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RPE65 deficiency: Natural history and insights from gene therapeutic trials

Mutations in the *RPE65* gene result in *RPE65* deficiency (LCA2) with diminished or abolished retinol recycling manifesting in severe night blindness from infancy on together with variably reduced cone function and progressive retinal degeneration. The wide range of phenotypes results from different degrees of enzyme malfunctioning and spans Leber Congenital Amaurosis LCA, early onset severe retinal dystrophy EOSRD and juvenile RP. Gene replacement therapy in dogs with naturally occurring mutations in *RPE65* was very successful. To date, at least 150 patients with *RPE65* deficiency have been treated with sub retinal AAV injections to reconstitute protein function in USA, England and in France. Treatment effects were generally much more modest than in the dog. To document smaller treatment effects, it has turned out that new readout parameters had to be defined and developed. With the current approaches, there is a clear rod effect but a much smaller cone effect. Also, some data would indicate that the degeneration process was not stopped by the currently used set-up. One of the challenges is to get novel readout parameters accepted by the authorities. Another challenge is to define disease stages where therapy is more likely to be effective as well as optimizing the sub retinal AAV injection technique and dosing the most efficient vector titer. Of note, efforts are made by Spark Therapeutics® to receive FDA and EMA approval for an AAV vector to treat *RPE65* deficient patients.

Biography

Birgit Lorenz was graduated from the University of Munich, Germany where she also specialized in Ophthalmology and Genetics and received her MD and PhD. She is currently a Professor of Pediatric Ophthalmology, Neuro Ophthalmology and Ophthalmic Genetics, Director and Chair of the Department of Ophthalmology at the Justus-Liebig University Giessen, Germany. She was the President of several scientific societies and is currently a President of the ISGEDR and elected Member of the AOI and EAO. She has lectured worldwide, published over 300 original papers (h-index 43), edited several books and was Reviewer for many peer-reviewed journals and grant applications. Her main research interests are inherited retinal disorders including gene therapy, development of new readout parameters and ROP.

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