Chapter 11:

NEUROLOGICAL DISORDERS

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This chapter covers the following topics:

- Non-accidental injury (NAI)
- Acute flaccid paralysis (ASD)
- Temper tantrums
- Night terrors
- Breath-holding attacks
- Microcephaly
- Hydrocephalus
- Movement disorders
- Seizure disorders
- Paediatric stroke
- Headache
- Myelomeningocoele/meningomyelocoele
- Spinal muscular atrophy (SMA)
- Neurocutaneous syndromes

NON-ACCIDENTAL INJURY (NAI)

It is a common problem and can be potentially life-threatening. Healthcare professionals have a duty to report any suspected or confirmed NAI to the relevant authorities e.g. senior clinical staff or child protection services.

Aetiology

NAI may be the result of maltreatment in the form of:

- Physical abuse
- Neglect
- Sexual assault
- Psychological abuse
- Medical abuse

Clinical Features

The child may present with:

- Pain
- Swelling
- Bruising
- Limping

Below (table 11.1) are some red flags which may indicate that the child has suffered an NAI:

Table 11.1: NAI Red Flags

Red Flags on History	Red Flags on Examination
● Variable/inconsistent history ● Injuries not consistent with history ● Delay in seeking treatment ● Multiple injuries ● Repeated admissions or presentations to the emergency unit ● Unexplained symptoms e.g. factitious	● Findings not in keeping with developmental age of child e.g. non-mobile child with bruising/fracture ● Bruises that resemble the shape of instruments or a hand ● Many bruises and bruises of different ages ● Bruises over soft tissue

	● Burns e.g. cigarette burns (common) ● Forced hot water immersion (will have glove and stocking distribution) ● Recurrent fractures (must exclude metabolic bone disease) ● Overbearing parent/guardian ● Fractures (especially metaphysical, posterior rib, skull, scapula or sternum) ● Retinal haemorrhage ● Signs of neglect

Investigations

They should include

- Skeletal survey (X-rays)
- CT scan
- Ophthalmologic assessment

Management

One must carefully record the child's injuries and contact the relevant authorities e.g. child protection services, senior staff, police.

ACUTE FLACCID PARALYSIS (AFP)

It is a clinical syndrome characterised by the rapid onset of weakness and reduced muscle tone without an obvious cause. Early diagnosis improves both morbidity and mortality.

Pathophysiology

The syndrome is the result of anterior horn cell injury which is usually secondary to viral infection (e.g. polio; however polio has nearly been completely eradicated) or immune-mediated peripheral pathologies.

Aetiology, Clinical Features and Investigations

Guillain Barré syndrome (GBS) is the most common cause of AFP in SA. It is classically preceded by a URTI and is characterised by symmetrical motor weakness of the lower limbs which ascends to involve the trunk and upper limbs to varying degrees. Fortunately, 80% of patients make a full recovery. Pertinent clinical signs include:

- Weakness of the limbs, face, bulbar muscles and respiratory muscles
- Reduced or absent reflexes
- Sensory loss in a glove and stocking pattern of distribution
- Autonomic instability (unstable BP, irregular HR, urinary incontinence)

An LP should be done as it will show isolated raised CSF protein in GBS and can be used to exclude other causes.

Management

One's immediate management priorities are to diagnose the child with AFP and provide:

    - Supportive management
    - Monitoring of respiratory status
    - Physiotherapy and occupational therapy (indicated in immobile children; help improve functioning)

IV gamma globulin is also used.

AUTISM SPECTRUM DISORDER (ASD)

See the *Disorders of Development* chapter.

TEMPER TANTRUMS

They are unpleasant and disruptive behaviours and emotional outbursts. They may be common in some communities and usually manifest between the ages of 15 months and 3 years.

Temper tantrums are associated with unmet needs or desires i.e. the child is not allowed to ‘have his/her own way’. They form part of the ‘negative stage of child development’ and are worsened by the child’s lack of vocabulary to fully express their feelings.

Clinical Features

Generally, the parent will give a history of the child lying down, kicking and screaming. The physical exam will be normal and is only done to reassure the parent.

