The most common syndromes and diseases of pediatric neurology/PGS

Paroxysmal disorders in children

Main points: Seizures or states of impaired consciousness often have already subsided during a medical examination, so the anamnesis is decisive for establishing a diagnosis, an accurate description of the process of the condition focusing on the activity that preceded the condition, triggering factors, duration of the condition, color change, eye movements, motor activity during the condition, the character of the condition's resolution, the child's behavior after the condition ends and the frequency of these conditions. A home video of recurring conditions is very beneficial, an essential examination is an EEG or video-EEG-monitoring.

Paroxysmal disorders in infants and toddlers

1. **Seizures with fever** – febrile convulsions, CNS infection (sepsis, meningitis, meningoencephalitis), epilepsy triggered by fever.
2. **Seizures without fever** – epileptic or provoked, the cause is an electrolyte imbalance, hypoglycemia, or intoxication.
3. **Apnea or breath holding:**
   - Neonatal apnea - usually caused by immaturity, rarely seizure.
   - Syncope with cyanosis, affective states - in up to 5% of infants, often a familial occurrence, provoked by anger, frustration or fear, crying before the state occurs, breath stops in expiration, followed by cyanosis and possibly unconsciousness. Onset of seizures is before 3 years, disappear between 4-8 yearthe s, prognosis is good.
   - Syncope with pallor - typically provoked by unexpected pain, rarely crying, sudden pallor, hypotonia and unconsciousness, usually caused by vasovagal syncope, prognosis is good, conditions resolve between 4-8 years.
4. **Migraine** - it is rare, it is manifested by paroxysms of vomiting, di,zziness or torticollis, consciousness is not impaired, the condition lasts for several minutes, accompanied by pallor, possibly nystagmus.
5. Furthermore, involuntary movements and stereotypies (see below).

Paroxysmal disorders in older children

1. **Seizures with fever – 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. Provoked seizures – systemic abnormalities (electrolyte abnormalities, hypoglycemia, intoxication, etc.)
3. **Migraine and migraine variants** – headache may not be present, positive family history is typical, the diagnosis is based on the history and clinical examination, other examinations only exclude other etiology.
   - acute migraine with confusion - age 6-16 years, episodes of confusion lasting several hours, differential diagnosis 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 the loss of consciousness and fall (convulsive syncope), short duration of unconsciousness and quick recovery are typical.
   - Vasovagal (reflex vasodilatation) – it can also be triggered by emotion, typically prolonged standing, painful stimuli, prodromes with a feeling of dizziness and fainting, accompanied by paleness, cold skin.
   - Cardiac arrhythmia – occurrence unprovoked, even when lying down or sitting, often familial.
   - Orthostatic hypotension - relation to the position.
   - Hyperventilation syndrome - provoked by irritation, paresthesia of the peripheral parts of the body, lips, headache, unconsciousness.
5. **Primary sleep disorders:**
   - Narcolepsy with cataplexy - falls during the day during emotions without disturbance of consciousness, falls during imperative falling asleep (rare in children), sleep paralysis during falling asleep or waking up, automatic actions while awake at night.
   - Parasomnias with disordered awakening reactions - night terrors, sleepwalking, awakening with confusion - patients are partially approachable, do not respond adequately, difficult to wake up, often automatic actions (urinating in the wrong places), amnesia for events, typically after the conditions occur within the first 2 hours after falling asleep.
6. **Staring, daydreaming** - quite common, patients approachable, EEG is normal.
7. **Psychogenic seizures** - often longer duration, bizarre manifestations, video EEG monitoring is often necessary.
8. **Paroxysmal involuntary movements:**
   - *Tic disorder* – transient or chronic, simple or complex movements, vocalization (Tourette syndrome), association with obsessive compulsive disorder.
   - *Benign paroxysmal torticollis in infancy* – initially the condition is more frequent (onset within 3 months of age), disappears by 5 years of age, most likely of vestibular etiology. The condition is accompanied by

