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### Genetic studies of hereditary retinal degenerations in northern Sweden

Blindness is a severe visual impairment with diminished or total vision loss. An average estimate of blind people in industrialized countries is 1-2/2000 compared to 5-10/1000 in developing countries. Disturbed anatomical structures and functions of cornea and retina are known to cause impaired vision. Retinitis pigmentosa, hereditary maculopathies and age related macular degeneration are part of a very heterogeneous group, called retinal dystrophies and degenerations associated with mutations in more than 250 loci and genes. In northern Sweden the presence of all hereditary disorders is relatively high due to a disposition of genetic abnormality in a geographically isolated area back in time. The frequency of retinitis pigmentosa (RP) is 1/3500 worldwide, however, in northern Sweden it is 1/2000 due to limited migration and a founder effect. RP shows autosomal dominant (10%), autosomal recessive (50-60%) and X-linked inheritance (15%). In our research using linkage analysis, copy number variants (CNV) analysis and next generation sequencing we uncovered genetic defects in autosomal recessive RP of Bothnia type (*RLBP1*), autosomal dominant RP with incomplete penetrance (RP11) (*PRPF31*), cone dystrophy (*CORD5*), Stargardt disease and leber congenital amaurosis (*ABCA4*, *CRB1*) and other severe RP forms (*MERTK*, *EYS*). Pathogenic mutations were detected in 53% of patients. Our data provide valuable information on the molecular mechanisms of RP evolvement and are useful in development of therapeutic strategies. Genetic diagnosis allows molecular genetic testing and genetic counselling of the patients and their families and also their inclusion to clinical trials of new treatments.

### Biography

Irina Golovleva is an Associate Professor at the Department of Medical Biosciences and a Clinical Scientist in Clinical Genetics Unit at University Hospital of Umeå, Sweden. She graduated in Russian State Medical University in 1982 and received PhD in 1989. She completed her Postdoctoral studies at Umeå University, Pasteur Institute, Paris, France and Great Ormond Street Hospital for Sick Children, London, UK. Since 1998 she conducts studies on identification of genetic causes and underlying molecular mechanisms in hereditary forms of retinal and corneal dystrophies in northern Sweden. She is a co-author of more than 70 papers published in peer-reviewed journals.