# The most common syndromes and diseases in paediatric neurology/PGS

## Paroxysmal disease among children

Key points: seizures or states of disturbance of consciousness have often resolved on medical examination, so a history, an accurate description of the course of the condition with a focus on the activity that preceded the condition, triggering factors, duration of the condition, color changes, eye movements, motor activity during the condition, the nature of the decline of the condition, the child's behavior after the condition has ended, and the frequency of these conditions are critical to making a diagnosis. Home video of recurrent conditions is very helpful; EEG, or video-EEG monitoring, is an essential investigation.

### Paroxysmal disease among infants and toddlers

1. **Seizure with fever** - febrile cramps, CNS infections (sepsis, meningitis, meningoencephalitis), epilepsy triggered by fever.
2. **Seizures without fever** - Epileptic or provoked, caused by electrolyte imbalance, hypoglycaemia, intoxication.
3. **Apnea or breath holding:**
   - Neonatal apnea - Usually caused by immaturity, rarely by seizure.
   - Syncope with cyanosis, affective states - up to 5% of infants, often familial occurrence, provoked by anger, frustration or fear, crying before onset of state, breath stops in expiration, followed by cyanosis and ev. Unconsciousness. Onset of seizures is before 3 years, disappears between 4-8 years, prognosis is good.
   - Syncope with pallor - provocation typically by unexpected pain, crying rarely, sudden pallor, hypotonia and unconsciousness, the cause is usually vasovagal syncope, prognosis is good, disappears between 4-8 years.
4. **Migraine** - is rare, manifested by paroxysms of vomiting, dizziness or torticollis, consciousness is not disturbed, the condition lasts several minutes, accompanied by pallor, event nystagmus.
5. Further **involuntary movements and stereotypies'** (see below).

### Paroxysmal disease in older children

1. **Fever seizures - febrile convulsions** (unlikely after 4-5 years of age), CNS infection, epilepsy triggered by fever.
2. **Seizures without fever - epileptic**: Absence, complex partial seizures, myoclonic seizures, benign and age-related epileptic syndromes. Seizures provoked - systemic abnomalities (electrolyte abnormalities, hypoglycemia, intoxication, etc.)
3. **Migraine and migraine variants** - headache may not be present, a positive family history is typical, diagnosis is based on history and clinical examination, other investigations only exclude other etiologies.
   - Acute migraine with confusion - age 6-16 years, episodes of confusion lasting several hours, diff. dg. non-convulsive status epilepticus, intoxication.
   - Basilar migraine - in adolescents, with episodes of ataxia, vomiting, tinnitus, paresthesia or alternating hemiplegia, followed by throbbing headache.
   - Other variants: retinal, ophthalmoplegic, global transient amnesia.
4. **Syncope** - may be accompanied by increased muscle tension or twitching after loss of consciousness and falling (spasmodic syncope), typically short duration of unconsciousness and rapid recovery.
   - *Vasovagal* (reflex vasodilatation) - may also be triggered by emotion, typically prolonged standing, painful stimuli, prodromes with a feeling of dizziness and fainting, accompanied by pallor, cold skin.
   - *Cardiac arrhythmia* - unprovoked, even when lying or sitting, often familial.
   - *Orthostatic hypotension* - related to standing.
   - *Hyperventilation syndrome* - induced by agitation, paresthesias of the acres, lips, headache, unconsciousness.
5. **Primary sleep disorders:**
   - Narcolepsy with cataplexy - daytime falls on emotion without disturbance of consciousness, falls on imperative falling asleep (rare in children), sleep paralysis on falling asleep or waking, automatic acting while awake at night.
   - Parasomnias with impaired waking reactions - night terrors, sleepwalking, waking with confusion - patients are partially addressable, not responding adequately, difficult to wake, often automatic actions (urinating in the wrong places), amnesia on episodes, typically occurring within the first 2 h after falling asleep.
6. **Enchantment, daydreaming** - total, normal, patients addressable, EEG normal on status.
7. **Psychogenic seizures** - often longer duration, bizarre manifestations, video EEG monitoring often required.
8. **Paroxysmal involuntary movements:**
   - **Tic disorder** - transient or chronic, simple or complex movements, vocalization (Tourette's sy.), association with obsessive compulsive disorder.
   - *Benign paroxysmal torticollis in infancy* - condition is initially more frequent (onset by 3 months of age), resolves by 5 years of age, most likely vestibular etiology. The condition is accompanied by agitation, pallor, vomiting, ataxia. The child is quiet only in an atypical position, duration is minutes to days.

