Case Report

Glycogen Storage Disease Type IX in a 6-year-old Male: A Case Report

Shahab Noorian[1], Hossein Moravej[2], Zhila Afshar[3], Afagh Hassanzadeh Rad[4], Setila Dalili[4*]

1. Department of Pediatrics, School of Medicine, Alborz University of Medical Sciences, Karaj, Iran.
2. Neonatal Research Center, Shiraz University of Medical Sciences, Shiraz, Iran.
3. Department of Pediatric Endocrinology, School of Medicine, Shiraz University of Medical Sciences, Shiraz, Iran.
4. Pediatric Diseases Research Center, Guilan University of Medical Sciences, Rasht, Iran.

Use your device to scan and read the article online

Citation Noorian Sh, Moravej H, Hassanzadeh Rad A, Dalili S. Glycogen Storage Disease Type IX in a 6-year-old Male: A Case Report. Journal of Pediatrics Review. 2024; 12(4):369-376. http://dx.doi.org/10.32598/jpr.12.4.584.7

http://dx.doi.org/10.32598/jpr.12.4.584.7

Article info:
Received: 12 May 2024
First Revision: 13 Jun 2024
Accepted: 28 Jun 2024
Published: 01 Oct 2024

Key Words:
Glycogen storage disease (GSDs), Child, Genetic therapy

ABSTRACT

Background: Glycogen storage diseases (GSDs) are a group of inherited metabolic disorders caused by enzyme deficiencies affecting glycogen synthesis or breakdown. Glycogen, stored mainly in the liver and muscles, is crucial for maintaining glucose levels during fasting or physical activity. GSDs lead to abnormal glycogen accumulation or impaired mobilization, causing symptoms, such as hypoglycemia, hepatomegaly, and muscle weakness. Each type of GSD results from a specific enzyme deficiency, requiring tailored management.

Case Presentations: A case study of a 6-year-old boy with GSD type IX is presented, highlighting recurrent hypoglycemia, growth delay, and elevated liver enzymes. Genetic testing confirmed a *PHKA2* mutation, and the patient's management included frequent meals, cornstarch therapy, and regular liver function monitoring. The discussion emphasizes the importance of early diagnosis, genetic testing, and personalized treatment in managing GSDs.

Conclusions: Future therapies, such as gene therapy and enzyme replacement, aim to address the root causes of GSDs rather than merely managing symptoms. Family education on hypoglycemia recognition and dietary restrictions is crucial for improving patient outcomes. Ongoing research into the molecular mechanisms of GSDs offers hope for more effective treatments, especially for GSD type IX, where individualized care can prevent complications like liver cirrhosis and promote better quality of life.

** Corresponding Author:*
Setila Dalili, Professor.
Address: *Pediatric Diseases Research Center, Guilan University of Medical Sciences, Rasht, Iran.*
Tel: *+98 (911) 1411463*
E-mail: *setiladalili1346@yahoo.com*

Introduction

Glycogen storage diseases (GSDs) represent a diverse group of inherited metabolic disorders that arise from deficiencies in the enzymes responsible for the synthesis or degradation of glycogen. Glycogen is a critical polysaccharide, stored primarily in the liver and muscles, that serves as a rapid source of glucose when the body requires energy. The ability to mobilize this glucose efficiently is vital for maintaining blood sugar levels and providing energy during fasting or increased physical activity [1]. The process of glycogen formation, known as glycogenesis, is a multi-step pathway that begins with glucose being phosphorylated to glucose-6-phosphate. This is followed by its conversion to glucose-1-phosphate and subsequently to uridine diphosphate glucose, which is the active form of glucose used by glycogen synthase to elongate the glycogen chain. Branching enzymes further modify this structure, increasing the solubility and storage capacity of glycogen [2, 3].

However, in individuals with GSDs, genetic mutations lead to enzyme deficiencies that disrupt these processes [4, 5]. The result is the accumulation of abnormal glycogen or an inability to mobilize stored glycogen, leading to a range of symptoms that can include hypoglycemia, hepatomegaly (enlarged liver), muscle weakness, and growth retardation. Each type of GSD is associated with a specific enzyme deficiency and presents a unique clinical profile, which necessitates a tailored approach to diagnosis and management [6, 7].

