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A child with LMNA-NTRK1 rearranged spindle cell neoplasm

Mirjana Gajic-Veljic*[1], Martina Bosic[2], Jovan Lalosevic[1], Branislav Lekic[1], Milos Nikolic[1]

[1]School of Medicine University of Belgrade, Clinic of Dermatovenereology, University Clinical Center of Serbia, Belgrade, Serbia, [2]Institute of Patology, School of Medicine University of Belgrade, Belgrade, Serbia

Introduction & Objectives:

A subset of mesenchymal neoplasms with NTRK /Neurotrophic Tyrosine Receptor Kinase/ gene fusions represents locally aggressive neoplasms, typically occurring in subcutaneous tissue, with a characteristic immunophenotype (CD34+, S100+) and recurrent kinase gene fusions, most commonly the LMNA /LaMiN A/-NTRK1 fusion. The differential diagnosis includes fibroblastic tumors such as lipofibromatosis and dermatofibrosarcoma protuberans.

Materials & Methods:

We present an 11-month-old girl with a slow-growing erythematous plaque with central atrophy, measuring 1x1.5 cm, on the scalp, which appeared a few months after birth. The parents denied the presence of any change at birth, as well as previous trauma. Lymph nodes were not enlarged. There were no other skin lesions and the child was otherwise completely healthy.

Results:

Routine laboratory analyses were normal. Scalp ultrasonography showed hypoechogenic subcutaneous plaque. Punch biopsy showed neoplastic proliferation consisting of elongated spindle cells, strongly CD34+, corresponding to plaque-like CD34+ dermal fibroma. A complete surgical excision was performed. Morphological, immunohistochemical and molecular analyses excluded dermatofibrosarcoma protuberans; the sarcoma fusion analysis by Next Generation Sequencing test, based on anchored multiplex PCR, demonstrated the presence of LMNA-NTRK1 gene fusion.

Conclusion:

All the findings confirmed the diagnosis of LMNA-NTRK1 rearranged spindle cell neoplasm, a recently described and rare molecularly-defined soft tissue tumor that may have a wide spectrum of morphologies and histological grades, with frequent co-expression of CD34 and S100. Due to the highly infiltrative growth pattern, the tumor has a propensity for local recurrence, if incompletely excised, but none has been shown to metastasize. In our patient, there was no recurrence more than 3 years after the excision. LMNA-NTRK1 fusion serves both as a diagnostic and therapeutic biomarker. Cases with advanced disease may be treated using tyrosine kinase inhibitors.