Neurological Conditions – Summary

Upper vs. Lower Motor Neurone Signs

Upper	Lower
Hypertonia	Hypotonia
Hyper-reflexia	Hypo-reflexia
Reduced power	Reduced power
Clonus	Fasiculation
Wasting	Wasting

Muscular Disorders

Disorder	Abnormality	Features	Gait	Tone	Power	Reflexes	Co-Ordination	Sensation
DMD Beckers	Dystrophin Gene	Wasting of distal muscles + pes cavus Pseudohypertrophy Contractures Scoliosis ↓ motor milestones Reduced cardiac + pulmonary function	Waddling Broad-based Difficulty with stairs High-stepping due to Achilles tightening Footdrop	Reduced - proximally	Reduced - proximally Facial sparing **Note: Limb-girdle is a differential Affects hips & shoulders + goes distally Easy to confuse with Beckers!**	Disappear early on except for ankle jerk	Normal	Normal
Facioscapulo-humeral		Shoulder girdle + facial muscles Winging of scapula Expressionless face	Normal	Reduced - proximally	Reduced - proximally	Reduced or normal	Normal	Normal

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Myotonic Dystophy (50-150 repeats)	Chromosome 19 Autosomal Dominant Anticipation	Failure of muscle to relax – myotonia Cataracts Balding Arrhythmias and cardiomyopathy Infertility Contractures Diabetes Mellitus Retardation Talipes	Wheelchair as it progresses Note Mum may be in a wheelchair	Hypotonia	Progressive weakness - face - jaw - neck - distal muscles Myopathic facies Ptosis	Reduced or normal	Normal	Normal
Congenital Myotonic Dystrophy (>2000 repeats)	Chromosome 19 Autosomal Dominant Anticipation	Floppy infant Myotonia later Facial diplegia and triangular facies Respiratory problems after birth Reduced fetal movements Polyhydramnios	May not walk if very severe	Hypotonia	Reduced ++ Myopathic facies – note that SMA I get facial sparing so helps to differentiate	Reduced or normal	Normal	Normal

Other differentials of proximal muscle weakness include: dermatomyositis, hypothyroidism, hyperthyroidism and chronic disease

Summary of Neurological Disorders

Location	Examples	Inspection/Other	Gait	Tone	Power	Reflexes	Co-Ordination	Sensation
Central **UMN**	Cerebral Palsy CVA Syndromes	Wasting Pseudobulbar palsy in quadriplegic so may be drooling Hyocine patch VP shunt Note type of chair	Hemiplegic Diplegic Quadriplegic Choreoi-Athetoid Dyskinetic	Increased	Reduced	Increased	Variable according to lesion	Normal
Spinal Cord **LMN**	Spina Bifida Transverse Myelitis **(keeps reflexes + have pain)**	Wasting Sensory/Motor Level Cutaneous Signs Sphincters Pes cavus High maternal AFP Hydrocephalus VP shunt due to AC malformation	Depends on site of lesion	Reduced (can be increased(	Reduced	Reduced	Varies with level of deficit	Sensory neuropathy

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Location	**Examples**	**Inspection/Other**	**Gait**	**Tone**	**Power**	**Reflexes**	**Co-Ordination**	**Sensation**
Anterior Horn Cell **LMN**	SMA I SMA III Autosomal Recessive Chromosome 5	SMA I very severe Floppy baby Frogs legs Weak cry Die within 2 years Wasting Alert expression Tongue fasciculation Gower's +ive Calf hypertrophy	Waddling gait Difficulty climbing stairs Gower +ive	Reduced	Reduced proximally - progressive	Absent knee jerks but persevered ankle and upper limb Similar to DMD and limb girdle muscular dystrophy = need EMG and muscle biopsy to differentiate	Normal	Normal
Nerve Fibre **LMN**	Guillain-Barre	Sensory loss is less marked than motor signs 7th nerve palsy is common – examine cranial nerves	Varies with weakness	Reduced	Reduced symmetrically ascending = distal weakness	Reduced or absent	Normal	Position and vibration lost first (then pain and light touch) Autonomic dysfunction
Nerve Fibre **LMN**	Poliomyelitis			Reduced	Reduced asymmetrically ascending = distal weakness	Reduced or absent	Normal	Intact

