EVI-Genoret integrating European vision research

Stefanie Petrou Binder MD
in Berlin

Researchers from across Europe are collaborating in a new project designed to hasten the application of genetic and genomic research to eye diseases that are presently difficult or impossible to treat.

Sponsored by the European Commission and coordinated by the European Vision Institute, the EVI-Genoret programme is bringing together partners from industry and academia to conduct research for the prevention and treatment of diseases that affect the retina, such as AMD and inherited retinal degeneration, said EVI-Genoret scientific coordinator José Sahel MD, Institut de la Vision, Hôpital St-Antoine, Paris, France.

“The number of people suffering from serious visual impairment is actually growing.”

José Sahel MD

“People are living longer and many of them need glasses, spectacles, or lenses to maintain their vision. But the number of people suffering from serious visual impairment and blindness is growing. We need to find new treatments and cures for these diseases.”

EVI-Genoret researchers believe that the mechanisms for understanding retinal degeneration are genetic, but the physiological representation of gene mutations is context dependent. Interrelationships between affected and non-affected genetic loci, as well as interactions within functional protein networks determine the risk and penetrance of disease. It is therefore the interplay between different genes and proteins networks that create regulatory networks determining the molecular nature of blindness.

Dr Sahel explained that EVI-Genoret addressed the systemic analysis of gene regulatory and protein networks by studying regulatory mechanisms that guide transcription, large-scale chip-based transcriptome analysis, proteomics, protein-interactome analyses, functional cellular and biochemical assays, and data integration through bioinformatics and model organisms.

Genes and their expression

EVI-Genoret researchers believe that their efforts will lead to the identification of a large number of genes for degenerative retinopathies. According to EVI-Genoret member Shomi Bhattacharya PhD, Institute of Ophthalmology, University College London, such work offers important clues to the molecular basis of gene-related sub-RPE deposits in AMD.

Identifying new genetic targets for AMD therapy

He noted that related research efforts targeted identifying new candidate genes for AMD by identifying the proteins specifically interacting with the C1QTNF5 short-chain collagen and its proposed RPE signalling pathway, as well as identifying the molecules, genes, and RPE products and components of RPE

Dr Bhattacharya noted that research addressing a variety of photoreceptor and retinal pigment epithelial (RPE) cell diseases as seen in patients with retinitis pigmentosa and macular dystrophies, including age related macular degeneration, was on EVI-Genoret’s agenda. This includes identifying genes expressed specifically in RPE (implicated in both monogenic and genetically complex retinal dystrophies, including AMD), and identifying proteins that interact with RPE-expressed gene products and components of RPE signalling pathways, which may point towards new candidate disease genes.

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