

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

Brigitte Bader-Meunier, Hélène Cave, Nadia Jeremiah, Frédéric Rieux-Laucat

▶ **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, 2013, 11 (Suppl 2), pp.P343. inserm-00914639

HAL Id: inserm-00914639

https://inserm.hal.science/inserm-00914639

Submitted on 5 Dec 2013

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

Bader-Meunier et al. Pediatric Rheumatology 2013, 11(Suppl 2):P343
http://www.ped-rheum.com/content/11/S2/P343

PEDIATRIC RHEUMATOLOGY

POSTER PRESENTATION Open Access

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

B Bader-Meunier[1*], H cave[2], N jeremiah[3], F Rieux-Laucat[3]

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 or 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), autoimmune 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.

Authors' details
[1]Hôpital Necker, France. [2]hôpital R.Debré, France. [3]INSERM U768, Paris, France.

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.

[1]Hôpital Necker, France
Full list of author information is available at the end of the article

BioMed Central

© 2013 Bader-Meunier et al.; licensee BioMed Central Ltd. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (http://creativecommons.org/licenses/by/2.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited. The Creative Commons Public Domain Dedication waiver (http://creativecommons.org/publicdomain/zero/1.0/) applies to the data made available in this article, unless otherwise stated.