

Cohen Syndrome (CS) is a rare autosomal recessive disorder caused by loss-of-function mutations in VPS13B. This is a transmembrane protein thought to function in vesicle-mediated transport and sorting. Individuals with CS present diverse clinical features including intellectual disability, developmental and motor planning challenges, microcephaly, hypotonia, joint laxity, truncal obesity, intermittent neutropenia, progressive high myopia and retinal dystrophy. Loss of vision generally begins in early childhood and advances to legal blindness over time.

One \$162,161 grant or two \$81,080 grants are available*. While research opportunities in this area are broad in scope, priority will be given to grants that cover one of the following areas:

1. Studying the functions of VPS13B and underlying pathways to understand the molecular basis of CS
2. Development of potential therapeutic interventions including drug repurposing, small molecules, oligonucleotides, gene and cell therapies or protein replacement therapies
3. Collection of clinical and genetic data from at least 50 CS patients worldwide to assess phenotypic variability and to evaluate the effect of various treatments on the relevant symptoms.

*Please submit a proposal for the total amount of \$162,161. The ODC may choose to fund two awards at \$81,080 each, at which point we will request a revised work plan and budget.