Laterocollis, retrocollis or torticollis is the most common manifestation; however, the trunk or limbs may also be affected. Other examinations tend to be normal.
- *Paroxysmal tonic upward deviation of the eyeballs* - onset during the first months of life, episodes last several hours, vertical nystagmus appears when attempting to look down, condition resolves in sleep and worsens during wakefulness. Other examinations are usually normal, the condition resolves within a few years.
- **Paroxysmal dyskinesia - *episodes of*** chorea**, *balismus*** *or* ***dystonia****, accentuation after alcohol or caffeine intake, during physical exertion or stress.*
- **Hyperekplexia** - dystonia states after startle, exaggerated reaction with prolonged motor persistence, often associated with more frequent nocturnal myoclonic jerks. Furthermore, impaired habituation to startle stimuli.
- *Episodic ataxias* - often familial, after physical exertion, may be associated with hypokalemic conditions, these are genetic ion channel disorders, often acetazolamide treatment is effective.
- **Stereotypes** - purposeless repetitive monotonous movements with no apparent voluntary control, unlike tics they are easily suppressed by willpower, without a compulsive component.
- *Other manifestations* - secondary paroxysmal dyskinesias with cortical, basal ganglia or spinal cord involvement.

# Disorders of psychomotor development

## Delay in psychomotor development

**Definition:** static, non-progressive significant developmental delay in 2 or more of the following skills: 1. gross or fine motor skills, 2. speech, 3. cognitive function, 4. social or personal interaction, 5. activities of daily living. The term mental retardation is used for children from 5 years of age. The overall prevalence of delay in only one component is 5-10%, global delay is present in 1-3% of children. The important distinction is from developmental regression, i.e., the loss of skills that a child already had. If the delay has a maximum in impaired motor development or language development, which have more specific causes.

#### Speech Development Disorders

1. **Hearing impairment** is a very common cause of speech development disorder requiring most phoniatric care.
2. **Autism** - developmental disorder evident between 1-2 years of age, apart from speech impairment, social interaction is severely affected, predilection for stereotyped activities.
3. **Acquired aphasia** (Landau-Kleffner's syndrome) - epileptic syndrome, regression of speech development is frequent, seizures in some patients, very heterogeneous EEG correlate, typically during nocturnal video EEG monitoring or polysomnography spikes occur in changing localizations, typical manifestation is status epilepticus electricus in NREM sleep.
4. **Bilateral structural involvement of the perisylvian regions**.
5. **Developmental dysphasia with normal findings** (normal nonverbal intellect and healthy hearing) - prevalence of 1-1.2% in preschool children. Low birth weight, prematurity, mental retardation or developmental dysphasia in family history, multilingual upbringing. Appropriate to rule out epileptic disease - EEG or nocturnal video-EEG monitoring.

#### Motor Development Disorder

1. Cerebral palsy - acquired non-progressive disability of the brain during its development, typically by pre- and perinatal influences - intrauterine infections, asphyxia, ischemic conditions, acquired brain malformations. The most common forms are: hemiplegia, spastic diplegia, quadriplegia, extrapyramidal form and cerebellar form with ataxia. The concomitant cognitive deficit is of a highly variable degree.
2. Myopathy congenital, spinal muscular atrophy - regression or slow progression also present. Metabolic causes are also in the differential diagnosis.
3. Neuropathies - hereditary, also possible within metabolic disease, may also progress.

