In Saudi Arabia, a Hospital Uses Genomics to Rebuild the Map of Rare Diseases

RIYADH, SAUDI ARABIA, November 28, 2025 /EINPresswire.com/ -- In a region where hereditary diseases appear more frequently than in many parts of the world, emerging genomic programs are helping clarify why certain conditions recur across extended families and how early intervention can influence long term outcomes.

As part of this effort, King Faisal Specialist Hospital and Research Centre has expanded its work in rare disease genomics through programs that blend prevention, population specific sequencing, and focused investigation of inherited disorders. The approach reflects a broader strategy to ease the burden of genetic disease while improving diagnosis and treatment pathways.

One of the central components of this work is the hospital's Preimplantation Genetic Diagnosis program, which screens embryos for more than fifteen hundred genetic disorders. By identifying unaffected embryos before pregnancy, the program prevents hereditary diseases from being passed to future generations and offers families a preventive model that is uncommon in the region.

Alongside this preventive effort, researchers are conducting large scale Whole Genome Sequencing to identify genetic markers relevant to populations in the region, where consanguinity contributes to higher rates of inherited conditions. The data generated through this sequencing supports earlier diagnosis and more individualized care for both rare disorders and widely prevalent illnesses such as cancer, cardiovascular disease, and diabetes.

Recent findings illustrate the impact. Investigators diagnosed hereditary osteomalacia in thirty eight members of a single family across three generations, an insight that is shaping future clinical approaches and highlighting how complex conditions can remain undetected without genomic tools. The hospital also contributes nearly ten percent of all global entries in the Online Mendelian Inheritance in Man database, underscoring its role in documenting rare disorders

worldwide.

Its genomic work extends into infectious disease as well. Through whole genome sequencing, scientists identified a previously unknown species of antibiotic resistant bacteria known as Stenotrophomonas riyadhensis. The discovery adds to the global effort to understand antimicrobial resistance and informs research into emerging threats.

King Faisal Specialist Hospital and Research Centre is ranked first in the Middle East and Africa and fifteenth globally among the top two hundred fifty academic medical centers for 2025. It is also recognized as the most valuable healthcare brand in the Kingdom and the Middle East according to Brand Finance 2025 and appears on Newsweek's lists of the World's Best Hospitals 2025, Best Smart Hospitals 2026, and Best Specialized Hospitals 2026.

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