UC Irvine and PDx collaborate, 5/16

May 2016—The Spooner Girls Foundation, which funds research toward treatments and a cure for mitochondrial complex I deficiency, along with the University of California Irvine School of Medicine and Population Diagnostics, announced a collaborative research initiative focused on the *NUBPL* gene. The Spooner family's decision to go public with their efforts to help their daughters and other patients diagnosed with mitochondrial complex I deficiency, an extremely rare disease caused by mutations in the *NUBPL* gene, has led to this groundbreaking collaboration.

PDx's proprietary gene discovery platform has revealed an *NUBPL* mutation that is believed to be linked to Parkinson's disease. Current data indicates that two *NUBPL* mutations need to be present in order to cause complex I deficiency, while the presence of only one *NUBPL* mutation may increase the risk for development of Parkinson's disease.

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