Leveraging network analytics to infer patient syndrome and identify causal mutations using patient DNA sequence and phenotype data

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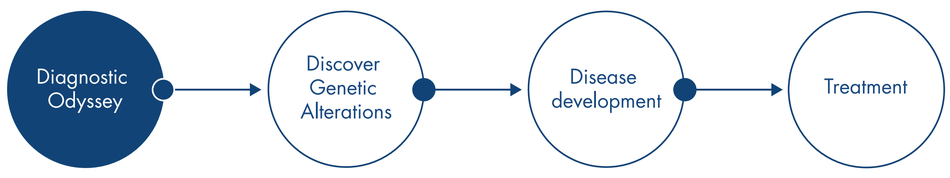
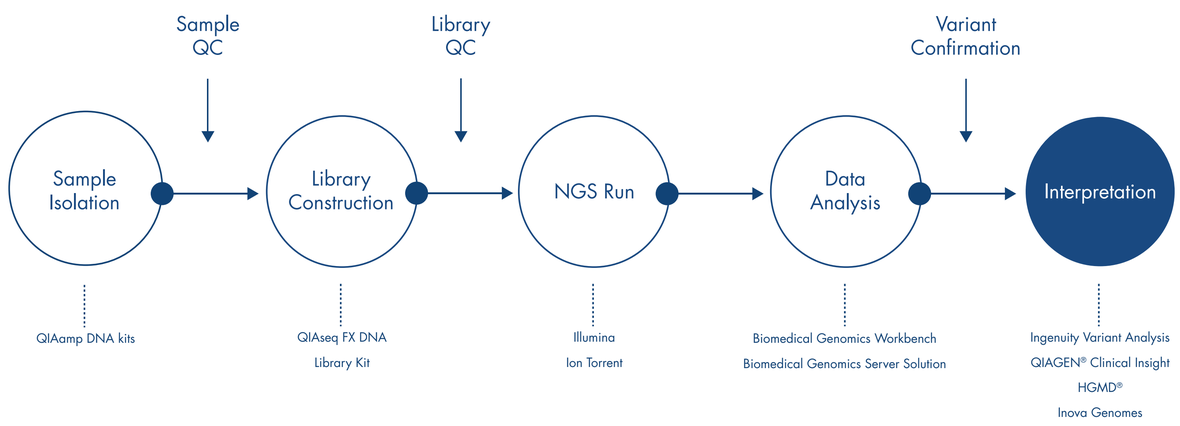
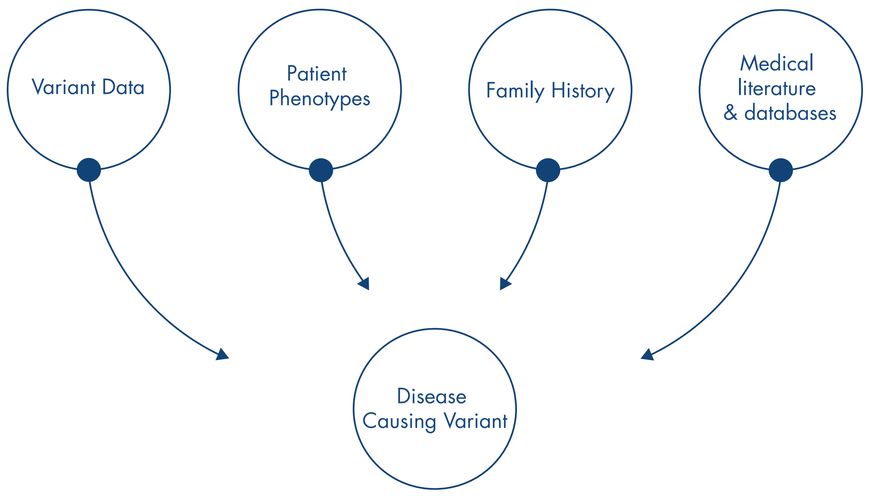
Clinical Genome and Exome Sequencing (CGES)





