

Notes from Dr. Leonide Saad's presentation at the CRB1 research symposium March 23, 2013

Leonide Saad, PhD, President and CEO of Alkeus Pharmaceuticals discussed Stargardt's macular dystrophy, the #1 cause of childhood macular degeneration. It affects 100,000 individuals, and starts around age 6. Vitamin A byproducts accumulate, seen as retinal autofluorescent flecks that lead to retinal degeneration. Although *ABCA4* is the most common gene behind Stargardt's, his work genotyping patients in the south of France revealed several additional genes. (Cells derived from human embryonic stem cells are in clinical trials to treat Stargardt's – <http://blogs.plos.org/dnascience/2012/09/27/human-embryonic-stem-cells-finally-reach-clinical-trials-mauries-story/> -- Ricki)

Dr. Saad described a new drug to treat transthyretin (TTR) familial amyloid polyneuropathy, based on a Portuguese family with mild disease and two mutations in the same gene – the disease-causing one, and a second mutation that prevents the disease. Discovering how the protection happens enabled researchers to develop the drug Tafamidis, marketed by Pfizer and approved in Europe. FDA is requiring additional data. He also talked about development of the cystic fibrosis drug Kalydeco, which helps about 5% of patients but is being teamed with a second compound to help others.