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MDDH, DFSP and Adenosine Deaminase Deficient Severe Combined Immunodeficiency

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To the Editor

We read with interest the report by Mutgi et al. [Epub ahead of print 2015 Dec 8]¹ regarding a 9 year-old girl with a CD34+ dermal plaque consistent with a medallion-like dermal dendrocytic hamartoma (MDDH). The authors provide a summary of the limited reports in the literature of other MDDH, noting that CD34 staining alone cannot differentiate between MDDH, a benign self-limited tumor, and dermatofibrosarcoma protuberans (DFSP), a low-grade sarcoma.

Although the child with MDDH presented by Mutgi et al. did not present with a history of immunodeficiency, we would like to alert pediatric dermatologists to a high risk of DFSP tumors resembling MDDH in children with adenosine deaminase deficient-severe combined immunodeficiency (ADA-SCID). At the NIH, we have now evaluated 15 pediatric ADA-SCID patients with one or more DFSP tumors. These tumors typically present as small (3–6mm), atrophic, hyperpigmented, indurated plaques. In several patients, multiple lesions have been detected (as many as 18 in an individual patient).² Although all ADA-SCID patients with DFSP tumors have molecular or cytogenetic confirmation of *COL1A1-PDGFB* gene fusion, ADA-SCID-associated DFSPs are relatively hypocellular and do not possess the classic storiform pattern which would facilitate histologic differentiation of DFSP from MDDH (Figure 1). As Kutzner et al. note in their comparison study of MDDH and plaque-like DFSP, both types of tumors present as small, brown, atrophic or indurated plaques in

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children, and both tend to occur on the neck, trunk and extremities.³ A similar distribution pattern of DFSPs has been detected in our ADA-SCID population.

The natural history and optimal management of atrophic/plaque-like DFSP in children with ADA-SCID is still uncertain, as is the pathophysiologic basis for susceptibility to multiple skin sarcomas in this immunodeficiency. Regardless, careful skin examination is critical when encountering this rare disease and a low threshold for skin biopsy is needed. The detection of a CD34+ spindled cell infiltrate in an ADA-SCID patient should prompt testing for the *COL1A1-PDGFB* fusion product to confirm DFSP, even in the absence of classic storiform histology.

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Figure 1.
Histology of DFSP in a 12 year old with ADA-SCID and multiple discrete atrophic lesions.
A. Stellate-shaped fibroblasts are present in the dermis, but lack high cellularity and classic storiform architecture; B. Intense CD34+ immunostaining in the fibroblast proliferation (Hematoxylin and eosin, x20, CD34 x40).