

CHOP Research Institute Summer Scholars Program



Ian Krantz, M.D.

Human Genetics and Molecular Biology

Translational Research: Research focused on bringing concepts in laboratory research directly into medical practice (may or may not involve human subjects)

Genomic Basis of Human Developmental Disorders

My research has two main areas of emphasis: 1) Understanding the genomic basis of syndromic and non-syndromic human developmental disorders. The identification of genes involved in human morphogenesis enables us to understand human embryonic development on a molecular level and enables us to begin the process that we hope will lead to improved diagnostics, management and eventually therapeutics for affected individuals, 2) Understanding the issues involved in introducing genomic medicine into clinical practice. There are multiple clinical and social issues involved in using genomic diagnostics in pediatric medicine (diagnostic yield, variant calling, secondary findings, integration into the electronic medical record, making this data portable and longitudinal etc...).

- **Potential summer research project:**
Specific projects currently underway involve: **(1)** the molecular etiology and genetics of Cornelia de Lange Syndrome, Pallister-Killian Syndrome, and diagnosing and understanding human developmental diagnoses through exome and genome sequencing. **(2)** Understanding the contribution that exome sequencing and genomic diagnostics makes in the management of various pediatric populations.
- **Students will learn the following techniques:**
(1) Polymerase chain reaction (PCR) based technologies and sequencing; **(2)** Exome and genome-scale analysis of samples from patients with various developmental disorders towards identification of candidate disease genes.