PEDIATRIC RHEUMATOLOGY

POSTER PRESENTATION Open Access

# PReS-FINAL-2053: Vitamin D receptor polymorphisms in a cohort of Italian patients with juvenile idiopathic arthritis

F Falcini[1*], F Marini[2], D Rigante[3], F Bertini[1], G Lepri[1], S Stagi[4], M Matucci Cerinic[5], ML Brandi[2]

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

## Introduction

A role for vitamin D has been hypothesized in generating disease activity for patients with juvenile idiopathic arthritis (JIA): specific polymorphisms of vitamin D receptor (*VDR*) gene have recently been associated with different biologic response to vitamin D itself.

## Objectives

To evaluate *VDR* polymorphisms in patients with JIA in comparison with unrelated healthy controls.

## Methods

We recruited 63 Italian children, adolescents and young adults with JIA (mean age 16.21 + 7.11 SD yrs, 51 female and 12 males, female/male ratio 4.25 from 1 Unit of Paediatric Rheumatology and 1 Unit of Rheumatology, Transition Clinic. After informed consent, during routine laboratory tests, their genomic DNA was extracted from peripheral blood leukocytes, to analyze *VDR* polymorphisms by PCR-based sequencing (*CDX2* in the promoter region) and PCR-based enzymatic digestions (*FokI* in exon 2, *BsmI* and *ApaI* in intron 8, and *TaqI* in exon 9). An Italian population of 2221 unrelated individuals without JIA was used as healthy controls.

## Results

The distribution of *FokI, BsmI, ApaI*, and *TaqI* polymorphisms did not show significant differences between children with JIA and controls. Regarding the *CDX2* polymorphism, we observed a statistical difference in the distribution of GG and GA genotypes, with the GG genotype more frequent in JIA subjects (Yates-corrected chi-square 6.97; Odds ratio = 2.08; $p = 0.008$) and the GA genotype in healthy controls (Yates-corrected chi-square 4.04; Odds ratio = 0.55; $p = 0.044$). Data about AA genotype were not significant due to their very low number (three) within the JIA population. G allele resulted to be more frequent in JIA subjects (Yates-corrected chi-square 6.51; Odds ratio = 1.82; $p = 0.011$).

## Conclusion

Pathogenetic mechanisms influencing the predisposition to JIA are poorly elucidated. Our analysis of *CDX2* polymorphisms located in the promoter region of the *VDR* gene has revealed that both GG genotype and G allele are more represented in patients with JIA. By our preliminary data, we can speculate that the G allele decreases *VDR* transcriptional activity with respect to the A allele, as well as the presence of GG genotype could explain a reduction of *VDR* activity, with subsequent decreased response to vitamin D and potential immunity deregulation leading to JIA.

## Disclosure of interest

None declared.

**Authors' details**
[1]Internal Medicine, Rheumatology Section, Transition Clinic, Italy. [2]Surgery and Translational Medicine, University of Florence, Florence, Italy. [3]Institute of Paediatrics, Università Cattolica Sacro Cuore, Rome, Italy. [4]Endocrinology Unit, Anna Meyer Children's Hospital, Florence, Italy. [5]Internal Medicine, Rheumatology Section, University of Florence, Florence, Italy.

Published: 5 December 2013

doi:10.1186/1546-0096-11-S2-P66
**Cite this article as:** Falcini *et al.*: PReS-FINAL-2053: Vitamin D receptor polymorphisms in a cohort of Italian patients with juvenile idiopathic arthritis. *Pediatric Rheumatology* 2013 **11**(Suppl 2):P66.

[1]Internal Medicine, Rheumatology Section, Transition Clinic, Italy
Full list of author information is available at the end of the article