HYPOHIDROTIC ECTODERMAL DYSPLASIA WITH MITRAL VALVE PROLAPSE
A Case Report

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Summary

Clinical and investigational features of a case of congenital hypohidrotic ectodermal dysplasia in a young boy are described. In addition, he was found to have mitral valve prolapse, which was confirmed by echocardiography. The available literature is discussed. Such an association, though probably fortuitous, has not been previously reported.

KEY WORDS: Ectodermal dysplasia, Hypohidrotic Ectodermal dysplasia, Mitral valve prolapse, Pili torti.

Introduction

Hypohidrotic ectodermal Dysplasia is an uncommon congenital disorder. Only a few reports are available from this country[1,2]. The disorder results from faulty evolution of the epiblastic layer of the blastoderm, probably due to an injury, during the third month of intra-uterine life[3]. It is characterised mainly by hypo-odontia, hypotrichosis and hypo-or anhidrosis[4]. Abnormalities of the eyes and the central nervous system have been documented[5]. The disease is transmitted as an X-linked recessive trait with full expression only in males.

Mitral valve prolapse is a recently recognised entity[6]. The aetiology is variable, though in some it is congenital[7]. Recent review shows the incidence to be around 17 percent of valvular diseases of the heart[8].

We report a patient with congenital ectodermal dysplasia of the hypohidrotic type with co-existing mitral valve prolapse.

Case Report

A 15 year old male (IP 4363/81) was admitted to the hospital on 4th February 1981 for inability to stand hot weather since childhood, occasional attacks of palpitation for 5 years and fever for 10 days.

The patient's main complaint was fever which was of insidious onset, being higher during the day. It was not associated with rigors, sweating, or other systemic symptoms.

Patient was born at term in a municipal hospital and had normal milestones. His father had noticed that whenever the child bathed with warm water he remained febrile for the day and cried

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listlessly. The child had no convulsions and his mother prudently avoided warm water.

At the age of ten years the boy noticed that he could not play in the open, because he tired easily and developed burning sensation all over with a feeling of unbearable heat in his head. He could lessen such discomfort by wearing clothes dipped in cold water. He had also noticed that he had only few teeth and sparse hair.

Palpitation was first noticed by him also around ten years of age, while drawing water from a well one day. It lasted for less than half a minute. Subsequent spells of palpitation each lasting for less than half a minute occurred after exertion. These paroxysms were never associated with symptoms referred to the central nervous system. There was no history of chest pain or a history suggestive of rheumatic fever in the past.

Patient's parents are first cousins and he is their first and only child. The mother sweated normally except on the palms where it was reduced. She and members of both the paternal and maternal families were in normal health. There was no family history of heart disease.

Examination revealed a thin boy 128 cms tall weighing 30 Kg. Scalp hair and eyebrows were sparse and of light colour. Pubic hair was also sparse. The scalp hair showed pili torti. The periorbital skin was hyperpigmented and wrinkled. There was frontal bossing and low set ears (Fig. 1). The lips were thick and everted, lower with a mucocoele. The eyes were normal.

The skin was generally thin, dark and dry. But not hyperelastic. Skin over the palms and soles was thick and dry. There was no telangiectasia, and the nails were normal.

Examination of the oral cavity revealed six widely set and peg shaped teeth, (Fig. 1). The lower jaw, gums, palate, tongue and pharynx were found to be normal.

Examination of the cardiovascular system showed a regular pulse with a rate of 86/minute. The blood pressure was 110/70 mm of mercury. The cardiac impulse was felt at the fourth intercostal space, half an inch medial to the mid-clavicular line. The first and the second heart sounds were normal. There was a mid-systolic click over the apical area. The click was of high pitch and well heard over the apex in the left lateral position and on standing. Moderate exercise changed the mid-systolic click into an early systolic one. There were no murmurs or adventitious sounds. The aortic, pulmonary and tricuspid areas were normal.

