Research Focus Areas:

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 $83,282 grant 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