Veronica van Heyningen

Veronica van Heyningen is a Section Head at the Medical Research Council's Human Genetics Unit – part of the Institute of Genetics and Molecular Medicine in Edinburgh. She maintains a wide interest in genetic mechanisms and broad aspects of biology that can be gleaned from the study of human genetic disease. For the past two decades she has studied the underlying genes implicated in human developmental eye anomalies. The identification of three major developmental regulator genes: PAX6, SOX2 and OTX2, all implicated in brain as well as eye development, have led to extensive mutational studies in humans and dissection of gene function in model organisms. Detailed patient analysis led to early observations of how chromosomal breakpoints some way outside certain genes can lead to altered expression of those genes, and to the study of distant DNA elements regulating gene expression. Observing the variability and occasional absence of phenotype in cases with known pathological mutations, led us to study mechanisms of phenotype modulation and gene-environment interactions. Comparisons between different model organisms, including humans, also revealed some of the mechanisms of evolutionary change. More than 200 publications along the way have led to funding awards, invitations to participate in interesting meetings and some honours. Veronica van Heyningen was a Beit Memorial fellow immediately after finishing her PhD on early human gene mapping in Oxford under the guidance of Walter Bodmer. From 1993, she was a Howard Hughes International Research Scholar for five years, allowing her to build up her group. Since 1995 she has been an Honorary Professor at the University of Edinburgh. She was elected a Fellow of the Royal Society of Edinburgh in 1997, a member of EMBO in 2002 and a Fellow of the Royal Society in 2007. She participated in the Human Genetics Commission, which provides advice to the UK government on social and ethical issues in genetics. She is a past President of the European Society of human Genetics and current President of the Genetics Society. She continues to enjoy the fast moving science of human molecular genetics and the mentoring of young scientists entering the field, and has acquired a renewed interest in human development through her grandchildren.