Anatomía Digital

ISSN: 2697-3391
Vol. 7 No. 1.3, pp. 85 – 98, March 2024

www.anatomiadigital.org

Enfermedades de almacenamiento de glucógeno. Glucogenosis tipo I

Glycogen storage diseases, Type I Glycogenosis

[1] Richard Franklin Avellan Zambrano https://orcid.org/0009-0007-4307-0569
Medical Surgeon, Technical University of Manabí, Portoviejo, Ecuador.
sanpanchouciasf@gmail.com

[2] Ana Maria Castro Zambrano https://orcid.org/0009-0007-3947-3285
Medical Surgeon, Technical University of Manabí, Portoviejo, Ecuador.
Anitamaria1208@hotmail.com

[3] JJesus Gregorio Loor Vera https://orcid.org/0009-0001-7856-547X
Medical Surgeon, Technical University of Manabí, Portoviejo, Ecuador.
jglv189@gmail.com

[4] Dayanara Aracely Lalangui Pinargote https://orcid.org/0009-0008-1153-3837
Medical Surgeon, Technical University of Manabí, Portoviejo, Ecuador.
dayiis_96lala@hotmail.es

[5] Andrea Isabel Quiroga Sabando https://orcid.org/0009-0008-1161-1698
Resident Physician, Fresenius Medical Care, Daule.
isa20011939@gmail.com

Scientific and Technological Research Article
Sent: 12/12/2023
Revised: 01/09/2024
Accepted: 10/02/2024
Published:22/03/2024
DOI: **https://doi.org/10.33262/anatomiadigital.v7i1.3.2954**

Please quote: Avellan Zambrano, RF, Castro Zambrano, AM, Loor Vera, JG, Lalangui Pinargote, DA, & Quiroga Sabando, AI (2024). Glycogen storage diseases. Glycogenosis type I. Digital Anatomy, 7(1.3), 85-98.https://doi.org/10.33262/anatomiadigital.v7i1.3.2954

DIGITAL ANATOMY is an electronic, quarterly journal that will be published in electronic format and has the mission of contributing to the training of competent professionals with a humanistic and critical vision who are capable of presenting their investigative and scientific results to the same extent that positive changes in society are promoted through their intervention.https://anatomiadigital.org
The journal is published by Editorial Ciencia Digital (a prestigious publisher registered with the Ecuadorian Book Chamber with membership number 663).www.celibro.org.ec

Anatomía Digital

ISSN: 2697-3391
Vol. 7 No. 1.3, pp. 85 – 98, March 2024

www.anatomiadigital.org

Palabras claves:
Enfermedad del almacenamiento de glucógeno; enfermedad del almacenamiento de glucógeno tipo I; glucosa-6-fosfatasa; glucosa-6-fosfatasa translocasa

Resumen

Introducción: El cuerpo humano, para la obtención de energía, descompone carbohidratos a partir de la dieta y los convierte en glucosa. Este proceso de aporte de glucosa al organismo se limita a las 2-3 horas postprandiales, por cuanto, es indispensable un mecanismo que garantice el aporte sostenido y suficiente de este recurso, fundamental para el metabolismo energético. Las glucogenosis son un grupo heterogéneo de enfermedades hereditarias, en su gran mayoría de herencia autosómica recesiva, que se caracterizan por un fallo en la ruta metabólica del glucógeno, también se las denomina enfermedades "raras" o "huérfanas", ya que pocas personas las padecen o han sido diagnosticadas. **Objeto:** sintetizar la más vigente información respecto a las glucogenosis tipo Ia y Ib. **Metodología:** Se realizó una revisión bibliográfica narrativa a partir de una búsqueda de artículos científicos en las bases de especializadas SciELO y PubMed. Los criterios de selección para la construcción del manuscrito son: artículos en inglés y/o español que incluyeran información respecto a la fisiopatología, manifestaciones clínicas, diagnóstico y abordaje terapéutico de la glucogenosis tipo Ia y Ib. **Resultados:** Se construyó un documento científico de fácil lectura y que aborda los principales tópicos de las glucogenosis Ia y Ib de forma accesible a todos los niveles de salud. **Conclusión:** La glucogenosis abarca una amplia gama de enfermedades relacionadas a anomalías enzimáticas específicas que, en función del tipo en concreto, resulta de mayor o menor complejidad su diagnóstico y abordaje. **Área de estudio general:** medicina**. Área de estudio específica:** pediatría y genética médica. **Tipo de estudio:** revisión narrativa.

