

Dyschromatosis Symmetrica Hereditaria Associated with Idiopathic Torsion Dystonia

A Case Report

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The authors report a case of a family in which a diagnosis of dyschromatosis symmetrica hereditaria was established by a clinical pattern of cutaneous lesions and by assessment of cellular DNA repair synthesis. The skin lesions were characterized by a mixture of hyperpigmented and hypopigmented macules and were localized on the back of the feet of three patients (two male brothers and one sister). All the patients also had small freckle-like pigmented macules on their face. The father presented large symmetrical hypopigmented vitiligo-like macules. In this patient, mottled pigmentation and depigmentation of the extremities had been present since childhood. In the four patients no cellular abnormalities in DNA repair ability were found. These data exclude a mild form of xeroderma pigmentosum. The daughter, a 9-year-old girl, had since the age of 7 also shown a neurological disorder diagnosed as idiopathic torsion dystonia. The authors emphasize the association of dyschromatosis symmetrica hereditaria, a rare cutaneous disease, with idiopathic torsion dystonia, a rare idiopathic neurological disease, in this young girl. **Key word:** *Deoxyribonucleic-acid repair*.

(Accepted August 30, 1993.)

Acta Derm Venereol (Stockh) 1994; 74: 135–137.

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Dyschromatosis symmetrica hereditaria (DSH) (1), also called symmetric dyschromatosis of the extremities (2) or acropigmentation symmetrical of Dohi (3), is a rare genodermatosis which appears to be caused by an autosomal dominant gene (2).

Pinpoint and pea-sized hyperpigmented and hypopigmented

macules are localized on the back of the hands and feet and sometimes on the limbs. Scattered small pigmented macules are localized on the face.

DSH appears in infancy or early childhood and usually becomes strongly marked in summer. Recently Nishigori et al. (4) reported a case of xeroderma pigmentosum with clinical features of DSH and proposed assessment of DNA repair levels to establish an accurate diagnosis of DSH and exclude a mild form of xeroderma pigmentosum. No cellular abnormalities in DNA repair ability were found in a previous study of three cases of DSH (5).

We report here the case of a family in which we established a diagnosis of DSH by assessment of DNA repair levels. The proband was also affected by neurologic symptomatology, represented by idiopathic torsion dystonia (ITD) (6, 7).

FAMILY CASE REPORT

The proband is a nine-year-old Caucasian girl, born after an uneventful pregnancy and delivery. Psychomotor development was normal. At the age of 7, she presented a left foot focal dystonia on walking, followed, one year later, by the appearance of a right writing cramp and a dystonic posture with athetoid movements on the left hand. On our first examination, at the age of 7, in addition to dystonia she exhibited a mixture of hyperpigmented and hypopigmented macules, irregular in size and shape, localized on the back of her hands (Fig. 1) and the top of her feet. The skin lesions had appeared at the age of 2 and had become more pronounced after sun exposure. Many scattered small freckle-like pigmented macules were localized on her face (Fig. 2). Since all biochemical, laboratory and instrumental examinations, as well as the girl's intelligence, were normal, we established a diagnosis of IDT for the neurological clinical pattern.

The girl's father, 37 years old, had noticed asymptomatic scattered small pigmented lesions on the back of his hands and top of his feet since childhood. On our examination he presented asymptomatic large symmetrical hypopigmented vitiligo-like macules, localized around his eyes and mouth, on his knees, on his penis and on the back of his hands. These hypopigmented macules had progressively widened after the age of 28. He was neurologically normal.

Two male brothers, aged 15 and 14, neurologically normal, showed freckles on their faces and limbs and mottled pigmentation and depigmentation on the back of their hands (Fig. 3) and the top of their feet but milder than in their sister. The lesions had first been noted in the early childhood.

Laboratory investigations concerning the members of this family were normal. There were no ophthalmologic abnormalities or other cutaneous and mucosal alterations. Hair and nails were normal.

Phototesting for minimal erythema dose, performed by using Waldmann-UV 800 with 10 UVB lamps Philips TL12 on all patients, showed a normal response.