Fig. 1
Photograph of the face of the patient showing the characteristic features.

Clinically there was no evidence to suggest cardiomegaly or cardiac failure. All the peripheral pulses were symmetrical and normal. Rest of the system review was normal.

Patient was investigated. Urine analysis and haemogram were within normal limits. Serum cholesterol was 119 mg percent. Examination of the urine for aminoacids was negative. Serum proteins and electrophoresis were normal. Slit lamp examination did not reveal arcus senilis or lenticular subluxation. The visual acuity was 6/6 in both the eyes. The fundi were normal. X-rays of the chest and the skull were normal. The electrocardiograms in supine and standing postures were normal. The ECG was repeated after moderate exercise. It did not reveal any T wave abnormalities. Blood VDRL was non-reactive. An audiogram was done which showed no hearing loss.

Skin biopsies were done on the palm as well as on the forearm. Microscopic examination revealed similar changes in both the specimens but were more marked in the forearm skin. The epidermis showed mild acanthosis and hyperkeratosis. The dermis contained fewer than normal sweat glands in the palmar skin and none in the forearm specimen. Pilosebaceous units were absent (Fig. 3). Patient's mother did not allow biopsies from her skin.

Discussion

Congenital hypohidrotic ectodermal dysplasia (CHED) as an entity has been recognised for more than 100 years[9], and exhaustively classified with the associated anomalies[10].

A combination of mitral valve prolapse and CHED has not been recorded[8,11]. We think the association to be fortuitous.

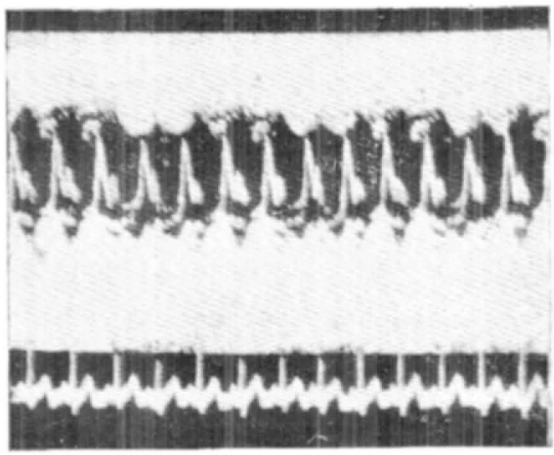

Fig. 2
Photograph of the echocardiograph showing mitral valve prolapse (after exercise).

An echocardiogram on a single crystal M-mode machine showed posterior excursion of the posterior mitral leaflet (PML) during ventricular systole. The posterior excursion of the PML occured during late systole when the patient was resting and became pan-systolic after moderate exercise (Fig. 2).

This combination has clinical importance as the patient not only faces the ill effects of CHED but also those of the mitral valve disease. The latter could result in sudden death[7,8]. The patient's mother with diminished palmar sweating, was obviously a carrier[12,13].

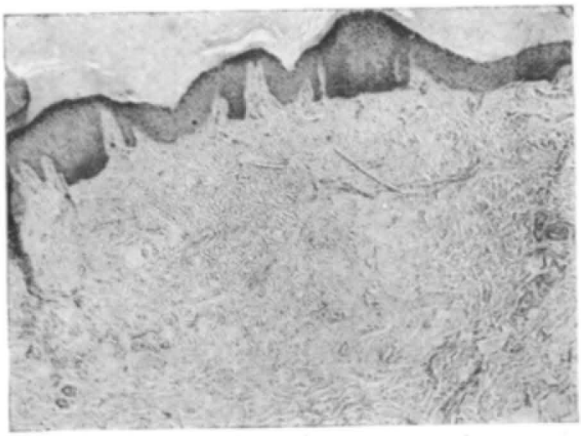

Fig. 3

Photomicrograph of the palmar skin specimen showing reduced number of sweat glands. Even those present (right extreme) show small ill defined duct system.

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