2. NEUROLOGY				
	a	**Hereditary Ataxias and Movement Disorders**		
	CAT	Ataxia Telengiectasia (Bieomycin Assay)	**6 ml blood Heparin**	3-4 wk
	MATCL	Ataxia Telangiectasia - Carrier Screening by Linkage	4 ml EDTA	2-3 wks
	MATLFP	Ataxia telangiectasia-prenatal diagnosis by linkage analysis	4 ml EDTA	2-3 wks
	MSCA	SCA -Spinal cerebellar ataxia - one gene	4 ml blood EDTA	3-4 wks
	MSCATG	SCA -Spinal cerebellar ataxia - two gene	4 ml blood EDTA	3-4 wks
	MJD	SCA3-Spino cerebellar ataxia -Machado - Joseph disease	4 ml blood EDTA	3-4 wks
	MSCA4	SCA - Spinal cerebellar ataxia, 4 genes , (1, 2, 3, 12)	4 ml blood EDTA	3-4 wks
	MSC4	SCA - Spinal cerebellar at. package (1, 2, 3, 6, 7, 12, 17)	4 ml blood EDTA	3-4 wks
	MSCAN	Spinocerebellar ataxia with peripheral neuropathy autosomal recessive SCAN 1 targeted mutation analysis	4 ml blood EDTA	3-4 wks
	MTDP1	SCA - with peripheral neuropathy autosomal recessive - TDP1 gene sequenicng	4 ml blood EDTA	3-4 wks
	MDYT	Dystonia 1 DYT 1 deletion analysis	4 ml blood EDTA	2-3 wks
	MDYT1	Dystonia -DYT1 gene sequencing	4 ml blood EDTA	4 to 6 wks
	MDYT5	Dystonia -DYT5 gene sequencing	4 ml blood EDTA	6-8 wks
	MATP1A3	Dystonia + parkinsonism (RDP) DYT12- ATP1A3 gene sequencing	4 ml blood EDTA	4 to 6 wks
	MMTS	Deafness Dystonia Optic Neuronopathy Syndrome- Mohr Tranebjaerg syndrome. TIMM8A gene sequencing	4ml blood EDTA	2 to 3 wks
	MDRPLA	Dentatorubral - Pallidoluysian atrophy (Triplet repeat)	4 ml blood EDTA	3-4 wks
	MFAMD	Friedreich ataxia Molecular diagnosis	4 ml blood EDTA	3-4 wks
	MPLA2G6	Infantile neuroaxonal dystrophy (INAD) PLA2G6 gene sequencing	4 ml blood EDTA	4 to 6 wks
	MHD	Huntington disease gene analysis	4 ml blood EDTA	3-4 wks
	b	**Hereditary Motor Sensory Neuropathy**		
	MCMT	Charcot - Marie tooth Disease (CMT)/ HNPP -PMP22 deletion/duplication	4 ml blood EDTA	3 to 4 wks
	MEGR2	Charcot - Marie tooth Disease/CMT-EGR2 gene sequencing	4 ml blood EDTA	6 to 8 wks
	MGDAP1	Charcot - Marie tooth Disease/CMT-GDAP1 gene sequencing	4 ml blood EDTA	6 to 8 wks

	MMFN2	Charcot - Marie tooth Disease/CMT-MFN2 gene sequencing	4 ml blood EDTA	6 to 8 wks
	MMPZ	Charcot - Marie tooth Disease/CMT-MPZ gene sequencing	4 ml blood EDTA	6 to 8 wks
	MPRX	Charcot - Marie tooth Disease/CMT-PRX gene sequencing	4 ml blood EDTA	6 to 8 wks
	MCMTX	Charcot-Marie-Tooth/CMT - X-Linked GJB1 gene sequencing	4 ml blood EDTA	2 wks
	MCIPA	Congenital Insensitivity to Pain with Anhidrosis (CIPA) - (Prenatal Diagnosis by Linkage), TRKA(NTRK1) gene	4 ml blood EDTA	3-4 wks
	MFDM	Riley Day Syndrome/Familial dysautonomia - markers	4 ml blood EDTA	3-4 wks
	c	**Neuromuscular**		
		Muscle biopsy and Immunohistochemistry		
	HMUDF	Muscle biopsy - Dysferlin staining	Muscle biopsy	
	HMUDT	Muscle biopsy - Dystrophin staining	Muscle biopsy	2 Wks
	HMUM	Muscle biopsy - Merosin staining	Muscle biopsy	2 Wks