agitation, pallor, vomiting, ataxia. The child is calm only in an atypical position, the duration is minutes to days. The most common manifestation is laterocollis, retrocollis, or torticollis, but the trunk or limbs can also be affected. Other examinations are usually normal.
- *Paroxysmal tonic upward deviation of the bulbs* - onset during the first months of life, episodes last several hours, vertical nystagmus appears when trying to look down, the condition disappears during sleep and worsens during wakefulness. Other examinations are usually normal, the conditions disappear within a few years.
- *Paroxysmal dyskinesia* – episodes of chorea, ballism or dystonia, accentuation after drinking alcohol or caffeine, during physical exertion or stress.
- *Hyperekplexia* – states of dystonia after startle, an exaggerated reaction with longer motor persistence, often associated with more frequent nocturnal myoclonic jerks. Furthermore, habituation disorder to startle stimuli.
- *Episodic ataxias* - often familial, after physical exertion, can be associated with hypokalemic states, these are genetic disorders of ion channels, treatment with acetazolamide is often effective.
- *Stereotypes* - purposeless repetitive monotonous movements without obvious voluntary control, unlike tics, they are easily suppressed with the help of will, without a compulsive component.
- Other manifestations – secondary paroxysmal dyskinesia when the cortex, basal ganglia, or spinal cord are affected.

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 functions, 4. social or personal interactions, 5. activities of daily living. The term mental retardation is used for children from 5 years of age. The overall prevalence of delay in just one component is 5-10%, global delay is present in 1-3% of children. It is important to distinguish it from developmental regression, i.e. the loss of skills that the child already had. If the delay has its maximum in a disorder of motor development or language development, which has more specific causes.

Disorders of speech development

1. Hearing impairment – a very common cause of speech development disorder requiring mostly phoniatric care.
2. Autism – a developmental disorder evident between 1-2 years of age, in addition to the speech disorder, social interaction and a fondness for stereotypical activities are also significantly affected.
3. Acquired aphasia (Landau-Kleffner syndrome) – epileptic syndrome, regression of speech development is also common, seizures in some patients, very diverse EEG correlate, typically spikes occur in changing locations during nighttime video EEG monitoring or polysomnography, status epilepticus electricus is a typical manifestation in NREM sleep.
4. Bilateral structural involvement of the perisylvian areas.
5. Developmental dysphasia with normal findings (normal non-verbal intelligence and healthy hearing) - prevalence 1-1.2% in preschool children. Risk factors: Low birth weight, prematurity, mental retardation or developmental dysphasia in the family history, multilingual education. Appropriate to rule out epileptic disease - EEG or night video EEG monitoring.

Motor skills development disorders

1. Cerebral palsy – acquired non-progressive impairment of the brain during its development, typically due to 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 concurrent cognitive deficit is of a very variable degree.
2. Congenital myopathy, spinal muscular atrophy - regression or slow progression is also present. Metabolic causes are also part of the differential diagnosis.
3. Neuropathy - hereditary, also possible as part of metabolic disease, can also progress.

Global delay in psychomotor development

1. **cerebral palsy** – as part of prenatal and perinatal impairment, significant prematurity, intracranial bleeding, hydrocephalus.
2. **chromosomal and genetic disorders**, often associated with craniofacial dysmorphism and congenital defects of other organs.
   - **Fragile X syndrome (FXS)** – expansion of trinucleotides in the FMR1 or FMR2 gene, typically affected men, with elongated faces, large ears, macroorchidism, and possibly autistic manifestations, with a very long expansion, women can also be affected. In men, this is the most common genetic cause of mental retardation.
   - **Rett syndrome** – dominant mutation in the MECP2 gene on the X chromosome, females are affected, retardation is evident by 1 year of age, developmental regression may also be present. Further progressing microcephaly, hypotonia, ataxia, epileptic seizures, autistic manifestations, irregular breathing, impaired free motor skills of the hands.
   - **Down's syndrome** - trisomy of chromosome 21, typical hypotonia, round face with flat nose and epicanthus, congenital heart defects, and sleep apnea syndrome are often present.
3. **structural malformation of the brain** - dysgenesis of the lobes, cortical dysplasia, agenesis of the corpus callosum, other dysmorphic features are often present. It is necessary to exclude a connection with a