There are over 20 recognized types of GSDs, each associated with a specific enzyme deficiency affecting glycogen metabolism and characterized by unique clinical presentations. While most GSDs primarily affect the liver, several types also involve other organs, such as muscles, heart, and nervous system. Some forms, such as GSD type IV (Andersen disease), can lead to liver cirrhosis and potentially fatal complications if untreated, due to progressive liver damage. Other types, notably GSD types II (Pompe disease) and III (Cori disease) are known for causing significant muscle-related symptoms, including myopathy and cardiomyopathy, due to glycogen accumulation in muscle tissues. Additionally, GSD type II can result in neurological involvement, particularly in the infantile form, due to glycogen buildup in neural tissues. Thus, GSDs represent a diverse group of metabolic disorders with multi-organ implications, each necessitating specific diagnostic and management strategies [1-4].

Theoritical foundation

A comprehensive literature review was conducted utilizing a variety of databases, including PubMed, EMBASE, and the Cochrane Library, to gather relevant information on GSDs from the year 2000 to 2024. The search was guided by specific keywords, which included “glycogen storage disease,” “GSD type IX,” “diagnosis,” “treatment,” and “case presentation.” This systematic approach provides an in-depth exploration of the different types of GSDs, their associated symptoms, methods of diagnosis, and treatment strategies.

In particular, the review placed a significant emphasis on GSD type IX, analyzing its unique characteristics, clinical presentation and variations in management approaches. The selection of articles was based on criteria that ensured a comprehensive understanding of both the common and rare manifestations of this condition, as well as the evolving treatment modalities. Through this literature review, the intention was to identify gaps in the current research, summarize key findings, and ultimately enhance clinical knowledge regarding GSDs, especially GSD type IX, to support improved patient care and outcomes.

Symptoms and treatment of GSDs

In terms of GSD type 0, the deficiency is glycogen synthase and the symptoms are fasting hypoglycemia and ketosis, postprandial hyperglycemia, hyperlactatemia, and no hepatomegaly. The treatment methods are a high-protein diet, and cornstarch therapy [8].

GSD type I (Von Gierke’s disease) results in glucose-6-phosphatase deficiency. The symptoms are severe hypoglycemia, lactic acidosis, hepatomegaly, hyperuricemia, hyperlipidemia, neutropenia in GSD type 1b and prone to adenoma. A galactose and sucrose-free diet is important in these patients. The treatment methods include frequent small meals, corn, and Cassava starch therapy, allopurinol for hyperuricemia, lipid-lowering medications, and granulocyte-colony stimulating factor in GSD type 1b, liver transplantation in severe cases [9, 10].

In terms of GSD type II (Pompe disease), the deficiency includes acid α-glucosidase. The symptoms are cardiomyopathy, hypotonia, muscle weakness, central nervous syndrome involvement, and respiratory failure. The treatment methods are enzyme replacement therapy with α glucosidase, respiratory support, physical therapy, and gene therapy [11-13].

In terms of GSD type III (Cori disease), the deficiency includes debranching enzyme and the symptoms are fasting hypoglycemia, hepatomegaly, myopathy and cardiomyopathy. Treatment methods are high-protein diet, cornstarch therapy, and regular cardiac monitoring [14, 15].

In terms of GSD type IV (Andersen disease), the deficiency is in branching enzyme and the symptoms are hepatosplenomegaly, liver cirrhosis and muscle weakness. Meanwhile, the treatment methods are symptomatic management, and liver transplantation for progressive liver disease [8, 16].

In terms of GSD type V (McArdle's disease), the deficiency is in muscle glycogen phosphorylase and the symptoms are exercise intolerance, muscle cramps, and myoglobinuria. The treatment methods are moderate exercise, high-protein diet, and creatine supplementation [17-19]. Glucose administration before exercise is needed.

In terms of GSD type VI (Hers disease), the symptoms are hepatomegaly, mild hypoglycemia, and growth retardation. Meanwhile, the treatment method is a high-protein, high-carbohydrate diet with frequent small meals to maintain blood sugar levels [8, 20].

In terms of GSD type VII (Tarui disease), the symptoms are exercise intolerance, muscle cramps, myoglobinuria, and hemolytic anemia and the treatment methods are avoiding strenuous exercise, a high-protein diet, and glucose administration during exercise [19].