#### Global psychomotor developmental delay

1. **DMO** - In the context of prenatal and perinatal disability, marked prematurity, intracranial hemorrhage, hydrocephalus.
2. **Chromosomal and genetic disorders**, often associated with craniofacial dysmorphism and congenital defects of other organs.
   - **Sy. Fragmented X** - trinucleotide expansion in the FMR1 or FMR2 gene, typically affects males, with elongated faces, large ears, macroorchidism and possibly autistic symptoms; females may also be affected if the expansion is very long. In males, this is the most common genetic cause of mental retardation.
   - **Rett syndrome** - dominant mutation in the MECP2 gene on the X chromosome, females affected, retardation evident by age 1 year, developmental regression may also be present. Furthermore, progressive microcephaly, hypotonia, ataxia, epileptic seizures, autistic symptoms, irregular breathing, impaired hand motor skills.
   - **Down's disease** - trisomy 21. chromosome, typical hypotonia, round face with flat nose and epicanthus, often present congenital heart defects and sleep apnea syndrome.
3. **Sturctural malformation of the brain** - lobar dysgenmesis, cortical dysplasia, corpus callosum agenesis, other dysmorphic features are often present. The association with chromosomal aberration must be excluded.

4. **Neurofakomatźy** (neurocutaneous syndromes):
    - Neurofibromatosis - learning disabilities, cognitive deficits. Typical features are neurofibromas, optic gliomas, caffé-au-latte spots (more than 8), bone malformations, positive family history (NF1), or bilateral neurinoma acoustica (NF2).
    - Tuberous sclerosis - beyond PM developmental delay, pharmacoresistant epilepsy, cortical malformations, subependymal hamartomas and retinal tumors, skin manifestations (even café-au-lait spots) including adenoma sebaceum, cardiac, renal, bone, renal or pulmonary cysts.
5. **Lead poisoning** - often only isolated cognitive impairment and high serum lead levels.
6. **Intrauterine infection**:
    - Toxoplasmosis - PMR manifestations after 1 year of life, also hydrocephalus, intracranial calcification, chorioretinitis, acute symptoms are in neonates (rash, fever, seizures, increased intracranial pressure, thrombocytopenia and icterus).
    - Rubeola - rarely encephalopathy - hypotonia, epileptic seizures, lethargy, also other organ involvement (icterus, chorioretinitis, cardiac malformations, deafness, anaemia, thrombocytopenia, rash, cataract).
    - Cytomegalovirus (CMV) - microcephaly, intracranial calcification, also rash, chorioretinitis, icterus, organomegaly.
    - Herpes simplex virus (HSV) - acute encephalitis with epileptic seizures, multiplication of elements in cerebrospinal fluid, PCR rather than serology is appropriate.
7. **Encephalopathy**.

## Regression of psychomotor development

The differential diagnosis of this symptom is very broad. In the history, it is important to assess the age at onset of the difficulties, as well as the involvement of parts of the CNS (white or grey matter, CNS compartments e.g. spinocerebellar atrophy), involvement of other organs or the peripheral nervous system. CNS + other organ or PNS involvement is indicative of spastic or mitochondrial disease. It is important to assess the course of regression of the development whether the deterioration is rapid and acute or coming in attacks or chronically progressive. Most of these diseases are not curable, but the diagnosis is important to determine further investigation and treatment plans and for genetic counseling, possibly for research reasons. Potentially treatable are disorders of metabolism of certain amino acids or carbohydrates, as well as progressive hydrocephalus, hypothyroidism or congenital infections (HIV).

### Primary gray matter involvement

1. **Autism** is the most common cause of developmental regression in children aged 18 to 26 months, individuals with lesser disabilities may begin to show impairments even later. Epileptic seizures are more frequent in older children, especially if there is evidence of a genetic basis.

The # **Rett syndrome** (see above) may also have developmental regression in its clinical picture and is the most common identifiable cause of mental regression and acquired microcephaly in girls.

Furthermore, neuronal ceroid-lipofuscinoses (visual impairment and myoclonia, diagnosis from leukocytes or skin biopsy), Menkes disease (copper transport disorder, low ceruoplasmin and total copper, phakmacoresistant myoclonic jerks), Infantile neuronal dystrophy (hypotonia, spasticity, optic atrophy), Lesch-Nyhan disease (hyperuricemia, initial hypotonia progressing to rigidity, torticollis, choratiform involuntary movements, autoaggression). In older children, Huntington's disease (5% of manifestations occur before 14 years of age, rigidity is the main symptom in childhood, followed by cognitive deterioration and seizures, and more rarely ataxia) and xeroderma pigmentosum (skin photosensitivity, deafness, microcephaly and spinocerebellar degeneration due to impaired DNA repair) are also considered.

