International Journal of Pediatrics and Adolescent Medicine (2015) 2, 96—97

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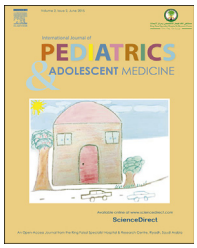

WHAT'S YOUR DIAGNOSIS

Hypertrichosis in a patient with hemophagocytic lymphohistiocytosis

Jeylan El Mansoury [a], Joyce N. Mbekeani [b,c,*]

[a] *Dept of Ophthalmology, KFSH&RC, Riyadh, Saudi Arabia*
[b] *Dept of Surgery, Jacobi Medical Center, North Bronx Health Network, Bronx, NY, USA*
[c] *Dept of Ophthalmology and Visual Sciences, Albert Einstein College of Medicine of Yeshiva University, Bronx, NY, USA*

Received 3 March 2015; received in revised form 15 April 2015; accepted 1 May 2015
Available online 30 May 2015

KEYWORDS
Cyclosporine A;
Hypertrichosis;
Trichomegaly;
Hemophagocytic lymphohistiocytosis;
Iatrogenic

1. A case of pediatric hypertrichosis and trichomegaly

A 19 month old baby boy was referred to ophthalmology for pre-operative assessment prior to bone marrow transplant (BMT) for hemophagocytic lymphohistiocytosis (HLH). This rare disorder of deregulated cellular immunity was diagnosed at 8 months when he presented with failure to thrive, hepatosplenomegaly, pancytopenia and elevated liver function tests. Bone marrow biopsy revealed multiple histiocytes with hemophagocytic activity compatible with the diagnosis of HLH. Molecular genetic testing was negative thus ruling out hereditary HLH. Intravenous cyclosporine A (CSA) and prednisolone were administered per HLH-2004 protocol for 2 weeks. He was discharged in stable condition on oral CSA 100 mg BID in preparation for subsequent allogeneic BMT. Ophthalmology examination revealed an alert, healthy looking baby boy with eyes that were central, steady, maintained and could fix and follow small objects; the anterior and posterior segments were normal. Abnormally long and thick crown of head hair, eyelashes (or trichomegaly) and a thick unibrow were noted (Fig. 1), along with diffuse downy facial hairs and even thicker sideburns and a moustache (Fig. 2) over normal colored skin. The increased facial hair and trichomegaly were reportedly noticed only after hospitalization and therapy for HLH.

* Corresponding author. Dept of Surgery, Jacobi Medical Center, 1400 Pelham Parkway, Bronx, NY 10461, USA. Tel.: +1 718 918 4784; fax: +1 718 918 7379.
E-mail address: jnanjinga@yahoo.com (J.N. Mbekeani).
Peer review under responsibility of King Faisal Specialist Hospital & Research Centre (General Organization), Saudi Arabia.

http://dx.doi.org/10.1016/j.ijpam.2015.05.001
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Figure 1 Hypertrichosis in an infant with eyelash trichomegaly and unibrow. Head hair, eyelashes and brow hairs are unusually long, thick and dark.

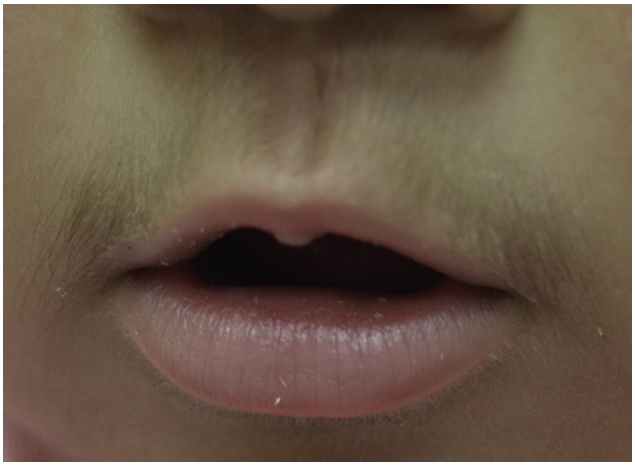

Figure 2 The same infant with hypertrichosis of upper lip and face manifesting as a moustache, sideburns and downy facial hairs.

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