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

Patsy Nishina, PhD, of the Jackson lab, described her work with a frameshift mutation in Crumbs in mice that generates a stop codon, leading to a truncated Crumbs protein. It is inherited as an autosomal recessive and causes retinal spotting. The defect is in a transmembrane protein necessary for forming adherens junctions between glial cells and photoreceptors.

In an effort to move all mutations onto the same genetic background (called black 6), so the mutations could be compared without adding effects from other genes, "the telltale spots went away, the photoreceptors were okay, and there was no dysplasia. Therefore *CRB1* is necessary but not sufficient to cause photoreceptor degeneration," Dr. Nishina explained. She added that the NIH has a knockout program, and by accident, all the cell lines used a black 6 that carries the *Crumbs* mutation that originated spontaneously, perhaps many years ago. She suggested looking at all papers that use black 6 to see if the authors attribute any aspects of the phenotype to the gene they are studying when it actually corresponds to the *Crumbs* mutation.