Identify Disease Causing Variants

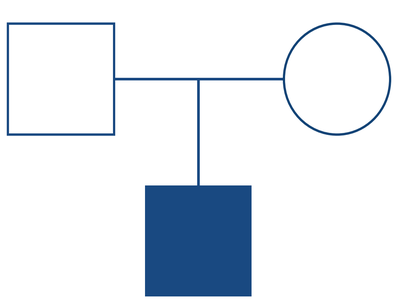


Causal Variant Discovery & Interpretation

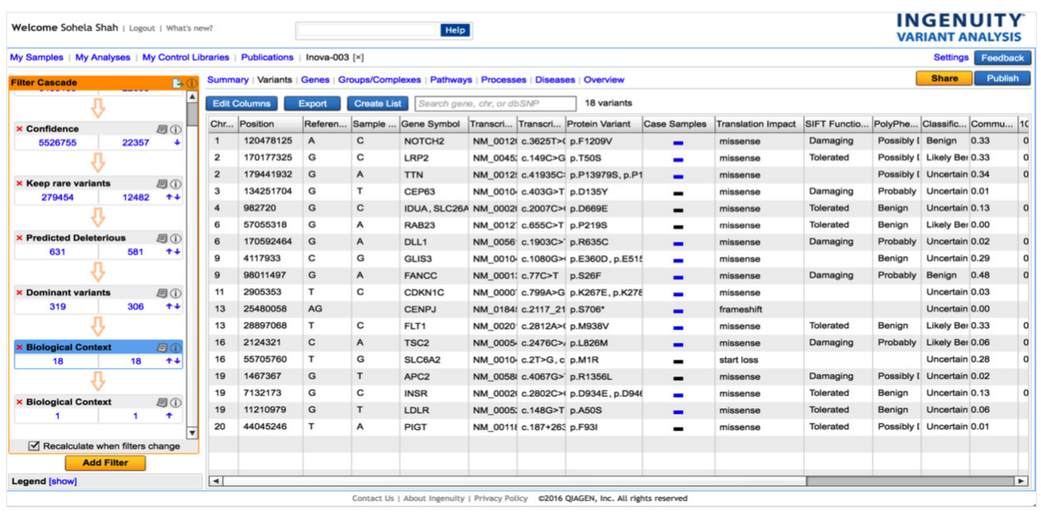
Confidence	Call Quality > 20 Variants not present in highly variable exonic regions
Common Variants	MAF < 0.5 1000G, NHLBI-EVS, ExAC, Allele Frequency Community
Predicted Deleterious	Pathogenic or Likely Pathogenic (ACMG) HGMD LoF and Missense
Genetic Analysis	Variant shared by proband and mother
Biological Context	Phenotypes- Prominent forehead; Macrocephaly; Hypospadia; Intrauterine growth retardation; Flat nasal bridge

INGENUITY® VARIANT ANALYSIS

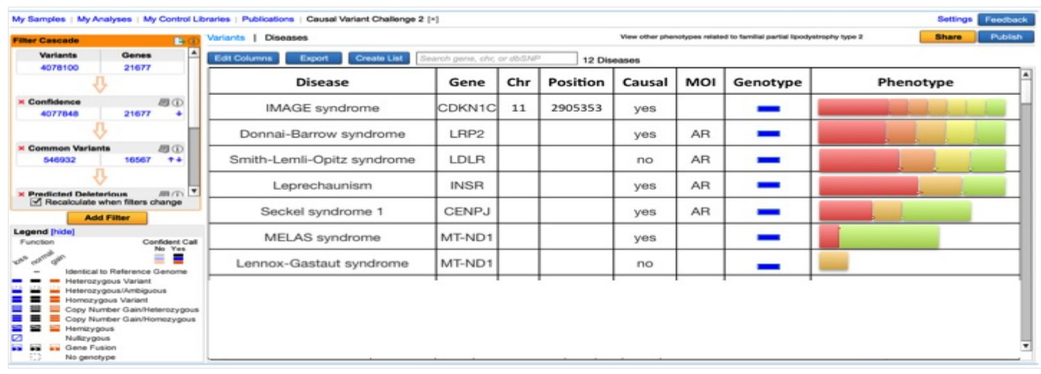
Biomedical Genomics Workbench | Ingenuity Variant Analysis | Knowledge Base

