ANHIDROTIC ECTODERMAL DYSPLASIA WITH DIABETES MELLITUS, HYPERTENSION, OBESITY AND HYPOGONADISM

Anhidrotic ectodermal dysplasia is a rare genetic disorder principally affecting the males. It need not always be determined by an X-linked recessive gene.[1] The salient clinical features include absent or reduced sweating, hypotrichosis and total or partial anodontia with characteristic facies.[2] Incomplete forms may occur. There may be many associated abnormalities. There are also many unclassified syndromes with ectodermal dysplasia.[2]

Recently, a 16-year-old male was admitted with cellulitis of the leg. He was a known case of essential hypertension and insulin dependent diabetes mellitus. His skin was dry, smooth and hypohidrotic. Scalp hair were normal but eyebrows, eyelashes and body hair were sparse. Moustache, beard, axillary hair and pubic hair were absent. Forehead was square and prominent, nasal bridge was sunken with lips being large and everted. There was hypodontia. Nails, mucosae, palms and soles were normal. Body weight was 82 kg, height 142 cm, span 144 cm with poorly developed penis and testes. His intelligence quotient was 45. Family members were normal.

Serum cortisol level and dexamethasone suppression test ruled out Cushing's syndrome. X-ray skull and contrast enhanced head scan showed no abnormality. T_3, T_4 and TSH levels were normal. FSH was 0.1 MIU/ml, LH 3.0 MIU/ml, prolactin 2.0 ng/ml, testosterone 10 ng/dl and growth hormone 1.2 ng/ml. Serum cholesterol was 190 mg%, triglyceride 259 mg%, HDL 32 mg%, VLDL 51.8 mg%, LDL 106.2 mg% and cholesterol : HDL ratio 5.9. These findings confirmed hypogonadotrophic hypogonadism and obesity of central origin. ECG showed left axis deviation. Investigations confirmed diabetes mellitus. Fundoscopy showed grade III hypertensive retinopathy with early background diabetic retinopathy. Skin biopsy showed normal epidermis without appendages.

The diagnosis of anhidrotic ectodermal dysplasia with diabetes mellitus, essential hypertension, hypogonadotrophic hypogonadism and obesity of hypothalamic origin was made. Presence of normal scalp hair, absence of episodes of hyperpyrexia and chest infections were features against the classical variety of the disorder. Also, such abnormalities have not so far been reported, in association with this condition except primary hypogonadism.[3]

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References

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