Ingenuity Variant Analysis, leveraging the Knowledge Base and HGMD, achieves over 30x enrichment in biologically relevant variants from whole genome and exome sequence data from patients with rare disease

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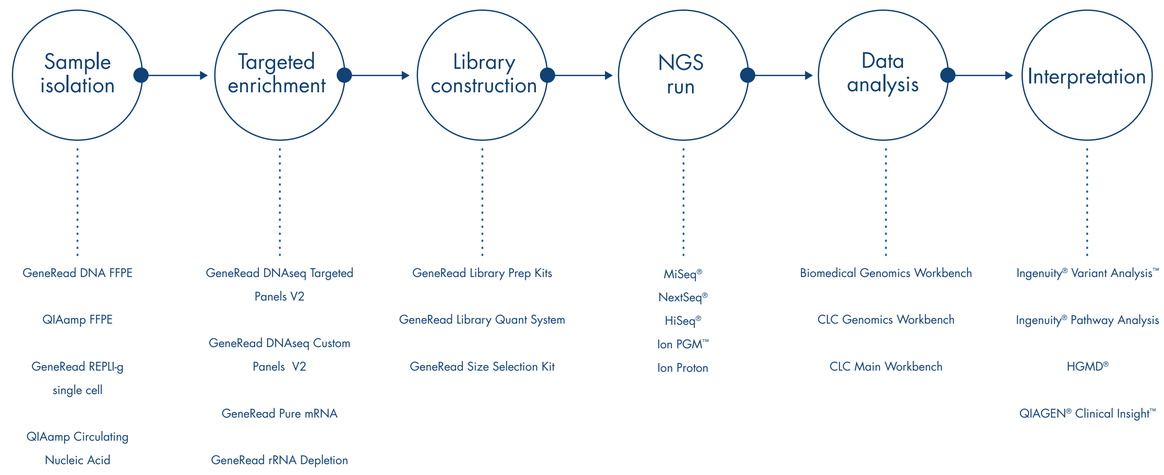
Sample to Insight

Automation solutions for seamless and cost-effective workflows, featuring:

- **Industry's most reliable sample technologies** – Top-quality assays and panels to accurately analyze and identify diseases and genetic variations.
- **Bioinformatics software and curated knowledge bases** – To transform your raw data into relevant, actionable findings.

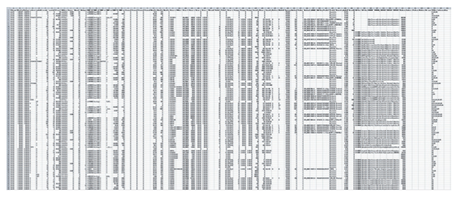


QIAGEN Ingenuity Variant Analysis

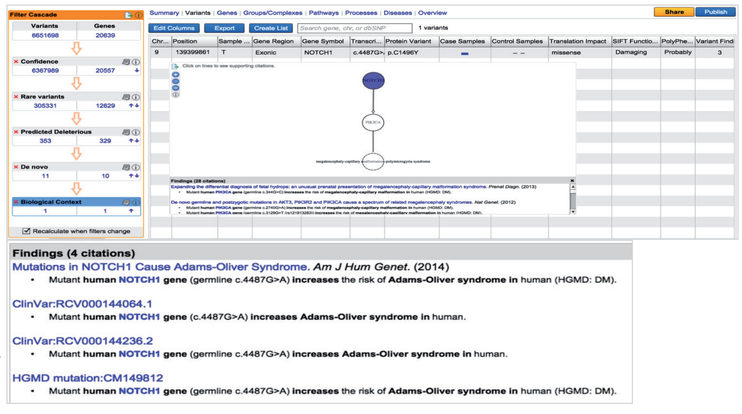
Identify causal variants from human sequencing data in just minutes.

- **Intuitive, user-friendly interface** – No bioinformatics skills needed.
  - **Interactive Filter Cascade** – Rapidly eliminate common and non-deleterious variants with a basic set of filters.
  - **Iterative analysis** – Apply a hypothesis, visualize and evaluate results in real-time.
- **Knowledge driven algorithms and analytics** – Genetics, Statistics, Functional Prediction.
  - **Curated up to date content at your fingertips** – Utilize the Ingenuity Knowledge Base™ containing millions of biomedical findings and mutations, accurate, up-to-date and curated by experts, from the literature and public databases.
  - **Causal Network Analytics** – Identify variant in genes within 1- or 2- network "hops" of upstream or downstream mutated genes.
- **Sharing and Publication Tools** – Easily collaborate with colleagues and peers. Export data and graphics to aid in manuscript preparation and publications.
- **Scalable processing capacity** – Analyze thousands of samples simultaneously.

