Question: 170

A 6-month-old baby whose family has just emigrated from Ecuador is brought to your office by the maternal grandmother. Physical examination reveals length at the 5th percentile, weight at the 10th percentile, head circumference at the 25th percentile, a sallow complexion with jaundice, hoarse cry, dry skin, and large tongue. The anterior fontanelle measures 3x4 cm. You diagnose primary hypothyroidism and start appropriate thyroid hormone replacement therapy.

Of the following, the MOST likely long-term outcome in this baby is

A. adrenal insufficiency
B. microcephaly
C. normal adult height
D. normal intellectual function
E. precocious puberty
The baby described in the vignette has congenital hypothyroidism and has not received early treatment with thyroid hormone. Fortunately, congenital hypothyroidism now usually is diagnosed soon after birth by newborn screening programs in the United States, but this screening may not be available in resource-limited countries. Children who have severe hypothyroidism at birth tend to have poorer intellectual outcomes than their siblings, even if therapy is started in the first few days after birth. However, earlier therapy with larger doses of thyroid hormone leads to improved intellectual outcomes, even in the most high-risk athyreotic group of infants. Despite the guarded prognosis for intellectual outcome, with continued and appropriate thyroid hormone therapy, this child should grow normally in height, and adult height should be appropriate for the family.

Congenital hypothyroidism is not associated with adrenal insufficiency, although acquired hypothyroidism because of chronic lymphocytic thyroiditis may be associated with the development of autoimmune adrenal insufficiency (Addison disease). Microcephaly is an unusual outcome for children treated appropriately with thyroid hormone. Indeed, brain catch-up growth in the first postnatal year and a somewhat more flattened basal skull can give affected children larger head circumferences, split sutures, and the appearance of macrocephaly with increased intracranial pressure. On the other hand, overtreatment with thyroid hormone can lead to more rapid fusion of the cranial sutures and microcephaly, so treatment must be monitored with careful and frequent laboratory measurements of thyroid-stimulating hormone and thyroid hormone (free thyroxine [T4] or total T4 if free T4 is not available). The potential for normal intellectual function in this child is guarded. At the least, he is likely to have learning disabilities and some cerebellar problems, including clumsiness and decreased fine motor skills. Precocious puberty is not an outcome of congenital hypothyroidism unless a child is overtreated with thyroid hormone and develops premature maturation, which is very unusual. Some children who have severe acquired hypothyroidism have manifested signs of sexual precocity that disappear after treatment of the hypothyroidism (Van Wyk-Grumbach syndrome).

References:


Postellon DJ, Bourgeois MJ, Varma S. Congenital hypothyroidism. eMedicine Specialties, Pediatrics: General Medicine, Endocrinology. 2007. Available at:
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