Pediatric Neurology 94 (2019) 82–83

Contents lists available at ScienceDirect

Pediatric Neurology

journal homepage: www.elsevier.com/locate/pnu

Visual Diagnosis

Macroglossia, Dry Skin, Developmental Delay, and Stippled Epiphysis: A Treatable Condition

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ARTICLE INFO

Article history:
Received 29 December 2018
Accepted 1 January 2019
Available online 4 January 2019

Keywords:
Congenital hypothyroidism
Endocrine disorder
Short stature
Epihyseal dysplasia

This two-and-a-half-year-old girl presented with delayed attainment of developmental milestones since late infancy. There was no history of seizures or developmental regression. She was born to non-consanguineous parents after an uncomplicated perinatal period. There was no similar illness in other family members. On examination, she had a developmental age of 12 to 15 months, short stature (72 cm, −5*z* score), normal weight and head size, wide open anterior fontanel, frontal bossing, delayed dentition, macroglossia, and dry skin (Fig A,B). Rest of the systemic examination was unremarkable. The possibility of lysosomal storage disorders and congenital hypothyroidism was considered.

Investigations showed markedly increased thyroid stimulating hormone of 1236 μIU/mL (normal value, 0.27 to 4.2), low total T4 of 0.756 μg/dL (normal value, 4.8 to 12.7), and total T3 of 0.559 ng/mL (normal value, 0.8 to 2). A radiograph of the knees showed small, stippled epiphyses (Fig C). She began levothyroxine (3 μg/kg/day). At her two-month follow-up, she had gained developmental milestones, and her thyroid stimulating hormone level was 12.2 μIU/mL.

Congenital hypothyroidism is one of the most common endocrine disorders with prevalence rate of 1:2000 to 4000 live births.[1] This condition is often underdiagnosed at birth because of subtle clinical manifestation. The clinical signs and symptoms include postdatism, high birth weight, neonatal hyperbilirubinemia, lethargy, hoarse cry, constipation, coarse facial features, large or late fusion of anterior and posterior fontanel, macroglossia, umbilical hernia, protuberant abdomen, and cold dry skin.[1] Thyroid hormone is also required for skeletal maturation and femoral epiphysis may be absent in about 50% to 60% affected newborns.[2] Delayed diagnosis can lead to devastating long-term effects, including developmental delay, short stature, and delayed puberty. Recognition of this potentially treatable disease by newborn screening can prevent long-term consequences and burden on the society.

References

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Conflict of interest: The authors declare no conflicts of interest.

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https://doi.org/10.1016/j.pediatrneurol.2019.01.001

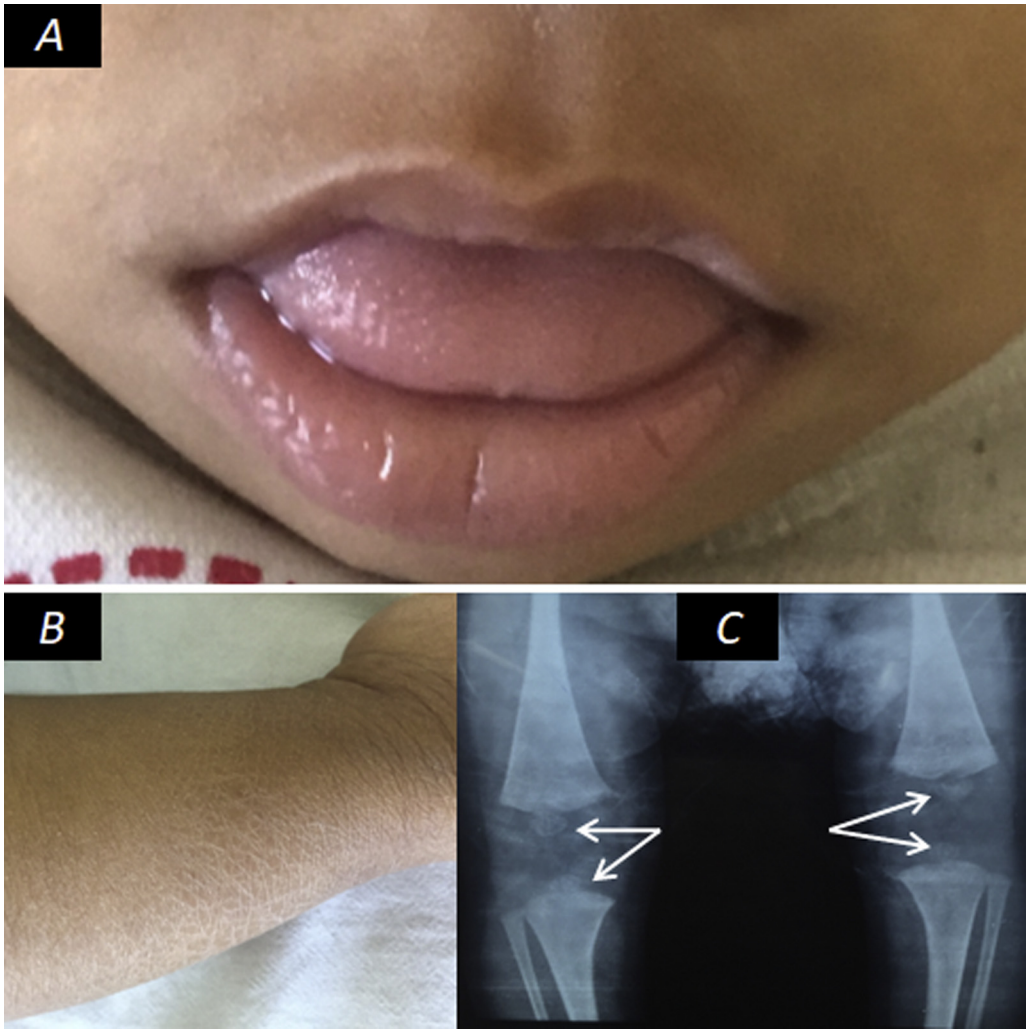

FIGURE. Photograph depicts a large protruded tongue (A) and dry skin (B). Plain radiograph of the knees shows small, stippled epiphyses (arrows) (C). The color version of this figure is available in the online edition.