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Elevated DHEAS and Acute Hair Loss in an Adult Male with Trichorhinophalangeal Syndrome Type 1: a Case of Male PCOS

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Background: Male androgenic alopecia is a common cause of hair loss in both men and women. Although genetic and hormonal factors likely play a role, the etiology is mostly unknown. PCOS is a highly heritable disease with elevated prevalence of reproductive abnormalities in first-degree relatives (FDRs) of patients with PCOS, which may be due to an underlying genetic defect in steroidogenesis. A recent meta-analysis revealed that male FDRs of patients with PCOS had significantly increased levels of DHEAS compared with controls. We present a young adult male who developed acute hair loss with biochemical evidence of hyperandrogenism. **Clinical Case:** A 21-year-old man presented for evaluation of hyperandrogenism after experiencing progressive loss of posterior patch of hair over 3 weeks. He also endorsed acne, intermittent headaches, striae, and difficulty losing weight for the past 2 years. He denied facial rounding, vision changes, and use of glucocorticoids or supplements. Physical examination was notable for height of 67 inches, BMI 30.5 mg/m 2, BP 120/78, recession of the frontotemporal and occipital hairline, sparse posterior scalp hair, bulbous nose, long philtrum, mild facial acne, 2, 3 toe syndactyly, and thin purple striae on bilateral flanks and axillae. Labs revealed elevated DHEAS 1065.7 mcg/dL (ref. 238.4-539.3) and normal 8 AM total testosterone of 382 ng/dL (240-871). Cosyntropin stimulation test revealed baseline cortisol level of 10.6 mcg/dL with ACTH of 22.2 pg/mL (5-46) and 60-minute cortisol level of 24.8 mcg/dL, 17-OH progesterone level 81 ng/dL (18-164), and 17-OH pregnenolone 1040 ng/dL (55-455). Workup for Cushing's revealed multiple normal midnight salivary cortisol levels and mild intermittent elevation of 24-hour urinary free cortisol levels. CT scan of the adrenals was negative for a mass. Exome sequencing revealed a heterozygous de novo pathogenic variant (c.1614_1615delTCinsAT: p. C538X) in the TRPS1 gene (NM_0141112.2). Family history was significant for a 20-year-old sister with PCOS and

elevated DHEAS of 443 ug/dL (51-321). Mid-parental height was 69.6 inches. **Conclusion:** Trichorhinophalangeal syndrome Type 1 (TRPS1) is a rare genetic disorder characterized by hair, craniofacial, and skeletal abnormalities such as androgenetic alopecia, bulbous nasal tip, and cone-shaped epiphyses. To our knowledge, the hair findings in TRPS are not associated with hyperandrogenism which prompted further investigation. Evaluation for endocrine causes of hyperandrogenism such as Cushing's disease, congenital adrenal hyperplasia, and adrenal tumor were negative. However, our patient's elevated DHEAS is likely due to male PCOS, as previously described in male FDRs of patients with PCOS. Thus, genetic susceptibility to PCOS and rare genetic syndromes should be considered in the differential diagnosis of young men with clinical and biochemical evidence of hyperandrogenism. Reference: Shan D et al. Reproductive Health in First-degree relatives of Patients with Polycystic Ovarian Syndrome: a Review and Meta-analysis. JCEM. 2022 Jan;107(1); 273-295.

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