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Susceptibility genes for Kawasaki disease.

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Kawasaki disease (KD) is a systemic vasculitis syndrome predominantly affecting infants younger than 5 years. We have been trying to identify susceptibility genes for KD by a genome-wide approach. A linkage study of affected sib pairs and subsequent association studies using single nucleotide polymorphisms (SNPs) identified SNPs in ITPKC at 19q13.2 and CASP3 at 4q35 which confer risk for KD in the Japanese and Caucasian children. Recently we performed a genome-wide association study (GWAS) and identified 3 additional susceptibility loci for KD (FAM167A–BLK, HLA classII and CD40). Associations of the SNPs in FAM167A–BLK and CD40 gene regions were also seen in a GWAS independently performed by a Taiwanese research group. A functional SNP of FCGR2A gene was associated with KD in a GWAS for European KD patients and we replicated the finding in our Japanese samples. Interestingly variations in FAM167A–BLK, CD40 and FCGR2A regions have been associated with several autoimmune diseases of adulthood. We believe further investigation of these loci will foster better understanding of the pathogenesis and pathophysiology of the disease.