Cockayne Syndrome

This article has been translated from WikiSkripta; ready for the **editor's review**.

Template:Infobox - genetická choroba **Cockayne syndrome** (also know as **CS**, **Weber-Cockayne syndrome** or **Neill-Dingwall syndrome**[1]) is a rare, autosomal recessive, multisystem disease characterized by **dwarfism**, **retinitis pigmentosa**, **"bird's" face** and **photosensitivity**.[2]

Pathophysiology

Cockayne syndrome is an **autosomal recessive disease** associated with a **defect in DNA** repair (nucleotide excision). As a result of this defect, patients are **photosensitive** to the UV part of the spectrum (similar to xeroderma pigmentosum or trichothiodystrophy).[2] The disease is caused by a mutation in the **ERCC6** or **ERCC8** gene.[3]

Cockayne syndrome is sometimes considered a variant of Pelizaeus-Merzbacher disease for a similar finding of **islet demyelination** of the central and peripheral nervous system. However, the pathogenetic mechanism is different.[4]

Calcifications occur in the globus pallidus, cerebellum and small arteries. Furthermore, bizarre **diffuse astrocyte proliferation** and the **formation of neurofibrillary nodules** can occur (similar to Alzheimer's disease).[4]

Forms

- **Cockayne syndrome I (Classic Cockayne's syndrome)** - the first symptoms appear at the end of the first decade of life with characteristic changes on the face and body, this subtype manifests itself primarily in progressive neurodegeneration. Death occurs in the second or third decade of life.[2]
- **Cockayne syndrome II** - manifests itself in perinatally rapid changes in the face and body, the patient dies at 6-7 years[2]
- **Cockayne syndrome III** - milder variant with late onset, little defined[3]
- **Xeroderma pigmentosum-Cockayne syndrome (XP-CS)**[5]

Clinical picture

Patient habitus is typical: **microcephaly**, **large nose** and **large ears** (reminiscent of Mickey mouse ®)[2]

The skin is **photosensitive**, **erythema**, **hyperpigmentation**, **telangiectasia** and **atrophy** appear. Subcutaneous Atrophy is responsible for sunken eyes and an old **progeric appearance**.[2]

Musculoskeletal system is affected by **microcephaly**, **short stature**, **long limbs** with **joint contractures**, patients have **large arms and legs**, **kyphosis**, **thickened skull**, **sclerotic pineal glands** and **osteoporosis**.[2]

Intracranial calcification and **diffuse demyelination** are evident **neurologically**. Manifestations are **ataxia**, **tremor** and the **gear effect**. **Mental retardation** and **progressive deafness** may occur.[2] **Reflexes may be poorly equipped**, resulting in combined central and peripheral damage. **Normotensive hydrocephalus** occurs non-constantly.[4]

Ophthalmically "salt and pepper", **miosis**, **cataracts**, **optic atrophy**, **corneal opacity**, **nystagmus** and **blepharoconjunctivitis** may appear on the retinal pigment. [2] **Tearing disorder** may occur.[4]

Tooth decay may be present.[2]

Endocrinological findings present **hypogonadism** in 30% of men and an **irregular menstrual cycle** in women.[2]

Diagnostics

Cockayne syndrome jes diagnosable on the basis of **clinical findings**, ie postnatal growth disorders and progressive neurological dysfunction. Atypical cases may require **DNA testing** for the presence of mutations in the ERCC6 (75% of cases) and ERCC8 (25% of cases) genes.[3]

Cells in Cockayne syndrome show lower DNA and RNA synthesis after **irradiation with UV** light. Fetal amniotic fluid cells can already be examined in this way. [2]

CT can show calcifications and cortical atrophy.[2]

Differential diagnosis

- **Xeroderma pigmentosum** - Unlike XP, Cockayne syndrome is not **associated with skin tumors**
- **Trichothiodystrophy** - similar to Cockayn's syndrome there is no increased risk of skin tumor, **but there is a sulfur deficiency in the hair** (sulfate groups) and they are **fragile**. **Progressive deafness** does not develop as in Cockayne syndrome or XP
- **Bloom's syndrome**
- **Hartnup disease**
- **Rothmund-Thomson syndrome**
- **UV-sensitive syndrome**
- **Werner syndrome**[2]

Therapy

There is no **curative therapy** (even casual) today. The therapy is only **symptomatic**. [4]

Photoprotection in the form of protective creams and clothing serves as a supportive treatment. A **cochlear implant** can at least partially compensate for the hearing loss.[2]

Links

Related articles

- Pelizaeus-Merzbacher disease
- Leukodystrophy

External links

- cockaynesyndrome.net (http://cockaynesyndrome.net/LOKUZ/OdOTZ/SQONj/main/AboutCS.aspx,)

Reference

1.
2.
3.
4.
5.

Kategorie:Neurology Kategorie:Genetics Kategorie:Pediatrics