JARMOŁOWICZ, Jakub, KRZESZOWSKA, Paulina, KUJAWA, Aleksandra, KROCHMAL, Karolina, SAMORAJ, Adrianna, ZAJĄCZKOWSKI, Jakub, MARRODÁN-WOJTCZAK, Karolina, WILK, Jan, STĘPNIAK, Kacper and KUŹMA, Radosław. Gaucher's disease - a review of the most important information about the disease in Paediatrics. Quality in Sport. 2025;43:61358. eISSN 2450-3118.
https://doi.org/10.12775/QS.2025.43.61358
https://apcz.umk.pl/QS/article/view/61358

The journal has been awarded 20 points in the parametric evaluation by the Ministry of Higher Education and Science of Poland. This is according to the Annex to the announcement of the Minister of Higher Education and Science dated 05.01.2024, No. 32553. The journal has a Unique Identifier: 201398. Scientific disciplines assigned: Economics and Finance (Field of Social Sciences); Management and Quality Sciences (Field of Social Sciences).
Punkty Ministerialne z 2019 - aktualny rok 20 punktów. Załącznik do komunikatu Ministra Szkolnictwa Wyższego i Nauki z dnia 05.01.2024 Lp. 32553. Posiada Unikatowy Identyfikator Czasopisma: 201398. Przypisane dyscypliny naukowe: Ekonomia i finanse (Dziedzina nauk społecznych); Nauki o zarządzaniu i jakości (Dziedzina nauk społecznych).
The authors declare that there is no conflict of interest regarding the publication of this paper.
Received: 25.05.2025. Revised: 08.07.2025. Accepted: 08.07.2025. Published: 12.07.2025.

Gaucher's disease - a review of the most important information about the disease in Paediatrics

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Abstract

Gaucher disease is an autosomal recessive spirochete disease caused by a deficiency of the enzyme glucocerebrosidase. A mutation in the GBA1 gene induces the accumulation of abnormal products in macrophages resulting in the transformation into Gaucher cells in many organs. There are three main types of the disease, 1 being the most common type (non-neuropathic). The 2nd and 3rd types of the disease are neuropathic types and these occur mainly in childhood. Among paediatric patients, Gaucher's disease manifests mainly with enlargement of the liver, spleen, thrombocytopenia, anaemia, but skeletal, ocular and central nervous system symptoms may be present. Diagnosis based on enzymatic and genetic tests - analysis of the GBA1 gene mutation - is essential to prolong patients' lives, improve their quality of life and select an appropriate form of treatment. Currently, care for patients with this disease is based on ERT (enzyme replacement therapy), SRT (substrate replacement therapy). The disease is characterised by rather non-specific symptoms and course, and can be interpreted by physicians as a haematological-proliferative disease due to frequent changes in the structure of the liver, spleen and changes in blood morphological parameters. Increased awareness among doctors of the symptoms of this disease would allow earlier detection and implementation of treatment. This paper presents the current diagnostic and therapeutic methods and the characteristic symptoms any paediatrician may encounter. The challenges of diagnosing this disease are also highlighted.

Keywords: Gaucher Disease, GBA1, Gaucher cells, ERT, SRT

Introduction

Gaucher disease is an inherited autosomal recessive lysosomal storage disease. Mutation in the GBA1 gene, located on chromosome 1q21, is responsible for the onset of the disease. [1][2] The GBA1 gene encodes the enzyme B-glucocerebrosidase (glucocerebroside) to glucose and ceramide.[3] Disruption of the enzyme leads to substrate accumulation in macrophages, which develop into the characteristic Gaucher cells.[4] The main site of these cells is the liver, spleen, bone marrow and in some cases in the central nervous system.[5]

The disease is characterised by considerable phenotypic heterogeneity, ranging from sparsely symptomatic forms that drag on for years to those manifesting with severe systemic symptoms.[6] The main classification of the disease distinguishes three types: Type I (non-neuropathic), which is the most common form, Type II, an acute neuropathic form that manifests a very rapid and severe course, and Type III, a chronic juvenile neuropathic form.[7]

Classification

Patients with GD are characterised by a wide range of clinical symptom levels, which depend on the degree of gene expression. The classification is based on three main grades.[8]

