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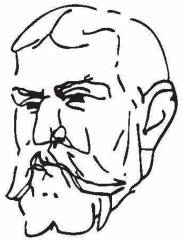

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CASE STUDY

GLYCOGEN STORAGE DISEASE TYPE II: A NARRATIVE LITERATURE REVIEW AND A CASE REPORT OF LATE-ONSET POMPE DISEASE IN A YOUNG WHITE CHILD

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ABSTRACT

At all ages, skeletal muscle weakness characterizes Pompe disease, causes mobility problems and affects the respiratory system. We aimed to provide a narrative review of terminology, etiology, epidemiology, clinical manifestations, complications, and prognosis of Pompe disease, supported with a clinical case presentation.

The clinical manifestation and complications of Pompe disease are illustrated with the clinical case presentation of a late-onset form in a white child. A comprehensive electronic literature search was performed on Ovid, Google Scholar, Scopus, PubMed, Embase, Cochrane Database, and World Health Organization databases to identify the articles that discussed Pompe disease.

KEY WORDS: Glycogen storage disease, Pompe disease, review, clinical case, late-onset

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INTRODUCTION

Glycogen storage disease (GSD) is a metabolic disorder caused by enzyme deficiencies affecting either glycogen synthesis, glycogen breakdown, or glycolysis (glucose breakdown), typically in muscles and/or liver cells. At least 15 types of GSD have been identified, all resulting in abnormal glycogen metabolism and an accumulation of glycogen in these cells. [1, 2]

The main types of GSD are categorized by number and name, as follows:

- Type I (Von Gierke disease; the most common type that accounts for 90% of all glycogen storage disease cases);
- Type II (Pompe's disease, acid maltase deficiency);
- Type III (Cori's disease);
- Type IV (Andersen's disease);
- Type V (McArdle's disease);
- Type VI (Hers' disease);
- Type VII (Tarui's disease);
- Type VIII disease.

Periodic acid-Schiff stain identifies glycogen and is useful in identifying these diseases.

GSD type II is an autosomal recessive metabolic disorder, caused by an accumulation of glycogen in the lysosome due to deficiency of the lysosomal acid alpha-glucosidase enzyme (GAA). [3] It is also called Pompe disease or an acid maltase deficiency and is a rare multi-system hereditary storage disease that is progressive, and often fatal. [4] Nowadays, more than 350 gene mutations causing this disease have been identified and their number is constantly growing. [5]

The exact prevalence of Pompe disease is unknown. According to various authors, the incidence of the disease, depending on the country and ethnicity, ranges from 1:40,000 to 1:300,000. A 'founder effect' cannot be excluded. [6]

THE AIM

The purpose of this study is to provide a comprehensive narrative review of terminology, etiology, epidemiology, clinical manifestations, complications, and prognosis of Pompe disease, illustrated by a clinical case presentation of late-onset Pompe disease in a white child.

MATERIALS AND METHODS

A complex clinical, neurological, laboratory, and instrumental analysis provided to a child, admitted to the Regional Clinical Center of Neurosurgery and Neurology, Uzhorod city, Ukraine, resulted in diagnosis of GSD type II.

A comprehensive electronic literature search on Ovid, PubMed, Scopus, Embase, Cochrane, Google Scholar, and World Health Organization databases was performed to identify articles that discussed the neurological manifestations, presentations, complications, and prognosis of Pompe disease. The applicable articles are cited and referenced. No limit placed on publication time or the language of the article. All the relevant articles were identified and screened by two authors (HP and OF), and disagreements were resolved by consensus and involvement of senior authors (MO, YH, OS, YF); the results are summarized narratively.

CLINICAL CASE

We provided a complex clinical and instrumental analysis of manifestations and complications of Pompe disease in a white child, admitted to the Regional Clinical Center of Neurosurgery and Neurology, Uzhorod city, Ukraine. Clinical case presentation is accompanied with a comprehensive narrative review of terminology, etiology, epidemiology, clinical manifestations, complications, and prognosis of Pompe disease, presented in the discussion.

We report a case of a late-onset form of Pompe disease in a child resident of the Transcarpathian region.

Ten-year-old boy brought to his primary care physician by his parents because of difficulties in walking and climbing stairs, rapid fatigue, and shin pain after exercise during the last year. Examination revealed a positive Hoover›s test. Routine hematological tests were within normal limits. Total creatine kinase level was elevated to 1381 units. On the EMNG there were signs of pronounced diffuse myopathic syndrome; more severe in the proximal muscles.

