



## **PW03-033 - SLC29A3 mutation: a new autoinflammatory condition**

Isabelle Melki, Karen Lambot, Laurence Jonard, Vincent Couloigner, Pierre Quartier, Bénédicte Neven, Brigitte Bader-Meunier

► **To cite this version:**

Isabelle Melki, Karen Lambot, Laurence Jonard, Vincent Couloigner, Pierre Quartier, et al.. PW03-033 - SLC29A3 mutation: a new autoinflammatory condition. *Pediatric Rheumatology*, BioMed Central, 2013, 11 (Suppl 1), pp.A259. <inserm-00881688>

**HAL Id: inserm-00881688**

**<http://www.hal.inserm.fr/inserm-00881688>**

Submitted on 8 Nov 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.



MEETING ABSTRACT

Open Access

# PW03-033 - SLC29A3 mutation: a new autoinflammatory condition

I Melki<sup>1,2\*</sup>, K Lambot<sup>3</sup>, L Jonard<sup>4</sup>, V Couloigner<sup>5,6</sup>, P Quartier<sup>1,5</sup>, B Neven<sup>1,5,7</sup>, B Bader-Meunier<sup>1,5,7</sup>

From 7th Congress of International Society of Systemic Auto-Inflammatory Diseases (ISSAID) Lausanne, Switzerland. 22-26 May 2013

## Introduction

Germline mutations in *SLC29A3* result in a range of clinically related, recessive syndromes: H syndrome, pigmented hypertrichosis with insulin-dependent diabetes mellitus (PHID) syndrome, Faisalabad histiocytosis (FHC), and sinus histiocytosis with massive lymphadenopathy (SHML). Main symptoms of these diseases are hyperpigmentation with hypertrichosis, sensorineural deafness, diabetes, short stature, uveitis and “Rosai-Dorfman-like” histiocytosis.

## Case report

We report the case of an eleven-month-old boy with early-onset recurrent episodes of unprovoked fever lasting 7 to 10 days associated with pericardial effusion, abdominal pain, diarrhea, and inflammation. Physical examination revealed hyperpigmentation with hypertrichosis, dysmorphic features and a spleen and liver enlargement. Failure to thrive, sensorineural deafness, psychomotor development delay, and a “Rosai-Dorfman like” cheek lesion further developed. Febrile attacks were not responsive to interleukin-1 and Tumor-Necrosis-Factor blocking agents. All known causes of genetic autoinflammatory syndromes were excluded by sequencing (*MEFV*, *NALP3*, *mevalonate kinase*, *NALP12*, *TNFRSF1*). Sequencing of *SLC29A3* gene revealed homozygous missense mutation c.1088G>A (p.Arg363Gln).

## Discussion

This case is the first description of a patient with an auto-inflammatory disorder due to a mutation in *SLC29A3* gene. Genetic defect of *SLC29A3* should be considered in patients with recurrent febrile attacks associated with any symptoms reminiscent of *SLC29A3* broad spectrum of

manifestations, especially hyperpigmentation with hypertrichosis.

## Disclosure of interest

None declared.

## Authors' details

<sup>1</sup>Unité d'Immunologie Hématologie et Rhumatologie Pédiatrique, Necker-Enfants Malades Hospital, Paris, France. <sup>2</sup>Service de Pédiatrie Générale, Robert Debré Hospital, France. <sup>3</sup>Department of pediatric radiology, Necker-Enfants Malades Hospital, France. <sup>4</sup>Department of genetics, Trousseau Hospital, France. <sup>5</sup>Institut IMAGINE, Paris Descartes University, Sorbonne Paris Cité, France. <sup>6</sup>Department of Microbiology, Necker Enfants Malades Hospital, France. <sup>7</sup>U768, INSERM, Paris, France.

Published: 8 November 2013

doi:10.1186/1546-0096-11-S1-A259

Cite this article as: Melki et al.: PW03-033 - SLC29A3 mutation: a new autoinflammatory condition. *Pediatric Rheumatology* 2013 11(Suppl 1):A259.

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at  
[www.biomedcentral.com/submit](http://www.biomedcentral.com/submit)



<sup>1</sup>Unité d'Immunologie Hématologie et Rhumatologie Pédiatrique, Necker-Enfants Malades Hospital, Paris, France  
Full list of author information is available at the end of the article