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

Stephen Tsang, MD, PhD, Associate Professor of Pathology and Cell Biology and Ophthalmology at Columbia University, discussed phenotyping *CRB1* patients. He does gene and stem cell therapies on albino mice that have human retinal genes.

"Crumbs retinopathies" (*CRB1*) include:
1. LCA
2. Early onset RP with coats-like exudative vasculopathy (RP12)
3. Early onset RD without preserved para-arteriolar RPE and without Coats

Dr. Tsang described para-arteriolar sparing -- areas around blood vessels that do not pick up stain for vitamin A. He suggested that the complex genetic situation – mutations in the same gene corresponding to different diseases, and the same disease resulting from mutations in different genes – could be why only 5 of thousands of compounds make it to clinical trials. "Maybe we're not doing clinical trials correctly, with too heterogeneous populations," he said, offering the examples of cystic fibrosis drugs and the breast cancer drug Herceptin. Growing iPS cells from patients will be a way to test for biomarkers.

Several attendees talked about how exome sequencing (now $750 for research) should be used to identify modifier genes, before gene therapy is attempted.