ASSOCIATION OF VAN WYK GRUMBACH AND DEBRE SEMELAIGNE SYNDROMES IN TWO CASES WITH SEVERE HYPOTHYROIDISM

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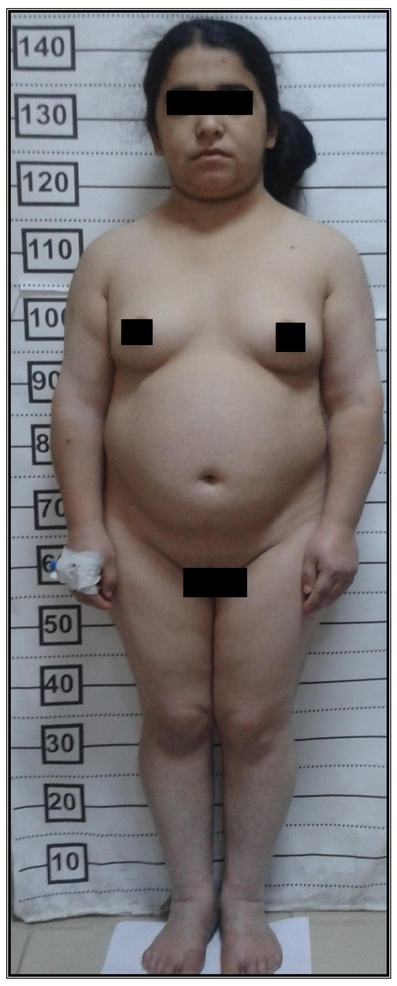
Introduction

Van Wyk Grumbach (VWG) and Kocher Debre Semelaigne (KDS) Syndromes are rare syndromes with clinical manifestation of hypothyroidism associated with precious pseudo puberty and myopathic pseudomuscular hypertrophy. We present two cases that have the characteristic of both VWGS and KDSS syndromes developed in association with a long term untreated hypothyroidism.

Case 1

Seventeen years old girl was referred due to menstrual irregularities and multicystic ovaries. In her medical history, she was assessed for vaginal bleeding, and diagnosed congenital hypothyroidism when she was 5. But she was noncompliant with her treatment. At the age of 11, her menstrual cycle started again, but she complains oligomenorrhea. On her physical examination, she had coarse facial appearance and marked hypertrophy of both calves. Her pubertal development was in stage 4, and she had all the clinical signs of severe hypothyroidism such as mental retardation and short stature (Figure 1,2). Her bone age was consistent with the age of 16. In laboratory evaluation TSH level was >100 mIU/mL, fT4 0.3 ng/dL, blood creatinine and creatinine kinase (CK) levels were 1.21 mg/dL and 254 U/L respectively.



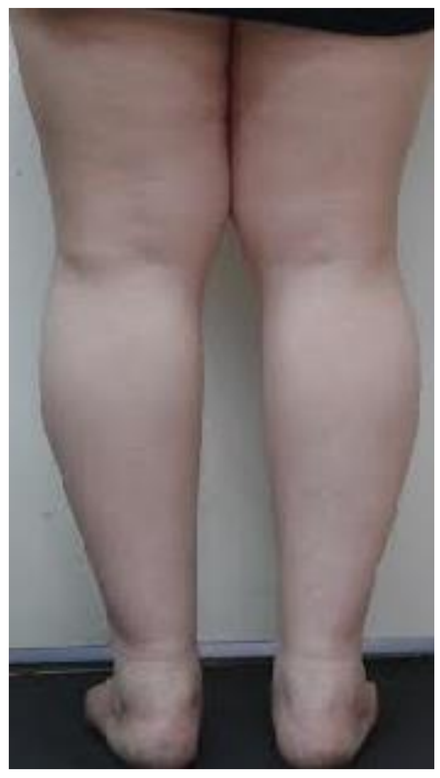
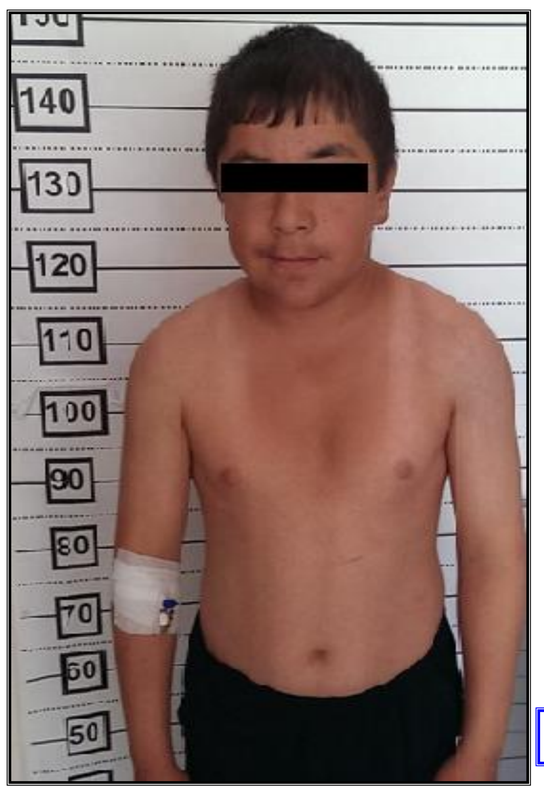


Figure 1,2: Appearances of case 1

Case 2

Thirteen years old boy was referred to our clinic because of severe muscle pain, hypothyroidism and goiter. His physical examination showed that his weight was 45 kg (10-25 p), and his height was 149 cm (3-10 p). He had dry skin, coarse facial features, grade 2 goiter and muscular pseudohypertrophy (Figure3,4). Both testes were 25 ml in size. Laboratory investigations showed TSH 100 mIU/mL, fT4 0.2 ng/dL, Thyroid peroxidase antibody >1072 IU/mL, LH 0.67 µIU/mL, FSH 3.22 mIU/mL, T. Testosteron 2.01 ng/mL, CPK 2537 U/L and serum creatinin level was 1,15 mg/dL.



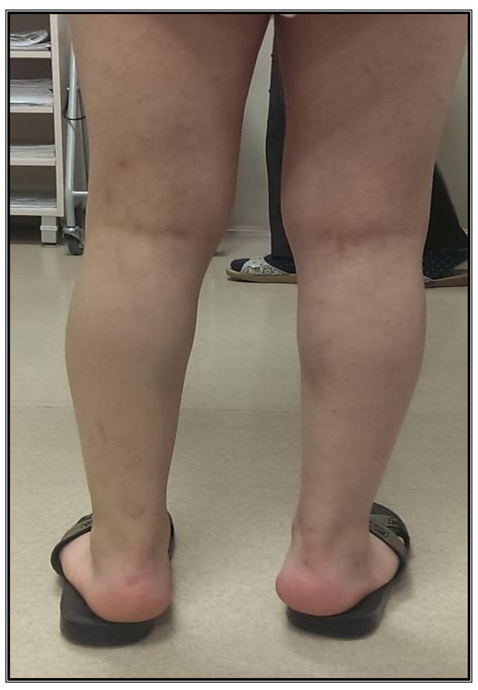


Figure 3,4: Appearenges of case 2

Conclusion

Due to severe hypothyroidism, pseudoprecocious puberty was diagnosed in the first case, whereas macroorchidism was diagnosed in the second one. Therefore, congenital and acquired hypothyroidism causes should be considered in differential diagnosis of primary muscle diseases, which are presented by muscle pain, muscle hypertrophy with elevated CK levels, and thyroid function tests should be controlled in those cases.