Type I is the non-neuropathic type, also known as the adult form. Symptoms in this group of patients can appear at different ages, from infancy to adulthood. [9] The patient may be fully asymptomatic for many years of life. The main symptoms that appear in patients include hepatomegaly, splenomegaly, osteolytic changes in the bones and haematological symptoms (thrombocytopenia, anaemia). In children, developmental - physical disorders and delayed puberty may occur.[10] The course of the disease in children is characterised by a much more severe and rapid course in contrast to adults. Noor Ul Ain et al. presents a case of a child with

type I gaucher disease, who started to develop symptoms such as nose bleeds, easy bruising and splenomegaly at the age of 2 years. Further diagnosis based on liver biopsy revealed the presence of Gaucher cells.[11][12]

Bone complications in Gaucher disease represent a significant impact in the course of the disease. They are a source of pain, disability and reduced quality of life for patients. Complications include osteoporosis, fractures and bone necrosis - the most common in type I Gaucher disease.[13]

Acute neuropathic type II affects young children. It affects the smallest children from infancy. The course of the disease is sudden and progressive, involving brainstem involvement (spasticity increasing in progression), hepatosplenomegaly and cachexia.[14]

Type III chronic neuropathic, represents an intermediate stage between type II and III.[12] The course of this type of Gaucher disease is associated with the development of mental retardation, myoclonus, convulsions and hepatosplenomegaly. Parents in the first years of a child's life may already observe symptoms that are suggestive of the disease. Di Costanzo et al. showed an example of such symptoms is the case described a case of a child whose parents noticed sudden ‘jerks’ in the child's head. Further neurological symptoms progressed as the child grew older.[15]

Typ I	Typ II	Typ III
- Asthenia -stunted growth - splenomegaly(90% cases) - hepatomegaly (80% cases) - osteopenia - pancytopenia	- infants 3-6 months - hepatosplenomegaly - severe neurological disorders (oculomotor paralysis, bilateral permanent strabismus with bulbar symptoms) - cachexy	- non-specific symptoms (type I and II symptoms occuring at different stages) - ophtalmoplegia -delayed puberty - hepatosplenomegalya - osteopenia

Tab. 1. Gaucher Disease classification

Genetic

Gaucher disease is characterised by a variety of phenotypic forms. Approximately >250 mutations in the GBA1 gene encoding glucocerebrosidase are responsible for their development and have been identified to date in this condition. However, there are far more factors involved in the pathogenesis of Gauscher's disease than the mutation of this gene alone. Oxidative stress, immune response, mitochondrial dysfunction, inhibition of macroautophagy, calcium dysregularion, accumulation of a-synculin in mitochondria, among others, are responsible for its development.[16][17][18]

Genetic testing on the family planning side plays a fairly important role in specialist counselling. Gaucher disease is inherited autosomal recessively. In this situation, the parents of the affected child are both heterozygotes, both having passed on a recessive gene that has phenotypically been shown to cause the disease. Genetic counselling of the child's parents allows the parents to help determine, among other things, the risk of the same phenotype and another child in the future.[17]

Diagnostic

The diagnostic process of Gauscher's disease is based on diagnosis algorithms that take into account different age groups.

Pediatric patients - the algorithm for the diagnostic procedure in this group of patients highlights the very important role of the general paediatrician, who is at the forefront of recognising the first symptoms of the disease. The first symptoms that should draw the attention of the doctor to whom a child under two years of age is referred include hepatomegaly and/or splenomegaly. Along with organ enlargement, there may be changes in the blood count: thrombocytopaenia or anaemia.[12][19][20][21] Alarm signals should be the indication of choice for referring the patient to a paediatric haematologist/ paediatric gastroenterologist. An organ biopsy (liver, spleen) is helpful if Gaucher cells are found in the histopathological material.

The recommended diagnostic elements for children and newborns with Gaucher disease type II and III are divided into different systems: gastrointestinal symptoms, development (motor, adaptive, cognitive, speech and language assessment), neurological (assessment of the presence of epileptic seizures, convulsions, visual involvement), musculoskeletal system (X-ray, MRI, DXA).[17][22][23]

Biochemical indicators in the blood: chitotriosidine, angiotensin-converting enzyme, ferritin, tartrate-resistant acid phosphatase.[24]

All the previously mentioned tests are helpful both at an early diagnostic stage and at later stages when it is necessary to verify the effectiveness of treatment.

