Identification of a novel locus for autosomal recessive retinitis pigmentosa (RP)

Hereditary retinopathies represent the major cause of adult genetic blindness in the western world. We identified a novel locus for autosomal recessive RP by localizing the disease gene in three large families (see figure below) to a 2.86-Mb interval on chromosome 14q by use of the “homozygosity mapping” approach. Genotyping was performed using the Affymetrix DNA Chip “Human Mapping 10K Array”. The identification of a novel disease gene will contribute to the understanding of the etiology and pathomechanism of this disabling condition, and may lead to future therapeutic concepts that may lead to prevention and/or cure of hereditary blindness.

Additional RP families were identified by us that will be first investigated for the involvement of known candidate genes. A whole genome scan will eventually be performed in case of the exclusion of major RP loci.

Figure 1: Families segregating retinitis pigmentosa

We further ascertained families segregating an endothelial corneal dystrophy, congenital sodium diarrhoea (MIM 270420), a cerebello-oculo-renal (Joubert-like) syndrome, Indian childhood cirrhosis (MIM 215600), Karsch-Neugebauer syndrome (MIM 183800), and KUFS disease (MIM 162350). Several projects are underway to map and identify the disease genes in these families.
In an ongoing project, patients with various forms of hearing loss are being screened for mutations in candidate genes.

All the work is being performed in collaboration with clinicians from the departments of pediatrics, hearing-, speech and voice disorders, ophthalmology, neurology, plastic surgery, and internal medicine of the University Hospital Innsbruck.

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