PAEDIATRIC DERMATOLOGY

MELANOCYTIC LESIONS AND RISK OF MELANOMA IN RASOPATHIES

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Introduction: The RASopathies are a clinically defined group of genetic syndromes caused by germline mutations in genes that encode components or regulators of the Ras/mitogen-activated protein kinase (MAPK) pathway. This group include: Noonan (NS), Costello (CS), Cardio-Facio-Cutaneous (CFCS) and Neurofibromatosis 1 (NF1) syndrome.

Objective: The basis of this study is to clarify if these patients have a major risk to have multiple nevi and melanoma, since their genetic heritage has the same mutations we find in sporadic melanoma.

Materials and Methods: This was a prospective cross sectional study including 103 patients affected by RASopathies recruited at the Center for Rare Disease and Birth Defects from the beginning of 2016 to the end of 2017. All participants underwent a dermatological evaluation aiming to count the melanocytic nevi and study their dermoscopic pattern.

Results: A total of 103 participants were classified according to the clinical and molecular diagnosis in 4 groups : 46 NS, 21 CFCS, 16 CS, and 20 NF1. Participants showing nevi were divided in three subgroups: a) patients with less than 10 nevi, including patients with none or lower count of nevi b) between 10 and 30 nevi, including patients with a normal count of nevi considering the median age of the patients and c) greater than 30 nevi, when multiple moles were present. Patients showing atypical lesion were monitored every three months if the atypia was mild or underwent surgical excision when the atypia was severe. Patients showing the greatest number of nevi and atypical lesions carried out mutations in BRAF and RAF1 gene. We noticed that, in our sample, the number of patients with >30 nevi is higher in the CFCS, especially with mutations in BRAF.

Conclusions:These preliminary results should represent a first step study to propose future

protocol of monitoring the melanocytic lesions in RASopathies.