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Are genetic variants of Caspase-1 and Cryopyrin associated with systemic JIA?

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Background

The systemic subtype of juvenile idiopathic arthritis (sJIA) can be the most severe, and unlike other forms of JIA is an autoinflammatory disease. There is evidence for the involvement of IL-1 in sJIA: treatment with the IL-1 receptor agonist, Anakinra, has shown dramatic improvement in some sJIA patients. Additionally we have shown a significant association with members of the IL-1 gene family and sJIA [1]. Caspase-1 is required to cleave IL-1β into its active form and Cryopyrin (NLRP3) is part if the IL-1β inflammasome, required for the activation of caspase-1. Mutations in *NLRP3* have been found in patients that have similar clinical features with sJIA, e.g. CINCA. Here we describe a candidate gene association study of *CASP1* and *NLRP3* in sJIA.

Materials and methods

Publicly available genotyping data and a tagging SNP(tSNP) approach were used to examine SNPs across the *CASP1* and *NLRP3* genomic regions. A total of 47 tSNPs were genotyped in 130 sJIA patients and 146 healthy controls. Analysis of the genotypes were performed using the software UNPHASED.

Results

There is no evidence of an association between *CASP1* and *NLR3* with sJIA.

Conclusion

These results indicate that while members of the IL-1 gene family have been shown to be associated with sJIA, there is no evidence for an association with SNPs in *CASP1* or *NLRP3*.

References

1. Stock CJ, Ogilvie EM, Samuel JM, Fife M, Lewis CM, Woo P: **Comprehensive association study of genetic variants in the IL-1 gene family in systemic juvenile idiopathic arthritis.** *Genes Immun* in press. 2008 Apr 17;