

HYPOTHYROIDISM IN A BOY WITH DOWN'S SYNDROME

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A 13-year-old boy (Panel A) with Down's syndrome was admitted to our hospital for evaluation of fatigue, palpitations and bouts of cyanosis. Shortness of breath and dyspnea on exertion appeared one month prior to admission. Physical examination showed clinical features typical for Down's syndrome, facial edema and dry and scaly skin. He was extremely obese with BMI 38.5 kg/m² (+3.9 SDS) and very short, with height 114.0 cm (-5.7 SDS). On palpation his thyroid gland was normal. His heart rate was 96 beats/min, with distant cardiac sounds, absent apex beat and normal blood pressure. The ECG showed QRS microvoltage with flattened T segments, a chest X-ray revealed cardiomegaly. An echocardiogram demonstrated a large pericardial effusion (diameter 11 mm) with collapse of the right atrium during diastole. Laboratory analysis revealed elevated cholesterol of 8.03 mmol/l and triglycerides of 1.95 mmol/l. The serum free thyroxin of 1 pmol/l (12-22 pmol/l), thyroid-stimulating

hormone of 942.9 mU/l (0.20-4.20 mU/l) and high titer of antithyroperoxidase antibodies of 1:303.4 U/ml (<35 U/ml) indicated hypothyroidism caused by autoimmune thyroid disease. Thyroid ultrasound was normal. His estimated bone age was 4 years. The patient was treated with L-thyroxine sodium, which was gradually increased to 75 µg daily. Within a few months he lost weight and became more alert; furthermore, the symptoms of hypothyroidism and the pericardial effusion were resolved. Regular evaluation of thyroid function tests is obligatory in children with Down's syndrome.

Key words: Hypothyroidism ▪ Down Syndrome ▪ Pericardial effusion

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