Keywords:
Glycogen storage disease; Glycogen storage disease type I; glucose-6-phosphatase; glucose-6-

Abstract

Background: The human body, to obtain energy, breaks down carbohydrates from the diet and converts them into glucose. This process of providing glucose to the body is limited to 2-3 postprandial hours, therefore, a mechanism that guarantees the sustained and sufficient supply of this resource, essential for energy metabolism, is essential. Glycogenosis is a heterogeneous group of hereditary diseases, mostly of

ISSN: 2697-3391
Vol. 7 No. 1.3, pp. 85 – 98, March 2024

www.anatomiadigital.org

phosphatase
translocase

autosomal recessive inheritance, which are characterized by a failure in the glycogen metabolic pathway. They are also called "rare" or "orphan" diseases, since few people have them. suffer or have been diagnosed. Objective: synthesize the most current information regarding glycogen diseases type Ia and Ib. Methodology: A narrative bibliographic review was carried out based on a search for scientific articles in the specialized databases SciELO and PubMed. The selection criteria for the construction of the manuscript are: articles in English and/or Spanish that include information regarding the pathophysiology, clinical manifestations, diagnosis and therapeutic approach of glycogenosis type Ia and Ib. Results: An easy-to-read scientific document was constructed that addresses the main topics of glycogenosis Ia and Ib in a way that is accessible to all health levels. Conclusion: Glycogenosis covers a wide range of diseases related to specific enzymatic abnormalities that, depending on the specific type, are more or less complex to diagnose and approach. General area of study: medicine. Specific study area: pediatrics and medical genetics. Type of study: narrative review.

Introduction

To obtain energy, the human body breaks down carbohydrates from the diet and converts them into glucose. This process of supplying glucose to the body is limited to 2-3 hours postprandially, so a mechanism is essential to ensure the sustained and sufficient supply of this resource, which is essential for energy metabolism.(1).

Glycogen, a branched polysaccharide of glucose, is the main form of glucose storage for the human body. It is found primarily in the liver and muscle cells, with muscle mass, given its greater volume in relation to the mass of the liver, being responsible for housing approximately three quarters of glycogen.(2)The essential difference between both storage locations is the purpose of this; liver glycogen, between meals and under fasting conditions of up to approximately 12-18 hours, allows the maintenance of glucose homeostasis, while muscle glycogen supplies glucose 1-phosphate locally during episodes of intense physical activity.(1–4).

The metabolic pathways involved in glycogen metabolism do not comprise two pathways of the same route, they are separate pathways.(3)Glycogenesis is the anabolic pathway

by which glycogen is obtained from glucose molecules, while glycogenolysis is the catabolic pathway by which glycogen is broken down into glucose.(1.4).

Glucogenesis starts with the production of glucose-6-phosphate from glucose and the enzyme glucokinase. The enzyme phosphoglucomutase mediates the transformation of glucose-6-phosphate to glucose-1-phosphate, the latter being activated by UDP-glucose-pyrophosphorylase, forming uridine diphosphate glucose (UDP-glucose). The incorporation of glucose into the pre-existing glycogen molecule is mediated by the enzyme glycogen synthase through a phosphoglucoprotein bond.α(1-4). 8 glucose residues are addedα(1-4) before the branching enzyme transfers 7 of these glucoseα(1-4) to a unionα(1,6) After this, the enzyme glycogen synthase continues the process of glucose adhesion with bondsα(1-4)(4).