Red flags include:

- Tantrums which worsen after 4 years of age (see related image here)
- Self-injury or damaging of property during the tantrum
- Breath-holding (especially if the child faints)
- Developmental regression
- Refusal to eat or sleep
- Headaches
- Development of anxiety

Management

Temper tantrums are usually a passing phase and these children generally respond well to counselling. Parents must be reassured and given encouragement. They should not reinforce the negative behaviour by acquiescing to the child, but need to be taught coping strategies. Principles of management include:

- Offering alternatives to the desired object or activity to avoid frustrating situations
- Ignoring or walking away from the child
- Not punishing the child (do not hit or argue with the child)
- Showing approval and giving attention when the child displays positive or acceptable behaviour
- Maintaining consistency

Problems occur when parental control is lacking or the family has a chaotic lifestyle. Important factors to consider when managing these patients are:

    - Child’s temperament

- Parent's temperament
- Inconsistency of the parent's behaviour towards the child

NIGHT TERRORS

This term refers to episodes when the child suddenly wakes up at night and is very upset.

Clinical Features

Night terrors generally occur during phase 3 non-REM (deep) sleep and usually occur early in the night. The episode tends to last 10-20 minutes and the child usually has 2-3 episodes weekly. Clinical features include:

- Screaming and jumping out of bed as if running away from something scary
- Hitting in all directions
- Having eyes wide open
- Disorientation on waking
- Failure to recognise someone who is trying to help him/her
- Failure to remember the episode the next day

Management

There is no specific treatment for night terrors. The child cannot be soothed during the episode but s/he will fall asleep after an episode. One must not try to wake the child.

If the night terrors take place at the same time every night, the child can be woken up 15 minutes before the time to try and prevent the episode. The child is then allowed to stay awake for a few minutes before falling asleep again.

BREATH-HOLDING ATTACKS

They are closely related to temper tantrums and are usually present between the ages of 6 and 18 months (rare in children >4 years).

Classification and Clinical Features

Breath-holding attacks may be of the cyanotic- or pale-type. Anoxia precipitates both types of attack and both types are associated with iron-deficiency anaemia.

- Cyanotic-type:
    - It is more common and is associated with temper tantrums. Episodes typically last 10-30 seconds.
    - The child's crying turns into hyperventilating and the child may become apnoeic and cyanosed. The child may even develop increased limb tone or clonic movements.
    - These clinical features cease when the child starts breathing normally. S/he will be apathetic but fully conscious afterwards.
- Pale-type:
    - It is associated with minor injury or fear.
    - The child develops severe pallor, bradycardia and may, in some instances, become asystolic (due to increased vagal tone).

Management

The parents must be reassured that the condition is benign and is unrelated to epilepsy. However, a full history must be done and thorough physical examination done. Iron-deficiency should also be corrected, if present.

ENURESIS

See the *Child and Adolescent Psychiatry* chapter.

ENCOPRESIS

See the *Child and Adolescent Psychiatry* chapter.

MICROCEPHALY

It is defined as an abnormally small head (head circumference >2 standard deviations below normal. Microcephaly (see related image here) may be isolated or

may be associated with congenital anomalies. It is a disorder of cell proliferation and brain growth as a whole is defective. The frontal lobes are usually more affected following perinatal insults. The cerebellum is usually relatively spared.

Aetiology

Causes of microcephaly include:

- Normal variant
- Hypoxic ischaemic encephalopathy (most common cause)
- Foetal alcohol syndrome (FAS) – see related features here
- Infections:
    - Toxoplasmosis
    - CMV
    - Zika virus
- Intrauterine growth restriction
- Inborn errors of metabolism
- Other genetic conditions

Clinical Features

The child may present with:

- Neurodevelopmental delay
- Hearing or vision problems
- Epilepsy
- Cerebral palsy

Management

No treatment is available to reverse microcephaly. Parents should be counselled and individual problems treated by the multidisciplinary team, such as audiology referral to manage for hearing impairment, and occupational therapist referral to teach the child skills which help him/her better integrate into society. Anticonvulsants may be prescribed for the child with seizures.

HYDROCEPHALUS

It is an abnormally large head with an increased CSF volume and/or pressure.