TESTS

Taking into account the patient's complaints, a genetic scan for Duchenne muscular dystrophy was included in the diagnostic search plan but no deletions of the dystrophin gene exons were detected.

Pompe disease diagnostic was conducted. In the dried spots, low activity of GAA was detected, in connection with which molecular genetic diagnostics of GAA gene was assigned. A mutation was detected in the heterozygous state p.32-13T>G and c.307T>G. Signs of the vital organ involvement that is usual for Pompe disease were absent, including cardiomegaly or cardiomyopathy according to ECG and echocardiography.

GENEALOGICAL ANAMNESIS

A dry blood spot testing was performed for the father, mother, and sister. GAA activity was within normal range.

DIAGNOSIS AND TREATMENT

Thus, the patient was diagnosed with the metabolic disorder from the group of lysosomal accumulation diseases: Pompe disease (E 74.0).

The patient was referred to the Orphan Disease Center for the purpose of life-long enzyme replacement therapy (Miozim at a dose of 20 mg/kg intravenously once every 2 weeks).

Glycogen storage disease type II (GSD II) is a classical lysosomal storage disorder, characterized by lysosomal accumulation of glycogen and tissue damage, primarily in muscle and heart. [7] It has a broad continuous clinical spectrum in terms of onset, the involvement of organs, and life expectancy. In addition to muscle and heart involvement, other tissues affected are liver, spleen, endothelium, lung, brain, anterior horns, and peripheral nerves. The underlying enzyme deficiency is acid maltase (also known as GAA).

DISCUSSION

DEFINITION

GSD II is an autosomal-recessive disorder that results from the deficiency of GAA, a lysosomal hydrolase, and is part of a group of metabolic diseases called lysosomal storage disorders [3].

The disease was first described by the Danish pathologist Joannes Cassianus Pompe in 1932 when he was presented with a 7-month-old girl who died after developing idiopathic hypertrophic cardiomyopathy. Pompe observed an abnormal accumulation of glycogen in all examined postmortem tissues. He described the cardinal pathologic features of this lysosomal storage disorder.

ETIOLOGY

Pompe disease is caused by mutations in a gene that produces an enzyme called GAA. Absence or deficiency of GAA, a lysosomal enzyme that is responsible for the cleavage of the α-1,4- and α-1,6-glycosidic bonds of glycogen to glucose, leads to the accumulation of glycogen in the lysosomes in numerous tissues, but clinical symptoms are primarily due to cardiac and skeletal muscles involvement. [4]

CLASSIFICATION

Classification of GSD II is based on the age of onset, organ involvement, severity, and the rate of disease progression. There are three forms of GSD II.

- *Classic infantile* form refers to the form of Pompe disease that was first described in 1932 and characterized by the onset of symptoms shortly after birth: generalized muscle weakness, and cardiomegaly in combination with excessive glycogen storage in virtually all organs. [1]
- *Non-Classic infantile* form or so-called 'childhood', and 'juvenile' forms of Pompe disease are introduced as the names for the less severe forms of Pompe disease, characterized by delayed onset and usually slower progression.
- *Adult-Onset* or so-called 'late-onset' Pompe disease differs from infant form with milder clinical manifestations and course, absence of multiple organ pathology (heart damage is extremely rare), and more recent respiratory complications due to the weakness of the diaphragm and intercostal muscles.

EPIDEMIOLOGY

According to various authors, depending on the country and ethnicity, the incidence of GSD II is generally placed at approximately 5000 to 10000 births worldwide. [6] It occurs in various populations and ethnic groups around the world.

Approximately a third of GSD II patients are infants. Occurrence in the Netherlands is one in 138,000 infants. In China, Taiwan, and among African-Americans occurrence is one in 14,000. [6-8]

Table 1. Clinical Presentations of Glycogen storage disease type II

Types of Pompe Disease	Onset	Findings	Prognosis
Classic Infantile Onset	First three months after birth	Failure to thrive Lung infections Feeding problems Hearing problems Heart defects Hypertrophic cardiomyopathy Skeletal muscles weaknes Diaphragm and other breathing muscles weakness Enlarged liver Large tongue	Fatal within the first year of life
Non-Classic Infantile Onset	Later than the classic form but still appears within the child's first year of life	Failure to thrive Myopathy Lung Infections Feeding problems Hearing problems Abnormally enlarged heart Progressive muscle weakness Delayed development of motor skills	Poor and is often Fatal
Late-Onset	Any age	Myopathy Progressive diaphragm weakness Mobility problems High chance of falls Breathing problems Shortness of breath Frequent lung infections Morning headaches Tiredness Weight loss Difficulty swallowing Difficulty hearing Scoliosis, or a curved spine.	Poor and is often Fatal

The exact incidence of late-onset Pompe disease worldwide is unknown. A study in the Netherlands estimates that one in 57,000 adults has late-onset Pompe disease. [8] In Ukraine seven patients (1 adult, 6 children) were diagnosed with Pompe disease.