Glycogenolysis begins with the activated glycogen phosphorylase isoenzymes (muscle, liver and brain), which break the bondsα(1-4) on the periphery of glycogen, releasing glucose-1-phosphate, which, through the intervention of the enzyme phosphoglucomutase, is isomerized into glucose-6-phosphate. At the liver level, this glucose-6-phosphate is hydrolyzed into glucose by glucose-6-phosphatase, allowing its incorporation into the blood. Glycogen phosphorylase exclusively cleaves glycosidic bonds.α(1-4), is unable to cleave glycosidic bondsα(1,6) norα(1-4) proximal to the branching pointα(1,6), four residues before the linkα(1,6) This is relieved by the debranching enzyme, with 4- activityα-glucan-transferase andα(1,6)-glucosidase. The debranching enzyme, by transferase activity, mobilizes a segment of branched glucoses in a bondα(1,6) to the linkα(1-4) and leaves 1 branched glucose residue, this is eliminated in the form of glucose by glucosidase activity, the result is a linear chain of glycosidic bondsα(1-4) which are cleaved by glycogen phosphorylase until they reach the proximity of a new branch pointα(1,6), where the described process will be repeated. About 90% of glucose is

Glycogenoses are a heterogeneous group of hereditary diseases, mostly autosomal recessive (AR) inheritance, which are characterized by a failure in the glycogen metabolic pathway. They are also called "rare" or "orphan" diseases, since few people suffer from them or have been diagnosed.(2,3,5–10)In this disease, there are failures in one of the enzymes responsible for forming or using up glycogen, which causes alternate metabolic pathways to be used; reflected in an abnormal accumulation and decrease in blood sugar levels available in the fasting state.(5,7,9)Symptoms mostly occur in childhood, but may also occur in the intrauterine phase.(5). Glucose-6-phosphatase deficiency (type I), lysosomal acid α-glucosidase deficiency (type II), debranching enzyme deficiency (type III) and hepatic PhK deficiency (type IX) being the most frequent in childhood and myophosphorylase deficiency (type V, or McArdle disease) in adults.(9).

The diagnosis is made through clinical examination and is subsequently confirmed with a liver and/or muscle biopsy, which shows abnormal glycogen storage.(7.9)To classify this disease, the genetic abnormality must be identified; it has been observed that its estimated frequency is approximately one in one hundred thousand inhabitants.(5,11).

Its clinical manifestations vary from asymptomatic and with only exercise intolerance, to those that can cause death. The most important glycogenosis in order of frequency are type I, III, IX, II, V, VII, corresponding to about 95% of the total.(9,12).

Historically, they have been classified in the order in which the enzymatic defects were identified, and also according to the affected organ (liver, muscle, heart or both), clinical phenotype and histopathological findings (hepatic glycogenosis, muscular glycogenosis and generalized glycogenosis).(9,11)Currently there are 10 types of glycogenosis, whether involving the muscles, liver or mixed, with glycogenosis of hepatic origin being the most common.(5). Thus, glycogenosis types Ia and Ib will be addressed in the development of this work.

Methodology

A narrative bibliographic review was conducted based on a search for scientific articles in the specialized databases SciELO and PubMed. The selection criteria for the construction of the manuscript are: articles in English and/or Spanish that included information regarding the pathophysiology, clinical manifestations, diagnosis and therapeutic approach of glycogen storage disease type Ia and Ib.

For the initial search, the following descriptor combinations were used: “glycogen storage disease”; “glycogen storage disease type I”; “glucose-6-phosphatase”; “glucose-6-phosphatase translocase”. Approximately 437 articles, books and other forms of scientific production were found, of which 31 were selected that met the selection criteria and were most relevant to the objective of the review, based on a quick review of the title, keywords and abstract.

Results and discussion

Glycogenosis Type I

Generalities

GSD type I is arare, inherited metabolic disorder with an incidence of approximately 1/100,000 to 1/400,000 live births in the general population of the Caucasus, the disorder occurs inGlucose-6-phosphatase, which mainly affects the liver, kidneys and intestinal mucosa(9,13)In this disease there may be an abnormality in theglucose-6-phosphatase enzyme (Type Ia) or in the translocase that transports glucose-6-phosphate across the microsomal membrane (Type IB)(9).

Glycogenosis Type Ia

By definition, it is an alteration in glycogen metabolism, it is rare and the alteration is found in the G6PC and G6PT1 gene, which is expressed biochemically as a deficiency of glucose-6-phosphatase (G6Pase).or its carrier proteins, this enzyme is found mainly in the liver and kidneys(14–16)

Epidemiology

The age of presentation is between three to six months and its incidence is 1 in 20,000 births.(14,17).