chromosomal aberration.
4. **phakomatoses** (neurocutaneous syndromes):
   - Neurofibromatosis – learning disabilities, cognitive deficits. Typical features are neurofibromas, optic gliomas, caffé-au lati spots (more than 8), bone malformations, positive family history (NF1), or bilateral acoustic neuroma (NF2).
   - Tuberous sclerosis – in addition to delayed psychomotor development, drug-resistant epilepsy, cortical malformations, subependymal hamartomas, and retinal tumors, skin manifestations (including café-au-lait spots) incl. sebaceous adenoma, tumors of the heart, kidneys, bone, renal or lung cysts.
5. **lead poisoning** - often only isolated cognitive impairment and high serum lead levels.
6. **intrauterine infection:**
   - Toxoplasmosis - manifestations of PMR after 1 year of life, then hydrocephalus, intracranial calcification, chorioretinitis, acute symptoms are in newborns (rash, fever, seizures, increased intracranial pressure, thrombocytopenia, and icterus).
   - Rubella - rarely encephalopathy - hypotonia, epileptic seizures, lethargy, further involvement of other organs (icterus, chorioretinitis, malformation of the heart, deafness, anemia, thrombocytopenia, rash, cataract).
   - Cytomegalovirus (CMV) – microcephaly, intracranial calcification, also rashes, chorioretinitis, icterus, organomegaly.
   - Herpes simplex virus (HSV) – acute encephalitis with epileptic seizures, multiplication of elements in the cerebrospinal fluid, suitable for PCR rather than serology.
7. **encephalopathy.**

Regression of psychomotor development

The differential diagnosis of this symptom is very broad. In anamnesis, it is important to evaluate the age at the onset of problems, as well as the involvement of part of the CNS (white or gray matter, CNS sections, e.g. spinocerebellar atrophy), involvement of other organs or the peripheral nervous system. Affecting the CNS + other organs or the PNS indicates a stage or mitochondrial disease. It is important to assess the progress of the regression of development, whether the deterioration is rapid and acute, or comes in attacks or is, chronically progressive. Most of these diseases are not curable, but the diagnosis is important to determine the next plan of examination and treatment and for genetic counseling, possibly also for research reasons. Potentially treatable are disorders of the metabolism of certain amino acids or carbohydrates, as well as progressive hydrocephalus, hypothyroidism or c,ongenital infection (HIV).

Primary gray matter involvement

1. **Autism** is the most common cause of developmental regression in children between the ages of 18 and 26 months, individuals withfewers disabilities may begin to show disorders even later. Epileptic seizures are more frequent in older children, especially if a genetic basis is proven.
2. **Rett syndrome** (see above) can also have developmental regression in its clinical picture, it is the most common identifiable cause of mental regression and acquired microcephaly in girls.

Also, neuronal ceroid-lipofuscinoses (visual impairment and myoclonus, diagnosis from leukocytes or skin biopsy), Menkes disease (copper transport disorder, low ceruoplasmin and total copper, pharmacoresistant myoclonic convulsions), Infantile neuronal dystrophy (hypotonia, spasticity, optic atrophy), Lesch -Nyhan's disease (hyperuricemia, initial hypotonia turns into rigidity, torticollis, choratiform involuntary movements, autoaggression). In older children, Huntington's disease (in 5% of manifestations before 14 years of age, in childhood the main symptom is rigidity, then cognitive deterioration and seizures, less commonly ataxia) and xeroderma pigmentosum (skin photosensitivity, deafness, microcephaly, and spinocerebellar degeneration on the background) come into consideration DNA repair disorders).

Progressive hydrocephalus

Macrocephaly, protruding fontanelles, lethargy, vomiting, the sign of the setting sun (sclera visible above the iris), the result of congenital malformations, intracranial expansions (especially in the posterior cranial fossa), the condition after bleeding or infection. Ventriculoperitoneal shunt treatment.

Disorders of amino acid metabolism

Phenylketonuria (vomiting, irritability), homocystinuria (thromboembolism, lens dislocation, osteoporosis), leucinosis (acute or episodic progress – encephalopathy with seizures, spasticit,y and hypoglycemia, ataxia), disorders in the urea cycle (intermittent encephalopathy with vomiting and seizures, hyperammonemia) and X-linked transcarbamoylase deficiency (with manifestation at a later age in girls), organic aciduria (encephalopathy with vomiting and hypotonia, abnormal sweat odor), Lowe's syndrome. (myopathy, neuropathy, congenital cataract or glaucoma, renal acidosis).

Hypothyroidism

Along with developmental regression, a wide small fontanelle, constipation, thermoregulatory disorder, icterus, macroglossia, and umbilical hernia are present in infants. Early treatment is crucial to the patient's prognosis.