### Progressive hydrocephalus

Macrocephaly, prominent fontanelles, lethargy, vomiting, sign of the setting sun (sclera visible above the iris), consequence of congenital malformations, intracranial expansions (especially in the posterior fossa), condition after hemorrhage or infection. Treatment by ventriculoperitoneal shunt.

### Disorders of amino acid metabolism

Phenylketonuria (vomiting, irritability), homocystinuria (thromboembolism, lens dislocation, osteoporosis), leucinosis (acute or episodic course - encephalopathy with seizures, spasticity and hypoglycaemia, ataxia), urea cycle disorders (intermittent encephalopathy with vomiting and seizures, hyperammonemia) and X-linked transcarbamoylase deficiency (with manifestation later in life in girls), organic aciduria (encephalopathy with vomiting and hypotonia, abnorm. Lowe syndrome (myopathy, neuropathy, congetinal cataract or glaucoma, renal acidosis).

### Hypothyroidism

Along with developmental regression, wide small fontanelle, constipation, impaired thermoregulation, icterus, macroglossia and umbilical hernias are present in infants. Early treatment is essential for the prognosis of the patient.

### Lysosomal storage diseases

Craniofacial dysmorphia and hepatosplenomegaly are common. Diagnosis is made by metabolic testing especially urine and enzymatic testing of cutlivated fibroblasts, or in some cases peripheral blood leukocytes are sufficient. Screening tests are chitotriosidase activity in peripheral blood, cytological examination of bone marrow punctate and imaging of the spleen and liver.

The most common representatives are gangliosidoses GM1 (visual impairment), GM2 (with hepatosplenomegaly), Gaucher disease (sucking disorder, oculomotor disorders), I-cell disease (heart failure), mucopolysaccharidoses type I (skeletal abnormalities) and type III (mental retardation may be the only symptom, esp. Speech impairment), type II (craniofacial dysmorphia, neuropathy), and VII (frequent dysmophia without corneal manifestations) are typical later in life; some types are treatable by bone marrow transplantation or enzyme replacement therapy, Neimanm-Pick disease (but often later onset of symptoms).

#### Mitochondrial disease

Often abnormal ratio of lactateu to pyruvateu, fasting, after physical or food stress. Multiple systems are typically affected, and difficulties can arise at any age. Mental regression is part of the encephalopathy; epileptic seizures are common. Diseases are often conditioned by defects in mitochondrial DNA; the most useful investigation is muscle biopsy.

The most common syndromes are MELAS (focal neurological deficits similar to stroke, deafness and myopathy), Leigh syndrome (respiratory disorders, oculomotor disorders), Alpers syndrome, MERRF (also with myopathy), Kearns-Sayre disease (also oculomotor disorders).

#### Primary white matter disorders

Conditions manifested mainly by spasticity, focal neurological findings and visual impairment.

The most common include - galactosemia (cerebral edema, vomiting, hepatomegaly), Canavan disease (macrocephaly, optic atrophy), Alexander disease (macrocephaly, ep. seizures), Krabbe's disease (visual impairment, opisthotonus with hyporeflexia), adrenoleukodystrophy (hyperreflexia, ep. seizures), Pelizaeus-Merzbacher disease (spasticity, nystagmus, choreoathetosis, BAEP abnormalities), Metachromatic leukodystrophy (peripheral neuropathy).

#### Peroxisomal diseases

1. **Zellweger syndrome** - dysmorphia, hypotonia, arthrogryposis, also biliary cirrhosis, polycystic kidneys, retinal degeneration, cerebral malformations.
2. **Refsum's disease** - early onset, blindness, deafness.

