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Exzellenz im Norden

Inflammation at Interfaces
human development in landscapes
ozean der zukunft
DIE KIELER MEERESWISSENSCHAFTEN

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Lay summary

Genome-wide association studies (GWAS) have identified 16 primary sclerosing cholangitis (PSC) susceptibility loci. For most loci, the variants that cause disease are not known and the genes affected by these variants have not been identified. We aim to identify variants that cause PSC through detailed sequencing, genetic association and functional studies. We sequenced whole exomes of 48 PSC patients with severe disease, covering 20,000 genes and more than 200,000 exons. In total, 2.72 billion reads of unique mappable sequence have been generated and mapped to target region for the 48 individuals. Validation and replication of the variants discovered by whole-exome sequencing will be carried out through sequencing and genotyping in (at a maximum of) ~2,700 PSC cases and 5,300 healthy controls to determine disease association and frequency of the variants, and basic characterizations of the newly identified and replicated loci/variants will be carried out.