GSD type IX (GSD IX) is among the most common GSDs, resulting from a deficiency in phosphorylase kinase (PhK), an enzyme essential for glycogen breakdown. It is primarily inherited in an X-linked recessive pattern, affecting males more frequently, though some forms can be autosomal recessive, affecting both sexes. The hallmark symptoms of GSD IX include hepatomegaly (enlarged liver), growth retardation and mild hypoglycemia. Hepatomegaly arises due to glycogen accumulation in the liver, often detected early in childhood, while growth delays stem from episodes of hypoglycemia and the body's inability to efficiently access stored glycogen. Mild hypoglycemia is typically managed well but can worsen during illness or prolonged fasting. In rare cases, particularly with mutations affecting the muscle isoform of PhK, individuals may experience muscle weakness and exercise intolerance.

Treatment focuses on dietary management to stabilize blood glucose levels and support growth. A high-carbohydrate, high-protein diet, frequent meals, and cornstarch therapy are central to this approach. Protein supports growth and provides substrates for gluconeogenesis, while uncooked cornstarch acts as a slow-release carbohydrate, helping maintain glucose levels overnight and reducing nocturnal hypoglycemia. With these dietary interventions, many children with GSD IX experience significant symptom improvement, often outgrowing some symptoms, such as hepatomegaly and growth delays, by adolescence. Regular follow-up is essential for monitoring liver function, growth, and overall metabolic health, allowing most individuals with GSD IX to lead relatively normal lives with few severe complications [8].

GSD IXa is a glycogen storage disorder caused by mutations in the *PHKA2* gene, located on the Xp22.13 chromosomes, which follows an X-linked recessive inheritance pattern. The subtypes are GSD IXa1 (XLG1) which primarily affects the liver and blood and GSD IXa2 (XLG2) which predominantly affects the liver and is recognized as the most common form of GSD IXa. The symptoms of GSD IXa include the following items: Hypoglycemia which is a condition characterized by abnormally low blood glucose levels, often leading to fatigue, confusion, and irritability; ketosis which is the production of ketone bodies due to increased fat metabolism, occurring when glucose availability is limited; hepatomegaly which is the enlargement of the liver, which can lead to abdominal swelling and discomfort; chronic liver disease that is long-term liver dysfunction that may progress to cirrhosis or other liver-related complications; liver fibrosis which is the excessive accumulation of extracellular matrix proteins, leading to scarring of liver tissue; hyperlipidemia is an increase in lipid levels in the bloodstream, which can contribute to cardiovascular problems; elevated liver enzymes which is the increased levels of liver enzymes in the blood, indicating liver inflammation or damage; growth retardation which is the delayed growth and development in children, often requiring monitoring and intervention.

Meanwhile, the symptoms of GSD IXa can vary in severity, and early diagnosis is crucial for managing the condition and minimizing complications. Management often includes dietary modifications, glucose supplementation, and monitoring of liver function to mitigate the impact of the disorder on overall health [21].

GSD type IXb (GSD IXb) is a metabolic disorder caused by mutations in the *PHKB* gene, located on the 16q12.1 chromosome and it follows an autosomal recessive inheritance pattern. This condition is characterized by abnormal glycogen metabolism due to the dysfunction of the glycogen PhK enzyme, which plays a critical role in

glycogen breakdown. The affected tissues are the liver and muscle tissues, resulting in a range of metabolic disturbances. The symptoms of GSD IXb can vary among individuals but typically includes the following symptoms: Hepatomegaly is the most common symptom and refers to the enlargement of the liver and can lead to abdominal swelling and discomfort, as well as potential complications related to liver function; growth retardation is that children with GSD IXb often experience growth delays, which can affect overall physical development and this may necessitate nutritional interventions and regular monitoring of growth parameters to ensure that affected individuals meet developmental milestones; mild or absent muscle disease is that unlike some other forms of GSD, muscle involvement in GSD IXb is usually mild or may be absent altogether, and affected individuals may not exhibit significant muscle weakness or pain, and their muscle function can remain relatively intact [22].