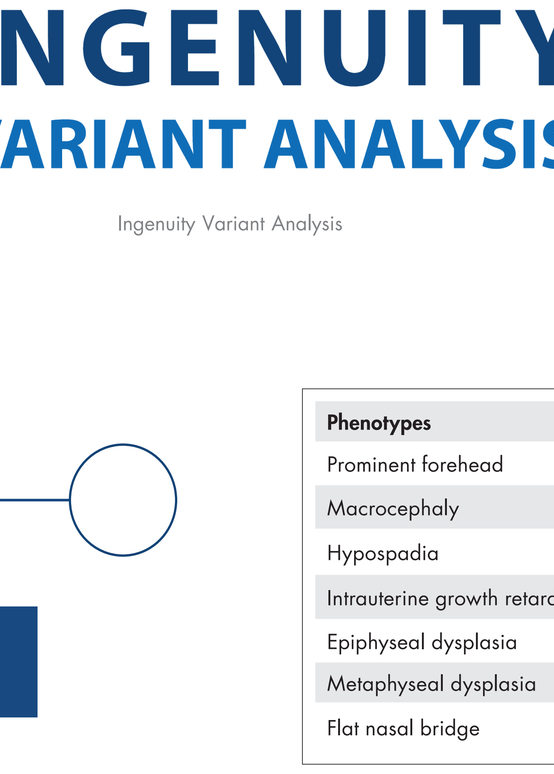


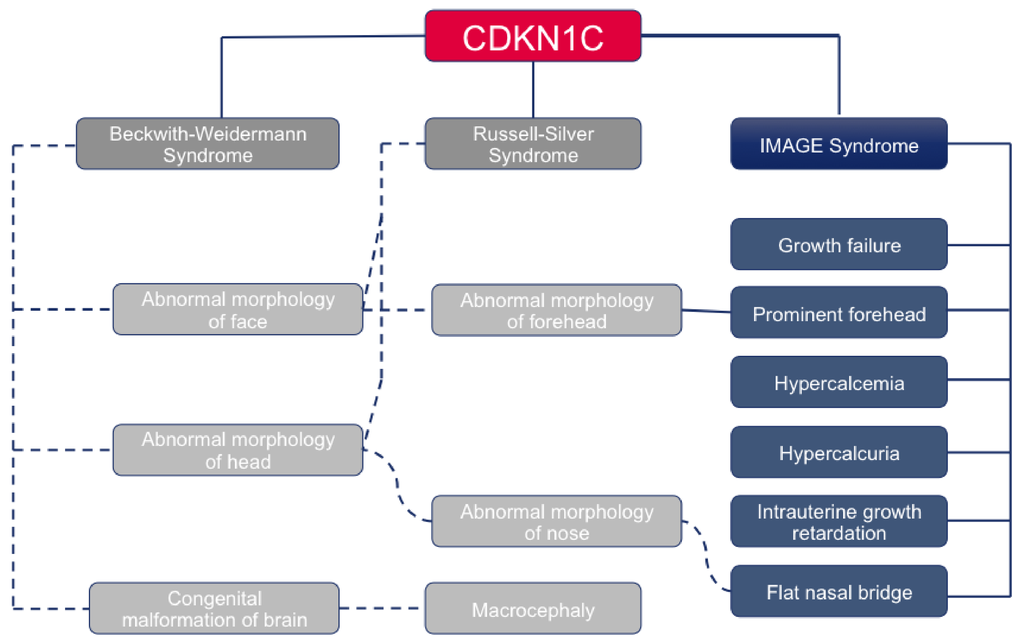
Phenotypes
Prominent forehead
Macrocephaly
Hypospadia
Intrauterine growth retardation
Epiphyseal dysplasia
Metaphyseal dysplasia
Flat nasal bridge



Syndrome Inference







Summary

- Fastest & most accurate application with built-in false positive check
- Leverage peer-reviewed literature content and network analysis to infer syndromes from patient phenotypes
- Discover known and novel causal variants