WERNER'S SYNDROME

S Pradeep Nair, M Vijayadharan, Avirup Gupta

A 41-year-old unmarried man presented with erosions, ulcers and keratosis of tendoachilles. He was short statured and had features of premature aging, sclerosis of the skin of the limbs and hypogonadism. The head was small with a beak -like nose. Investigations revealed diabetes mellitus, generalised osteoporosis, calcification of blood vessel walls and microsplanchia of kidneys. All the features suggested a diagnosis of Werners's syndrome.

Key words: Pangeria, Werner's syndrome, Premature aging.

Werner's syndrome (WS) or Pangeria is an adult premature aging syndrome of autosomal recessive inheritance affecting connective tissues through out the body.[1] The exact aetiopathogenesis remains obscure even though biochemical and connective tissue abnormalities have been postulated.[2] The disease involves multiple systems of the body and may be associated with internal malignancy.

Case Report

A 41-year-old unmarried male patient presented with erosions, ulcer and keratosis of both tendoachilles. He also complained of premature greying of scalp hair and baldness, along with hoarseness of voice. For the past 15 years he was getting recurrent erosions and ulcers of the feet. He had undergone bilateral cataract surgery at the age of 29 years. The family history of the patient indicated a second degree consanguinity of his parents. His younger brother had similar disease and had died of coronary heart disease 7 months back. His other three siblings including one sister were normal.

The physical examination of the patient showed a short stature with muscular atrophy of the extremities, a weight of 20 kg and he appeared old for his age.

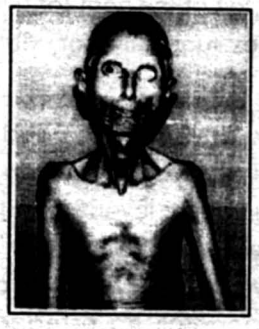

Figure-1. Werner's syndrome. Note corneal leucoma, beak-like nose.

The head was small with a beak-like nose and the scalp had scanty grey hair (Fig. 1).

From the Department of Dermatology and Venerology, Medical College Hospital, Thiruvananthapuram-655 011, India.

Address correspondence to Dr. S Pradeep Nair

The left eye had a corneal leucoma and the right eye was aphakic. The skin of the limbs was sclerosed with thin tapering digits and keratosis of both elbows(Fig.2).

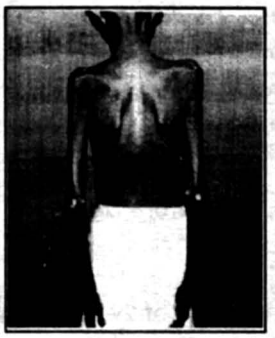

Fig.2 note keratosis of elbows and wasting of muscles of upper limbs

The skin of the trunk and limbs also showed areas of hyperpigmentation and depigmentation. The patient had hypogonadism with scanty axillary and pubic hair. The region of the tendoachilles showed keratosis, multiple erosions, and secondary eczematisation. The feet were small with dystrophic nails and shortening of the first and fifth digit of the right foot. There were no features suggestive of Hansen's disease.

Investigations recorded a normal blood hemogram, liver and renal function tests. Blood sugar levels indicated diabetes mellitus . Serological tests for syphilis and HIV were negative.X-Rays of the limbs showed generalised osteoporosis and blood vessel wall calcification. Ultrasound of abdomen showed microsplanchia of the kidneys, but no evidence of malignancy. ENT examination showed reduced movement of left vocal cord. Skin biopsy showed epidermal atrophy and nonspecific features of the dermis. The patient was given symptomatic treatment.

Discussion

The presentation with classical clinical features of short stature, premature aging, sclerodermatous skin, eye lesions, hoarseness of voice, hypogonadism, non healing ulcers of feet, d iabetes, X-ray finding of osteoporosis of limbs and vessel wall calcification and ultrasound finding of microsplanchia enabled us to make a diagnosis of Werner's syndrome. WS is relatively a rare disorder of low incidence. About 400 cases have been reported world wide since its first publication.[3] The tissues of patients with WS show non-uniform premature aging, atheroma of the blood vessels and microsplanchia. In addition, altered connective tissue metabolism and abnormalities of immune and endocrine systems have been described.[4] There is increased incidence of malignancy in WS, the usual malignancy being fibrosarcoma, though other sarcomas and organ specific malignancies have been reported.[5] However in our case there was no evidence of malignancy but careful and periodic follow up of the patient may be required as malignancies can occur later on . The cells of the patient s with WS show chromosomal instability and a spontaneous hypermutability with a high rate of deletions.[6] This factor may be responsible for

the premature aging and high incidence of malignancy in these patients. The sclerodermatous skin and blood vessel wall calcification contribute to the development of non healing ulcers and keratosis of the limbs. it i s mandatory and important to recognise the clinical features of WS as early as possible to rule out internal malignancies and for the purpose of genetic counselling.

References

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