GSD type IXc (GSD IXc) is a metabolic disorder resulting from mutations in the *PHKG2* gene, located on chromosome 16p11.2, and it follows an autosomal recessive inheritance pattern. This condition is characterized by a deficiency in glycogen PhK, which is essential for the regulation of glycogen metabolism, leading to abnormal accumulation of glycogen in tissues, particularly in the liver. The affected tissues are the liver, where the pathological processes of glycogen accumulation manifest in various ways. The clinical features of GSD IXc tend to be more severe compared to other forms, particularly GSD IXa2 and can include the following items: Marked hepatomegaly which is related to individuals with GSD IXc typically presents with significant liver enlargement due to the accumulation of glycogen this enlargement can cause abdominal distension and discomfort and it may have implications for liver function; recurrent hypoglycemia is the hallmark of GSD IXc, recurrent episodes of low blood glucose levels can occur, especially during fasting or periods of increased physical activity and symptoms of hypoglycemia may include irritability, confusion, fatigue and in severe cases, seizures or loss of consciousness; hyperlipidemia is an increase in lipid levels in the blood is often observed, which can contribute to metabolic complications and cardiovascular risks and this occurs as the body attempts to mobilize fat stores due to impaired glycogen metabolism; markedly elevated liver enzymes is that liver function tests typically reveal significantly increased levels of liver enzymes, indicating hepatocellular damage or inflammation and this can be a critical sign of the disease's progression and severity; liver fibrosis/cirrhosis is that over time, the chronic accumulation of glycogen and resultant liver damage may lead to fibrosis, which is the excessive formation of connective tissue, potentially progressing to cirrhosis, meanwhile, cirrhosis can result in serious complications, including portal hypertension and liver failure [23].

GSD type IXd (GSD IXd) is a rare metabolic disorder caused by mutations in the *PHKA1* gene, located on the Xq13.1 chromosome and it follows an X-linked recessive inheritance pattern. This condition primarily affects muscle tissue, leading to disturbances in glycogen metabolism that manifest in various symptoms, especially during physical activity. GSD IXd predominantly impacts muscle, where the impaired function of glycogen PhK disrupts normal glycogen utilization during exercise. The clinical presentation of GSD IXd is characterized by several key features as follows: Cramps and exercise intolerance is that individuals with GSD IXd commonly experience muscle cramps during physical exertion and this occurs due to the inability to adequately mobilize glycogen stores for energy production during exercise, leading to muscle fatigue and discomfort; middle age onset is that symptoms of GSD IXd typically begin to manifest in middle adulthood, which can delay diagnosis and this late-onset can complicate recognition, as individuals may attribute symptoms to normal aging or other conditions; raised creatine kinase levels is that Patients often present with elevated serum creatine kinase levels, which indicate muscle damage or stress, meanwhile, elevated creatine kinase is a common marker in muscle disorders and is used in the diagnostic process to evaluate muscle integrity; normal red cell and liver PhK activity is the one distinguishing feature of GSD IXd that red blood cell liver glycogen PhK activity remains normal and this contrasts with other GSDs where PHK activity may be significantly reduced or absent in these tissues, in addition, the preservation of PHK activity in red blood cells and liver is a critical factor for differentiating GSD IXd from other related disorders [24].

In terms of GSD XI (Fanconi-Bickel syndrome), the glucose transporter 2 is affected and the symptoms are hepatomegaly, glucose and galactose intolerance, fasting hypoglycemia, proximal tubular nephropathy, and severe short stature. Meanwhile, there is no specific treatment [25].

Case Presentation

The patient in this case is a 6-year-old boy, the first child of an unrelated family. He was born with a birth weight of 2800 g through a natural vaginal delivery. At the age of 4 months, he began experiencing hypoglycemic attacks, which were concerning and required medical evaluation.

During this time, episodes of metabolic acidosis and seizures were also noted, prompting further investigation.

Laboratory findings

Laboratory evaluations during hypoglycemic episodes revealed elevated lactate levels and findings consistent with metabolic acidosis. Despite a normal liver size reported by the age of 3 years, hepatomegaly developed thereafter, raising suspicion of an underlying metabolic disorder. Further diagnostic work revealed elevated liver enzymes and abnormal lipid profiles, suggesting a metabolic disturbance.

Imaging studies and additional biochemical testing failed to identify a specific disorder. The patient had been treated for a long time under the suspicion of organic acidemia, with inconclusive results from urine organic acid analysis and other metabolic evaluations. Whole-exome sequencing was requested to identify the underlying condition.