	HMMP	Merosin staining - Prenatal diagnosis (Immunostain and DNA Studies)	Muscle biopsy	2 Wks
	HMA	Merosin / Dysferlin staining / Alpha dystroglycan	Muscle biopsy	2 Wks
	MMLL	Merosin prenatal diagnosis by linkage studies	4 ml EDTA	10 to 14 days
	HMDSP	Muscular dystrophy staining profile (Muscle biopsy - Dystrophin staining and Sacrogylcan staining)	Muscle biopsy	2 Wks
	HMUS	Muscle biopsy - Sacrogylcan staining	Muscle biopsy	2 Wks
		Molecular analysis for Muscular Disorders		
	MCAL	Calpainopathy Common Mutations	4 ml EDTA	2-3 wks
	MCAPN	Calpainopathy Gene Sequencing	4 ml EDTA	4-6 wks
	DMDMLPA	Duchenne muscular dystrophy (DMD) deletion analysis by MLPA (79 exons)	4 ml blood EDTA	2 -3 wks
	MDD	Duchenne muscular dystrophy (DMD), deletion testing by multiple PCR	4 ml blood EDTA	2-3 wks
	MDC	Duchenne mus dyst (DMD) carrier test - linkage studies	4 ml blood EDTA	2-3 wks
	MDDF	DMD - Dosage Studies in Females	4 ml blood EDTA	2-3 wks
	MDP	Duchenne muscular dystrophy (DMD) prenatal diagnosis	CVS + 4 ml EDTA blood Couple/ Affected subject	1-2 wks
	MEMD	Emery Driefuss Muscular Dystrophy - EMD gene sequencing	4 ml blood EDTA	6-8 wks

	MFSHDL	Facioscapulohumeral muscular dyst (FSHD) Linkage analysis	4 ml blood EDTA	3-4 wks
	MFSHDPND	Facioscapulohumeral muscular dystrophy- PND by Linkage analysis	CV+ 4 ml EDTA blood from two affected members	10-14 days
	MFKRP	Fukutin related protein-Fukuyama congenital muscular dystrophy- FKRP gene sequencing	4 ml blood EDTA	6-8 wks
	MFKTN	Fukuyama congenital MD-FKTN gene sequencing	4 ml blood EDTA	6-8 wks
	RAPSN	Congenital Myasthenic Syndrome-RAPSN gene Common mutation(p. N88K)	4 ml blood EDTA	2 to 3 wks
	MLAMA2	LAMA2 gene deletion/ duplication	4 ml blood EDTA	3 to 4 wks
	MLGMD2E	LGMD2E common mutations (exons 19 & 22)	4 ml EDTA	2 to 3 wks
	MPYGM	McArdle disease gene sequencing - PYGM gene	4 ml blood EDTA	3-4 wks
	MAMPD1	Myoadenylate Deaminase Def.-AMPD1 exon 2 &3 gene seq	4 ml blood EDTA	3-4 wks
	MDM	Myotonic dystrophy - Type 1 (19q 13.3)	4 ml blood EDTA	3-4 wks
	MMDT2	Myotonic dystrophy - Type 2 (3q 21)	4 ml blood EDTA	3-4 wks
	MPM	Paramyotonia, 2 mutations Arg 1448 His Thr 1313 Met	4 ml blood EDTA	3-4 wks
	BPOM	Pompe Disease- α- glucosidase enzyme assay	**6 ml Heparin blood / DBS**	3-4 days
	MGAA	Pompe disease – GAA gene sequencing	4 ml blood EDTA	6-8 wks
	MSMA	Spinal Muscular Atrophy - SMA - molecular diagnosis	4 ml blood EDTA	3-4 wks
	MSMNS	Spinal Muscular Atrophy - SMN 1 Gene sequencing	4 ml blood EDTA	3-4 wks
	MSMC	Spinal muscular atrophy -SMN2 copy number analysis	4 ml blood EDTA	3-4 wks
	SMAMLPA	SMA carrier screening by MLPA	4 ml blood EDTA	3-4 wks
	MSMARD	SMA with respiratory distress1- SMRD 1 Gene Sequencing	CVS + 4 ml EDTA blood couple/ Affected subject	3-4 wks
	MSBM	Spinal Bulbar muscular atrophy (CAG Repeats)	4 ml blood EDTA	3-4 wks
