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Glucose and galactose malabsorption: A new case in Spain☆

Malabsorción de glucosa y galactosa. Nuevo caso en España

Dear Editor:

Congenital diarrhoea refers to a heterogeneous group of enteropathies that cause symptoms from the first few days of life, and it may be the only manifestation or one of the symptoms of a systemic disease. In most cases, early treatment is necessary to prevent dehydration, given the potential complications of the latter. The first step in the differential diagnosis is to differentiate between secretory and osmotic diarrhoea: in the former, the diarrhoeal output does not improve with a nil per os trial, whereas in the latter it does.[1]

We present the case of a male newborn aged 2 days admitted to the neonatal unit with hyperbilirubinaemia. The parents were Pakistani and reported consanguinity. The newborn was a product of a monochorionic-diamniotic twin pregnancy and had been born second at 36 weeks' gestation with a weight of 2460 g. He was receiving artificial formula following the wishes of the mother. On admission to the neonatal unit, the physical examination revealed a 15% weight loss, jaundice extending to Kramer's zone 3 and a dry oral mucosa, with no other abnormal findings. Blood tests revealed hyperbilirubinaemia on account of elevation of indirect bilirubin and dehydration with hypernatraemia and hyperchloraemia (urea, 49 mg/dL; creatinine, 1.5 mg/dL; sodium, 163 mEq/L; chloride, 132 mEq/L) and metabolic acidosis (pH, 7.30; bicarbonate, 18.7 mM; base excess, 7 mM; lactate, 3.9 mM). Phototherapy was initiated, along with placement of a peripheral catheter for intravenous fluid replacement, while the patient continued to receive artificial formula. The diarrhoeal output continued to be very high, leading to prescription of nil per os, which achieved resolution of the diarrhoea. The analysis of stool samples revealed an acidic pH and the presence of reducing bodies. In the following days, several attempts were made to reintroduce oral feedings, first with elemental formula and later with a soy-based formula, with diarrhoea recurring in both instances as the feeding volume increased. Eventually, the newborn was given a fructose-based formula with no other carbohydrates, to which he responded favourably. Increases in feeding volume did not create problems, and the boy started producing normal stools and exhibiting adequate weight gain.

In this case, since the patient improved with fasting, the diarrhoea was classified as osmotic, and the presence of reducing bodies in the stool suggested that the congenital diarrhoea could be due to carbohydrate malabsorption. Since the patient did not improve with the soy formula, we suspected glucose–galactose malabsorption,[1] as soy formulas do not contain galactose but do contain glucose. In fact, upon switching to a formula in which the sole carbohydrate was fructose, the newborn responded well, which supported our clinical suspicion.

Congenital glucose–galactose malabsorption (cGGM) is a rare autosomal recessive disorder characterised by changes in the *SLC5A1* gene that encodes the protein SGLT1, responsible for transporting glucose and galactose from the intestinal lumen into intestinal cells.[2] Sequencing of this gene in the patient revealed the homozygous mutation c.875G>A (p.C292Y), previously described as a pathogenic variant, as it impacts the location and function of the protein,[3] thus confirming the diagnosis. Microsatellite genotyping confirmed that the twins were dizygotic, which explained the clinical differences between the two.

Congenital glucose–galactose malabsorption is characterised by severe diarrhoea and dehydration from the first day of life, which can quickly result in the death of the infant if glucose, galactose and the disaccharides that contain them are not removed from the diet.[4] Approximately 300 cases have been diagnosed worldwide.[5] Treatment consists in the elimination of glucose and galactose from the diet, substituting a fructose-based formula, which achieves resolution of diarrhoea. Some authors have proposed that cGGM improves over time as the intestinal flora adapts, and it appears that administration of *Lactobacillus acidophilus* helps shorten the time that carbohydrates need to be restricted.[6] However, there are no prospective studies in the literature establishing how to identify which patients will develop tolerance and which will remain intolerant for life, so we currently recommend that older children and

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adults consume a diet where fructose is the sole source of carbohydrates until tolerance can be evaluated.

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Advances in the diagnosis of ocular toxoplasmosis: Use of optical coherence tomography☆

Novedades para el diagnóstico de la toxoplasmosis ocular: uso de la tomografía de coherencia óptica

Dear Editor:

Ocular toxoplasmosis (OT) is the leading cause of posterior uveitis in immunocompetent patients. The primary infection is asymptomatic in most cases, and therefore OT is commonly diagnosed in subsequent reactivations manifesting with characteristics chorioretinal scars (white-yellow lesions with greyish edges, usually unilateral).

From a clinical standpoint, paediatric patients may present with retinochoroiditis and be asymptomatic, with blurry vision due to vitreitis or with bloodshot eyes and ocular pain, frequently associated with anterior uveitis. The pathogenesis of such reactivations remains unclear.[1]

The diagnosis is made based on the described clinical manifestations combined with compatible serological findings. Most patients have low IgG titres and negative IgM titres, so IgG avidity or increased titres may be useful for diagnosis.[2] Challenging cases may require the use of invasive techniques, such as polymerase chain reaction (PCR) analysis for detection of *Toxoplasma gondii* in aqueous or vitreous humour[3]; however, this is considered an aggressive technique.

We present 3 cases of acquired OT in paediatric patients, describing characteristic signs that can be assessed by optical coherence tomography (OCT) and that contributed to the early and accurate diagnosis of the disease.

Case 1: girl aged 8 years from Equatorial Guinea that presented with acute unilateral anterior uveitis manifesting with eye pain and redness. The patient underwent immunological testing and screening for infections including serological tests for antibodies against Toxoplasma, with a negative result for IgM antibodies and a positive result for IgG antibodies. The fundus appeared normal in the initial examination. In subsequent evaluations, the patient had elevation of IgG antibodies and the OCT revealed retinitis with stalagmite-like raised lesions in the inner perimacular surface, suggestive of toxoplasmosis[4] (Fig. 1).

Case 2: boy age 12 years of Spanish descent presenting with headache and loss of visual acuity (0.8). Examination of the fundus revealed focal chorioretinitis focal with vitreitis, compatible with toxoplasmosis, while serological testing for antibodies against Toxoplasma was negative for IgM and positive for IgG. Subsequent evaluations revealed a 4-fold increase in IgG titres. Treatment started with antibiotherapy and systemic steroid therapy. The outcome was parafoveal scarring with absence of photoreceptors and small retinal cysts, with normal visual acuity (Fig. 2).

Case 3: girl aged 10 years of Moroccan descent presenting with panuveitis with acute granulomatous anterior uveitis in the left eye. The ophthalmological evaluation by means of OCT revealed areas of inflammation, signs of vitreitis and inflammation of the inner layers of the retina. Ocular toxoplasmosis was suspected, leading to serological testing, which was positive for IgG, with a titre of more

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