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# Transforming the future of clinical genetic testing

Dr Ahmad Abou Tayoun is focused on improving diagnosis for patients with rare diseases in the Middle East. Find out how he has uncovered 13% more potential diagnoses[1], changing the face of clinical genetic testing.

> ***...our results demonstrate the potential of long-read sequencing as a single unified assay for routine clinical genetic testing and the discovery of novel rare disease variation[1]***

**Dr Ahmad Abou Tayoun**
Clinical Molecular Geneticist

## REVEALING MORE TO TRANSFORM HUMAN HEALTH

## Ask bolder questions

Identifying pathogenic variants in patients with rare disease is essential for timely treatment. With current methodologies, patients can wait for years to receive a diagnosis, and around half remain undiagnosed[1,2]. The diverse spectrum of disease-causing variants requires a flexible platform to provide scalable and cost-effective solutions.

## Ultra-rich data

**13%**
more potential diagnoses[1]

**Novel SMA**
methylation profile detected[1]

**<48 hours**
time to answer[3]

## Reveal more biology

Dr Tayoun *et al*. found that variants of any length and methylation patterns could be identified in a single, whole-genome nanopore sequencing run, uncovering pathogenic changes in 13% of individuals with rare diseases and prior inconclusive tests[1]. He also explored nanopore sequencing for its capacity as a single screening test for SMA, using a cost-efficient, targeted approach[3].

> ***Importantly, the assay is cost-effective and scalable, showing potential for broad implementation in diagnostic and screening programs[3]***

Read more about clinical research with nanopore sequencing


**References**

1. Tayoun, A.A. et al. *Research Square* (2024). DOI: https://doi.org/10.21203/rs.3.rs-4235049/v1
2. Tayoun, A.A. Presentation. Available at: https://nanoporetech.com/resource-centre/london-calling-2023-nanopore-sequencing-potential-diagnostic-tool-genetic-diseases [Accessed 26 April 2024]
3. Hall, B... Tayoun, A.A. *medRxiv* (2024), https://www.medrxiv.org/content/10.1101/2024.02.22.24303180v1


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