Abstract Title: Genetic Frontiers: Navigating Rare Diseases through Genome Sequencing

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Abstract: The landscape of genomic medicine has undergone a profound transformation facilitated by exome sequencing (ES) and genome sequencing (GS). While ES has become widely accessible in clinical settings, GS, with its potential to replace ES and chromosomal microarray (CMA) testing, is less prevalent because of various constraints. However, GS is now emerging prominently as the cost of sequencing rapidly decreases. As innovators in the field, we were among the early adopters of clinical genome sequencing, providing diagnostic solutions for patients across the developmental spectrum—from fetal to newborns, pediatric, and adult patients suspected of genetic diseases.

Distinguishing our program is its comprehensive detection capability, covering all variant types reported by individual genetic tests. Our methodical approach involves rigorous patient selection, employing a phenotype-agnostic bioinformatics pipeline, and subsequent manual curation of variants. The integration of phenotypic information is a collaborative effort between clinicians and the laboratory, fostering the exploration of intriguing variants.

Due to its effectiveness, scalability to GS, and resource efficiency, specific elements of our approach can be seamlessly integrated into existing clinical settings. Alternatively, a hybrid model can be implemented within health systems with established genomic medicine programs, encompassing prenatal, postnatal, and adult genetics clinics across various subspecialties. This ensures the provision of genome sequencing testing in a scientifically rigorous yet pragmatic setting, contributing to the advancement of precision medicine.

Area of expertise: Precision Medicine, Genomics