Pathophysiology

Hydrocephalus is the result of:

- Obstruction of CSF flow
- Overproduction of CSF
- Decreased absorption of CSF

Aetiology

Hydrocephalus may be caused by:

- Infection e.g. TB, other causes of meningitis
- Genetic factors
- Malignancy (rare)
- Trauma
- Congenital disease e.g. aqueduct stenosis, Arnold-Chiari Malformations

See related image here.

Clinical Features

The child may present with:

- Macrocephaly (diagnosed at birth or antenatally)
- Setting-sun eyes
- Bulging fontanelles
- Anorexia
- Vomiting
- Irritability
- Lethargy

Management

Serial head circumference measurements should be taken and plotted to track progression. The child should be referred early for the insertion of a shunt

(ventriculoperitoneal shunt or third ventriculostomy). Once a shunt has been placed, the child must be regularly followed up to ensure that it has been correctly inserted.

MOVEMENT DISORDERS

They include:

- Ataxia
- Chorea
- Dystonia
- Athetosis:
    - This term refers to slow, writing movements of limbs.
    - It may occur in patients with chorea or hypoxic ischaemic encephalopathy resulting in cerebral palsy.
- Tics (see *Child and Adolescent Psychiatry* chapter)
- Tremor:
    - A tremor is an involuntary, oscillatory movement with a fixed frequency.
    - One must exclude hypothyroidism.
- Myoclonus:
    - These rapid muscle jerks have decreased frequency and severity when the patient is asleep.
    - However, unlike tics, they do not disappear in sleep.

Ataxia

It is unco-ordination of postural control and gait, as well as unco-ordination of skilled movements involved in fine hand movements and speech.

Aetiology

Disturbances in cerebellar function generally lead to ataxia. One must be able to differentiate acute causes from chronic conditions.

- Acute causes:
    - Infection:
        - Viruses (which cause meningitis) e.g. VZV, coxsackie virus, Epstein-Barr virus, influenza, mumps, measles

        - Bacteria (which cause meningitis or cerebral abscesses) e.g. diphtheria, pertussis, typhoid, scarlet fever
    - Alcohol
    - Toxins or metabolic disease e.g. hypoglycaemia, lead, glue, mercury
    - Drugs e.g. phenytoin, carbamazepine, phenobarbitone
    - Posterior fossa tumours
    - Trauma (cerebellar haemorrhage)
    - Basilar migraine
    - Pseudo-ataxia (non-convulsive status epilepticus)
- Chronic causes may be perinatal or postnatal

Table 11.2: Perinatal and Postnatal Causes of Ataxia

Perinatal Causes	Postnatal Causes
• Birth asphyxia • Congenital malformations • Primary cerebellar hypoplasia • FAS • Friedrich's ataxia • Hydrocephalus • Meningitis • Metabolic disease (hypoglycaemia, hyperbilirubinaemia)	• Hypoxic insults • Hypoglycaemia • Thyroid deficiency • Chronic phenytoin use • Thiamine deficiency • Trauma • Genetic causes e.g. ataxia telangiectasia, Friedrich's ataxia

Clinical Features

The cerebellum matures with age, therefore signs which are considered pathological in the adult may be physiological in the child. Clinical features may include:

- Truncal ataxia – poor posture, exaggerated joint angles, head lag, truncal hypotonia
- Volitional ataxia – dysmetria, dysdiadochokinesia, nystagmus, dysarthria, staccato speech
- Hypotonia

Chorea

Choreas are rapid, brief, jerky movements that are irregular and unpredictable. They are often associated with hypotonia. It is thought that the caudate nucleus and subthalamic nuclei are involved in the pathophysiology of chorea.

One must always consider rheumatic fever as a cause of the chorea (Sydenham's chorea).

Sydenham's Chorea

It is characterised by abrupt, irregular and purposeless movements. ARF is the most common cause of acquired chorea in the young (see *Cardiovascular Diseases* chapter). Sydenham's chorea is common in the developing world.