CLINICAL PRESENTATION

GSD II has a broad clinical spectrum. First symptoms can occur at any age from birth to late adulthood. Earlier onset compared to later onset is usually associated with faster progression and greater disease severity (Tabl. 1). At all ages, skeletal muscle weakness causes mobility problems and affects the respiratory system.

In table one we have narratively summarized the main clinical representations and outcomes of different forms of Pompe disease.

CLASSIC INFANTILE ONSET POMPE DISEASE

The infantile-onset form of the disease is the result of complete or near-complete deficiency of GAA. It is caused by mutations that lead to the production of less than 2 percent of functional GAA. [9, 10]

Symptoms begin in the first months of life and are characterized by a severe progressive course and rapid development of multiple organ pathology. Early symptoms include difficulty gaining weight and failure to grow at a normal rate, known as "failure to thrive." These infants experience feeding problems, poor weight gain, muscle weakness, breathing problems, lung infections, floppiness, and head lag. Many infants with Pompe disease also have enlarged liver and tongue.

Symptoms of malnutrition are due to the weakness of the facial muscles. Respiratory difficulties are due to the weakness of the diaphragm. Intercostal muscles are often complicated by lung infections. The heart is grossly enlarged due to hypertrophic cardiomyopathy that results in heart failure.

The condition is often fatal within the first year of life, but rapid treatment can reduce the risk of heart failure. Without timely treatment, most babies die from cardiac or respiratory complications before their first birthday. [11]

NON-CLASSIC INFANTILE-ONSET POMPE DISEASE

Non-classic infantile-onset Pompe disease begins later than the classic form but still appears within the child's first year. Despite the abnormally enlarged heart, the chance of

heart failure is lower compared to classic-infantile-onset form. Progressive muscle weakness leads to the delayed development of motor skills such as rolling over and sitting. Infants with this type of Pompe disease often experience severe respiratory problems due to damage and weakness in the muscles involved in breathing. [12]

The condition has a poor prognosis and is often fatal.

LATE-ONSET POMPE DISEASE

Late-onset Pompe disease is the result of a partial deficiency of GAA. The onset can be as early as the first decade of childhood or as late as the sixth decade of adulthood. [13, 14]

Late-onset Pompe disease is often milder and progresses more slowly than the infantile forms. In general, the later the disease appears, the slower the symptoms progress. It differs from infant form with milder clinical manifestations and course, absence of multiple organ pathology, and more recent respiratory complications due to the weakness of the diaphragm and intercostal muscles. [15] The primary symptom is muscle weakness progressing to respiratory weakness and death from respiratory failure after a course lasting several years.

Heart involvement is reduced in most cases of late-onset Pompe disease, but some patients may experience an irregular heartbeat or an enlarged heart. [16] However, as the disease progresses, breathing problems may increase and the most common cause of death is lung failure. Increased muscle weakness will often result in patients having to use mobility assistance, such as wheelchairs.

Late-onset Pompe disease patients usually die from respiratory failure and infectious pulmonary complications, depending on the time of onset and subsequent course of the disease. It may occur in childhood, adolescence, adulthood, or old age.

DIAGNOSTIC METHODS

A diagnosis of Pompe disease can be confirmed by screening for the common genetic mutations or measuring the level of GAA enzyme activity in a blood sample. [1, 17] Once Pompe disease is diagnosed, testing of all family members and a consultation with a professional geneticist are recommended. Carriers are most reliably identified via genetic mutation analysis.

CONCLUSIONS

Along with other lysosomal diseases, in the case of Pompe disease, it is possible to carry out pathogenetic enzyme replacement therapy which allows us to modify the course of the disease significantly, improve the quality of patients' lives and prevent the development of critical complications. The key to the successful use of enzyme replacement therapy is the early diagnosis of GSD II. A careful study of the target categories of patients suffering from myodystrophy and other myopathic syndromes at any age is required to identify late-onset Pompe disease so that pathogenetic treatment is started as early as possible.

We presented a comprehensive narrative review of the terminology, etiology, epidemiology, clinical manifestations, complications, and prognosis of Pompe disease, accompanied by a clinical case report of late-onset Pompe disease, to raise awareness about the GSD II.

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