Editorial
https://doi.org/10.6065/apem.24224014edi03
Ann Pediatr Endocrinol Metab 2024;29:141-141

Annals of Pediatric Endocrinology & Metabolism
apem

# Commentary on "Long-read next-generation sequencing for molecular diagnosis of pediatric endocrine disorders"

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The review article,[1] "Long-read next-generation sequencing for molecular diagnosis of pediatric endocrine disorders," provides a comprehensive overview of the benefits of long-read next-generation sequencing (NGS) in the detecting structural variants, repeat expansions, and its applications in difficult-to-sequence regions. This review particularly highlights the importance of haplotype phasing in understanding the genetic underpinnings of autosomal recessive diseases and the parental origin of de novo mutations and emphasizes the role of long-read NGS for enhancing haplotype phasing. For haplotype phasing, binary alignment map (BAM) format from FASTQ files of NGS are analyzed using various methods including HapCUT2, WhatsHap, and SAMTools. Notably, SAMTools is a well-known software tool for processing short-read NGS data for reading, writing, editing, indexing, viewing and phasing BAM formats.[2] However, short-read NGS has limitations due to the short-read length of 100–200 bp, which making it difficult to provide linkage information between distantly spaced single nucleotide polymorphisms. Additionally, it faces challenges with read errors and accurately analyzing complex structural variants and repetitive regions.

Long-read NGS is significantly enhancing haplotype phasing. The extended read lengths enable more accurate analysis of complex structural variants and repetitive regions. This capability is critical for identifying and characterizing structural variations and repeat expansions with high precision.[1] Recent studies have demonstrated that long-read NGS can accomplish complex variant analyses that were previously unattainable with short-read NGS.[3] Additionally, platforms like Oxford Nanopore Technologies have introduced methods for direct haplotype phasing using long-read data, further proving the superiority of this technology for comprehensive genomic analysis.[4] Thus, enhancing haplotype phasing with long-read NGS is expected to play a crucial role in the future of pediatric endocrine disorder diagnosis.

**Conflicts of interest:** No potential conflict of interest relevant to this article was reported

See the article "Long-read next-generation sequencing for molecular diagnosis of pediatric endocrine disorders" via https://doi.org/10.6065/apem.2448028.014.

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ISSN: 2287-1012(Print)
ISSN: 2287-1292(Online)