Clinical Presentation

This major manifestation of ARF is characterised by:

- Insidious onset (4 months post initial infection)
- Migratory chorea
- Hypotonia
- Emotional lability
- Fidgeting

Management

One must investigate to exclude cardiac pathology. All affected children should be given penicillin V followed by prophylaxis, if ASOT titres and anti-DNAse levels are raised. The chorea is treated with the lowest-tolerated dose of haloperidol (see related image here) or with sodium valproate. The child should be referred to a paediatrician if his/her activities of daily living have been affected.

Dystonia

It is simultaneous, sustained contraction of agonist and antagonist muscles, leading to posture disturbances and involuntary movements of the trunk, limbs and face. These movements are slow, laboured and repetitive in nature (alternate between flexion and extension).

It may be caused by:

- Drugs e.g. haloperidol, metoclopramide
- Bilirubin encephalopathy:
    - Severe neonatal jaundice leads to the deposition of bilirubin in the basal ganglia (will see kernicterus on pathological examination).

- These children often present with dyskinetic movements.

SEIZURE DISORDERS

Seizures are caused by abnormal and excessive neuronal activity in the brain, and lead to transient signs and symptoms

Aetiology

Seizures may be caused by:

- Infection
- Hypoxia during gestation and/or after birth
- Intracranial haemorrhage
- Hypoglycaemia
- Drug withdrawal
- Developmental abnormalities

Diagnosis and Investigations

A seizure disorder may be diagnosed if the child has had >2 unprovoked seizures. However, a thorough and reliable history is very important, especially eyewitness reports (video footage can be very helpful). Other investigations which may be helpful include:

- Brain MRI (to look for lesions in the brain which may be causing the seizures)
- Electroencephalogram (EEG; can be used to rule-in seizures but cannot be used to rule them out)

See related image here.

Classification and Clinical Features

Seizure disorders may be classified into syndromes based on clinical features:

- Generalized seizures e.g. tonic-clonic, myoclonic, absence, atonic
- Focal seizures
- Other e.g. neonatal seizures
  - Neonatal seizures are subtle – due to relatively underdeveloped cortex

- Facial grimacing, nystagmus, sudden loss of time, eye blinking and chewing

Febrile Seizures

They are generalised seizures which occur because of an extracranial cause of fever. They affect children aged 6 months to 5 years and do not usually last longer than 15 minutes. There is often a family history of febrile seizures.

Classification

There are two categories of febrile seizures:

- Simple febrile seizures:
  - They are generalised seizures which last <15 mins and do not recur within a 24-hour period.
  - They are most commonly generalised tonic-clonic seizures.
- Complex febrile seizures:
  - They have a focal onset, last >15 mins and recur within 24 hours

Infantile Spasms

They mainly occur in children within the first year of life and are characterised by

- Epileptic spasms (flexor, extensor or mixed flexor-extensor spasms)
- Hypsarrhythmia on EEG

Aetiology

Causes of infantile spasms include:

- Neurocutaneous syndromes e.g. tuberous sclerosis
- Hypoxia
- Infections
- Trauma
- Hypoglycaemia
- Inborn errors of metabolism

When the diagnosis of infantile spasms is made, one must assess the child for West syndrome. West syndrome is diagnosed based on the presence of the following triad:

- Flexor or extensor spasms
- Hypsarrhythmia on EEG
- Mental retardation

Management

Infantile spasms are a neurological emergency, therefore an EEG must be urgently done. However, they are strongly resistant to conventional anti-epileptic treatment. The treatment of choice is with a combination of steroids and vigabatrin. Vigabatrin alone may be given to patients with tuberous sclerosis.

Table 11.3: Status Epilepticus

Status Epilepticus
It is an epileptic seizure which is sufficiently long or regularly repeated to produce a varying and enduring epileptic condition, in terms of convulsions or mental state i.e. the child has seizures lasting 30 mins or longer with no recovery of consciousness between seizures. Status epilepticus is common in the first two years of life.

General Management of Seizures

The aim of management is to maintain vital functions. Thus, one must identify the cause and treat it according to the relevant protocol.

General management principles include:

- Attempting to control symptoms with monotherapy (although resistant forms may need 2 or 3 anticonvulsants before control is gained)
- Gradually titrating up doses of new anticonvulsants (weekly)
- Monitoring the child for side effects

If one is unable to control seizures with the correct agent that is being given at the appropriate dose, the child should be referred to a paediatrician or paediatric neurologist.