#### Infectious diseases

1. **AIDS** - microcephaly, regression of PM development or dementia, spasticity, less commonly ataxia, involuntary movements and myoclonic seizures, opportunistic CNS infections may be a distant cause of deterioration.
2. **Congenital syphilis** - in children under 2 years of age Hutchinson's trias (deafness, interstitial keratitis, barrel-shaped incisors), in newborns condylomata lata, rashes and periostitis or osteochondritis. Treatment with ATB has a good effect, testing for HIV co-infection is necessary.
3. **Subacute sclerosing panencephalitis**.

#### Untreated or pharmacoresistant epilepsy

In older children, it already causes signs of cognitive regression. The cause may be a neurodegenerative disease, neurofacomatosis or idiopathic (Lennox-Gastaut, West's syndrome, Landau-Kleffner, and other childhood epileptic syndromes).

Disorders of glycoprotein glycosylation (CDG) ====

A very large group of disorders including encephalopathies, with a highly variable phenotype. Screening test is a level of low sialylated transferrin (non-valid results in alcohol abuse).

# Movement disorders - hypotonia in children

Main signs: the cause may be either central or peripheral. On examination, head lifting, poor posture in horizontal or vertical suspension, various reduced reflexes in the limbs, bulbar syndrome may be present. Diseases with the earliest onset manifest as congenital hypotonia, with later onset of disease as regression of motor development. If the disease is acute or subacute, ventilatory support may be necessary (e.g., spinal injuries, botulism, sy. Spinal muscular atrophy and myopathy require ventilatory support only with prolonged disease duration.

## Hypotonia in infants

#### Cerebral hypotonia

Often associated with delayed and mental development, epileptic seizures are present, primitive reflexes persist, and malformations of other organs are present. With the passage of time, hypotonia may progress to spasticity

1. **Non-progressive encephalopathy**
   - Brain malformation - abnormal size and shape of the head, often accompanied by craniofacial dysmorphism.
   - DMO - perinatal asphyxia, infections, CNS haemorrhage, trauma.
2. **Chromosomal aberrations**
   - Prader-Willi syndrome - deletion of the maternal part of chromosome 15q11-13, periodically impaired sucking, hypogonadism, PM retardation, hyperphagia with obesity in later life, sleep disorders (Sec. narcolepsy, sleep apnea).
   - Down syndrome (trisomy 21)
3. **Inherited metabolic disorders**
   - Peroxisomal disorders - Zellweger's syndrome (arthrogryposis, cereberohepatorenal syndrome, epilepsy), neonatal form of adrenoleukodystrophy (epilepsy, hepatomegaly, retinal degeneration)
   - Lowe's syndrome (Oculocerebrorenal sy) - congenital cataract or glaucoma, renal impairment.
   - Familial dysautonomia (Riley-Day) - impaired sucking, autonomic dysfunction, insensitivity to pain, recurrent vomiting.
   - Acid maltase deficiency - myopathy with cardiomyopathy
   - GM1 gangliosidosis - PM retardation, visual impairment
4. **Benign idiopathic congenital hypotonia** - spontaneous relapse with further normal development.

**Spinal Cord Disease**

1. **Perinatal and birth trauma** - impaired ventilation may be present with involvement of the medulla oblongata or high cervical spinal cord lesions. Diagnosis of a defect in sensation can be difficult in such young children.
2. **Hypoxic-ischemic myelopathy in the context of perinatal asphyxia**

**Neuromuscular transmission disorders**

1. **Botulism in infants** - after home-preserved vegetables or honey - constipation, proximal muscle weakness, no impairment of consciousness, dysphagia, photoreaction disorder, weak crying. Possibility of respiratory clamping, incrememten in EMG on repetitive stimulation, effective treatment with antitoxin.
2. **Myasthenia**
   - Congenital - defect in epsilon subunit of Achr, sucking and respiratory disturbances, ptosis, arthrogryposis, less commonly ophthalmoplegia, worsening in intercurrent infection.
   - **Neonatal myasthenia** - in 10-20% of births to mothers with myasthenia gravis - sucking disorder, fatigue, arthrogryposis, weak crying. Caused by passive transfer of anti-Achr antibodies, spontaneous adjustment.

**Polyneuropathy -Guillain-Barré syndrome**

Hereditary sensorimotor neuropathy (most commonly HMSN III), congenital hypomyelinating neuropathy.