Genetic defect

The alteration is located in the genes G6PT1 and G6PC, located on chromosome 17q21 and encoding the G6Pase catalytic subunit, which causes loss of G6Pase function and accounts for approximately 80% of GSD(13,14). The functional G6Pase-α/G6PT complex maintains interprandial blood glucose homeostasis; itsThe main function of the G6PT protein in vivo is to translocate G6P from the cytoplasm to the lumen of the ER, taking it to the catalytic site of G6Pase-α or G6Pase-β for hydrolysis into glucose and phosphate.(18).

Greater impact

In an early process, the organ most affected is the liver and when focusing on long-term complications, we have that the disease focuses on three organs mainly, which are the liver (adenomas and hepatocellular carcinomas), the kidneys (proteinuria, kidney failure, stones) and the bone (osteopenia, osteoporosis).(9,15).

Clinical manifestations

The clinical presentation becomes apparent from three to six months with hepatomegaly and signs and symptoms of hypoglycemia; or during the neonatal period with hypoglycemia and lactic acidosis.(17)Symptoms and signs include severe fasting intolerance, failure to thrive and hepatomegaly, nonketotic hypoglycemia, hyperlactidemia, hyperuricemia, and hyperlipidemia.(15);As a result of the accumulation of glucose-6-phosphate (G6P) in hepatocytes and renal cells, there is an excess storage of glycogen and triglycerides in the liver, causing fatty liver similar to non-alcoholic fatty liver disease (NAFLD).(19).

Diagnosis

It is performed with the clinical picture and abnormalities in the levels of lactate and lipids in the plasma; but genetic analysis of mutations is the option to establish a definitive diagnosis.(9).

Treatment

The treatment is aimed at maintaining normoglycemia and consequently normalizing triglyceride (TG) levels; it basically consists of implementing meals at short intervals, rich in carbohydrates, with slow-release glucose preparations, such as uncooked corn starch, which is used to prolong the fasting period.(16,19).

Continuous nocturnal gastric drip feeding in children maintains normal glucose levels, allowing rest during the night(14,16).In a case reportRaza et al reported that "strict adherence to dietary therapy leads to improved quality of life, increases survival and minimizes complications, and liver transplantation can reverse biochemical abnormalities."(17).

In a 2013 meta-analysis, which identified five controlled trials (49 participants) with follow-up from 2 days to 14 years comparing intermittent administration of uncooked cornstarch with continuous overnight feedings of dextrose, with modified uncooked cornstarch and dextrose, and an uncooked cornstarch blend, twenty-six participants (three trials) receiving uncooked cornstarch showed a significant increase in blood glucose concentration, 21 (two trials) increased serum insulin, and 22 (three trials) increased plasma total cholesterol compared with continuous overnight feedings of dextrose and twenty-eight subjects (three trials) showed decreased plasma lactate, leading to the conclusion that "short- and long-term uncooked cornstarch prevents nocturnal hypoglycemia in GSD-1a children more effectively than continuous overnight feedings of dextrose."(20)

Standard glucose self-regulation, generally pre-meal, should be encouraged, since it is observed that the fear of hypoglycemia leads to overeating and then to obesity, which generates a clear risk of atherosclerosis and cardiovascular diseases.(16).

Glycogenosis Type Ib

Epidemiology

G6PT deficiency (GSD-Ib) accounts for 20% of GSD-I cases and Ashkenazi Jews have a 5-fold higher prevalence compared to the rest of the population.

Genetic defect

In this subtype there is a deficit in the ubiquitously expressed G6PT or also called G6Pase-β, in contrast we have that a functional G6Pase-β / G6PT complex maintains energy homeostasis and functionality in neutrophils and macrophages.(18,21)It is encoded by the SLC37A4 gene located on chromosome 11q23 and is a member of the solute carrier family 37 (SLC37)(22–24)

Greater impact

The organs most affected in this case are a little more extensive because the affected enzyme is expressed ubiquitously, the glycogen deposit is mostly seen in the liver and kidney where it is expressed as megalias, in addition to this we have marked defects, when it is expressed, in the white series (neutropenia). The latter is also reflected in the gastrointestinal tract as colitis and in the liver with a greater propensity to neoplasias.(18,23,25).