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Location	Examples	Inspection/Other	Gait	Tone	Power	Reflexes	Co-Ordination	Sensation
Nerve Fibre **LMN**	Peroneal Muscular Atrophy (HSMN) Seven types Autosomal dominant Chromosome 17 **Sensory neural hearing loss**	Distal wasting Pes cavus Hypertrophic myelin (onion-bulb) Tremor Motor > Sensory Claw hand Kyphoscoliosis Normal life span Most patients remain ambulant	Footdrop (S1) Possibly ataxic due to sensory loss (compare with Freidriechs)	Normal	Reduced distally esp dorsiflexion	Reduced distally Upper limb reflexes preserved - downward plantars	Normal	Proprioception and vibration affected Cold feet **later** with loss of hair Foot deformities due to lost sensation
NMJ **LMN**	Myaesthenia Gravis Neonatal or autoimmune	Fatiguability Autoimmune disorder affecting ACh receptor Dysphagia Dysarthria	Normal	Normal	Facial and eye muscle weakness ++ Tensilon test Pyridostigmin e Steroids	Normal	Normal	Normal

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Location	Examples	Inspection/Other	Gait	Tone	Power	Reflexes	Co-Ordination	Sensation
Spinocerebellar Degeneration **LMN**	Friedreich's Ataxia *Metabolic disorder of Krebs Cycle* Anticipation (Trinucleotide repeat)	Distal wasting Progressive Pes Cavus Kyphoscoliosis HOCM/Diabetes Optic atrophy Autosomal recessive Chromosome 9	Ataxic	Reduced	Reduced	Reduced Upward plantars	Difficult Cerebellar signs	Sensory ataxia (Rombergs +ive) Loss of position and vibration sense Peripheral neuropathy
Spinocerebellar Degeneration **LMN**	Ataxic Telangectasia *Ask about milestones as motor delay in infancy + increased risk of lymphoma* **High AFP**	Immature behaviour Progessive Developmental delay Café-au-lait patches Immunodeficiency (usually low IgA) Autosomal recessive Chromosome 11 Bulbar telangectasia	Ataxic	Reduced	Normal	Reduced (with ↑ age) Normal (down) plantars	Difficult Cerebellar signs Conjugate gaze difficult	Proprioception and vibration intact

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Location	Examples	Inspection/Other	Gait	Tone	Power	Reflexes	Co-Ordination	Sensation
Muscular Dystrophies	See Chart above	See Chart above	Wheelchair Waddling Depends on type	Reduced	Reduced - proximally +	Reduced or normal (check DMD)	Normal	Normal
Other Central	ADEM MS	Mixed UMN and LMN signs Cranial nerve palsy Ataxia Optic neuritis			Reduced	Increased	Normal	Usually intact

Differentials of Footdrop and Pes Cavus

Footdrop	Pes Cavus
HSMN I (Peroneal Muscular Atophy)	HSMN I (Peroneal Muscular Atophy)
Lateral Popliteal Nerve Palsy	Spina Bifida
Polio	DMD
	Friedrich's Ataxia

Older child with ataxia and positive Romberg's = **Freidriech's Ataxia**

HSMN and Friedreich's can look similar – due to ataxia and loss of proprioception and vibration sense with pes cavus. Distinguish because other features of Friedreich's may be present and there are upgoing plantars and it is Romberg's +ive.

Tumours in Neurocutaneous Syndromes

Tuberous Sclerosis	Neurofibromatosis I	Neurofibromatosis II
Cerebral hamartoma	Astrocytoma	Meningioma
Astrocytoma	Optic glioma (Type I)	Acoustic neuroma (bilateral)
Cerebral Glioma	Lisch nodule (Iris hamartoma)	Vestibular schwanomas
Retinal phakoma	Rhabdomyosarcoma	Optic glioma
Rhabdomyosarcoma (tongue)	Medullary thyroid carcinoma	
Rhabdomyoma (heart)	Neurofibrosarcoma (skin)	
Angiomyolipoma (kidneys)	Wilms tumour (kidney)	
Rectal polyps	Phaeochromocytoma (adrenal cortex)	
	Neuroblastoma (sympathetic chain)	
	Leukaemia	

Neurological Diseases

Upper Motor Neurone Signs	Lower Motor Neurone Signs
Cerebral palsy	SMA
CVA (Intraventricular Haemorrhage)	Guillain-Barre
Neurodegenerative disorders • Leukodystrophies	HSMN
ADEM	Muscular Dystophies

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