NGS)	19 days
66	MECP2 gene sequencing		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
67	SACS gene sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
68	TIA1 and DES genes sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
69	MFSD8 gene sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
70	SETX gene sequencing		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
71	Fukuyama Congenital Muscular Dystrophy (FKTN) gene sequencing (does not include repeat expansions)[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
72	TBC1D24 gene analysis[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
73	Alkaptonuria (HGD) gene analysis[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days

74	Neurotransmitter disorders gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
75	Adrenoleukodystrophy (ABCD1) gene analysis[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
76	Giant axonal neuropathy-1 (GAN) gene analysis[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
77	PMP22 gene analysis (inflammatory demyelinating polyneuropathy screen)[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
78	Alexander disease (GFAP) gene analysis[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
79	Brown Vialetto-Van Laere syndrome gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
80	Spinal muscular atrophy gene panel (does not include SMN1/SMN2)[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
81	ATRX gene analysis[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
82	TSC1 & TSC2 gene analysis[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
83	Neurofibromatosis type 2 (NF2) gene analysis[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
84	Neurofibromatosis (NF1 and NF2) gene analysis[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days

85	Tay-Sachs disease (HEXA) gene analysis[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
86	Rett Syndrome gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
87	Pontocerebellar hypoplasia gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
88	NOTCH3 (CADASIL) gene analysis[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
89	Neuronal migration disorder gene panel[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
90	Neuronal ceroid lipofuscinosis gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
91	Microcephaly gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
92	Lissencephaly gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
93	Joubert syndrome gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
94	Hereditary spastic paraplegia gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
95	Familial hemiplegic migraine gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days

96	Cystic megalencephaly (MLC1) gene analysis[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
97	Comprehensive neurology panel[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
98	Aicardi-Goutieres syndrome gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
99	4H syndrome gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
100	Myotonia congenita gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
101	Muscular dystrophy & congenital myopathy gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
102	Duchenne muscular dystrophy (DMD) gene analysis[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
103	Charcot-Marie-Tooth and sensory neuropathies gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
104	Arthrogryposis & congenital myasthenic syndrome gene panel[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
105	Neurodegeneration with brain iron accumulation gene panel[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
106	Hyperekplexia gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days

107	Episodic ataxia gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
108	Early-onset juvenile parkinsonism gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
109	Dystonia gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
110	Ataxia-telangiectasia (ATM) gene analysis[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
111	Metachromatic leukodystrophy gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
112	Leukodystrophy gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
113	Krabbe disease (GALC) gene analysis[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
114	Hypomyelination syndrome gene panel[Expedited TAT]		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
115	Canavan disease (ASPA) gene analysis[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
116	Progressive myoclonic epilepsy gene panel[Expedited TAT]		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	14 days
117	Familial female mental retardation/epilepsy gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days
118	Epileptic encephalopathy gene panel[Expedited TAT]		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	14 days

119	Dravet syndrome (SCN1A) gene analysis[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
120	Benign infantile epilepsy gene panel[Expedited TAT]		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days
121	Pyridoxine-dependent epilepsy (ALDH7A1) gene analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
122	Fukuyama Congenital Muscular Dystrophy (FKTN) gene sequencing (does not include repeat expansions)		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
123	ABCD1 gene analysis	ABCD1	Amniotic fluid (AF),Peripheral Blood in EDTA (Purple Top),Cord blood in EDTA (Purple Top),Fetal DNA,Chorionic Villus Sample (CVS),Product of conception (POC),DNA (POC),Cardiac Blood in EDTA (Purple Top),Placenta,DIRECT DNA	Next Generation Sequencing (NGS)	19 days
124	TBC1D24 gene analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
125	Alkaptonuria (HGD) gene analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
126	Neurotransmitter disorders gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
127	Adrenoleukodystrophy (ABCD1) gene analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
128	Autosomal dominant adult-onset demyelinating leukodystrophy (LMNB1) deletion/duplication analysis		Cord blood in EDTA (Purple Top),Peripheral Blood in EDTA (Purple Top),Product of conception (POC),Chorionic Villus Sample (CVS),DNA (POC),Amniotic fluid (AF),Cord Tissue / Umbilical Cord,DIRECT DNA,Placenta	MLPA	14 days

129	Giant axonal neuropathy-1 (GAN) gene analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
130	Myotonia congenita (CLCN1) deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
131	Charcot-Marie-Tooth type 4C (SH3TC2) deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
132	DCX & PAFAH1B1 deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
133	PMP22 gene analysis (inflammatory demyelinating polyneuropathy screen)		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
134	Alexander disease (GFAP) gene analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
135	Spinocerebellar ataxia repeat expansion analysis: SCA1, SCA2, SCA3, SCA6, SCA7,SCA12		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	21 days
136	Spinocerebellar ataxia repeat expansion analysis: Any two of (SCA1, SCA2, SCA3, SCA6, SCA7, SCA12)		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
137	Spinocerebellar ataxia 6 (CACNA1A) repeat expansion analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
138	Spinocerebellar ataxia 3 (ATXN3) repeat expansion analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
139	Spinocerebellar ataxia 2 (ATXN2) repeat expansion analysis		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Fragment Analysis (FA)	14 days
140	Spinocerebellar ataxia 12 (PPP2R2B) repeat expansion analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
141	Rett Syndrome gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
142	Pontocerebellar hypoplasia gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days