Lysosomal storage diseases

Craniofacial dysmorphia and hepatosplenomegaly are frequent co-occurrences. The diagnosis is established using a metabolic examination, especially of urine, and enzymatic examinations of cultured fibroblasts, possibly in some cases leukocytes of peripheral blood will suffice. Screening tests include chitotriosidase activity in peripheral blood, cytological examination of a bone marrow punctate, and imaging methods 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 can be the only symptom, especially a speech disorder), in later life type II (craniofacial dysmorphia, neuropathy) and VII (partial dysmorphia without corneal disorders) are typical, some types can be treated with bone marrow transplantation or enzyme replacement therapy, Neimanm-Pick disease (often, however, later onset of symptoms).

Mitochondrial diseases

Often an abnormal ratio of lactate to pyruvate, on an empty stomach, after exerci,se or after eating. Multiple systems are typically affected, difficulties can arise at any age. Mental regression is part of encephalopathy, epileptic seizures are common. Diseases are often caused by defects in mitochondrial DNA, the most profitable examination is a muscle biopsy.

The most common syndromes are MELAS (focal neurological deficits similar to cerebrovascular accidents, deafn,ess and myopathy), Leigh syndrome (breathing disorders, oculomotor disorders), Alpers syndrome, MERRF (also with myopathy), Kearns-Sayre disease (also oculomotor disorders).

Primary white matter disorders

The conditions are mainly manifested by spasticity, focal neurological findings and visual impairment.

Among the most common are - galactosemia (brain edema, vomiting, hepatomegaly), Canavan's disease (macrocephaly, optic atrophy), Alexander's disease (macrocephaly, seizures), Krabbe's disease (visual impairment, opisthotonus with hyporeflexia), adrenoleukodystrophy (hyperreflexia, epi. seizures), Pelizaeus-Merzbacher disease (spasticity, nystagmus, choreoathetosis, BAEP abnormalities), metachromatic leukodystrophy (peripheral neuropathy).

Peroxisomal disorders

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

Infectious diseases

1. **AIDS** – microcephaly, regression of psychomotor development or dementia, spasticity, less often ataxia, involuntary movements, and myoclonic seizures, another cause of deterioration can be opportunistic CNS infections.
2. **congenital syphilis** – in children under 2 years of age, Hutchinson's triad (deafness, interstitial keratitis, barrel-shaped incisors), in newborns condyloma lata, rashes and periostitis, possibly osteochondritis. ATB treatment has a good effect, an examination for HIV co-infection is necessary.
3. **subacute sclerosing panencephalitis.**

Untreated or drug-resistant epilepsy

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

Glycosylation disorders of glycoproteins (CDG)

A very numerous group of diseases including encephalopathies, with a very variable phenotype. The screening examination is the level of low-sialylated transferrin (invalid results in case of alcohol abuse).

Movement disorders - hypotonia in children

Main signs: The cause can be either central or peripheral. During the examination, lifting of the head is difficult, poor holding in a horizontal or vertical suspension, various reduced reflexes on the limbs, bulbar syndrome may also be present. Diseases with the earliest onset are manifested as congenital hypotonia, with later onset of the disease as regression of motor development. If the disease is acute or subacute, ventilation support may also be necessary (e.g. in spinal injuries, botulism, Guillain-Barré syndrome, myasthenia grav,is or in the context of congenital myopathy). Spinal muscular atrophy and myopathy require ventilatory support only when the disease lasts longer.

Hypotonia in infants

Cerebral hypotonia

Often associated with a delay in mental development, epileptic seizures are present, primitive reflexes persist, malformations of other organs are also present. Over time, hypotonia can turn into spasticity

1. **Non-progressive encephalopathy**
   - brain malformation – abnormal size and shape of the head, often accompanied by craniofacial dysmorphism.
   - cerebral palsy – perinatal asphyxia, infection, bleeding into the CNS, trauma.
2. **Chromosomal aberrations**
   - Prader-Willi syndrome - deletion of the maternal part of chromosome 15q11-13, regularly impaired sucking, also hypogonadism, psychomotor retardation, hyperphagia with obesity in later life, sleep disorders (sec. narcolepsy, sleep apnea).
   - Down syndrome (trisomy 21)
3. **Hereditary metabolic disorders**
   - Peroxisomal disease - Zellweger's sy. (arthrogryposis, cereberohepatorenal sy., epilepsy), neonatal form of adrenoleukodystrophy (epilepsy, hepatomegaly, retinal degeneration)
   - Lowe's syndrome (oculocerebrorenal system) – congenital cataract or glaucoma, renal function disorder.
   - Familial dysautonomia (Riley-Day) - sucking disorder, vegetative dysfunction, insensitivity to pain, repeated vomiting.
   - Maltase acid deficiencies - myopathy with cardiomyopathy
   - GM1 gangliosidosis – psychomotor retardation, visual impairment
4. **Benign idiopathic congenital hypotonia** - spontaneous resolution with further normal development.

