



Raquel Gur and 9 Universities on “Dissecting the effects of genomic variants on neurobehavioral dimensions in CNVs enriched for neuropsychiatric disorders.”

On the translational science front, *Raquel Gur* is leading 9 universities in a U01 funded, innovative large-scale collaborative study focused on neuropsychiatric manifestations of Rare Genetic Disorders (RGDs), which often evince negative effects on brain development and functioning throughout the lifespan. Because copy number variants (CNVs) at 22q11.2 and 16p11.2 loci are among the most common RGDs impacting developmental psychopathology, this far-ranging scientific team of investigators in North America and Europe will capitalize on highly informative samples and integrate prospective dimensional and categorical phenotyping with whole genome sequencing across these reciprocal CNVs. The goal is to identify convergent risk mechanisms for developmental neuropsychiatric disorders that have relevance to the broader population. The effort builds on established collaborations between Penn and CHOP in the study of 22q11.2 that led to a previously funded international consortium that now expands to other CNVs.