**Muscular diseases**

1. **Congenital myopathies** - arthrogryposis, contractures, scoliosis, ophthalmoplegia, respiratory and swallowing difficulties, hip dislocations. Muscle biopsy is diagnostic.
2. **Congenital myotonic dystrophy** - respiratory difficulties, hypomimia, sucking disorders, cardiomyopathy, arthrogryposis, later in life mental retardation, cataract, endocrinopathy, later baldness. Mothers are often also affected to a lesser or greater extent.
3. **Congenital muscular dystrophy**
4. **Metabolic myopathies** - acid maltase deficiency (cardiomyopathy), cytochrome c oxidase deficiency (lactic acidosis, creatine kinase elevation), kinase deficiency and phophyllos (muscle cramps and exercise intolerance).

**Spinal muscular atrophy (defect with SMN gene)**

1. **Acute (SMA I)** - onset by 6 months of age, proximal weakness, areflexia. Respiratory difficulties, no arthrogryposis, development of atrophy, fasciculations, often aspiration, rapid progression
2. **Chronic (SMA II)** - onset between 3-18 months of age, normal newborn at birth, with faster or slower progression, diagnosis is based on EMG findings and genetic testing.

## Muscle strength disorders in older children =

Main symptoms: gait disturbance, tiptoeing, often other development may be normal, apart from EMG, MRI examination of LS area is important. However, the most common cause is idiopathic shortening of the Achilles tendon.

- **Structural disorder of the lumbosacral plexus or spinal cord** - Often the neurological findings are abnormal, the cause may be fixed spinal cord syndrome.
- **Muscular dystrophies** - Often dystrophinopathy, also cardiomyopathy, variable intellectual disability.

- **DMO**
- Autism and idiopathic shortening of the Achilles tendon.

# Ataxia

## Dizziness and vertigo in children

**Infections**

- **Bacterial mesotheitis or labyrinthitis** - vomiting, nausea, hearing loss. The consequence may be a cholesteatoma penetrating the scalene and even possibly the labyrinth, typically manifested by vertigo on coughing, sneezing or pressure on the ear.
- **Bacterial meningitis** - fever, disturbance of consciousness, meningeal irritation, may be due to mastoid enlargement or other otogenic infections.
- **Vestibular neuritis** - isolated vertigo with good prognosis, improvement starts within 48 h, most often viral etiology.

**Medication and drug side effects** - Mostly antiepileptics, neuroleptics, and some antibiotics (hearing loss may occur).

**Kinetosis** - Disparity between visual and vestibular input, most commonly in children seated in the rear of a vehicle at a small window.

**Migraine** - May be a cause of vertigo even without headache, benign paroxysmal vertigo in children may be a variant of migraine.

**Trauma** - After head trauma often occurs within 3 days, vestibular coma manifests as long-lasting vertigo triggered by head movements. It is necessary to rule out fractures of the skull base.

**Epilepsy** - Vertigo may be aura before seizure, often accompanied by nausea, often followed by arrest of activity or disturbance of consciousness.

**Other rare causes** - Structural lesions of the brainstem, Mernière's disease and congenital defects of the inner ear.

## Acute transient ataxia

**Poisoning or adverse effect of medication** is a very common cause of ataxia.

**Autoimmune and parainfectious causes**

1. **Miller-Fischer syndrome** - also ophthalmoplegia of varying degrees, hyporeflexia, respiratory monitoring required.
2. **Acute perainfectious cerebellitis (rhombencephalitis)** - very common in children.
3. **Multiple sclerosis cerebrovascularis.**

**Migraine** - also common, a variant is basilar migraine or benign paroxysmal vertigo.

**Traumata** - post coma syndrome persists for weeks, dif. Dg. intracranial hematoma or dissection to occlusion in the vertebrobasilar basin.

**Intracranial expansion** - acute onset of difficulties with tumor hemorrhage, decompensation of hydrocephalus (often including cephalea, vomiting, nausea, impaired consciousness), paraneoplastic manifestations in neuroblastomas or medulloblastoma (possible spinal localization of primary lesion) - typically opsoclonus (chaotic eye movements) and myoclonus.