Spinal diseases

1. **Perinatal and birth traumas** – ventilation disorder may be present when the medulla oblongata is affected or a high lesion of the cervical spinal cord is present. Diagnosing a defect in sensitivity can be difficult in such young children.
2. **Hypoxic-ischemic myelopathy in perinatal asphyxia**

Disorders of neuromuscular transmission

1. **Botulism in infants** - after home-canned vegetables or honey - constipation, proximal muscle weakness, without impaired consciousness, dysphagia, impaired photoreaction, weak crying. Possibility of respiratory failure, increase in EMG during repetitive stimulation, effective treatment with antitoxin.
2. **Myasthenia**
   - congenital – defect in the epsilon subunit Achr, sucking disorders and respiratory difficulties, ptosis, arthrogryposis, less often ophthalmoplegia, worsening during intercurrent infection.
   - neonatal myasthenia – in 10-20% of births of mothers with myasthenia gravis – sucking disorder, fatigue,arthrogryposisa, weak crying. The cause is the passive transfer of anti-Achr antibodies, spontaneous modification.

Polyneuropathy – Guillain-Barré syndrome

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

Muscle diseases

1. Congenital myopathy - arthrogryposis, contractures, scoliosis, ophthalmoplegia, respiratory and swallowing difficulties, hip dislocation. A muscle biopsy is diagnostic.
2. Congenital myotonic dystrophy – respiratory difficulties, hypomimia, sucking disorders, cardiomyopathy, arthrogryposis, mental retardation in later life, cataracts, 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 and phosphorylase deficiency (muscle spasms and exercise intolerance).

Spinal muscular atrophy (defect in the SMN gene)

1. **Acute (SMA I)** – onset before 6 months of age, proximal weakness, areflexia. Respiratory difficulties, without arthrogryposis, development of atrophy, fasciculations, often aspiration, rapid progression
2. **Chronic (SMA II)** – onset between 3-18 months of age, a normal newborn at birth, with faster or slower progression, diagnosis based on EMG findings and genetic examination.

Disorders of muscle strength in older children

Main symptoms: Gait disorder, walking on tiptoe, often other development can be normal, in addition to EMG, MRI examination of the LS area is also important. However, the most common cause is idiopathic shortening of the Achilles tendons.

- structural disorder of the lumbosacral plexus or spinal cord - often the neurological findings are abnormal, the cause can also be tethered spinal cord syndrome.
- muscular dystrophy – Often dystrophinopathy, then cardiomyopathy, variable intellectual disability.

- cerebral palsy
- autism and idiopathic shortening of the Achilles tendons.

Ataxia

Dizziness and vertigo in childreInfections

Infecions

- **bacterial mesotitis or labyrinthitis** - vomiting, nausea, hearing impairment. The result can be a cholesteatoma penetrating the petrous bone and possibly the labyrinth, which is typically manifested by vertigo when coughing, sneezing or pressure on the ear.
- **bacterial meningitis** - fever, impaired consciousness, meningeal irritation, the cause may be the spread of mastoiditis or other otogenic infections
- **vestibular neuritis** - isolated vertigo with a good prognosis, improvement begins within 48 hours, most often viral etiology.

Side effects of medicines and drugs – most antiepileptics, neuroleptics, and some antibiotics (may also cause hearing impairment).

Kinetosis – Disproportion between visual and vestibular inputs, most often in children sitting in the back of a vehicle near a small window.

Migraine – Vertigo can be the cause even without a headache, benign paroxysmal vertigo in children can be a variant of migraine.

Traumas – After a head injury, it often occurs within 3 days, coma of the vestibular system is manifested by long-lasting vertigo triggered by head movements. It is necessary to exclude fractures of the cranial base.

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

Other rare causes – Structural lesions of the brainstem, Mernier's disease and congenital defects of the inner ear.

Acute transient ataxia

Poisoning or an adverse effect of treatment is a very common cause of ataxia.

