



POSTER PRESENTATION

Open Access

# Impact of mevalonate kinase deficiency (MKD) on the quality of life in children and young adults: a national multicentre study

Silvia Federici<sup>1\*</sup>, Alberto Tomasini<sup>2</sup>, Antonella Meini<sup>3</sup>, Matteo Doglio<sup>1</sup>, G Calcagno<sup>4</sup>, Francesco Zulian<sup>5</sup>, Rita Consolini<sup>6</sup>, Martina Finetti<sup>1</sup>, Luciana Breda<sup>7</sup>, Roberta Caorsi<sup>1</sup>, Laura Obici<sup>8</sup>, Romina Gallizzi<sup>9</sup>, Donato Rigante<sup>10</sup>, Mariolina Alessio<sup>11</sup>, Alberto Martini<sup>1</sup>, Marco Gattorno<sup>1</sup>

From 18th Pediatric Rheumatology European Society (PReS) Congress Bruges, Belgium. 14-18 September 2011

## Background

MKD is an autosomal recessive disease caused by mutations in the mevalonate kinase (MVK) gene.

## Aim

To analyze the long term follow-up and health related quality of life (HRQL) in MKD.

## Methods

MVK gene was analyzed in 950 consecutive patients with periodic fever. 40 MKD patients were identified. Spontaneous disease course was classified as follows: i) resolution (no episodes in the last 6 months), ii) improvement (reduction of more than 30% of fever episodes) iii) stationary iv) worsening (increase frequency of fever episodes or appearance of new major clinical manifestation). The Child Health Questionnaire (CHQ-PF 50) was used to assess the health related quality of life (HRQL). An international sample of 3315 healthy children (52.2% female), with a mean (SD) age of 11.2 (3.8) years constituted the healthy control group.

## Results

Data on follow-up are available for 31 patients. The mean follow-up was 12.9 years (range 2.3-38.2). Steroid on demand was effective in treating fever episodes. 15 patients showed a significant spontaneous reduction of the frequency of fever episodes. Complete resolution was observed in 3 patients. In 9 patients the disease was stable, in 4 worsened. When compared to healthy age-

matched individuals, HRQL of MKD patients was generally affected, particularly for global health, general health perception, mental health, parental-impact emotion and self-esteem ( $p < 0.001$ ).

## Conclusions

Even if a relevant percentage of MKD patient show a spontaneous amelioration of the disease, most of them display a tendency towards a persistence of fever episodes that significantly affect their quality of life.

## Author details

<sup>1</sup>Gaslini Institute, Genova, Italy. <sup>2</sup>IRCCS Burlo Garofolo, Dipartimento di Pediatria, University of Trieste, Trieste, Italy. <sup>3</sup>Dipartimento di Pediatria, Unità di Immunologia e Reumatologia Pediatrica, Spedali Civili E University Of Brescia, Italy. <sup>4</sup>Sezione di Reumatologia Pediatrica, AOU „G. Martino,, Messina, Italy. <sup>5</sup>Dipartimento A.I. di Pediatria, University of Padua, Padova, Italy. <sup>6</sup>Dipartimento di Medicina della Procreazione e dell'Eta' Evolutiva, Pisa, Italy. <sup>7</sup>Clinica Pediatrica, Divisione reumatologia, Ospedale Policlinico di Chieti, Chieti, Italy. <sup>8</sup>Laboratorio di Biotecnologie, IRCCS Policlinico San Matteo, Pavia, Italy. <sup>9</sup>Divisione di Immunologia e Reumatologia Pediatrica, università di Messina, Messina, Italy. <sup>10</sup>Università Cattolica del Sacro Cuore, Roma, Italy. <sup>11</sup>Dipartimento di Pediatria, Università Federico II, Napoli, Italy.

Published: 14 September 2011

doi:10.1186/1546-0096-9-S1-P24

Cite this article as: Federici et al: Impact of mevalonate kinase deficiency (MKD) on the quality of life in children and young adults: a national multicentre study. *Pediatric Rheumatology* 2011 **9**(Suppl 1):P24.

<sup>1</sup>Gaslini Institute, Genova, Italy

Full list of author information is available at the end of the article