

At present, there are several areas of pharmacological research, such as c-kit inhibitors, anti-IgE (omalizumab) and interferon alpha.^{3,6} During acute exacerbations, the dosage of maintenance medications must be increased, adding topical or systemic corticosteroids (1–2 mg/kg/day) and topical sodium cromoglycate or antibiotics, if needed.

Patients must be provided with intramuscular adrenaline due to the risk of anaphylactic reactions, and adrenaline auto-injectors are commercially available.⁴

In conclusion, early diagnosis and a multidisciplinary followup comprising paediatric primary care, dermatology, allergy and haematology/oncology, along with REMA, are needed to improve the management of these patients.

References

1. Rueda ML, Yarza ML, Colina V, Gallego S, Pereira S, Navacchia D, et al. Mastocitosis cutánea: revisión de 10 años de experiencia en el Servicio de Dermatología del Hospital General de Niños Pedro de Elizalde. *Dermatol Argent*. 2011;17:32–9.
2. Lange M, Niedoszytko M, Renke J, Glen J, Nedoszytko B. Clinical aspects of paediatric mastocytosis: a review of 101 cases. *J Eur Acad Dermatol Venereol*. 2013;27:97–102.
3. Heide R, Beishhizen A, de Groot H, den Hollander J, van Doormaal J, Demonchy J, et al. Mastocytosis in children: a protocol for management. *Pediatr Dermatol*. 2008;25:493–500.
4. Molina Garrido MJ, Guillén Ponce C, Molina MJ, Guirado Risueño M. Mastocitosis sistémica. Revisión sistemática. *An Med Interna*. 2008;25:134–40.
5. González de Olano D, de la Hoz Caballer B, Núñez López R, Sánchez Muñoz L, Cuevas Agustín M, Diéguez MC, et al. Prevalence of allergy and anaphylactic symptoms in 210 adult and pediatric patients with mastocytosis in Spain. A study of the Spanish Network on Mastocytosis (REMA). *Clin Exp Allergy*. 2007;37:1547–55.
6. Morren AM, Hoppé A, Renard M, Debiec Rychter M, Uyttebroeck A, Dubreuil P, et al. Imatinib mesylate in the treatment of diffuse cutaneous mastocytosis. *J Pediatr*. 2013;162:205–7.

B. Selva Folch^{a,*}, R. López Almaraz^b,
R. Sánchez González^c, B. Martínez de las Heras^a

^a Servicio de Pediatría, Hospital Universitario de Canarias, La Laguna, Tenerife, Spain

^b Sección de Hematología/Oncología Pediátricas, Hospital Universitario de Cruces, Baracaldo, Vizcaya, Spain

^c Servicio de Dermatología (Dermatología pediátrica), Hospital Universitario de Canarias, La Laguna, Tenerife, Spain

* Corresponding author.

E-mail address: blancaselvafolch@gmail.com

(B. Selva Folch).

Different glycogen storage diseases presenting as abdominal distention and growth and weight retardation[☆]



Distensión abdominal y estancamiento ponderoestatural como forma de presentación de diferentes tipos de glucogenosis

Dear Editor:

Glycogen storage diseases comprise a heterogeneous group of rare hereditary disorders with an approximate incidence of one case per 20,000 live births a year, and are caused by genetic defects in proteins responsible for glycogen synthesis and storage. Several types of glycogen storage diseases have been described that are caused by different genetic changes and whose clinical expression is diverse, with predominance of hepatic and/or muscular involvement.¹

Despite the low prevalence of glycogen storage diseases in everyday paediatric practice, we present two illustrative clinical cases of patients diagnosed with different forms of glycogen storage disease:

Clinical case 1

Boy aged 9 months referred to the paediatrics clinic for evaluation of progressive abdominal swelling. The parents were not consanguineous and had no relevant medical history. There had been a progressive decrease in the weight and length percentiles of the patient from birth to the time of the evaluation, while his psychomotor development was normal. The patient had been exclusively breastfed on demand in the first two months of life, and was subsequently fed artificial formula, with frequent feedings at two to three hour intervals with no breaks at night. Physical examination revealed a weight of 8 kg (3rd–10th percentile), height of 67 cm (3rd–10th percentile), Waterlow height-for-age of 94.3%, weight-for-height z-score of –0.44 (WHO standards), and marked abdominal swelling with hepatomegaly of 9 cm. These findings led to performance of an abdominal ultrasound scan, which revealed uniform hepatomegaly, and a chemistry panel with the following key findings: AST, 1071 U/L (normal range [NR], 0–34); ALT, 573 U/L (NR, 10–49); glucose 23 mg/dL (NR, 70–100); and triglycerides, 772 mg/dL (NR, <110). The patient did not respond to the glucagon stimulation test, with a glucose level before stimulation of 38 mg/dL, followed by 40 mg/dL 30 min after glucagon administration, with no hyperlactaemia. Genetic testing was performed due to suspicion of glycogen storage disease, identifying a defect in the α subunit of phosphorylase kinase, which confirmed the diagnosis of glycogen storage disease type IXa. The patient required continuous nocturnal gastric feeding until age 5 years, when the family succeeded in achieving adequate metabolic control.

[☆] Please cite this article as: Sierra-Poyatos R, Gavela-Pérez T, Blanco-Rodríguez M, Soriano-Guillén L. Distensión abdominal y estancamiento ponderoestatural como forma de presentación de diferentes tipos de glucogenosis. *An Pediatr (Barc)*. 2016;84:288–289.