Clinical manifestations

It shares the same metabolic phenotype of impaired glucose homeostasis with GSD-Ia, but carries the additional complications of neutropenia and myeloid dysfunction such as severe congenital neutropenia syndrome type Ia.(18)The hallmark of the GSD-Ib metabolic phenotype is hypoglycemia after a brief fast.(24).

The activity depends on its ability to form a functional complex with a G6Pase, which, in the absence of this, the G6P transport activity is minimal. Consequently, G6PT is important for maintaining both interprandial blood glucose homeostasis and energy homeostasis of myeloid cells, making GSD-Ib a metabolic and immunological disorder; despite which there does not seem to be a strict relationship between genotype and phenotype.(24)

The defective G6PT/G6Pase-α complex impairs ER transport and G6P hydrolysis, leading to elevated levels of cytoplasmic G6P and loss of blood glucose homeostasis. In gluconeogenic organs lacking functional G6PT, this stimulates competing pathways that use elevated cytoplasmic G6P, ultimately giving the clinical manifestations of fasting hypoglycemia, hepatomegaly, nephromegaly, hyperlipidemia, hyperuricemia, and lactic acidemia characteristic of this entity.(21,24)Hepatomegaly and nephromegaly are the result of an accumulation of glycogen and neutral fat.(24).

Diagnosis

It presents a clinical picture very similar to that of GSD Ia, however, hepatomegaly, doll-like face, short stature, and chronic fatigue are more severe, all of this added to neutrophil dysfunction and neutropenia that makes one susceptible to frequent bacterial infections.

Neutropenia is not reliable as a differential for GSDIa since it is periodic, never develops in GSD-Ib or is normal in the first two years of life.(22,26)

The presence of hepatomegaly plus hypoglycemia should prompt a battery of tests including lactate, uric acid, liver profile including liver function tests, cholesterol, triglycerides, basic chemistry panel, creatine kinase, complete blood cell count with manual white cell differential, plasma total and free carnitine, acylcarnitine amino acid profile, β-hydroxybuty4, and urine organic acids.(26). In the laboratory, hypoglycemia, lactic acidosis, hyperuricemia, hypercholesterolemia and hypertriglyceridemia will be found, the glucagon stimulation test should be avoided due to the risk of acute acidosis and decompensation.(27).

The first and best option to confirm the clinical suspicion and diagnosis are molecular genetic tests that allow complete sequencing of the G6PC (GSD Ia) and SLC37A4 (GSD Ib) genes and are non-invasive, their detection rate is estimated to be up to 100%.(22,27). In a systematic review of the literatureBeyzaeiZ et al. state that "targeted gene sequencing analysis can be considered as the first-line diagnostic method with valuable results and exome sequencing can be used to diagnose complex cases of GSD, and that these molecular methods are considered accurate forms of diagnosis."(25).

Another option that is increasingly less recommended is a liver biopsy where the G6Pase enzymatic activity is analyzed to confirm the diagnosis.(27).

Treatment

Nutritionally, the treatment is the same, but additional dietary intervention is required as a result of neutropenia that presents as enterocolitis similar to Crohn's disease.(26).

The feeding strategy consists of frequent small feedings high in complex carbohydrates (preferably those highest in fiber) spread evenly over 24 hours. The regimen should be structured with 60–70% of calories from carbohydrates, 10–15% calories from protein (to provide the recommended daily intake), and the remaining calories from fat (<30% for children older than 2 years).(26).

Conclusion

- Glycogen storage disease covers a wide range of diseases related to specific enzymatic abnormalities that, depending on the specific type, are more or less complex to diagnose and address.
- Glycogen storage disease type Ia is rare and the alteration is found in the G6PC and G6PT1 genes, which are biochemically expressed as a deficiency in glucose-

6-phosphatase (G6Pase) or its carrier proteins; while glycogen storage disease type Ib occurs with a deficiency in the ubiquitously expressed G6PT or also called G6Pase-β, in contrast we have that a functional G6Pase-β / G6PT complex maintains energy homeostasis and functionality in neutrophils and macrophages.

- Both pathologies tend to begin before the first year of age, affecting the white blood cell series and having the liver and kidneys as target organs.