The level of UV-induced DNA repair synthesis was evaluated in lymphocytes and fibroblasts of the four patients. This study was carried out in the laboratories of the Institute of Genetics of the National

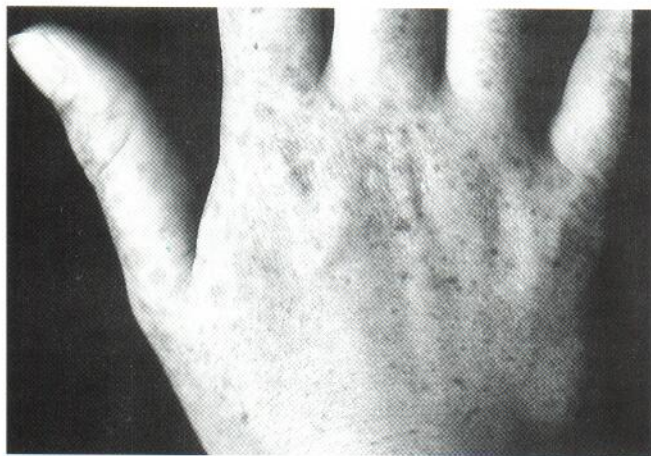


Fig. 1. Right hand detail of the 9-year-old girl. Note the irregularity of the macules in size and shape.

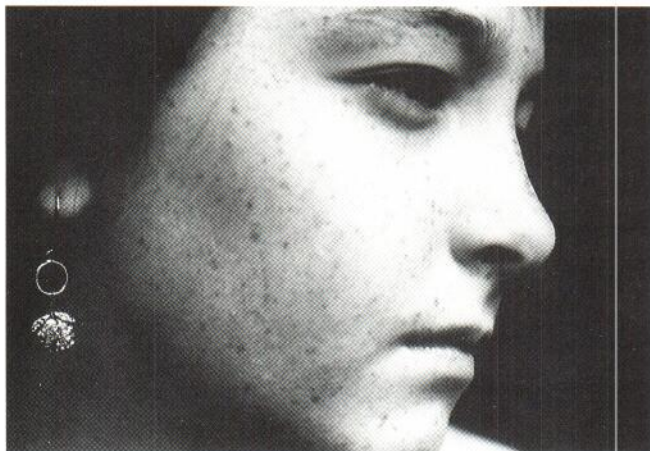


Fig. 2. Face of the 9-year-old girl. Note the scattered small freckle-like pigmented macules on her cheek.

Council of Research in Pavia. The method of research has been previously described (8, 9). All the cellular parameters analysed were within the normal range.

DISCUSSION

In 1986, Nishigori et al. (4) reported a case with the clinical appearance of DSH, but they accurately diagnosed the skin lesions as xeroderma pigmentosum studying the cellular DNA



Fig. 3. Back of the left hand of the 15-year-old brother. Note the less strongly marked mottled pigmentation and depigmentation macules.

repair capacity. These authors proposed that only patients with normal DNA repair levels and typical clinical features should be diagnosed as having DSH. This report clearly pointed out the infrequency of DSH. In fact cases clinically diagnosed as DSH may be mild clinical features of xeroderma pigmentosum with complementation group such as E (10) or F (11) or G (12).

The family members in our study were investigated by lymphocytic DNA synthesis and evaluation of survival and RNA synthesis of dermal fibroblastic cells after UV irradiation, which ruled out the diagnosis of xeroderma pigmentosum.

The infrequency of DSH in the Caucasian race (13) justifies the report on this family, in which DSH shows an autosomal dominant pattern of inheritance, as previously reported in most cases (2, 5). Skin manifestations were typical and limited to sun-exposed areas in all patients, but the vitiligo-like cutaneous lesions of the father were very atypical and represented a possible clinical evolution of the disease or an association with a true vitiligo in this patient. In the daughter (proband of the family-tree) the association of a rare cutaneous disease (DSH) with a rare idiopathic neurological disease (ITD) is very interesting and has not previously been reported in the literature as far as we know.

ACKNOWLEDGMENTS

We are particularly grateful to Prof. P. Nuzzo, director of the Institute of Genetics of the National Council of Research in Pavia, and her collaborators M. Stefanini, P. Lagomarsini and S. Ferrari for their contribution to this report. They studied the UV-induced DNA repair synthesis, the most important analysis in the diagnosis of DSH.

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