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Review Article

Oral Health and Dental Management of Children with Glycogen Storage Diseases - An Overview

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Abstract

Glycogen Storage Disorders (GSD) exist in a variety of forms with the common denominator resulting in a deficiency of one or more of the enzymes involved with the glycogen metabolic pathways GSDs, though rare, are generally diagnosed early in a patient's life. All GSDs involve the liver and cause hepatomegaly, except for types V and VII, which involve muscle and/or red blood cells except for types V and VII, which involve muscle and/or red blood cells. Clinical features are Low blood sugar, enlarged liver, growth retardation and Muscle cramps. This article discusses about etiological factors, clinical features along with dental management of children with glycogen Storage disease.

Keywords: Children; Dental management; Glycogen storage; Oral health

Inroduction

Glycogen Storage is a group of defects of glycogen metabolism which are genetically determined but which are not common. The overall prevalence of GSD has been reported to account for 1:20.000 to 1:25.000 live births [1]. Glycogen is a polysaccharide made up of glucose units for 'storage' purposes in the liver or muscles. The latter can only take up glucose, store it as glycogen and break it down into pyruvic and lactic acids, or to carbon dioxide and water. The liver, in addition to this, can synthesize glycogen from the circulation, store it and release it as glucose. A large number of enzymes are necessary in all stages of these changes and sometimes deficiencies occur which interfere with the balance of the process [2,3]. Fascinatingly, there is great phenotypic variance and versatile clinical courses even when a specific enzyme is castrated by mutation (Table 2) [5-9].

The symptoms of Glycogen storage disease in children are based on the its type, which are depicted in Table 1 [4]. Type I GSD, also known as Von Gz'erke's disease is the commonest of these defects to occur and is due to a deficiency of enzyme activity in the liver and kidney with decreased release of glycogen. This leads to an accumulation in 'store' and hypoglycaemia especially in the morning after night fasting when the liver and kidneys are enlarged because of the accumulated glycogen. Such patients tend to be of short stature and a number of them have a tendency to serious bleeding with thrombocytosis. This latter suggests platelets of slower than normal rate of destruction which are therefore poor at initiating haemostasis. An estimated incidence reported to vary from 1 :200,0002 to 1:400,000 [10,11].

Treatment is with regular meals of high protein content and a limited amount of carbohydrates. Prolonged fasting must be avoided because of the hypoglycaemia. While a number of these patients die in childhood, many survive and require regular supervision. Other defects of glycogen metabolism are even less common. In Pompe's disease, cardiac muscle as well as the liver and skeletal muscle is involved in the storage of glycogen, but the patient does not survive infancy. Forbe's disease affects liver and skeletal muscle, producing muscular weakness. Other manifestations are similar to those of von Gierke's disease, though milder. In Her's disease, where there is a deficiency of liver phosphorylase, the manifestations are also similar to those of von Gierke's disease but milder [12-17].

Clinical Features

Hyperlipidemia may also occur because of the altered glucose metabolism resulting in formation of xanthomatous deposits. These deposits are present in 10% to 20% of the affected individuals [18]. Gout and gouty nephropathy can develop from possible hyperuricemia. Clinically, patients with GSD I are characterized by a cherubic or "doll-like" face, shoulder adiposity, protuberant abdomen, thin skin, prominent veins, and growth retardation. [19].Varying degrees of mental retardation may be present although neurologic examinations are typically unremarkable [20].

Characteristic oral lesions of GSD Ib are oral ulcers, gingivitis and periodontal disease, that may be related to the limited intake of glucose into polymorphonuclear leucocytes. [21-23]. Frequent epistaxis, ecchymoses and prolonged bleeding can be complications of minimal trauma or after dental procedures, as a result of the acquired reduced platelet adhesiveness and aggregation [21,24]. The published data on dental manifestations associated with GSD Ib, such as dental caries, delayed dental maturation and eruption, and taurodontism are limited to case series or isolated case reports [26-28].

The distance between the central fossae of maxillary right and left first primary molars has to match the distance between the distobuccal cusps of mandibular right and left primary first molars in primary dentition particularly in Type III

There are no special dental features associated with these diseases. Vitamin supplements and calcium also should be included in the diet to ensure normal skeletal growth and to prevent long-term complications, such as osteopoenia [29].

Dental Treatment

Routine care is important because an acute infection can be

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Table 1: The symptoms of Glycogen storage disease in children are based on its type, which are depicted in [4].

