A novel basic carrier test for all ethnicities using long read sequencing and an artificial intelligence platform.

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Purpose: To develop a novel basic carrier test for all ethnicities using long read sequencing and an artificial intelligence platform.

Background: The prevalence of genetic disorders in the global population is expected to continue increasing. Through development of technologies for early detection, including for carriers of genetic disorders, advancement can be made in efforts to contribute to better disease management.
In this poster presentation, we will focus on Basic Carrier Test, which, as recommended by the American College Obstetricians Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) identifies adult carriers of all ethnicities of genetic disorders Spinal Muscular Atrophy (SMA), Cystic Fibrosis (CF) and Fragile X,

Methods: We developed an Artificial Intelligence (AI) platform for real-time and automated simultaneous analysis and detection of genetic diseases. It enables extensive numerical and visualization analysis and accurate, systematic and timely diagnosis significantly improving disease management.

Results: This platform can be applied directly on the stream of base-called DNA reads generated by the nanopore device. It exceeds the limits of the real-time monitoring and analysis per DNA sample, which can significantly reduce the overall cost. The calculated throughput of the analysis pipeline is 4120 reads per sec measured on a referent hardware architecture using thread parallelism of 10. This platform managed to correctly identify CF, SMA and Fragile X. Following testing and validation on a mix of real and synthetically prepared samples, the sensitivity and specificity exceeded 90%.

Conclusions: This work is a groundbreaking proof of concept that the nanopore low-cost sequencing platform can serve as a certified diagnostic tool with AI-based disease classification for mass-screening significantly reducing time and cost of future diagnostics for any genetic disorder. At the same time, this work opens an opportunity to substitute older diagnostic tools with more accurate, faster and cost-effective technology, and therefore will contribute to improvement of life for people with genetic disorders by improving the current practices in healthcare.