Join us at the *Kids First Poster Session: Accelerating Pediatric Genomics Research through Collaboration*

Tuesday, October 15th 6:00 - 9:00 pm CT

Hilton Americas-Houston– Lanier Ballroom J&K - Level 4

*Space is limited, please register in advance through Eventbrite

Recommended Posters, Oral Presentations, & Booths:

Booths: 233 - The Children's Hospital of Philadelphia: Kids First Data Resource Center

714- Broad Institute Genomic Services (Kids First Sequencing Center)

310 - St. Jude Children's Research Hospital (HudsonAlpha Kids First Sequencing Center collaborator)

Tuesday, Oct 15th

Ancillary Event: The All of Us Research Program: Cohort Diversity and Data Diversity. Agenda.

10:45 am – 12:45 pm. Hilton Americas – Houston. Lanier Ballroom G&H - Level 4

Wednesday, Oct 16th

Ancillary Event: The Early Career Researcher's Guide to the NIH Funding Process. Registration Page

7:15 - 8:45 am. Hilton Americas – Houston. Rm 337AB - Level 3

Poster Session: Mendelian Phenotypes

Exhibit Hall - Level 1/Convention Center.

2:00 - 3:00pm:

- PgmNr 1113/W: Overcoming challenges in identification, annotation, and interpretation of variants in DSD genes. Kids First X01 author: Emmanuele Delot. Kids First Dataset: Disorders of Sex Development (phs001178)

Poster Session: Cancer Genetics

Exhibit Hall - Level 1/Convention Center.

2:00pm - 4:00pm

- PgmNr 765/W: A germline susceptibility variant in VHL in a patient with Maffucci syndrome. Kids First X01 author: Nara Sobreira. Kids First Dataset: Enchondromatoses

Platform: Improved Structural Variation Detection Leads to New Insights into Disease and Development

Room 361D - Level 3/Convention Center Session #33.

5:15 - 5:30pm:

- PgmNr 80: Integration of optical genome mapping and sequencing technologies for identification of structural variants in disorders/differences of sex development (DSD). Kids First X01 author: Eric Vilain Kids First Dataset: Disorders of Sex Development (phs001178)

Thursday, Oct 17th

Platform: Methods and Resources in Large-scale Population Data

Hall B - Level 1/Convention Center Session#40

9:00 - 9:15am:

- PgmNr 96: Open access to dbGaP new aggregated allele frequency for variant interpretation

Poster Session: Cardiovascular Phenotypes

Exhibit Hall - Level 1/Convention Center.

2:00pm - 3:00pm:

- PgmNr 2779/T: DataSTAGE: Leveraging cloud technology to accelerate scientific discovery for heart, lung, blood, and sleep research.

Poster Session: Cancer Genetics
Exhibit Hall - Level 1/Convention Center.
2:00pm - 3:00pm:

- PgmNr 787/T: Identification and functional characterization of germline variants predisposing patients to congenital anomalies and childhood cancer. Kids First X01 authors: Sharon Plon & Philip Lupo

Poster Session: Complex Traits and Polygenic Disorders
Exhibit Hall - Level 1/Convention Center.
3:00pm - 4:00pm:

- PgmNr 1990/T: Identification of de novo variants by WGS of European and Latino trios and their contribution towards nonsyndromic orofacial clefts. Kids First X01 authors: Mary Marazita, Elizabeth Leslie, Elearnor Feingold. Kids First Datasets: phs001168 & phs001420

Poster Session: Mendelian Phenotypes
Exhibit Hall - Level 1/Convention Center.
3:00pm - 4:00pm:

- PgmNr 1390/T: Mondo Disease Ontology: Harmonizing disease concepts across the world. Kids First DRC author: Julie McMurry

Poster Session: Bioinformatics and Computational Approaches
Exhibit Hall - Level 1/Convention Center.
3:00pm - 4:00pm:

- PgmNr 1588/T: Quantifying phenotype similarity for complex harmonized disease cohorts. Kids First DRC author: Deanne Taylor

Friday, Oct 18th

Data CoLab: Using dbGaP Aggregated Allele Frequency and other large data sets in dbSNP to improve human genetic variation interpretation. Exhibit Hall - CoLab Theater 1 #345/Convention Center. 10:00am – 10:30am

Exhibit Hall - Level 1/Convention Center.
1:00pm - 2:00pm

Poster Session: Mendelian Phenotypes

- PgmNr 1217/F: Metaphyseal enchondromatosis with D-2-hydroxyglutaric aciduria and variants in IDH1, IDH2, and EX. Kids First X01 author: Nara Sobreira. Kids First Dataset: Enchondromatoses

Poster Session: Molecular and Cytogenetic Diagnostics

- PgmNr 2453/F: Identifying causal genes from abnormal clinical microarray results in patients with heterotaxy-spectrum congenital heart defects. Kids First X01 author: Stephanie Ware. Kids First Dataset: **Laterality Birth Defects**

Poster Session: Statistical Genetics and Genetic Epidemiology

- PgmNr 1565/F: Gabriella Miller Kids First Data Resource Center: Collaborative platforms for accelerating research in genetic etiology of pediatric diseases. Kids First DRC author: Allison Heath
- PgmNr 1673/F: Accelerating research with the NCBI Sequence Read Archive on the commercial cloud.

Poster Session: Complex Traits and Polygenic Disorders

- PgmNr 2285/F: Developing a data resource for pediatric research: The Gabriella Miller Kids First Pediatric Research Program (Kids First).

Saturday, Oct 19th

Platform: New Insights into Rare Skeletal, Growth, and Vascular Disorders
Room 360D - Level 3/Convention Center Session #106.
10:45 - 11:00pm:

- PgmNr 348: Variants in the HIF-1 pathway are associated with Ollier disease and Maffucci syndrome. Kids First X01 author: Nara Sobreira. Kids First Dataset: Enchondromatoses