	d	**Mental retardation & Neurodevelopmental Disorders**		
	MANG	Angelman syndrome, DNA test	4 ml blood EDTA	3-4 wks
	CBLD	Chromosomal analysis	**3 ml Heparin blood**	2-3 wks
	MDCX	Lissencephaly- DCX gene sequencing	4 ml EDTA	6 to 8 wks

	MFXS	Fragile X syndrome screen	4 ml blood EDTA	3-4 wks
	MFXM	Fragile X syndrome (Methylation test - confirmatory)	4 ml blood EDTA	3-4 wks
	MSXF	Fragile X in Females	4 ml blood EDTA	3-4 wks
	MRX	Mental Retardation - X Linked - Panel Deletions / Duplications	4 ml blood EDTA	3-4 wks
	MR1	Mental Retardation Panel1 - Multiple Deletions/Duplications	4 ml blood EDTA	3-4 wks
	MR2	Mental Retardation Panel 2 - Multiple Deletions/Duplications	4 ml blood EDTA	1-2 wks
	CMRA	Microarray (180 K)	4ml blood EDTA	4-6 wks
	MRSPQBP1	Renpenning Syndrome-PQBP1 gene sequencing	4 ml EDTA	6 to 8 wks
	MRTSQ	RETT Syndrome MECP2 Deletion/Duplication	4 ml blood EDTA	3-4 wks
	MRSS	Rett syndrome MECP2 Sequencing	4 ml blood EDTA	3-4 wks
	MRS	Rett syndrome (MECP2 sequencing and Deletion / duplication)	4 ml blood EDTA	3-4 wks
	MKIAA1279	Goldberg-Sphrintzen syndrome-KIAA1279 gene sequencing	4 ml blood EDTA	6-8 wks
	MLN	Lesch Nyhan syndrome sequencing	4 ml EDTA	6 to 8 wks
	FISHMDLG	Miller Dieker syndrome FISH-LIS gene	**3 ml Heparin blood**	3-7 days
	MMWSDD	Mowat Wison syndrome deletion/duplication	4 ml blood EDTA	3-4 wks
	MMWS	Mowat Wison syndrome-ZEB2 gene sequencing	4 ml blood EDTA	6-8 wks
	MTELD	Subtelomeric Deletions / Duplications - All Autosomes	4 ml EDTA	2 to 3 wks
	FISHSM	Smith Magenis FISH test	**3 ml Heparin blood**	3-7 days
	e	**Leukodystrophies**		
	MANE	Acute necrotizing encephalopathy (ANE) RANBP2 gene T585M	4 ml blood EDTA	3-4 wks
	MXALD	Adrenoleukodystrophy (X-linked ALD) sequencing of ABCD1 gene	4 ml blood EDTA	6-8 wks
	MSCG	Canavan disease gene sequencing	4 ml blood EDTA	6-8 wks
	MLC1	Cystic megalencephaly MLC 1 gene mutation	4 ml blood EDTA	3-4 wks
	MLCS	Cystic Megalencephaly - MLC1 Gene Sequencing	4 ml blood EDTA	6-8 wks
	MPEPACAM	Cystic Megalencephaly- HEPACAM Gene Sequencing	4 ml blood EDTA	6-8 wks
	MDARS2	DARS 2 gene sequencing	4 ml EDTA	6 to 8 wks

	MPLA2G6	Infantile neuroaxonal dystrophy (INAD) PLA2G6 gene sequencing	4 ml blood EDTA	6-8 wks
	MGCDH	Glutaric Aciduria-GCDH gene sequencing	4 ml blood EDTA	6-8 wks
	MHSC	Hallervorden Spatz - Common mutation	4 ml blood EDTA	2 to 3 wks
	MHSDSEQ	Hallervorden-Spatz Syndrome - PANK2 gene sequencing	4 ml blood EDTA	6 to 8 wks
	MHCF	Hypomyelination with Congenital Cataract- FAM126A gene sequencing	4 ml blood EDTA	4 to 6 wks
	MPLA2G6	NBIA- Neurodegeneration with brain iron accumulation - PLA2G6 gene sequencing	4 ml blood EDTA	6-8 wks
	MPZD	NBIA- Neurodegeneration with brain iron accumulation- Pelizaeus Merzbacher Deletion/Duplication	4 ml blood EDTA	3-4 wks
	MPMD	NBIA- Neurodegeneration with brain iron accumulation Pelizaeus-Merzbacher disease-PLP1 gene sequencing	4 ml blood EDTA	3-4 wks
