

Van Wyk-Grumbach syndrome – rare manifestation of a common disease

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INTRODUCTION Van Wyk-Grumbach syndrome (VWGS) is characterized by prolonged primary hypothyroidism, isosexual precocious pseudopuberty, delayed bone age with either enlarged multicystic ovaries in girls or enlarged testes in boys. Pathophysiology is most likely based on similarity of TSH and FSH glycoproteins and its binding to FSH receptors leading to precocious pseudopuberty. **CASE REPORT** A 8.5-year old girl was referred to endocrinologist due to hypothyroidism and obesity. She experienced fatigue, weakness, weight gain and growth retardation for 1.5 years prior to referral. At the examination she was short 110 cm(-3.7 SDS), obese 40.5 kg(+1.75 SDS) with bradycardia c/p 58/min; edematous, with dry skin, thin hair and hoarse voice. Breasts were Tanner II-III, with no pubic or axillary hair. Menarche occurred at the age of 8.3 years. Labs: TSH >100 mU/L(ref. 0.5-4.7), fT4 2.6 nmol/L(ref. 9-19), FSH 3.9 IU/L(ref. 0.4-3.5), anti-TG 18 IU/mL(ref. <4.1), anti-TPO 250 IU/mL(ref. <5), LH 0.1 IU/L(ref. 0.7-2.2), E 2 62 pmol/L(ref.<37), prolactin 43.3 µg/L(ref. 4.2-23.3). Estimated bone age was 6 years. Thyroid ultrasound revealed atrophic autoimmune thyroiditis. Pelvic ultrasound showed endometrial thickening with enlarged cystic ovaries. Subsequently, she was diagnosed with VWGS and L-thyroxine substitution was introduced. After 5 months on L-thyroxine she grew 7.5 cm, lost 7 kg and was clinically euthyroid. **CONCLUSION** Autoimmune thyroid disease is one of the most common endocrine disorders, characterized by gradual onset and relatively mild symptoms. VWGS represents its rare complication. Early recognition of VWGS and introduction of treatment is important in order to prevent unnecessary diagnostic and surgical procedures, and achieve excellent prognosis.