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Severe neurodevelopmental and growth delay in immigrant child



Grave retraso del neurodesarrollo y estatura en niña inmigrante

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We present the case of a girl newly arrived from Morocco, a country without universal newborn screening, who presented in the emergency department at age 9 years with severe global neurodevelopmental delay and dwarfism. The parents reported chronic constipation and delayed milestones, with head control and sitting achieved at age 4 years, standing at age 7 years, the first steps at 9 years and no speech to date. Her height was 90 cm (< 1st percentile; z score -7.6), her weight 15 kg (< 1st percentile; z score -2.55), her body mass index18.5 kg/m² (55th percentile; z score, +0.15), and she exhibited hearing loss, facial myxedema, macroglossia, prominent lips, loss of the outer third of the eyebrows, abdominal distension, umbilical hernia, hyporeflexia and flexion contracture of the lower extremities (Fig. 1). She received a diagnosis of thyroid agenesis with a free thyroxine level of less than 0.08 ng/dL, a thyrotropin level greater than 1000 mU/L, a thyroglobulin level of less than 0.04 ng/mL and absence of tracer uptake on scintigraphy. She also had elevation of low-density lipoprotein (LDL) cholesterol (185 mg/dL). Treatment with levothyroxine achieved normalization of bowel movements,

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Figure 1 Phenotype observed in the girl: loss of outer third of the eyebrows, facial edema, macroglossia, abdominal distension, umbilical hernia.

improvement of contracture and walking and resolution of myxedema and hypercholesterolemia.

The arrival of immigrants who had no access to newborn screening and presenting with intellectual disability and slow linear growth should spur suspicion of congenital hypothyroidism. Thyroid hormone replacement therapy can resolve some of its manifestations and is available at any level of care.¹⁻³

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Declaration of competing interest

The authors have no conflicts of interest to declare.

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