vcf



QIAGEN Ingenuity Variant Analysis



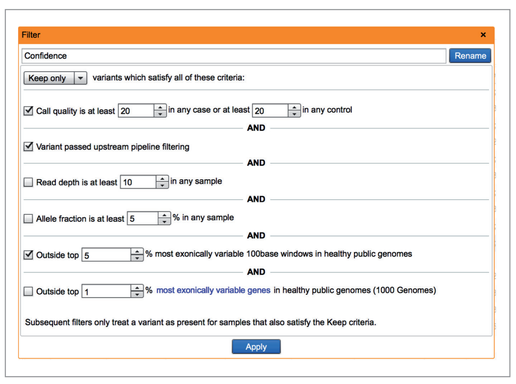
Single Sample Analysis

Total number of samples: 80

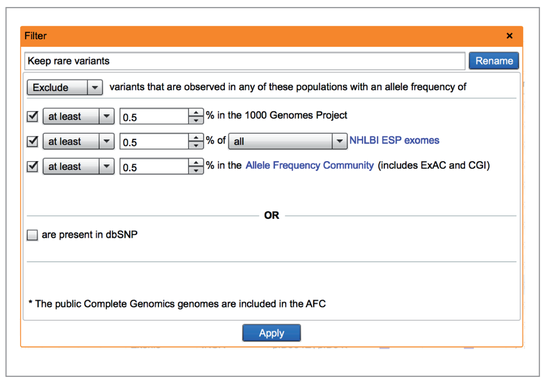
Source	Samples
Inova Health Systems, Virginia	15
Children's National Medical Center, Washington, DC	28
Children's Hospital of Eastern Ontario	33
Legacy Ingenuity Demo cases	5

Filter Cascade:

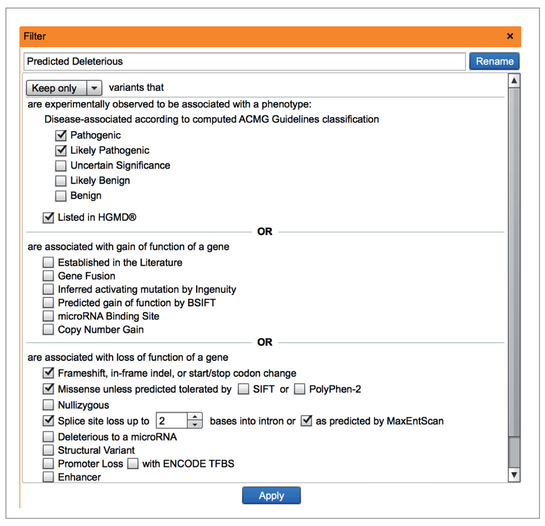
Confidence



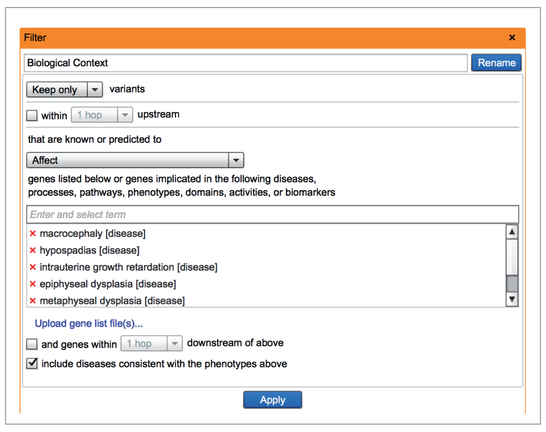
Common Variants



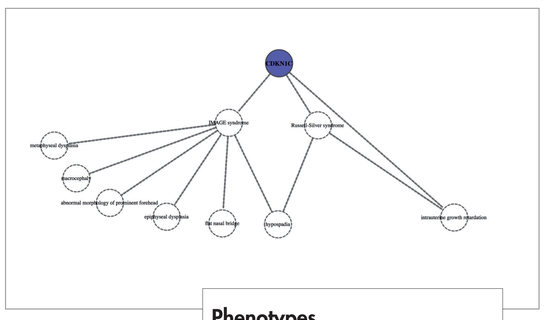
Predicted Deleterious



Biological Context (Example)

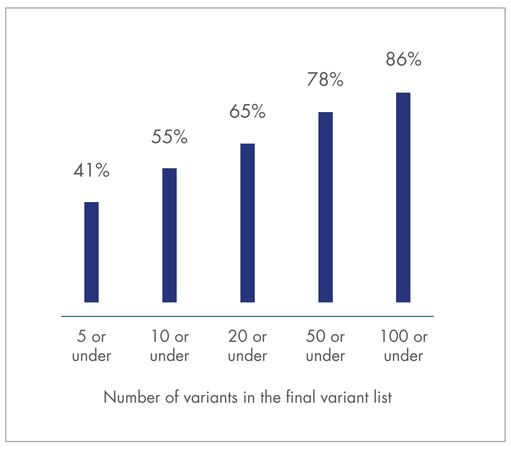


Path-to-Phenotype



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

Number of cases solved:



Summary:

- All 80 cases were solved using Ingenuity Variant Analysis, by re-adjusting the pre-configured filter settings.The causal variant was selected based on the number of phenotypes linked to the gene and/or consistent with known syndromes.
- In 86% cases, we were able to narrow the list of variants to <100, with a 30x enrichment in variants linked to one or more phenotypes.
- In 5% cases, the reported causal variant was >0.5% MAF in the population.

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