Conflict of interest

The authors declare that they have no conflicts of interest that could compromise, in whole or in part, the results of this work or its publication.

Authors' contribution statement

RKAZ and AIQS conceived the research idea, defined the problem and conducted an initial information search.

AMCZ and JGLV conducted the non-systematic search to construct the article database and designed the first draft under the supervision of JLI and KGP.

AIQS supervised the development of the second draft by AMCZ and DALP.

JGLV applied corrections to the second and third drafts.

RKAZ and AIQS approved the final manuscript.

Bibliographic references

1. Baynes JW, Dominiczak MH. Medical Biochemistry. 5th ed. Barcelona, Spain: Elsevier; 2019.

2. Ejarque Doménech I, Marín Reina P, García-Miñaur Rica S, Chirivella González I, Martínez Martínez MT, García Rodríguez AM, et al. Criteria for referral to clinical genetics from Primary Care. Consensus document. Aten Primary [Internet]. 2022;54(12):102501. Available at: http://dx.doi.org/10.1016/j.aprim.2022.102501

3. Murray RK, Kennelly PJ, Bender DA, Rodwell VW, Botham KM, Weil PA.Harper. Biochemistry Illustrated. 29th ed. 2012.

4. González Hernández Á. Principles of clinical biochemistry and molecular pathology. 2nd ed. Barcelona, Spain: Elsevier; 2014.

5. Glycogen storage disease. Cooperative University of Colombia. 2017. Available at: https://repository.ucc.edu.co/server/api/core/bitstreams/aed9b490-0418-4cd6-b609-0a9b989832d0/content

6. Sibrián Beltrán DP. Clinical-epidemiological profile of patients with suspected diagnosis of hepatic glycogen storage disease aged 1 to 18 years at the Benjamín Bloom National Children's Hospital in the period 2005 to 2015. [Faculty of Medicine]: University of El Salvador; 2016. Available at: https://pesquisa.bvsalud.org/portal/resource/pt/biblio-1247348

7. Demczko M. Glycogen storage disorders [Internet]. MSD Manual Professional Version. [cited 10 Feb 2024]. Available from: https://www.msdmanuals.com/en/professional/pediatrics/hereditary-metabolism-disorders/glycogen-storage-disorders?autoredirectid=22092

8. World Health Organization. Frequently Asked Questions: How does WHO define health?[Internet]. [cited 2020 Jun 19]. Available from: https://www.who.int/es/about/who-we-are/frequently-asked-questions

9. Kishnani PS, Chen YT. Glycogen storage disease and other inherited disorders of carbohydrate metabolism. Harrison's Principles of Internal Medicine, 20e. 2018;

10. Mitanchez D. Neonatal hypoglycemia and hyperglycemia. EMC - Pediatr [Internet]. 2022;57(4):1–8. Available at: http://dx.doi.org/10.1016/s1245-1789(22)47084-3

11. Funes Rivera K. Pediatric Anthropometry Protocol. University of El Salvador Publishing House. Available at: http://www.medicina.ues.edu.sv/habilidadesydestrezas/documentospdf4/ANTROPOMETRIA%20PEDIATRICA%202017.pdf: University of El Salvador; 2017.

12. Glycogen storage diseases and other inherited disorders of carbohydrate metabolism. Medicine. 2016;12(19):1082–93.Available at: https://dialnet.unirioja.es/servlet/articulo?codigo=5678352

13. Ai J, He W, Huang X, Wu Y, Lei Y, Yu C, et al.A case report of acute pancreatitis with glycogen storage disease type IA in an adult patient and review of the literature. Medicine. 2020 Oct 16;99(42):e22644. Available at: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7571931/

14. Torra Balcells R. Nephropathies due to Hereditary Metabolic Disorders with Renal Involvement. Nephrology Today [Internet]. 2023 [cited on February 10, 2024];23. Available at: https://www.nefrologiaaldia.org/es-articulo-nefropatias-por-trastorno-metabolico-hereditario-con-308

15. Peeks F, Steunenberg TAH, de Boer F, Rubio-Gozalbo ME, Williams M, Burghard R, et al. Clinical and biochemical heterogeneity among patients with glycogen storage disease type IA: the added value of CUSUM for metabolic

control. J Inherit Metab Dis. 2017 Sep 10;40(5):695–702.Available at: https://pubmed.ncbi.nlm.nih.gov/28397058/

