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Current pharmacotherapy and diagnostic methods of Pompe Disease in Poland

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Abstract: Pompe disease is estimated to happen in 1 out of 40 000 borns. It is rare metabolic disease connected to autosomal recessive genetic mutation. Disease is characterised by deficit of α-glucosidase (GAA) which is lysosomal glycogen hydrolizing enzyme acid. Decreased activity of enzyme leads to glycogen storage in lysosomes which appears as disfunction of tissues, especially cardiac muscle and skeletal muscles.

In Poland, we have two common and very simple methods to diagnose Pompe disease. First test is dried blood spot (DBS), second is full peripheral blood test.

Currently, in Poland, drug containing alglucosidase alpha is refunded. Drug is available as powder for infusion. Periodic assessment of therapy effectiveness is performed at least every 6 months based on rating the patient's clinical condition and assessment of the effectiveness of the therapy used.

Keywords: Pompe disease, alglucosidade alpha, chromosome 17, glycogen storage disease type ii

Introduction:

Pompe disease (glicogenosis type II) is rare metabolic disfunction caused by autosomal recessive genetic mutation. It is estimated to happen in 1 out of 40 000 borns. Gene is localised on chromosome 17. It has been identified about 150 different mutations of this gene, which appears as different clinical symptoms of the same condition. Disease is characterised by deficit of α-glucosidase (GAA) which is lysosomal glycogen hydrolizing enzyme acid. Decreased activity of enzyme leads to glycogen storage in lysosomes which appears as disfunction of tissues, especially cardiac muscle and skeletal muscles. Currently, there is no fully effective therapy for Pompe disease. Enzymatic therapy can delay appearance of clinical symptoms [1].

Material and method:

We analysed most current research and informations about Pompe disease found in Google Scholar and on Science Direct website.

Types of Pompe disease and main symptoms:

There are two types of this disease. Infantile – onset Pompe disease, which is diagnosed before one year old, and late – onset Pompe disease, after one year of age [2]. It is estimated, that in Poland there are 15 patients with late-onset type of the sickness, but there is no data about numer of infantile-onset patients [1].

The main symptoms of infantile-onset Pompe disease are cardiomegaly with progressive cardiomyopathy, respiratory infections, apnea, muscle laxity, feeding difficulties and impaired physical development. Most of the patients do not live to the age of one.

The late-onset Pompe disease has a slower course. Symptoms can occur in any age, even after the age of 50. A characteristic symptom is the weakening of proximal muscles of the lower limbs, back and diaphragm. Patients have difficulties in maintaining proper body posture and as the disease progresses, they need help with moving. They may also need mechanical ventilation when sickness develops. We don’t observe cardiomegaly in this type of the disease. Some of the patients may have deformations of spine, such as scoliosis or lordosis, frequent respiratory infections, headaches and dizziness and reduction of muscle mass.

Table 1. Pompe Disease: glycogen accumulation and consequences

Glycogen accumulation in:	**Consequences:**
proximal skeletal muscle	limb-girdle myopathy
intercostal muscles	respiratory failure
smooth muscle	abdominal pain/nausea/vomiting/diarrhea/urinary incontinence
cerebral vasculature	cerebral aneurysm
genioglossus	tongue weakness
extraocular muscles	unilateral or bilateral ptosis

An important prognostic factor is genotype. Knowing it, we can predict how the disease may progress and what to expect when it comes to therapy.

Diagnostic methods:

We can diagnose the disease based on deficit of GAA enzyme activity in muscle, fibroblasts, or dried blood spots. Most effective is muscle biopsy, which has typical picture of the disease. It is also possible to diagnose GAA mutations [3].

In Poland, we have two common and very simple methods to diagnose Pompe disease. Because of blood analysis, we can detect GAA deficiency. First test is dried blood spot (DBS), we can collect and protect blood and send it to laboratory for analysis. If the DBS test is positive (reduced level of GAA or no GAA), we still have to perform independent test to confirm existence of the disease. Second test is full peripheral blood test – an alternative analysis of level of GAA in full blood, it can be only determined in selected laboratories [4].

Pharmacotherapy:

Currently, in Poland, drug containing alglucosidase alpha is refunded. Pharmaceutic is made with recombinant DNA, it is a form of human alpha glucosidase. Drug is available as powder for infusion. The most common side effect of alglucosidase alpha is anaphylactic shock, so it should be administered by an experienced physician specializing in the treatment of rare metabolic diseases. Periodic assessment of therapy effectiveness is performed at least every 6

months based on rating the patient's clinical condition and assessment of the effectiveness of the therapy used [1].

Summary:

We still need much more research connected to this disease. Especially when it comes to pharmacotherapy. Disease is very rare so people working in the hospital should be educated and prepared for this kind of cases, so they can take proper care of their patients.

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