	BARA	Metachromatic leukodystrophy/MLD - Arylsulfatase A enzyme assay	**6 ml blood in heparin**	3-4 days
	MMLD	Metachromatic Leukodystrophy gene sequencing	4 ml blood EDTA	6-8 wks
	BKRB	Krabbe disease- Galactocerebrosidase enzyme assay	**6 ml Heparin blood**	3-4 days
	MKD	Krabbe disease - Common deletion	4 ml blood EDTA	3-4 wks
	MGALC	Krabbes Disease - GALC gene sequencing	4 ml blood EDTA	6-8 wks
	BNIL1	NCL Infantile- Palmitoyl Protein Thioesterase enzyme assay	**6 ml Heparin blood**	3-4 days
	MNCLH	NCL - Infantile (2 Mutations)	4 ml EDTA	2 to 3 wks
	MNCL	Neuronal Ceroid Lipofuschinosis-NCL Linkage	4 ml blood EDTA	3-4 wks
	BNIL2	NCL Late Infantile - Tripeptidyl Peptidases/TPP	**6 ml blood in heparin**	3-4 days
	MTPP1	NCL - Late Infantile (CLN2).- TPP1 gene sequencing	4 ml EDTA	4 to 6 wks
	MNCLL	NCL - Late infantile (2 Mutations)	4 ml blood EDTA	3-4 wks
	MNCLJ	NCL - Juvenile (1 Mutation - common deletion)	4 ml blood EDTA	3-4 wks
	MPZD	Pelizaeus Merzbacher Deletion/Duplication	4 ml blood EDTA	3-4 wks
	MPMD	Pelizaeus-Merzbacher disease-PLP1 gene sequencing	4 ml blood EDTA	3-4 wks
	BVLC	Peroxisomal Disorders- VLCFA analysis	4 ml EDTA	3-4 wks

	MXALD	X-linked ALD-Adrenoleukodystrophy ABCD1 gene sequencing	4 ml blood EDTA	6-8 wks
	f	**Neurocutaneous Syndromes**		
	MDTS1	Tuberous sclerosis (TSC1) deletion / duplication	4 ml blood EDTA	3-4 wks
	MTSC1	Tuberous sclerosis (TSC1) Sequencing	4 ml blood EDTA	3-4 wks
	MDTS2	Tuberous sclerosis (TSC2) deletion / duplication	4 ml blood EDTA	3-4 wks
	MTSC2	Tuberous sclerosis (TSC2) Sequencing	4 ml blood EDTA	3-4 wks
	MNL	Neurofibromatosis linkage (prenatal diagnosis)	4 ml blood EDTA	1-2 wks
	CVHL	Von Hippel Landua dis (VHL Gene) Deletion/ Duplication	4 ml blood EDTA	3-4 wks
	CVHS	VHL gene mutation 3 exons by sequencing	4 ml EDTA	3 to 4 wks
	g	**Miscellaneous**		
	MNOT	CADASIL- NOTCH3 gene sequencing -5 exons	4 ml blood EDTA	6-8 wks
	MMAPT	Dementia - Frontotemporal (MAPT) exons 9-13) sequencing	4 ml blood EDTA	6-8 wks
	MSCN	Epilepsy - SCN1a gene sequencing	4 ml blood EDTA	6-8 wks
	HB1502	HLA-B 1502 Genotyping, Carbamazepine, Phenytoin	4 ml blood EDTA	3-4 wks
	MFVL	Factor V Leiden mutation	4 ml blood EDTA	3-4 wks
	MFOL	Folate polymorphism 3'5 MTHFR - (677C>T, 1298A>C)	4 ml blood EDTA	3-4 wks
	MASPM	Primary Microcephaly -ASPM gene sequencing	4 ml blood EDTA	6-8 wks
	MPQBP1	Primary Microcephaly -PQBP1 gene sequencing	4 ml blood EDTA	6-8 wks
	SGLY19	Parkinson Gly19 ser mutation	4 ml blood EDTA	6-8 wks
	ML1CAM	X-Linked Hydrocephalus - L1CAM gene sequencing,	4 ml blood EDTA	6-8 wks
	MXLL	X-Linked Hydorcephalus - MASA Syndrome - Linkage studies	4 ml blood EDTA	3-4 wks
	MEPM1	Unverricht–Lundborg disease	4 m EDTA	4 to 6 wks
	MSPG11 A	Hereditary Spastic Paraparesis-SPG11 common mutations	4 ml blood EDTA	2 to 3 wks