Genetic findings

Genetic testing ultimately confirmed the presence of a pathogenic variant in the PHKA2 gene, leading to the diagnosis of GSD IX. This condition is characterized by the body's inability to properly metabolize glycogen, resulting in hepatic involvement, hypoglycemia, and growth abnormalities. This diagnosis explained the patient's clinical symptoms and guided future management and treatment strategies tailored to his condition.

The case underscores the diagnostic challenges associated with non-specific clinical features and inconclusive initial evaluations. Differential diagnoses included inborn errors of metabolism, mitochondrial disorders, and rare endocrine conditions. Targeted genetic testing (e.g. whole-exome sequencing) and thorough metabolic evaluations were essential in reaching the diagnosis. Eventually, GSD IX should be considered in any patient presenting with recurrent hypoglycemia, metabolic acidosis, and inconclusive metabolic evaluations.

Discussion

This case of a 6-year-old male with GSD type IX offers valuable insights into the variability and complexity of GSDs. GSD type IX, caused by mutations in the *PHKA2* gene that encodes the α subunit of phosphorylase kinase, typically presents with milder symptoms compared to other GSDs. However, as demonstrated in this case, the disease can still lead to significant clinical challenges, including recurrent hypoglycemia, hepatomegaly, and growth retardation [22].

The stepwise diagnostic approach starts with laboratory assessments, including liver function tests, blood glucose levels, lactic acid, triglycerides, and uric acid, as these markers can provide initial clues. However, confirming GSD IX relies heavily on genetic testing, with next-generation sequencing or targeted gene panels able to detect mutations in the *PHKA2* gene, which encodes the α subunit of PhK [4]. A confirmed genetic diagnosis not only validates the clinical suspicion but also guides the management plan, including dietary adjustments. Liver biopsy is less commonly required now but may still be performed in certain cases to assess enzyme activity, particularly in challenging diagnostic scenarios [26].

Once diagnosed, GSD IX management requires a multidisciplinary approach with regular follow-up to monitor liver, cardiac, and neurologic health, as these systems can be variably affected. Liver function tests and imaging (such as ultrasound) should be conducted periodically to monitor hepatomegaly and assess for fibrosis, especially since liver enlargement and related complications can persist into adulthood in some cases. Growth monitoring is essential, as growth retardation may persist without strict dietary management. Cardiac evaluation, including echocardiography, is recommended since GSD IX can sometimes impact cardiac function, even if asymptomatic initially. Neurological assessments, although less critical in GSD IX than in other forms, may be warranted if the patient exhibits developmental delays or muscle weakness [27].

This case illustrates that a personalized approach to dietary management, including a high-carbohydrate and high-protein diet with frequent meals and cornstarch therapy, is essential to stabilize blood glucose and support growth. Educating the family about recognizing hypoglycemia symptoms is crucial, as acute episodes can have significant effects on the child's development and quality of life [28]. The potential for advanced treatments, such as gene therapy and enzyme replacement therapy, also presents exciting prospects for the future, aiming to address the disease's root cause rather than merely managing its symptoms. Research into the molecular mechanisms of GSD continues to expand therapeutic possibilities, offering hope for improved outcomes in GSD type IX and other forms of GSDs [29].

Conclusion

GSD type IX represents a less severe but still significant form of GSD. The case discussed demonstrates the importance of early genetic diagnosis, personalized dietary management, and continuous monitoring to

prevent complications. Advances in research, including gene therapy and enzyme replacement therapy, offer promising future treatment options that may further improve outcomes for patients with GSD type IX.

Lifelong dietary management and regular monitoring are essential to prevent complications like liver cirrhosis and growth failure. In metabolic diseases, potential future treatments may include gene therapy or new enzyme replacement therapies. Educating the family on disease management, dietary restrictions, and hypoglycemia symptoms is crucial for effective home care and improving the patient's quality of life. Ongoing research into genetic therapies and personalized medicine offers hope for more effective treatments for GSD type IX and other metabolic disorders.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.

Funding

This research did not receive any grant from funding agencies in the public, commercial, or non-profit sectors.

Authors contributions

All authors equally contributed to preparing this article.