**Infection** - typically rhombencephalitis.

**Genetically linked and metabolic diseases**:

- **Episodic ataxia** - ion channel disorder, good effect of acetazolamide.
- **Hartnup's disease** (also encephalopathy and rash).
- **Intermittent varians leucinosa** (hereafter encephalopathy).
- **Pyruvate dehydrogenase deficiency** - further episodes of lactic acidosis, hypotonia, encephalopathy.

**Vascular disease** - cerebellar haemorrhage, rarely ischaemia in this area, more commonly Kawasaki's disease (systemic vasculitis with cardiac manifestations, fever, limb oedema and lymphadenopathy, may also be aseptic meningitis).

**Pseudoataxia in epilepsy, as well as conversion and psychogenic disorders**

## Chronic ataxia

**DMO and other motor retardation** (see above)

**Congenital malformations of the brain** - Basilar impression, cerebellar hypoplasia, Arnold-Chiari malformation (type I - often also with cephalea).

**Intracranial expansion** - Apart from ataxia, there are also signs of intracranial hypertension.

**Genetic diseases:**

1. **Autosomal dominant spinocerebellar atrophy** - many types.
2. **Autosomal recessive ataxia - Friedreich's ataxia** (areflexia, py. irritation, also pes cavus, cardiomyopathy and diabetes), Hypobetalipoproteinemia (vit E deficiency), Ataxia-telagiectatica (also recurrent respir. Infections, high alpha-fetoprotein), Hartnup's Chroroba, GM2 gangliosidosis (tremor, dysarthria), Leusinosa, Metachromatic Leukodystrophy (epilepsy, dementia), Marinesco-Sjörgen's Syndrome (progressive ataxia, congenital cataract, mental retardation). Pyruvate dehydrogenase deficiency, progressive myoclonic epilepsy, Refsum's disease (polyneuropathy, retinitis pigmentosa, hearing loss, cataracts, cardiomyopathy, pes cavus).
3. **X-linked ataxia** - adrenoleukodystrophy (typical white matter image on MRI), Leber's optic atrophy (mitochondrial disease visual impairment).

# Headache in children

## Acute headache

- **Feverish state in extracranial infection**
- **Post-traumatic state, post-ictal syndrome** - usually persistent, then with a good prognosis; conversely, progressive, with signs of increased intracranial pressure, more suggestive of intracranial lesions.
- **Sinusitis** - often with fever, sinus pain and chronic rhinitis.
- **Aseptic meningitis** - with fever, meningeal irritation, more likely in older children.
- **Migraine** - episodes of throbbing pain, nausea, vomiting visual disturbances, photo- and phonophobia, typically positive family history, duration is shorter in children than in adults (up to 1 h).
- **Tension headache** - diffuse in nature, sensation of pressure.
- **Seizure pain in patients with epilepsy**
- **Rare causes** - intracranial expansion, arterial hypertension, intoxication, intracranial haemorrhage, hydrocephalus, bacterial meningitis, acute metabolic disease (hypoglycaemia, porphyric crisis), cluster-headache (very rare in children), post-puncture syndrome, neuralgia (rare in children), vascular (pheochromocytoma, carcinoid).

## Chronic headache

- **Migrainous cephalea**,
- **Tension headache** - usually chronic, poorly localized pain of prolonged duration.
- **Post-ictal syndrome** - can last for weeks.
- **Seizure pain** - may occur repeatedly, the patient has amnesia about his or her own seizures and the surroundings do not notice them.
- **Intracranial hypertension** - vascular cause, hydrocephalus, abscess, pseudotumor cerebri.
- **Adverse drug reactions, substance abuse, withdrawal syndromes**;
- **Vertebrogenic aetiology** common cause of headache, especially occipitally, where spasms of the short extensors of the head are palpable, also typically present with disturbances of spinal dynamics. Treatment with gentle myorelaxants (Mg lactici) and rehabilitation;
- **Sinusitis**;
- **Ocular cause (glaucoma, astigmatism), odontogenic cause, neuralgia, post-puncture syndrome, cluster-headache, metabolic (hypoglycemia, profusion), hypoxia (sleep apnea, hypoventilation)**;
- **Psychogenic and purposeful.**