UNIVERSITÀ di VERONA | Dipartimento di BIOTECNOLOGIE

"Whole-genome sequencing as a first-tier genetic test in pediatric field"

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Abstract

Genetic testing is a fundamental diagnostic component in pediatric medicine. The standard of care is often a time-consuming, gradual step-by-step approach, involving the analysis of chromosomal arrays and targeted gene sequencing panels, which can be costly and inconclusive. Whole genome sequencing (WGS) provides a complete analytical platform with the potential to simplify the genetic evaluations, but its clinical implementation is limited by challenges in data interpretation.

We have prospectively recruited a set of pediatric patients with a clinical phenotype indicative of a genetic disease, and compared the diagnostic yield and coverage of WGS with those of conventional genetic tests. Here, we report the comparative results between analysis based on gene panels, exome sequencing (WES) and WGS and other common tests, in pediatric patients with different phenotypes.

The lecture will take place at the **Department of Biotechnology,** Cà Vignal 2, Strada Le Grazie 15 **– at 10.30, Aula H.**

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