Unusual facial lesions in H syndrome

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Abstract

H Syndrome is a rare genodermatosis. It may include facial involvement such as : facial telangiectasia, both hypo- and hyperpigmented lesions, hirsutism, swollen cheeks due to subcutaneous infiltration and eczematous lesions. We describe a new facial phenotype with dermoscopic and histological features in the spectrum of non-Langerhans cell histiocytosis.

Introduction :

H Syndrome is an autosomal recessive genodermatosis due to mutation in SLC29A3 with multisystem involvement. Skin lesions are characteristic with hyperpigmentation, hypertrichosis and progressive skin sclerosis of the lower part of the body. We report a new skin manifestation in the face of a young girl with H Syndrome.

Case report :

A 12 year-old girl, born from consanguineous marriage, presented with a 5-year progressing bilateral symmetrical hyperpigmented and thickened patches with hypertrichosis. These lesions were present in the inner thighs as well as pubic and lumbar regions. On examination, she had swelling with hyperpigmentation of labia majora, finger and toe flexion contracture (camptodactily and hallux valgus) (Figure 1). She also had been explored for hearing loss in the early childhood with hearing aids. Ophtalmological examination showed chorioretinal atrophy. Biological data showed low hemoglobin (9,5g/dl) and high erythrocyte sedimentation rate (75mm/h). The rest of laboratory tests including thyroid function tests, lipid profile, liver function tests, renal function tests and antinuclear antibodies profile were all normal. Echocardiography and electrocardiogram were normal. Abdominal ultrasound found small uterus and ovaries with thickened skin and subcutis of vulva. Genetic analysis revealed homozygous missense mutation c.1088G>A ; p.Arg363GIn in exon 6 in linkage disequilibrium with the same non-pathogenic variant at intron 2 c.300+3A>G. H Syndrome was diagnosed based on histological, immunohistochemical (IHC) examination and molecular analysis. Erythematous, annular and figurate lesions slightly keratotic without atrophy was present in cheeks and nose (Figure 2). Dermoscopy revealed multiple telangiectasias drawing a reticulated network (Figure 3). Mycological test was negative. A 4-mm punch biopsy showed perifollicular focal para-keratotic hyperkeratosis of the epidermis. The dermis was slightly edematous and contain ectatic capillaries. There was a lymphocytic and histiocytic infiltrate (Figure 4). IHC was positive for CD68 (Figure 5). The diagnoses of porokeratosis of Mibelli and sub acute lupus erythematous were excluded. We concluded in a clinical feature of H Syndrome.

Discussion :

A wide range of cutaneous lesions were reported in patients with H Syndrome. Based on increased CD68+ histiocytes, this syndrome is considered as a histiocytic disorder (non-Langerhans cell histiocytosis). (1) (2) The commonest are hyperpigmentation, hypertrichosis and skin induration involving especially lower limbs but also other parts of the body including the face. (3,4) Authors described less common facial manifestations. These features include : facial telangiectasia (4) (2) (5), both hypo- and hyperpigmented lesions (6), hirsutism (1), swollen cheeks due to subcutaneous infiltration (4) and eczematous lesions (1).

Our case is particular. The girl had not only classic features of H Syndrome but also additional manifestations. To our knowledge, annular keratotic and figurate lesions of the face has not been described before. The histiocyte CD68+ infiltrate indicates a novel phenotype of the same pathologic process. Our patient presented gluteal lipodystrophy with cutaneous and subcutaneous thickening which is a less frequent feature in this syndrome (4). The reduced size of uterus and ovaries may be secondary to the deep histiocyte infiltrate. Chorioretinal atrophy is also an unusual ophtalmological abnormality.

Conclusion :

H Syndrome is a rare genodermatosis and approximately 100 cases had been reported in the world. We describe a new facial phenotype with dermoscopic and histological features in the spectrum of non-Langerhans cell histiocytosis.

Author contribution:

RM and BM: wrote the manuscript. BE and SK: revised the manuscript. CS and BT: wrote parts of the manuscript related to the histopathological aspects of the disease. RM, BE, SK and TH: contributed to the management of the patient and revised the article. CH: wrote parts of the manuscript related to genetics. TH: critically reviewed the manuscript and gave final approval. All authors have read and approved the final manuscript and agree to take full responsibility for the integrity and accuracy of the work.

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