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A "Family Project" to fight Usher, a rare disease leading to deaf-blindness

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It was 1994 when physicians told us, that our two sons - deaf from birth - will lose their eye sight also to Usher, a rare genetic disease. With inspiring support from motivated scientists, private equity, National- and European Agencies we moved from fragmented basic research in identifying the gene, the molecular mechanisms of the protein and the specific mutation to shared objectives in disease specific research for a therapy. We founded a Family-Foundation to finance our activities, a medical device company to develop a retina implant, a chip for the eye and worked in EU- and US organisations to bring science ahead, e.g. the Foundation Fighting Blindness (FFB), Fondation Voir & Entendre, Pro Retina Foundation. We have already identified a cell-based treatment by a drug which, down the road, has the potential to put on hold the progression of our son's disease.

Therefore we have focused our activity with a view to strengthening research with the European TREATRUSH project within FP-7 Health, participated in networks for fund-raising and pursued tests to see, if the drug (PTC 124) will stop the degeneration process in the retina with the assumption that what works in Duchenne disease will also work in Usher 1C, and raise awareness of companies to invest in therapy with PTC 124 for patients with nonsense mutations in retinal degenerative diseases including Usher 1C. Unlike the early days we see a clear road to a therapy and are planning clinical trials with interdisciplinary teams. There is also a critical mass of expertise and substantial management by a biotech company for the benefit of all with Usher 1C. Yes, we can.

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