Conflicts of interest

The authors declared no conflict of interest.

Acknowledgements

The authors acknowledge their colleagues for their warm cooperation.

References

1. Gümüş E, Özen H. Glycogen storage diseases: An update. World J Gastroenterol. 2023; 29(25):3932-63. [DOI:10.3748/wjg.v29.i25.3932] [PMID]

2. Ellingwood SS, Cheng A. Biochemical and clinical aspects of glycogen storage diseases. J Endocrinol. 2018; 238(3):R131-41. [DOI:10.1530/JOE-18-0120] [PMID]

3. Neoh GKS, Tan X, Chen S, Roura E, Dong X, Gilbert RG. Glycogen metabolism and structure: A review. Carbohydr Polym. 2024; 346:122631. [DOI:10.1016/j.carbpol.2024.122631] [PMID]

4. Koeberl DD, Koch RL, Lim JA, Brooks ED, Arnson BD, Sun B, et al. Gene therapy for glycogen storage diseases. J Inherit Metab Dis. 2024; 47(1):93-118. [DOI:10.1002/jimd.12654] [PMID]

5. Yu J, Ling X, Chen L, Fang Y, Lin H, Lou J, et al. Genotypic and phenotypic features of 39 Chinese patients with glycogen storage diseases type I, VI, and IX. Clin Genet. 2024; 106(3):267-76. [DOI:10.1111/cge.14530] [PMID]

6. Chen MA, Weinstein DA. Glycogen storage diseases: Diagnosis, treatment, and outcome. Transl Sci Rare Dis. 2016; 1(1):45-72. [DOI:10.3233/TRD-160006]

7. Anam S, Parakash A, Merchant A. Clinical, biochemical and radiological findings in children with glycogen storage disease. Prof Med J. 2023; 30(10):1324-7. [DOI:10.29309/TPMJ/2023.30.10.7667]

8. Massese M, Tagliaferri F, Dionisi-Vici C, Maiorana A. Glycogen storage diseases with liver involvement: a literature review of GSD type 0, IV, VI, IX and XI. Orphanet J Rare Dis. 2022; 17(1):241. [DOI:10.1186/s13023-022-02387-6] [PMID]

9. Patil SB, Gadad PC. Novel approaches in glycogen storage disease type I management: Harnessing the potential of micronutrients and macro molecules. Int JPharm Investig. 2024; 14(1). [Link]

10. Küçükçongar Yavaş A, Engin Erdal A, Bilginer Gürbüz B, Ünlüsoy Aksu A, Kasapkara ÇS. Assessment of the diagnosis, treatment, and follow-up of a group of Turkish pediatric glycogen storage disease type 1b patients with varying clinical presentations and a novel mutation. J Pediatr Endocrinol Metab. 2023; 36(11):1092-9. [DOI:10.1515/jpem-2023-0336] [PMID]

11. Rakib TM, Islam MS, Tanaka S, Yabuki A, Pervin S, Maki S, et al. Novel mutation in the feline GAA gene in a cat with glycogen storage disease type II (Pompe disease). Animals (Basel). 2023; 13(8):1336. [DOI:10.3390/ani13081336] [PMID]

12. Colella P. Advances in pompe disease treatment: From enzyme replacement to gene therapy. Mol Diagn Ther. 2024; 28(6):703-19. [DOI:10.1007/s40291-024-00733-x] [PMID]

13. Angelini C. Evaluating avalglucosidase alfa for the management of late-onset Pompe disease. Expert Rev Neurother. 2024; 24(3):259-66. [DOI:10.1080/14737175.2024.2306855] [PMID]

14. Jain S, Shah I. Glycogen storage disease-Type 3. Pediatr Oncall J. 2023; 21:45-7. [DOI:10.7199/ped.oncall.2024.8]

15. Patterson MC. Diseases associated with primary abnormalities in carbohydrate metabolism. In: Swaiman KF, Ashwal S, Shevell MI, editors. Swaiman's pediatric neurology-E-Book: Principles and practice. Amsterdam: Elsevier; 2024. [DOI:10.1016/B978-0-323-37101-8.00039-4]