Clinical case 2

Girl aged 19 months referred to the paediatrics clinic for failure to thrive between 15 and 18 months of life. The parents were not consanguineous, were originally from Bangladesh, and had no relevant medical history. The patient had a varied diet adequate for her age, and she continued to breastfeed on demand. Her psychomotor development was normal. Physical examination revealed a weight of 9.045 kg (3rd–10th percentile), height of 77 cm (3rd–10th percentile), Waterlow height-for-age of 95%, weight-for-height z-score of -0.73 SDS (WHO standards), and marked abdominal swelling with hepatomegaly of 8 cm. The abdominal ultrasound scan evinced a uniform hepatomegaly. The salient findings of the chemistry panel were AST, 1858 U/L; ALT, 1029 U/L; glucose, 56 mg/dL; and triglycerides, 257 mg/dL. In the glucagon test, the glucose level before administration of glucagon was 28 mg/dL, and at 30 min it was 40 mg/dL without hyperlactacidaemia. Genetic testing was requested for suspected glycogen storage disease, which identified amylo-1,6-glucosidase deficiency, leading to diagnosis of type IIIa glycogen storage disease. She required continuous nocturnal gastric feeding for a period of two weeks. Subsequently, thanks to extensive family involvement, the patient achieved adequate metabolic control with feedings at 4 h intervals in the daytime, and two cornstarch feedings during the night.

The presence of progressive abdominal swelling in an infant requires ruling out hepatomegaly, among other conditions. In some cases, considerable liver enlargement that can reach as far as the iliac crest is difficult to feel with palpation and may go unnoticed,² as described in the two cases.

A differential diagnosis of the multiple aetiologies of hepatomegaly can be performed by history taking, physical examination and diagnostic tests.² In the cases presented here, in addition to hepatomegaly and the marked abdominal swelling, the failure to thrive and the presence of hypertransaminasaemia, hyperlipidaemia and hypoglycaemia with abnormal results in the glucagon test led us to suspect a diagnosis of glycogen storage disease.³

Glycogen storage disease type III is caused by a deficiency of amylo-1,6-glucosidase, also known as glycogen debranching enzyme, an enzyme involved in glycogenolysis whose deficiency leads to accumulation of limit dextrins. In 85% of cases, this deficiency affects the liver and muscle tissues

(subtype IIIa).⁴ Glycogen storage disease type IX results from a defect in the activation of phosphorylase kinase, which is also involved in glycogenolysis. Different mutations may occur in the genes of each of the subunits that compose the enzyme (α , β , γ , δ) with variable presence in different tissues. X-linked glycogen storage disease type IXb (XLG) is the most frequent form and only involves the liver.⁵

The main goal of treatment is to prevent hypoglycaemia. This requires avoiding prolonged fasting periods by the frequent intake of slow-release carbohydrates throughout the day, and in some cases, especially in infants, nocturnal gastric feedings.⁶

References

1. Cabral A. Glucogenosis. In: Sanjurjo P, Baldellou A, editors. *Diagnóstico y tratamiento de las enfermedades metabólicas hereditarias*. Madrid: Ergon; 2001. p. 161–72.
2. Wolf AD, Lavine JE. Hepatomegaly in neonates and children. *Pediatr Rev*. 2000;21:303–10.
3. Wolfsdorf JI, Weinstein DA. Glycogen storage diseases. *Rev Endocrinol Metab Disord*. 2003;4:95–102.
4. Hershkovitz E, Forscher I, Mandel H, Spiegel R, Lerman-Sagie T, Anikster Y, et al. Glycogen storage disease type III in Israel: presentation and long-term outcome. *Pediatr Endocrinol Rev*. 2014;11:318–23.
5. Beauchamp NJ, Dalton A, Ramaswami U, Niinikoski H, Mention K, Kenny P, et al. Glycogen storage disease type IX: high variability in clinical phenotype. *Mol Genet Metab*. 2007;92:88–99.
6. Smit GPA, Rake JP, Akman HO, DiMauro S. The glycogen storage diseases and related disorders. In: Fernandes J, Saudubray JM, Walter JH, editors. *Inborn metabolic diseases*. 4th ed. Berlin: Springer; 2006. p. 101–20.

R. Sierra-Poyatos^a, T. Gavela-Pérez^b,
M. Blanco-Rodríguez^b, L. Soriano-Guillén^{b,*}

^a *Servicio de Endocrinología y Nutrición, Instituto de Investigación Sanitaria Fundación Jiménez Díaz, Universidad Autónoma de Madrid, Madrid, Spain*

^b *Servicio de Pediatría, Instituto de Investigación Sanitaria Fundación Jiménez Díaz, Universidad Autónoma de Madrid, Madrid, Spain*

*Corresponding author.

E-mail addresses: lsoriano@fjd.es, leansom4@hotmail.com (L. Soriano-Guillén).

Elective extubation during skin-to-skin contact in the extremely premature newborn[☆]



Extubación electiva durante el contacto piel con piel en el prematuro extremo

[☆] Please cite this article as: Camba F, Céspedes MC, Jordán R, Gargallo E, Perapoch J. Extubación electiva durante el contacto piel con piel en el prematuro extremo. *An Pediatr (Barc)*. 2016;84:289–291.

Dear Editor:

As evidence has been growing on the benefits of kangaroo care,¹ the practice of skin-to-skin contact has spread through neonatal units and is being implemented in more patients, including extremely preterm newborns.²

Cardiorespiratory parameters are more stable during skin-to-skin contact.^{3,4} Studies in extremely preterm newborns have demonstrated the safety of skin-to-skin contact during mechanical ventilation,⁵ and one study conducted in term newborns that had undergone surgery found greater stability in cardiorespiratory parameters following extubation in infants that had been put in skin-to-skin contact with their parents.⁶