

PReS-FINAL-2353: Are rasopathies new monogenic predisposing conditions to the development of systemic lupus erythematosus?

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► **To cite this version:**

Brigitte Bader-Meunier, Hélène Cave, Nadia Jeremiah, Frédéric Rieux-Laucat. PReS-FINAL-2353: Are rasopathies new monogenic predisposing conditions to the development of systemic lupus erythematosus?. *Pediatric Rheumatology*, BioMed Central, 2013, 11 (Suppl 2), pp.P343. <inserm-00914639>

HAL Id: inserm-00914639

<http://www.hal.inserm.fr/inserm-00914639>

Submitted on 5 Dec 2013

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POSTER PRESENTATION

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PreS-FINAL-2353: Are rasopathies new monogenic predisposing conditions to the development of systemic lupus erythematosus?

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From 20th Pediatric Rheumatology European Society (PreS) Congress
Ljubljana, Slovenia. 25-29 September 2013

Introduction

RASopathies (Noonan syndrome (NS) and Noonan-related syndromes) are neurodevelopmental syndromes resulting from germline mutations in genes that participate in the rat sarcoma/mitogen-activated protein kinases (RAS/MAPK) pathway (*PTPN11*, *SOS1*, *RAF*, *KRAS* or *NRAS* and *SHOC2*). Some monogenic conditions are associated with the development of systemic lupus erythematosus (SLE), and a few reports described the association of SLE, with NS.

Objectives

We aim to search for a relationship between RASopathy and the development of SLE.

Methods

We reported for the first time on a 13-year-old boy with NS with loose anagen hair (NSLAH) resulting from mutation in *SHOC2* who developed an autoimmune disorder which fulfilled four American College of Rheumatology (ACR) criteria for the classification of SLE (polyarthritis, pericarditis, antinuclear antibodies, anti-DNA antibodies). The case report then prompted a literature review by a systematic search for English and French articles on the subjects of RASopathies and SLE that had English abstracts in PubMed from 1966 to 2012.

Results

We identified seven additional patients with RASopathy and SLE. The male-to-female ratio was 1:1, and age at onset of SLE ranged from 5 to 32 years. The most common features were polyarthritis (7/8 patients), auto-

immune cytopenia (4/8 patients) and pericarditis (4/8 patients) while only one patient presented with skin involvement.

Conclusion

The association of two rare diseases in eight patients suggests that RASopathies may be associated with the development of SLE, which is characterized by a higher male-to-female ratio, a lower rate of skin involvement and a higher rate of pericarditis than “classic” SLE.

Disclosure of interest

None declared.

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Published: 5 December 2013

doi:10.1186/1546-0096-11-S2-P343

Cite this article as: Bader-Meunier et al.: PreS-FINAL-2353: Are rasopathies new monogenic predisposing conditions to the development of systemic lupus erythematosus? *Pediatric Rheumatology* 2013 11(Suppl 2):P343.

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