16. Jelaska BK, Ostojić SB, Berović N, Kokić V. Continuous glucose monitoring in the treatment of obesity in patients with glycogen storage disease type Ia. Endocrinol Diabetes Metab Case Rep. 2014 Jan 17;2014(1).Available at: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3965284/

17. Raza M, Arif F, Giyanwani PR, Azizullah S, Kumari S. Dietary Therapy for Von Gierke's Disease: A Case Report.Cureus. 2017 Aug 8;9(8). Available at: https://pubmed.ncbi.nlm.nih.gov/29018645/

18. Chou JY, Jun HS, Mansfield BC. Type I glycogen storage diseases: disorders of the glucose-6-phosphatase/glucose-6-phosphate transporter complexes. J Inherit Metab Dis. 2015;38(3):511–9.Available at: https://pubmed.ncbi.nlm.nih.gov/25288127/

19. Waskowicz LR, Zhou J, Landau DJ, Brooks ED, Lim A, Yavarow ZA, et al. Bezafibrate induces autophagy and improves hepatic lipid metabolism in glycogen storage disease type Ia. Hum Mol Genet.2019 Jan 1;28(1):143–54. Available at: https://academic.oup.com/hmg/article/28/1/143/5106899

20. Shah KK, O'Dell SD.Effect of dietary interventions in the maintenance of normoglycaemia in glycogen storage disease type 1a: a systematic review and meta-analysis. Journal of Human Nutrition and Dietetics. 2013 Aug 1;26(4):329–39. Available at: https://pubmed.ncbi.nlm.nih.gov/23294025/

21. Jeon EH, Park TS, Jang Y, Hwang E, Kim SJ, Song KD, et al. Glucose-6-phosphate transporter mediates macrophage proliferation and functions by regulating glycolysis and mitochondrial respiration. Biochem Biophys Res Commun. 2020;524(1):89–95.Available at: https://pubmed.ncbi.nlm.nih.gov/31980167/

22. Beyzaei Z, Geramizadeh B. Molecular diagnosis of glycogen storage disease type I: A review. Vol. 18, EXCLI Journal. Leibniz Research Center for Working Environment and Human Factors; 2019. p. 30–46.Available at: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6449677/

23. Wicker C, Roda C, Perry A, Arnoux JB, Brassier A, Castelle M, et al. Infectious and digestive complications in glycogen storage disease type Ib: Study of a French cohort. Mol Genet Metab Rep. 2020;23(March):100581.Available at: https://pubmed.ncbi.nlm.nih.gov/32300528/

24. Chou JY, Cho JH, Kim GY, Mansfield BC. Molecular biology and gene therapy for glycogen storage disease type Ib. J Inherit Metab Dis. 2018;41(6):1007–14.Available at: https://pubmed.ncbi.nlm.nih.gov/29663270/

25. Beyzaei Z, Geramizadeh B, Karimzadeh S. Diagnosis of hepatic glycogen storage disease patients with overlapping clinical symptoms by massively parallel sequencing: a systematic review of literature. Vol. 15, Orphanet Journal of Rare Diseases. BioMed Central Ltd; 2020. p. 286.Available at: https://pubmed.ncbi.nlm.nih.gov/33054851/

26. Kishnani PS, Austin SL, Abdenur JE, Arn P, Bali DS, Boney A, et al. Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics. Genetics in Medicine 2014 16:11. 2014 Nov 6;16(11):e1–e1.Available at: https://pubmed.ncbi.nlm.nih.gov/25356975/

27. Glycogen Storage Disease Type I - StatPearls - NCBI Bookshelf [Internet].[cited 2020 Dec 28]. Available at: https://www.ncbi.nlm.nih.gov/books/NBK534196/

Anatomía Digital

ISSN: 2697-3391
Vol. 7 No. 1.3, pp. 85 – 98, March 2024

www.anatomiadigital.org

The published article is the sole responsibility of the authors and does not necessarily reflect the thinking of the Anatomía Digital Journal.

Ciencia Digital Editorial

latindex catálogo

Anatomía Digital

Indexaciones