Non-specialist management options may be grouped according to seizure type:

- Generalised or focal seizures – phenobarbitone, phenytoin (only use for status epilepticus in children), carbamazepine, sodium valproate
- Absence seizures – sodium valproate, ethosuximide
- Complex partial or focal seizures – carbamazepine, phenytoin

- Febrile seizures – diazepam (phenobarbitone and sodium valproate are seldom used for febrile seizures)
- Myoclonic seizures – sodium valproate, clonazepam

PAEDIATRIC STROKE

It may occur in a child with any of the following risk factors:

- Inflammation or infection e.g. meningitis (bacterial or mycobacterial), HIV
- Cardiac disease
- Haematological or thrombotic syndromes

Classification

Paediatric strokes may be classified according to

- Lesion type (haemorrhagic or ischaemic)
- Vessel type (arterial vs venous)

If the stroke is arterial, it may be further classified as embolic or thrombotic.

Aetiology

An ischaemic stroke may be the result of:

- Vascular disease:
    - Vasculopathies e.g. focal cerebral arteriopathy (may occur post-VZV infection)
    - Vasculitides e.g. meningitis, Takayasu's arteritis
- Intravascular disease:
    - Haematological conditions e.g. sickle cell, leukaemia, polycythaemia
    - Prothrombotic state, which may be due to:
        - Congenital conditions e.g. protein C or S deficiency, factor V Leiden mutations
        - Metabolic disease e.g. hyperlipidaemia, homocysteinaemia
- Embolic cause:
    - Congenital heart disease
    - Acquired heart disease e.g. rheumatic heart disease, infective endocarditis, prosthetic heart valve

A haemorrhagic stroke may be the result of:

- Vascular disease:
    - Congenital vascular anomalies e.g. arteriovenous malformation, venous angioma, cavernous malformation, intracranial aneurysm
    - Vasculopathies e.g. moyamoya disease, connective tissue disorders, sickle cell disease
- Intravascular disease (haematological disorders):
    - Idiopathic thrombocytopenic purpura (ITP)
    - Thrombotic thrombocytopenic purpura
    - Haemophilic states
    - Clotting factor deficiencies
    - Liver dysfunction
    - Vitamin K deficiency

Clinical Features

The child will present with a specific stroke syndrome based on the artery involved:

- Internal carotid artery:
    - Hemiplegia
    - Hemianopia
    - Aphasia (if the dominant hemisphere is affected)
- Middle cerebral artery (MCA):
    - Hemiplegia (arm and face are more affected than leg)
    - Hemianopia
    - Aphasia (if the dominant hemisphere is affected)
- Anterior cerebral artery (ACA):
    - Hemiplegia (leg is more affected than arm or face)
- Posterior cerebral artery (PCA):
    - Hemiparesis
    - Homonymous hemianopia
    - Ataxia
    - Vertigo
- Penetrating branches:
    - Pure motor involvement

    - Pure sensory involvement
    - Sensorimotor involvement
    - Speech involvement (occasional)

Approach

History

The child may present with:

- Sudden onset of neurological deficit (suggestive of an embolic event)
- Gradual onset of neurological deficit (suggestive of an arteriopathy or thrombotic event)
- History of recent illness (e.g. chicken pox) or trauma to the head
- Family history of young strokes

Examination

One must pay look for attention to:

- Changes in GCS
- Evidence of raised intracranial pressure
- Evidence of meningitis
- Neurological deficits (including aphasia/dysphasia)
- Evidence of bleeding abnormality or vasculitis
- Cardiovascular disease

Investigations

They should include:

- Blood tests:
    - FBC and differential count
    - ESR
    - Infection screen (HIV, TB, VZV, toxoplasma)
    - Hb electrophoresis (dependent on ethnic group)
    - Fasting lipogram
    - Lupus screen
- LP (to assess CSF for infection)
- Imaging:

- CT (to exclude haemorrhage and define the territory involved)
- MRI and magnetic resonance angiography (MRA)
- Conventional angiography

Differential Diagnoses

Paediatric stroke mimics include:

