Genotype-Phenotype Associations in Immunodeficient Children with 22q11.2 Deletion

Wednesday, October 21
2pm-3pm
283 EMRB (Seebohm Conference Room)

The 22q11.2 microdeletion syndrome (22q) is the most common chromosomal microdeletion syndrome clinically detected with an estimated incidence of 1 in 4000 births. Children with 22q can exhibit a wide range of conditions including congenital heart disease, palatal abnormalities, immune deficiency, neurocognitive disorders and electrolyte imbalances. Despite the common genetic lesion all of these children share, a deletion of approximately three million base pairs of DNA from one of their 22nd chromosomes, it is currently not possible to predict which children will develop the conditions listed above and which will not. The ability to predict with a high degree of confidence which patients will exhibit immune dysfunction, the second leading cause of death among children with 22q, would allow for enhanced prevention of secondary infectious complications and improve clinical outcomes in these children. This presentation will cover how we used an extreme phenotyping approach combined with whole exome sequencing to discover candidate modifier genes for the immunodeficient phenotype in patients with 22q.