

Clinical Quiz

What is the Diagnosis?

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Case Report – Presentation

We present the case of a three-year-old female child, who underwent evaluation in our paediatric endocrinology outpatient clinic due to "congenital hypothyroidism" and short stature.

Born at 37 weeks gestational age with low birthweight, she adapted poorly to extrauterine life, with constant groan and axial hypotonia, for which she was admitted to the neonatal intensive care unit. During her stay, endocrine studies showed progressively increasing high Thyroid-stimulating Hormone (TSH) levels (10 mIU/L; 14 mIU/L)

and decreasing low Free Thyroxine (FT4) levels (10.42 pmol/L; 9.14 pmol/L) - no anti-thyroid antibodies were detected and the thyroid ultra-sound was normal. Congenital hypothyroidism was diagnosed and, at 2 months of age, she began levothyroxine (LT4) therapy.

When evaluated at the age of three, even though short stature was not present (Height P25%) and growth was apparently not impaired (Growth Velocity P10-25%), the patient's physical examination unveiled several unique characteristics – obesity, brachydactyly, round facies and hard subcutaneous nodules (Figure 1). A complete family history revealed that all of these



Figure 1 Index patient. (A) General habitus; (B and C) Subcutaneous nodules; (D) Hands – brachydactyly with evident shortening of the distal phalanx of the thumb.

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physical characteristics were also present on other family members, particularly her mother (Figure 2), one maternal uncle and her maternal grandfather.

The re-evaluation of previous lab results as well as new lab, histologic and radiologic studies revealed important information: Neonatal thyroid hormone studies suggested

an insensitivity to TSH's action; current high Parathormone (PTH), low vitamin-D and hypocalcemia suggested an insensitivity to PTH action; Ossifications were identified on the subcutaneous nodules' histology; a hand X-ray confirmed the clinical evidence of brachydactyly (Figure 3).



Figure 2 Index patient and her mother. (A) General habitus; (B) Hands side-by-side – Brachydactyly with evident shortening of the distal phalanx of the thumb.

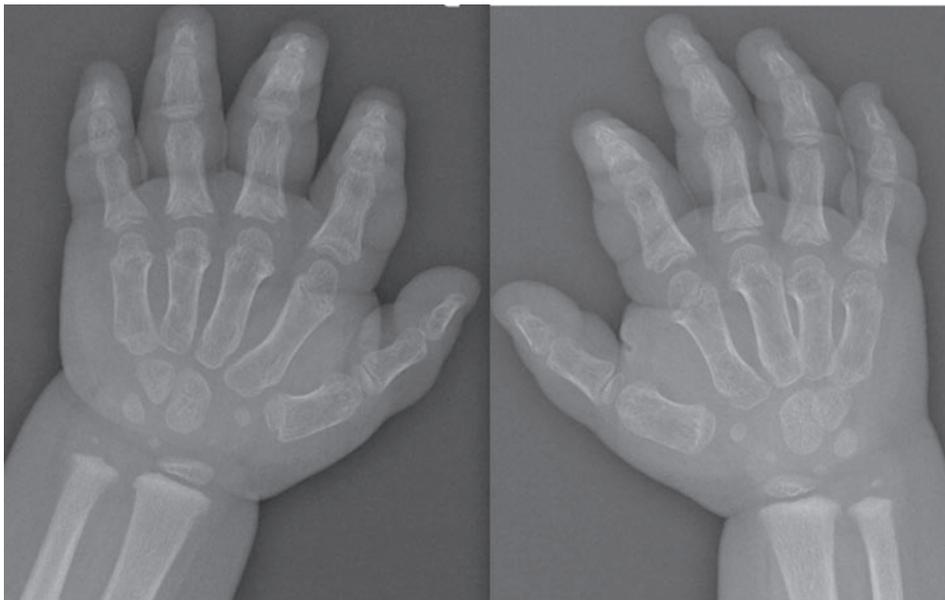


Figure 3 The index patient's hand X-ray showing the characteristic brachydactyly.

Answer to "Clinical Quiz" on Pages 262-265

N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.