- Seizures and epilepsy (Todd's paresis)
- Migraine
- Psychogenic disease
- Inflammatory disease
- Intracranial infections
- Metabolic causes

Management

The aim of acute management is to preserve the penumbra. This is done by:

- Maintaining a low-to-normal body temperature
- Ensuring euglycaemia
- Maintaining good oxygen saturation
- Ensuring adequate cerebral perfusion

Secondary prevention should be started in patients with underlying cardiovascular disease. Start the child on anticoagulation with low molecular weight heparin. It is recommended that these children are managed in consultation with the relevant specialist (paediatric neurologist, haematologist or cardiologist).

Rehabilitation is multidisciplinary and should involve a physiotherapist, occupational therapist, audiologist and speech therapist. One must address issues with feeding, nutrition, pain, communication, mobility and positioning.

HEADACHES

Headaches are common in children and their frequency increases with age. Most headaches are benign. However, one must take a detailed history of the pain (SOCRATES) and ask about social and psychological factors.

Aetiology

Headaches may be caused by:

- Primary headache syndromes:
    - Migraine
    - Tension headache
- Secondary headache syndromes:
    - Sinusitis
    - Meningitis
    - Encephalitis
    - Hydrocephalus
    - Sleep apnoea or hypoventilation
    - Benign intracranial hypertension
    - Haematoma
    - Brain abscess
    - Brain tumour

Approach

One must rule out pathological causes of headaches by asking about the following danger signs:

- Headache which interrupts sleep
- Headache present on waking
- Visual disturbances
- Altered level of consciousness
- Focal neurological deficits
- Hypertension

If any of the above features are present, one must investigate further with:

- Lumbar puncture (to exclude meningitis)
- CT scan

Patients with danger signs and who cannot be managed at the primary care level should be referred to a paediatrician or paediatric neurologist.

MYELOMENINGOCOELE/MYELOMENINGOCOELE

A myelomeningocoele (see related image here) is a midline defect of the skin and vertebral arch which contains both meninges and neural tissue. The lumbosacral region of the spinal cord is the most commonly affected area. One must look for sensorimotor impairment as well as bladder and bowel incontinence.

Aetiology

The aetiology of myelomeningocoeles is poorly understood. However, evidence suggests that there is an association with inadequate levels of folate before conception and during the first trimester. There is an increased risk of recurrence in subsequent pregnancies.

Arnold-Chiari malformations are the most commonly associated congenital anomalies (lead to hydrocephalus).

Diagnosis and Investigations

Myelomeningocoeles may be diagnosed:

- In-utero via ultrasound (termination may be offered at this point)
- At birth

Once diagnosed, one should perform a CT scan and cranial USS (to assess initial ventricular size so that subsequent monitoring can be done).

Management

Prenatal counselling should

- Prenatal counselling

One should attempt to diagnose the condition early – in-utero or at birth. If diagnosed in-utero, prenatal counselling should be performed. If diagnosed at birth, one should initiate emergency management:

- Keep the defect and tissues sterile and moist (place saline-soaked gauze over the defect)
- Refer the child for urgent surgical correction (within 48 hours of delivery)

Associated abnormalities must then be excluded. A multidisciplinary team must be involved in the care of this child (neurologist, orthopaedic surgeon/neurosurgeon, physiotherapist, urologist).

MENINGITIS

See *Infectious Diseases* chapter.

SPINAL MUSCULAR ATROPHY (SMA)

It is an autosomal recessive disorder which is characterised by progressive hypotonia and muscular weakness. It is a common genetic cause of mortality in children, with males being affected more commonly than females (2:1). There is a high prevalence in Central and Eastern Europe.

Pathophysiology

The primary pathology involves progressive degeneration of α-motor neurons on anterior horn cells in the spinal cord. This is caused by a mutation in the gene responsible for the survival of motor neurons.