Genetic testing for GBA1 mutations is helpful in the diagnosis of Gaucher disease. However, it must be borne in mind that the location of the mutation depends on the ethnicity and the region of the world the patient comes from.

Adrade-Campaos et al. Performed targeted screening for GD. He included patients after finding symptoms such as splenomegaly and thrombocytopenia. The test consisted of a high-throughput DBS enzyme test. This method proved to be very effective for making a correct diagnosis in a short time.[25]

DBA is a screening method to measure GBA activity from a dried drop of blood. However, the author points out that a genetic test to detect mutations in the GBA1 gene is needed to make a definitive diagnosis.

Bone complications

Bone complications in Gaucher disease are a significant impact in the course of the disease. They are a source of pain, disability and reduced quality of life for patients. Complications include osteoporosis, fractures and bone necrosis - the most common in type I Gaucher disease. Osteoclast activity in patients with Gaucher disease is significantly increased as a result of glucocerebrosidase dysfunction. Patients are characterised by a significantly higher incidence of reduced bone density compared to the healthy population.[4][12]

Therapeutic management

In the treatment of Gaucher disease, the greatest therapeutic success is achieved with enzyme replacement therapy (ERT).[26] The therapy consists of intravenous supply of recombinant glucocerebrosidase (e.g. imiglucerase, velaglucerase alfa, taliglucerase alfa). For some type I patients, substrate reduction therapy (SRT) is used to increase glucosylceramide production (miglstat, eliglustat).[27] Cases without neurological symptoms are treated with ERT or SRT with good results. However, these therapeutic modalities do not cross the blood-brain barrier and are therefore not as effective in relieving neurological symptoms.[26][28]

As with many rare diseases, the decision to undertake treatment as well as the choice of treatment regimen should be made on an individual basis. Failure to treat the disease leads to liver, spleen, bone marrow failure, portal hypertension, and some haematological diseases (multiple myeloma).

In addition to the commonly accepted therapies of ERT and SRT, symptomatic treatment is the cornerstone of management.[29] Treatment of complications of the disease, such as anaemia, fractures or pain management.

Therapy	Mechanism	Medicines	Indications
ERT	Administration of recombinant ß-glucocerebrosidase enzyme	Imigluceraza, taligluceraza alfa, velagluceraza alfa	Type I, rarely type III
SRT	Inhibiton of glucosyloceramide synthesis	Miglustat, eliglustat	Mild, local forms in case of contraindications to ERT
Symptomatic Treatment	Anemia, fractures, pain	Przetaczanie elementów morfotycznych krwi, analgezja	All types

Tab. 2. Gaucher Disease Therapy

Monitoring

The process of monitoring patients after the end of treatment according to ERT/SRT therapy should involve two types of regular examinations. The first group of regular examinations every 6-12 months: blood count, abdominal ultrasound (to check the size of the liver, spleen), MRI of the skeleton (to look for osteolytic changes in the bones), levels of disease markers (chitotriosidase or CCL18, glucosylsphingosine).[17][30][31][32]

Second group of examinations every 12-24 months: neurological examinations, bone densitometry (DXA- assessment of bone mineral density- determination of the presence of osteoporosis), assessment of lung function (in case of organ involvement).[27]

Discussion

Gaucher disease is a systemic disease giving symptoms from different systems. The first symptoms that may raise suspicion of this disease in a patient are unexplained hepatomegaly

and/or splenomegaly. It is important for the physician to be vigilant when these symptoms are present and to exclude a haematological aetiology for the existence of the symptoms in question. The course of the disease in symptomatic children can be very dramatic, so it is important to take decisive diagnostic action: ordering appropriate tests (chitotriosidase or CCL18, glucosylsphingolysin) or referring the patient to a reference centre specialising in the treatment of paediatric patients with Gaucher disease. In the literature, there is a perceived problem of practitioners' lack of knowledge regarding patients' knowledge of the manifestation of the disease.

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All authors have read and agreed with the published version of the manuscript.

FUNDING STATEMENT

This research received no external funding.

ISTITUTIONAL REVIEW BOARD STATEMENT

Not applicable.

INFORMED CONSENT STATEMENT

Not applicable.

DATA AVAILABILITY STATEMENT

Not applicable.

ACKNOWLEDGEMENTS

Not applicable.

CONFLICTS OF INTERESTS

The author declares no conflict of interest.

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