143	Neurodegeneration with brain iron accumulation gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
144	NOTCH3 (CADASIL) gene analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
145	Neuronal migration disorder gene panel		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
146	Neuronal ceroid lipofuscinosis gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
147	Myotonia congenita gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
148	Muscular dystrophy and congenital myopathy gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
149	Microcephaly gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
150	Metachromatic leukodystrophy gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
151	Lissencephaly gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
152	Leukodystrophy gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
153	Krabbe disease (GALC) gene analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
154	Hyperekplexia gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days

155	Hereditary spastic paraplegia gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
156	Familial hemiplegic migraine gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
157	Episodic ataxia gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
158	Aicardi-Goutieres syndrome gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
159	4H syndrome gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
160	Charcot-Marie-Tooth and sensory neuropathies gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
161	Arthrogryposis & congenital myasthenic syndrome gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
162	Early-onset juvenile parkinsonism gene panel		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
163	Dystonia gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
164	Ataxia-telangiectasia (ATM) gene analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
165	Ataxia-telangiectasia (ATM) deletion/duplication analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	MLPA	14 days
166	Hypomyelination syndrome gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days

167	Progressive myoclonic epilepsy gene panel		Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS), Amniotic fluid (AF), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
168	Familial female mental retardation/epilepsy gene panel		Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS), Amniotic fluid (AF), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
169	Epileptic encephalopathy gene panel		DIRECT DNA, Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS), Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
170	Dravet syndrome (SCN1A) gene analysis		DIRECT DNA, Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS), Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
171	Benign infantile epilepsy gene panel		Amniotic fluid (AF), DIRECT DNA, Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
172	TSC1 and TSC2 gene analysis		Chorionic Villus Sample (CVS), Amniotic fluid (AF), DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
173	Neurofibromatosis type 1 (NF1) deletion/duplication analysis		DIRECT DNA, Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS), Amniotic fluid (AF)	MLPA	14 days
174	Neurofibromatosis type 2 (NF2) gene analysis		Chorionic Villus Sample (CVS), Amniotic fluid (AF), DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
175	Neurofibromatosis (NF1 and NF2) gene analysis		Chorionic Villus Sample (CVS), Amniotic fluid (AF), DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
176	ATRX gene analysis		Amniotic fluid (AF), DIRECT DNA, Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
177	Brown Vialetto-Van Laere syndrome gene panel		Amniotic fluid (AF), DIRECT DNA, Peripheral Blood in EDTA (Purple Top), Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
178	Spinocerebellar ataxia 7 (ATXN7) repeat expansion analysis		DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days

179	Spinal Muscular Atrophy gene Panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
180	Neurodegeneration with brain iron accumulation 2B (PLA2G6) deletion/duplication analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	MLPA	14 days
181	Early infantile epileptic encephalopathy-4 (STXBP1) deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	MLPA	14 days
182	Charcot-Marie-Tooth type 4 (EGR2, GDAP1, NEFL, PRX) deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	MLPA	14 days
183	Spinocerebellar ataxia 1 (ATXN1) repeat expansion analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Fragment Analysis (FA)	14 days
184	TSC2 deletion/duplication analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	MLPA	14 days
185	TSC1 deletion/duplication analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	MLPA	14 days
186	X-linked spastic paraplegia-2 (PLP1) deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	MLPA	14 days
187	Tay-Sachs disease (HEXA) gene analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
188	Tay-Sachs disease (HEXA) deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	MLPA	14 days
189	Rett Syndrome (MECP2) deletion/duplication analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	MLPA	14 days
190	Joubert syndrome gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days

191	Cystic megalencephaly (MLC1) gene analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
192	Comprehensive neurology panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
193	Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	MLPA	14 days
194	Dysferlinopathy/LGMD2B (DYSF) deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
195	Duchenne Muscular Dystrophy (DMD) gene sequencing		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
196	Duchenne Muscular Dystrophy (DMD) deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
197	Congenital Muscular Dystrophy (LAMA2) deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
198	Charcot-Marie-Tooth 1A/HNPP (PMP22, COX10, TEKT3) deletion/duplication analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	MLPA	14 days
199	Krabbe disease (GALC) deletion/duplication analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	MLPA	14 days
200	Canavan disease (ASPA) gene analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
201	Dravet syndrome (SCN1A) deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	MLPA	14 days