Clinical Presentation

The child will present with:

- Abnormal posture (pronated arms)
- Proximal weakness with atrophy
- Cranial nerve involvement, especially bulbar signs (in some patients) e.g. tongue fasciculations
- Reduced tone
- Areflexia
- Bell-shaped chest
- Normal sensation
- Normal intelligence
- Sparing of involuntary muscles (heart, sphincters, GIT)

Classification

SMA is classified into four types according to the age of onset of clinical features and most advanced physical milestone achieved:

- Type 1 – the children typically present within the first six months of life; they are unable to sit unsupported
- Type 2 – the children typically present at 3-15 months old; they are able to sit without support but are never able to stand or walk
- Type 3 – the children typically present between 18 months old and adulthood; they achieve independent ambulation
- Type 4 – the patient presents after 30 years of age (latest onset disease); all motor milestones have been achieved and independent ambulation is usually maintained through life

The prognosis is worse the younger the age of onset is. The median age of death is 10 years old and is normally due to respiratory compromise.

Management

Children with SMA are all affected differently, therefore treatment should be catered to the individual. Management is mainly supportive and involves a multidisciplinary team (pulmonologist, orthopaedic surgeon, nutritionist, genetic counsellor, social worker, occupational therapist, physiotherapist and orthotist). Surgical interventions are considered to treat scoliosis, contractures and fractures.

Thus far, no cure has been found but research is ongoing. Currently, scientists are looking at inhibitors of GABA synthesis and genetic therapy which attempts to fix/replace the affected gene.

NEUROCUTANEOUS SYNDROMES

They are congenital disorders that are due to genetic changes. Tissues and organs derived from the ectoderm are generally affected (i.e. skin, nervous tissue, eyeballs and retina), however bones and visceral organs may also be affected. The conditions evolve slowly throughout childhood and the lesions have malignant potential.

Diagnosis

Neurocutaneous syndromes may be diagnosed based on the results of:

- Genetic tests
- MRI scan
- CT scan
- EEG
- Eye exam
- Lesion biopsy

Classification

Neurocutaneous syndromes include:

- Tuberous sclerosis
- Sturge-Weber syndrome
- Neurofibromatosis

Tuberous Sclerosis

It is an autosomal dominant with variable expression. Features evolve as the child grows. It is diagnosed based on the presence of two major features, or one major feature and two minor features. TSC1 gene mutations are familial and TSC2 gene mutations are associated with adult polycystic kidney disease.

Sturge-Weber Syndrome

In this syndrome angiomas cross the leptomeninges and involve the skin of the face, producing a characteristic skin lesion (typically in the ophthalmic or maxillary region of the trigeminal nerve). The associated mutation is sporadic and there are three subtypes of the syndrome.

Affected children may present with:

- Facial naevus (port wine stain)
- Seizures
- Hemiparesis
- Hemianopia
- Headaches
- Glaucoma

- Learning difficulties
- Developmental delay

Neurofibromatosis (NF)

There are two types – NF 1 and 2. Males and females are equally affected (1:1) in both types (see related image here).

Neurofibromatosis Type 1 (Nf1)/Von Recklinghausen's Disease

This is an autosomal dominant condition which is more common than NF2. The child may present with:

- Cafe-au-lait spots
- Benign tumours – neurofibromas (see image linked above)
- Lisch nodules
- Seizures
- Scoliosis
- Facial pain
- Numbness
- Variable degree of intellectual disability
- Attention deficit disorder

NF1 is diagnosed based on the presence of specific diagnostic criteria:

- First-degree relative with confirmed NF1
- ≥6 café-au-lait spots
- Neurofibromas
- Optic glioma
- Axillary freckling
- Lisch nodules
- Bone lesions e.g. sphenoid dysplasia

Neurofibromatosis Type 2 (Nf2)

The child may present with:

- Hearing loss (due to vestibular schwannomas)
- Meningiomas
- Spinal tumours

- Neuropathies
- Headaches
- Facial pain
- Problems with balance and walking
- Neurofibromas
- Café-au-lait spots

Management

There is no cure for neurocutaneous syndromes. Management depends on the individual child's needs and requires input from a multidisciplinary team:

- Paediatric neurologist
- Plastic surgeons
- Genetic counsellor
- Neurosurgeon
- Ophthalmologist
- Dermatologist
- Oncologist
- Occupational therapist
- Speech therapist
- Physiotherapist

CEREBRAL PALSY

See *Disorders of Development* chapter.