Autoimmune and parainfectious causes

1. Miller-Fischer sy. – further ophthalmoplegia of varying degrees, hyporeflexia, necessary monitoring of respiratory functions.
2. Acute perinfectious cerebellitis (rhombencephalitis) - very common in children.
3. Multiple sclerosis of the cerebrospinal cord.

Migraine – An intermittent variants are basilar migraine or benign paroxysmal vertigo.

Traumas – post-concussion syndrome persists for weeks, differential diagnosis intracranial hematoma or dissection to occlusion in the vertebrobasilar basin.

Intracranial expansion - acute onset of problems due to bleeding into the tumor, decompensation of hydrocephalus (often including headache, vomiting, nausea, impaired consciousness), paraneoplastic manifestations in neuroblastoma or medulloblastoma (spinal localization of the primary lesion is also possible) - typically opsoclonus (chaotic eye movements) and myoclonus .

Infections – typically rhombencephalitis.

Genetically linked and metabolic diseases:

- Episodic ataxia - disorder of ion channels, good effect of acetazolamide.
- Hartnup's disease (hereafter encephalopathy and rashes).
- Intermittent variant of leucinosis (hereafter encephalopathy).
- Deficiency of pyruvate dehydrogenase - further episodes of lactic acidosis, hypotonia, encephalopathy.

Vascular disease – bleeding into the cerebellum, rarely also ischemia in this area, more often Kawasaki disease (systemic vasculitis with cardiac symptoms, fever, swelling of the limbs and lymphadenopathy, it can also be aseptic meningitis).

Pseudoataxia in epilepsy and conversion and psychogenic disorders

Chronic ataxia

Cerebral palsy and other motor retardation (see above)

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

Intracranial expansion – In addition to 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-telagiectatic (also recurrent respiratory infections, high alpha-fetoprotein), Hartnup's disease, GM2 gangliosidosis (tremor, dysarthria), leucinosis, metachromatic leukodystrophy (epilepsy, dementia), Marinesco-Sjörgen syndrome (progressive ataxia, congenital cataract, mental retardation). Pyruvate dehydrogenase deficiency, progressive myoclonic epilepsy, Refsum's disease (polyneuropathy, retinitis pigmentosa, hearing impairment, cataract, cardiomyopathy, pes cavus).
3. **X-linked ataxia** - adrenoleukodystrophy (typical image in the white matter on MRI), Leber's optic atrophy (mitochondrial disease, visual impairment).

Headache in children

Acute headache

- **Febrile state** in extracranial infection
- **Post-traumatic condition, post-concussion syndrome** - usually persistent, then rather with a good prognosis, on the contrary, progressive, with signs of increased intracranial pressure, more indicative of intracranial lesions.
- **Sinusitis** - often with fever, pain in the paranasal sinuses, and chronic runny nose.
- **Aseptic meningitis** - with fever, meningeal irritation, more in older children.
- **Migraine** – episodes of throbbing pain, nausea, vomiting, visual impairment, photo- and phonophobia, typically positive family history, duration is shorter in children than in adults (up to 1 hour).
- **Tension headaches** - of a diffuse nature, feeling of pressure.
- **Post-seizure pain in patients with epilepsy**
- **Rare causes** - intracranial expansion, arterial hypertension, intoxication, intracranial hemorrhage, hydrocephalus, bacterial meningitis, acute metabolic disease (hypoglycemia, porphyric crisis), Cluster-headache (very rare in children), post-puncture syndrome, neuralgia (rare in children), vascular (phaeochromocytoma, carcinoid).

Chronic headache

- Migraine headaches,
- Tension headaches – mostly chronic, poorly localized pain of longer duration.
- Post-concussion syndrome – can last for weeks.
- Post-seizure pain - it can appear repeatedly, the patient has amnesia for his own seizures and those around him do not notice them.
- Intracranial hypertension - vascular cause, hydrocephalus, abscess, pseudotumor cerebri.
- Adverse effects of drugs, use of addictive substances, withdrawal syndromes;
- Vertebrogenic etiology is a frequent cause of headaches, especially in the occipital area, where there are palpable spasms, especially of the short extensors of the head, and disorders of the spine dynamics are also typically present. Treatment with gentle myorelaxants (Mg lactici) and rehabilitation;
- Sinusitis;
- Ocular cause (glaucoma, astigmatism), odontogenic cause, neuralgia, postpuncture syndrome, cluster-headache, metabolic (hypoglycemia, porphyria), hypoxia (sleep apnea, hypoventilation);
- Psychogenic and purposeful.