16. Koch RL, Soler-Alfonso C, Kiely BT, Asai A, Smith AL, Bali DS, et al. Diagnosis and management of glycogen storage disease type IV, including adult polyglucosan body disease: A clinical practice resource. Mol Genet Metab. 2023; 138(3):107525. [DOI:10.1016/j.ymgme.2023.107525] [PMID]

17. Stefanik E, Dubińska-Magiera M, Lewandowski D, Daczewska M, Migocka-Patrzałek M. Metabolic aspects of glycogenolysis with special attention to McArdle disease. Mol Genet Metab. 2024; 142(4):108532. [DOI:10.1016/j.ymgme.2024.108532] [PMID]

18. Reason SL, Voermans N, Lucia A, Vissing J, Quinlivan R, Bhai S, et al. Development of continuum of care for McArdle disease: A practical tool for clinicians and patients. Neuromuscul Disord. 2023; 33(7):575-9. [DOI:10.1016/j.nmd.2023.05.006] [PMID]

19. Lucia A, Martinuzzi A, Nogales-Gadea G, Quinlivan R, Reason S; International Association for Muscle Glycogen Storage Disease study group. Clinical practice guidelines for glycogen storage disease V & VII (McArdle disease and Tarui disease) from an international study group. Neuromuscul Disord. 2021; 31(12):1296-310. [DOI:10.1016/j.nmd.2021.10.006] [PMID]

20. Hahn JW, Lee H, Seong MW, Kang GH, Moon JS, Ko JS. Clinical and genetic spectrum of GSD type 6 in Korea. Orphanet J Rare Dis. 2023; 18(1):132. [DOI:10.1186/s13023-023-02750-1] [PMID]

21. Lin WD, Tsai FJ, Wang CH. Identification of a novel deletion mutation in PHKA2 in a taiwanese patient with type IXa glycogen storage disease. Pediatr Neonatol. 2023 ; 64(4):465-7. [DOI:10.1016/j.pedneo.2023.03.005] [PMID]

22. Renuka IV, Ramamoorthy S, Vijayalakshmi B, Srilaxmi N, Kanth BS, Bhimireddy V. Glycogen storage disorder type ixb: exploring clinical patterns and genetic insights into a rare phosphorylase kinase B (PHKB)-associated case. cureus. Cureus. 2024; 16(7):e65474. [DOI: 10.7759/cureus.65474] [PMID]

23. Kido J, Mitsubuchi H, Watanabe T, Sugawara K, Sasai H, Fukao T, et al. A female patient with GSD IXc developing multiple and recurrent hepatocellular carcinoma: A case report and literature review. Hum Genome Var. 2021; 8(1):45. [DOI:10.1038/s41439-021-00172-8] [PMID]

24. Huang K, Duan HQ, Li QX, Luo YB, Bi FF, Yang H. Expanding the clinicopathological-genetic spectrum of glycogen storage disease type IXd by a Chinese neuromuscular center. Front Neurol. 2022; 13:945280. [DOI:10.3389/fneur.2022.945280] [PMID]

25. Noorian S, Aghamahdi F, Rad SS. A novel deletion mutation in the glut 2 gene in a patient with Fanconi Bickel Syndrome. Paper presented at: 57th Annual Meeting of the European society for paediatric endocrinology (ESPE). 27 - 29 September 2018; Athens, Greece. [Link]

26. Szymańska E, Jóźwiak-Dzięcielewska DA, Gronek J, Niewczas M, Czarny W, Rokicki D, et al. Hepatic glycogen storage diseases: Pathogenesis, clinical symptoms and therapeutic management. Arch Med Sci. 2019; 17(2):304-13. [DOI:10.5114/aoms.2019.83063] [PMID]

27. Mathew P, Thoppil D, McClinton T. Hypoglycemia (Nursing). 2022 Dec 26. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024. [PMID]

28. Liu X, Chen H, Zhong Y, Lee TY, Han W, Yu D, et al. Diet therapy in patients with rare diseases: A scoping review. J Hum Nutr Diet. 2023; 36(3):742-53. [DOI:10.1111/jhn.13116] [PMID]

29. Kishnani PS, Sun B, Koeberl DD. Gene therapy for glycogen storage diseases. Hum Mol Genet. 2019; 28(R1):R31-41. [DOI:10.1093/hmg/ddz133] [PMID]

This Page Intentionally Left Blank