Role of PTPN22 C1858T Gene Polymorphism in Pediatric Polyautoimmunity

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Background:

Children with more than one autoimmune disease (AID) may have a stronger genetic component than children with a single AID. *PTPN22* C1858T single nucleotide polymorphism (SNP) has been associated with multiple different AIDs in adults and children, but it has not been studied in pediatric polyautoimmunity.

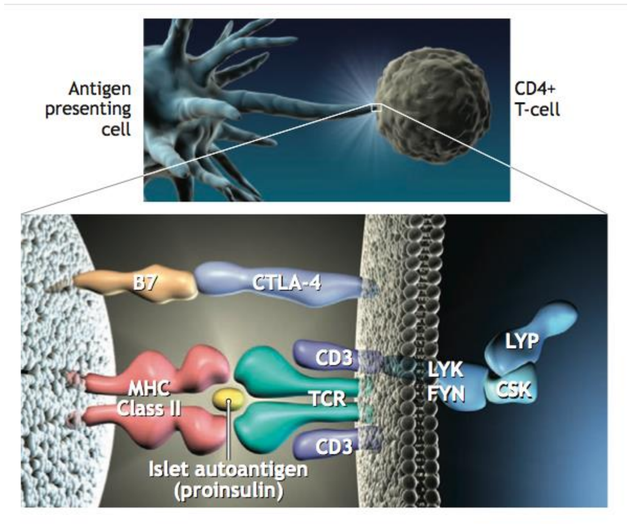

Figure 1: LYP encoded by PTPN22 gene involved un inmune response signaling pathway . Garth L. Burn et al.

Objective and hypotheses

To evaluate the association of *PTPN22* C1858T gene polymorphism with pediatric polyautoimmunity. We hypothesized that children with polyautoimmunity have a higher frequency of the PTPN22 *C1858T* SNP.

Method

A cross-sectional study was performed in 128 subjects with AIDs of pediatric-onset recruited at pediatric endocrinology, rheumatology and gastroenterology clinics at the Pontificia Universidad Católica de Chile Health Network and 98 healthy controls. Children with single AIDs included in this study had either juvenile idiopathic arthritis, type 1 diabetes, or autoimmune thyroid disease. Pediatric polyautoimmunity was defined as >1 AID. Genotyping of the rs2476601 (C1858T) *PTPN22* gene SNP was performed using TaqMan SNP genotyping assay by Real Time PCR.

Results

Mean age of children with AID was 12±4.4 years and 69% were female. The C1858T allele frequencies of cases and controls showed no deviation from Hardy-Weinberg equilibrium. Genotypes CC, CT, and TT of the *PTPN22* C1858T polymorphism presented frequencies of 85.2%, 13.3% and 1.6%, respectively, in the AID group, and 83.7%, 12.8%, and 2.3% in the control group (P=0.18).

Alelle	Mono	Poli	Total
CC	83 (89.2%)	26 (74.3%)	109 (85.2%)
CT+ TT	10 (10.8%)	9 (25.7%)	19 (14.8%)
Total	93 (100%)	35 (100%)	128 (100%)

Table 1: C1858T polymorphism in de los 128 patients sampled

The T-allele frequency was higher among patients with pediatric polyautoimmunity than children with single AID (26% vs 11%, OR=2.87, 95%CI=1.05-7.83, P=0.04).

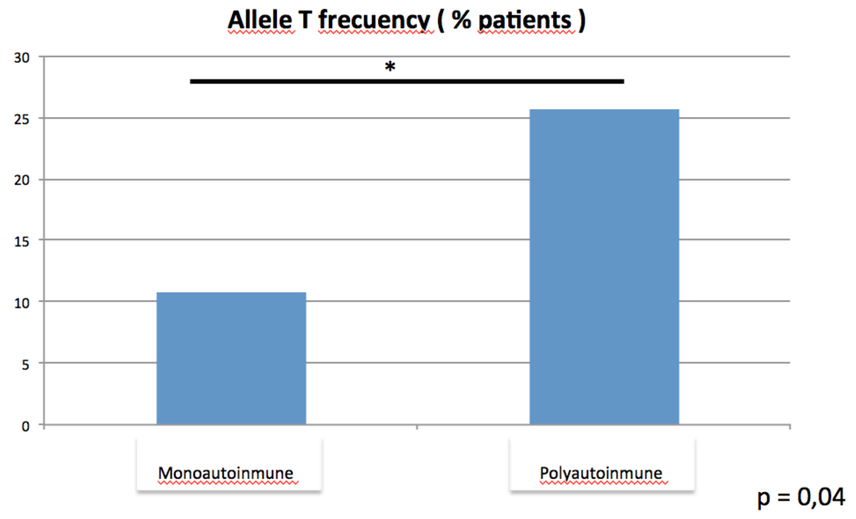

Figure 2: T-allele frecuency in patients with pediatric autoinmune

No significant differences were found in the age of onset of autoimmunity between mono and polyautoimmune subjects (*P*=0.44) or between subjects with CC genotype vs. CT+TT genotypes (*P*=0.81).

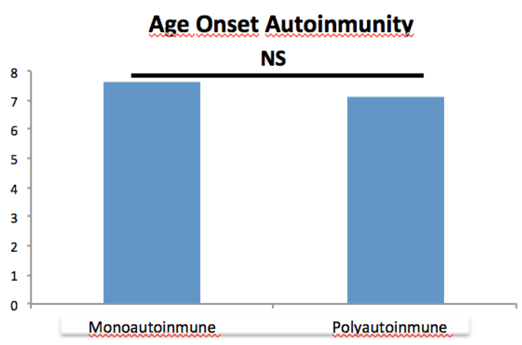

Figure 3: Autoinmunity age onset in patients with pediatric autoinmunity

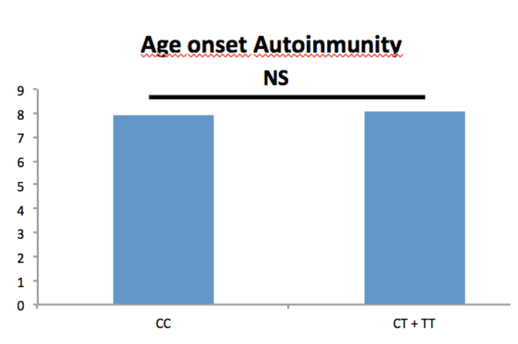

Figure 4: Autoinmunity age onset in patient with T-allelle presence

Conclusion

Children with polyautoimmunity have a higher prevalence of the *PTPN22* C1858T polymorphism, suggesting that this variant may be a risk factor for polyautoimmunity in children with AID.

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