TYPES	NAMES	SYMPTOMS
Type -I	Von Gierke Disease	Enlarged liver and kidneys, Low blood sugar, High levels of lactate, fats, and uric acid in the blood, Impaired growth and delayed puberty, Bone thinning from osteoporosis, Increased mouth ulcers and infection
Type -II	Pompe' Disease	• Enlarged liver and heart, In severe cases, muscle weakness and heart problems develop. • In severe cases, infants may suffer heart failure by the age of 18 months • Milder forms of type II may not cause heart problems
Type -III	Cori 'Disease	• Swollen abdomen due to an enlarged liver,Growth delay during childhood, Low blood sugar, Elevated fat levels in blood and • Possible muscle weakness
Type V	McArdle's Disease	• Muscle cramps during exercise, Extreme fatigue after exercise a • Burgundy-colored urine after exercise
Types VI, IX	Hers' Disease	• Liver enlargement occurs, but diminishes with age • Low blood sugar
Type VII	Tarui's Disease	• Muscle cramps with exercise • Anemia
Type VIII		• Muscle weakness, Anemia • Increased levels of uric acid

Table 2: Fascinatingly, there is great phenotypic variance and versatile clinical courses even when a specific enzyme is castrated by mutation [5-9].

Types of Storage disease	Enzyme deficiency	Inheritance
GSD 0 (Aglycogenosis)	Glycogen synthase (12p12.2)	AR
GSD I (von Gierke disease) GSD 1a	Glucose-phosphatasetranslocase/transporter (17q21)	AR
GSD 1b	Glucose-6-phosphatase translocase/transporter(11q23)	AR
GSD 1c	Phosphatasetranslocase/transporter(11q23-24.2)	AR
GSD II (Pompe disease)	Alpha-1-4-glucosidase (acid maltase) (17q25.2-25.3)	AR
GSD III (Cori disease, Forbes disease, limit dextrinosis, debranching enzyme disease)	Amylo-1-6-glucosidas(1p21)	AR
GSD III a and b	Phosphorylase limit dextrin accumulation	AR
GSD III c	Glucosidase Activity Loss	AR
GSD III d	Transferase activity loss	
GSD IV (Andersen disease,glycogen phosphorylase deficiency,brancher deficiency, amylopectinosis, glycogen branching enzyme deficiency)	Amylo-1,4 to 1,6-transgluosidase (3p12)	AR
GSD V (McArdle disease) • Rapidly fatal neonatal form • Mild form with congenital myopathy form • Benign classic with myalgia and dark urine form	Glycogen phosphorylase (11q13) (myophosphorylase	AR
GSD VI (Hers disease)	Liver glycogen phosphorylase E (14q21-q22)	AR
GSD VII (Tarui disease) • Muscle isoform (12q12) • Liver isoform (21q23) • Platelet/fibroblast isoform (10p15.2- p15.3)	Phosphofructokinase enzyme (12q13)	AR
GSD with phosphorylase activation system defects (VIII, IX) • X-linked liver phosphorylase kinase(α subunit, Xp22.2-22.1) • Autosomal liver and muscle phosphorylase kinase (beta subunit, 16q12-q13) • Autosomal liver phosphorylase kinase (gamma subunit, 16p12.1)	X-linked	AR AR AR
GSD X	Cyclic 3', 5' AMP-dependent kinase (17q23-24)	AR
GSD XI (Fanconi-Bickel syndrome, GLUT2 deficiency)	Glucose transporter 2 (3q26.1-q26.3)	AR
GSD XII (aldolase A deficiency)	Aldolase A (16q22-24)	AR

Table 2 Genetics of glycogen storage disease.

responsible for the development of ketosis and acidosis, and vomiting and drowsiness may be the prelude to coma or convulsions. Chronic infection is not acceptable in any patient with renal involvement in case of blood-born transference of infection or exacerbation of a pre-existing infection. Thus, dental care should be directed towards elimination of any potential dental sepsis andthe maintenance of this

position [17].

Extractions may create a problem for two reasons. Since some patients may have haemorrhagic complications, the blood must be checked before surgery is contemplated with particular reference to bleeding and clotting time and platelet count. The second point is that of general anaesthesia. This must be done as an in-patient as hypoglycaemia is a real possibility in such circumstances of fasting. There is no contraindication to local anaesthesia. Local infiltrate anesthesia is preferable to an inferior alveolar nerve block because of the risk of bleeding along the pharyngeal fascial spaces up to the patient's airways and mediastinum [Ralls and Marshall, 1985].

As no patient can be treated property unless adequate information is available about his condition [29]. Dental prevention necessities to be the basis for patients with GSD. Improved oral hygiene instruction, fluoride supplementation, and frequent dental recalls should institute to lessen the risk of dental sequential from accrued cariogenic influence. Besides, anti-bacterial mouth rinses like chlorhexidine gluconate is a helpful adjunct to maintain the oral hygiene of these patients [18].

Conclusion

Certain basic cognitive process to aseptic proficiency, normoglycemia, and acid base condition, and cognizance of the expectation of bleeding and hematomata establishment are necessary to overwhelm these condition. Hence interdisciplinary approach is important to improve their quality of their life.

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