Premature ageing syndromes

Premature ageing syndromes are rare genetic diseases. The main manifestations of the disease are visible signs typical of the **ageing process,** which appear in early childhood. The patients are therefore mainly **children**, who often live to a maximum of 20 years. Their physical appearance often corresponds to the elderly around 80 years.

The time when the disease breaks out may vary, but it can be diagnosed in several months old children. The first signs of the disease most often appear between 18 and 24 months of age.

Symptoms

Early symptoms include general **failure to thrive**. Growth slows down to stop, **alopecia** occurs. Patients tend to have a fragile body constitution and characteristic small wrinkled faces. As the disease progresses, other organ complications appear - for example: the development of atherosclerosis, cataracts or cardiovascular diseases.

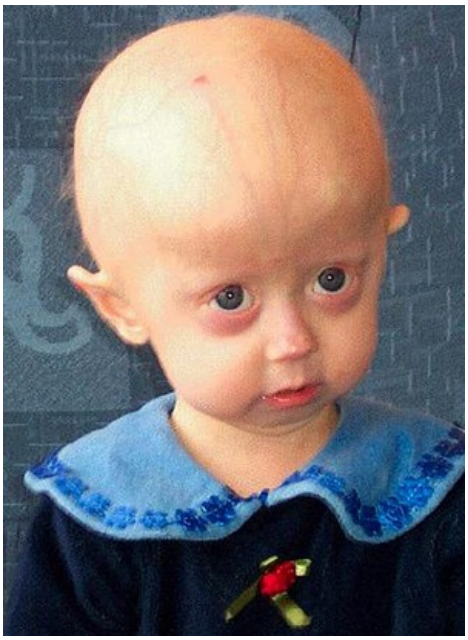
Treatment and prognosis

At present, no treatment is known and the approach to therapy is based only on the alleviation of disease manifestations (treatment of atherosclerosis, cardiovascular system, etc.). Patients often die between the ages of 13 and 15 from a myocardial infarction or stroke (due to atherosclerotic processes). In 2005-2006, studies were published on **farnesyltransferase inhibitors**, as a possible drug of the future for those affected by progeria. However, testing is currently only performed on animal models.

Examples of syndromes of premature ageing

Progeria (Hutchinson-Gilford syndrome)

Hutchinson-Gilfordův syndrome (OMIM: 176670 (http://omim.org/entry/176670)) is caused by a de novo mutation, replacing one pyrimidine nucleobase: cytosine with thymine. As a result of this mutation, the transcription of the **LMNA** gene (1q21.2; OMIM: 150330 (http://omim.org/entry/150330)) changes and a special form of **lamin A** - progerin is formed. Laminae are a type of intermediate filament that is involved in the structural and transcriptional functions of cell nuclei. The word progeria itself comes originally from Greek and means "prematurely old". The disease was first described in Great Britain in the late 19th century.



A child with Hutchinson-Gilford syndrome

Werner syndrome (adult progeria)

It is an AR inherited disease caused by a mutation in the **WRN** gene (8p12-p11.2; OMIM: 604611 (http://omim.org/entry/604611)), which encodes a DNA helicase. Due to this mutation, an **unstable genome** is likely to form. The influence of helicase on the production of inactive forms of telomerases is considered. Telomerase is a ribonucleoprotein complex that serves to protect the final parts of chromosomes (telomeres). These normally shorten during replication.

Manifestations often appear at puberty.

Cockayne syndrome

also Weber-Cockayne syndrome or Neill-Dingwall syndrome

AR hereditary disease, where premature aging is only one of many symptoms. Others include photosensitivity, disorders of the nervous system, sight or hearing. It probably arises from the principle of disruption of DNA repair processes.

More detailed information can be found on the page Cockayne syndrome

Xeroderma pigmentosum

More detailed information can be found on the page Xeroderma pigmentosum

Links

External links

- Progeria Research Foundation (https://www.progeriaresearch.org/)