Pigmented xerodermoid - report of three cases

Jayanta Kumar Das, Asok Kumar Gangopadhyay
Department of Dermatology, Vivekananda Institute of Medical Sciences and Ramakrishna Mission Seva Pratisthan Hospital and 99, Sarat Bose Road, Kolkata - 700 026, India.

Address for correspondence: Dr. Jayanta Kumar Das, Flat BE 3, 193 Andul Road, Howrah - 711109, W.B., India. E-mail: jayanta_das@hotmail.com

ABSTRACT

Pigmented xerodermoid, a rare genodermatosis, presents with clinical features and pathology similar to xeroderma pigmentosum, but at a later age. DNA repair replication is normal, but there is total depression of DNA synthesis after exposure to UV radiation. Two siblings in their teens and a man in his thirties with features of pigmented xerodermoid, e.g. photophobia, freckle-like lesions, keratoses, dryness of skin, and hypo- and hyper-pigmentation, are described. Although classically the onset of pigmented xerodermoid is said to be delayed till third to fourth decade of life, it seems the disease may appear earlier in the tropics. Early diagnosis and management could be life-saving.

KEY WORDS: Xeroderma pigmentosum.

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease in which the patient typically lacks the normal capacity to repair UV radiation-damage to DNA.[1] XP is a genetically and clinically heterogeneous disease. Genetically, there are at least eight subtypes designated complementation groups A-G, and XP variant (pigmented xerodermoid). Pigmented xerodermoid, reported in the early seventies,[2,3] is aptly described as xeroderma pigmentosum of late onset.[1] In pigmented xerodermoid, the onset is classically delayed till the third or fourth decade of life; and repair replication is normal, but there is depression of DNA synthesis after exposure to UV radiation.[1] Three cases of pigmented xerodermoid are presented here for the rarity of this disease, especially in the Indian literature.[4]

CASE REPORTS

Case 1
A 16-year-old boy coming from a poor rural household presented with the complaint of gradually increasing number of ‘freckles’ on the face for about 5 years. Initially, the lesions were on the forehead and nose; then they gradually spread to the cheeks, neck, forearms, trunk, and legs. There was also history of photophobia and burning sensation of the skin on sun-exposure. There was no history of consanguinity in the family. The younger sister of the patient (case 2) had similar but milder lesions. The parents as well as the other brother (aged 18 years) did not have similar skin lesions.

On examination, he was found to have numerous
localized small (1-4 mm) hyperpigmented macules on the face as well as on all sun-exposed areas, e.g. extensors of the upper extremities, trunk (more on the upper part), and legs (Figure 1). There were hypopigmented macules interspersed with the hyperpigmented macules. A few of the hyperpigmented lesions were hyperkeratotic and looked clinically like seborrheic keratoses. Additionally, there was dryness of the skin and a diffuse brown-black pigmentation on the exposed parts for about the same duration. All the skin changes were more marked on areas that were exposed to sunlight. Ophthalmological examination was normal except conjunctival redness in both the eyes. General examination including neurological examination was normal. Histopathological examination of a hyperkeratotic lesion resembling seborrheic keratosis showed acanthosis, hyperkeratosis and papillomatosis, and that of one hyperpigmented macule showed elongation of rete ridges, increased melanin in melanocytes and basal cells, and melanophages in the upper dermis.

Case 2
A 13-year-old-girl, sister of case 1, came with the complaint of ‘freckles’ on the face for about 2 years. The lesions were mostly on the forehead and nose; only a few pigmented macules were found on the cheeks, neck, and forearms; and there was a diffuse brown-black pigmentation on the exposed parts for about the same duration. There was history of photophobia and burning sensation of the skin on sun-exposure. Ophthalmological examination showed conjunctival redness, but was otherwise normal. General examination including neurological examination was normal. Histopathological examination of a hyperpigmented macule showed elongation of rete ridges, increased melanin in melanocytes and basal cells, and melanophages in the upper dermis.

Case 3
A 33 year-old-male presented with the complaints of freckle-like lesions and spotty hypopigmentation over the face and sun-exposed areas of the body and generalized dryness of the skin developing for the past 12 years. There was also history of photophobia and burning sensation of the skin on sun-exposure. Initially, the skin lesions were on the forehead and nose; then they spread to the cheeks, neck, forearms, trunk, and legs (Figure 2). There were a few hyperkeratotic hyperpigmented papules that looked clinically like seborrheic keratoses. There was no history of consanguinity in the family. There was no history of similar skin lesions in the family, including the parents and the only sister. Ophthalmological examination showed...
conjunctival redness, dryness, and pigmentation. General examination including neurological examination was normal. Histopathological examination of a hyperpigmented macule showed features similar to case 2.

In all the three cases, palms, soles, hair, nails, and oral and genital mucosae were normal. Haemogram, routine urine analysis, and blood biochemistry were within normal limits. There was no history of arsenic contamination in the drinking water of the patients’ home-towns, and no arsenic was found in the hair, nail, and urine of the patients, or in the water they used to drink. The patients were treated with strict avoidance of direct sunlight, sunscreens and glycolic acid topically, 6% on the face and 12% on the body.

DISCUSSION

In classical XP, the median age of onset of the cutaneous symptoms is between 1 and 2 years.[5] The sun-exposed skin becomes dry, pigmented and parchment-like, with freckle-like hyperpigmented macules. Premalignant actinic keratoses develop at an early age. Patients with XP under 20 years of age have greater than 1000-fold increased risk of cutaneous basal cell or squamous cell carcinoma or malignant melanoma.[5]

Our cases fit into the classical description of pigmented xerodermoid or ‘xeroderma pigmentosum of late onset’.[1] Although the first two cases had symptoms appearing from the second decade, rather than the third or the fourth, of life, this might be attributed to the fact that they belonged to a poor rural peasant family, and that entailed a lot of recreational as well as occupational sun-exposure from early life. The clinical presentation of these siblings was very similar, the younger sister having milder disease that could be accounted for by the 3-year age difference and the fact that girls generally cover a greater part of their trunk. The third case was classical in having symptoms in the third decade. None of them had any actinic keratoses, or malignancies, or neurological manifestations, but long-term follow up will be needed. Genetic studies to pinpoint the exact nature of defect in our cases could not be done due to lack of facility.

REFERENCES