RE-DEFINE GENETIC TESTING WITH

ORION
Intelligent Genomics

Powered by

Neuberg DIAGNOSTICS | CENTER FOR GENOMIC MEDICINE

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With an aim to bridge the gap between Phenotype first v/s Genotype first approaches, the Neuberg Center for Genomic Medicine presents:

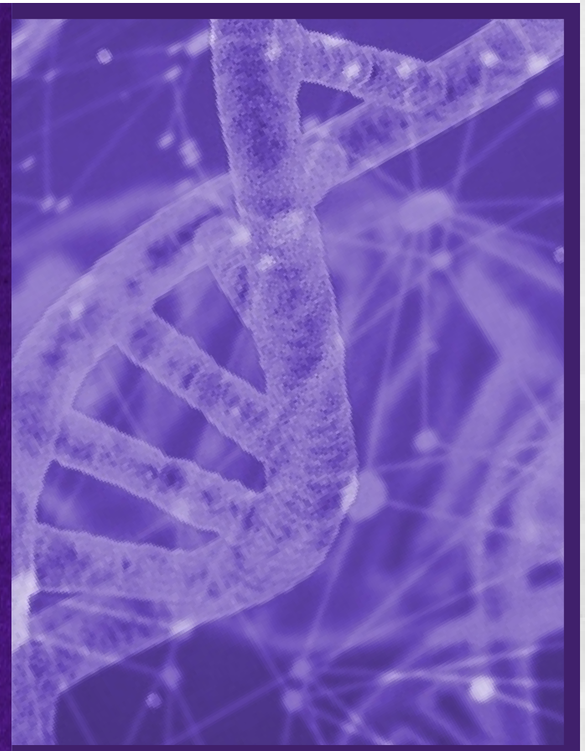
ORION
Intelligent Genomics

COMPREHENSIVE • CUSTOMIZED • SCALABLE

Phenotype based genetic test

ORION HIGHLIGHTS:

- Performed on a backbone of High Quality Clinical Grade Whole Exome Sequencing.
- AI based Phenotype-specific gene and variant curation.
- In-depth variant review by a core clinical team.
- Precision in variant classification by ORIONSeek a proprietary algorithm.



A COMPREHENSIVE BACKBONE

- Clinical grade Whole Exome backbone sequencing in all cases.
- Up-to 20,000 genes sequenced at a mean depth of 100X.
- >95% gene coverage at 20X.
- Sequencing of common clinically relevant intronic regions.
- Sanger backfill of low covered areas when precisely indicated.

CUSTOMIZED ANALYSIS

(Phenotype First + Genotype First Approach)

- **PHENOTYPE FIRST** approach: Phenotype based gene curation by in house expert team of : clinical geneticists, PhD scientists and genetic counselors in conjunction with the referring clinician
- AI based state -of -art algorithms for real time gene updation on a daily basis.
- Custom-developed bio-informatics pipeline for enriched variant calling.
- **GENOTYPE-FIRST** approach : Scaling up analysis to include all genes in negative cases.

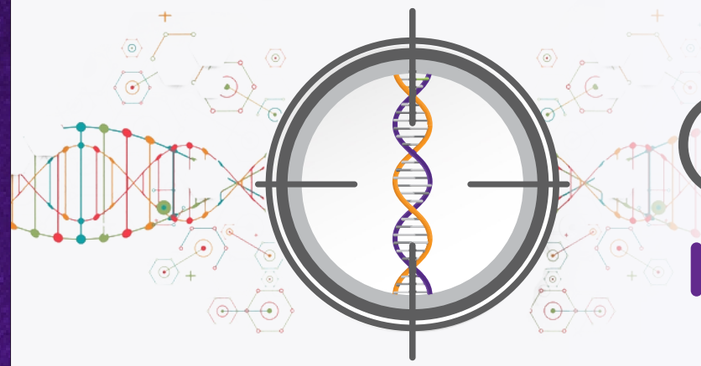
IN-DEPTH VARIANT REVIEW & CLASSIFICATION

- Rigorous analysis of gene-disease relationship.
- Minimizing inter-curator variability with-

ORIONSeek :
A proprietary variant scoring system modeled on the ACMG criteria.

FINAL REPORTING

- Analyzed by a team of clinical geneticists and molecular scientists each report provides : Clinical details, search terms utilized, phenotype -specific gene lists and coverage metrics.
- Reasons for variant classification along with sub-classification of variants of unknown significance.
- Recommendations for further analysis along with access to raw data.



ORION INDICATIONS

- Uniquely designed, ORION combines a phenotype -first approach to a comprehensive. Exome backbone analysis to serve as a BEST TEST-FIT FOR ALL.
- **Applicable to any patient with a suspected genetic disorder irrespective of the phenotype!***
- **INFORMATION REQUIRED :** Informed consent, patient symptoms, investigations performed and family history.

*Excluding triplet repeat expansion, copy number variation, intronic variations & methylation anomalies.

ORION SUCCESS STORIES

- Recently we analyzed a fetus with clinically suspected OI (Osteogenesis Imperfecta) on ORION.
- **Details provided:** Non consanguineous couple. Fetus at 13 weeks gestation with cystic hygroma.
- Previous sibling succumbed to suspected osteogenesis imperfecta: had multiple fractures.

- Osteogenesis Imperfecta panel- No mutation detected.
- Analysis of OI-like disorders: No mutation detected.
- (Hypophosphatasia, hypophosphatemic rickets etc)
- Customized panel curation by in-house team.
- Search terms: fractures OR decreased mineralisation.
- Homozygous previously reported pathogenic variant in TRIP4.

Diagnosis :
Spinal muscular atrophy with congenital fractures

Hunting for Hidden Answers in Your DNA!

Your PARTNERS in HEALTH

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