Pigmented hypertrichotic dermatosis: manifestations of a rare syndrome

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DESCRIPTION

A Moroccan 19-year-old patient, from a non-consanguineous marriage, with a history of diabetes mellitus since the age of 8 years, presented at the age of 14 year with hypertrichosis and hyperpigmentation on the upper inner thighs, with involvement of the genitalia, trunk and limbs. The physical examination showed symmetrical pigmented hypertrichotic skin patches with induration of thighs and lower limbs with sparing of knees and popliteal fossa (figure 1), an orbital proptosis, musculoskeletal abnormalities including flat feet, scoliosis, clinodactyly and short stature, inguinal lymphadenopathy and hepatosplenomegaly.

The patient had low haemoglobin level at 9 g/dL, elevated laboratory markers of inflammation included an elevated erythrocyte sedimentation rate (ESR), C-reactive protein (CRP) and raised serum immunoglobulin levels. He had had a positive anti-glutamic acid decarboxylase (anti-GAD) 65 antibody titre consistent. Biopsy specimens from the skin showed a chronic inflammatory perivascular infiltrate composed of lymphocytes and plasma cells, with no histological sign in favour of an autoimmune disease. No cardiac abnormalities were detected on echocardiography, ultrasound morphology of inguinal lymph nodes showed benign lymphadenopathy and radiography found pes planus (figure 2).

In view of associated cutaneous lesions and articular distortions, an H syndrome was suspected and genetic counselling was requested. The karyotype study did not demonstrate chromosomal abnormalities. Molecular analysis was carried out: the patient was not carrying the mutations already described in two Moroccan patients (c.243delA at exon 2 and 300+1G>C at the second intron of the SLC29A3 gene) and sequencing of the entire gene could not be achieved due to lack of resources. Nevertheless, the diagnosis of H syndrome was retained in light of the concordance with all the abnormalities outlined above.

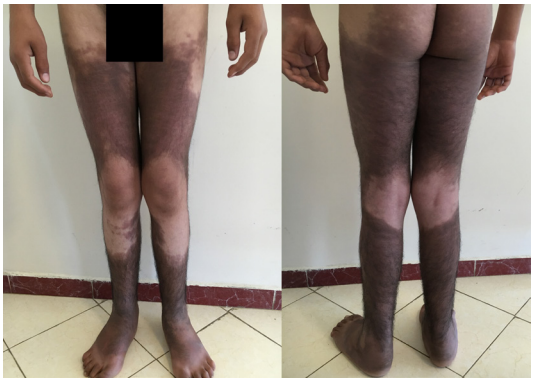

Figure 1 Hyperpigmented patches over thighs and limbs with associated hypertrichosis.

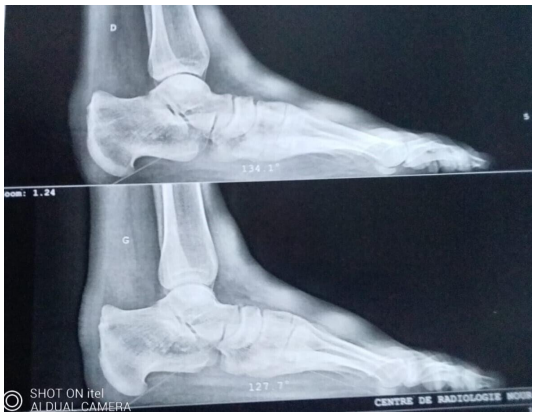

Figure 2 Weightbearing lateral view on radiography showing pes planus with angle of the longitudinal arch increased to more than 130°.

First described in Israël in 2008,[1] H syndrome is an autosomal recessive genodermatosis, whose manifestations are induced by mutations in the SLC29A3 gene encoding the nucleotide transport protein hENT3 and the underlying aetiology remains unknown.[2] To date, 130 cases have been reported in the literature around the world and most of them come from low-income population groups where consanguinity is common.[3] This entity is mainly characterised by hyperpigmented, indurated and hypertrichotic skin with variable systemic features including insulin-dependent diabetes mellitus (IDDM), sensorineural hearing loss, hepatosplenomegaly, hypogonadism, heart

Learning points

- H syndrome is a rare autosomal recessive disorder which is one of a spectrum of diseases all resulting from a mutation in the SLC29A3 gene.
- Diagnosis should be made at an early stage to avoid unnecessary immunosuppressive treatment and provide an appropriate genetic counselling, moreover.
- It would be necessary to develop a simplified process to make genetic analysis more affordable for patients from low-income countries.

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anomalies, flexion contractures of the interphalangeal joints, short stature, hallux valgus, foot deformities, episcleritis, exophthalmos, lymphadenopathy and anaemia. Sometimes, certain of these conditions may lack in 85%.[4]

Reports have shown that along with H syndrome, three others diseases are caused by recessively mutations in the SLC29A3 gene: pigmented hypertrichosis with IDDM syndrome, familial Rosai-Dorfman disease and Faisalabad histiocytosis.[5] Supportive care is recommended considering the absence of specific treatment.

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