

KFSHRC Advances Rare Disease Care Through Pioneering Genomics Programs

TOKYO, JAPAN, September 26, 2025 /EINPresswire.com/ -- King Faisal Specialist Hospital & Research Centre (KFSHRC) has reinforced its global leadership in rare disease care by advancing pioneering genomics programs that are transforming the understanding, diagnosis, and treatment of hereditary conditions. These initiatives reflect the hospital's strategic vision to reduce the burden of genetic diseases and to build a healthier society through innovation and early intervention.

At the core of these efforts is the hospital's Preimplantation Genetic Diagnosis (PGD) program, which screens for more than 1,500 genetic disorders. By enabling the selection of healthy embryos before pregnancy, the program prevents hereditary diseases from being passed to future generations, establishing a new preventative model in public health.

Complementing PGD is KFSHRC's large-scale Whole-Genome Sequencing (WGS) initiative, which seeks to identify genetic markers unique to the Saudi population, where high rates of consanguinity contribute to increased prevalence of inherited disorders. This approach holds transformative potential for precision medicine, enabling early diagnosis, individualized treatment plans, and targeted therapies for both rare and common conditions such as cancer, cardiovascular diseases, and diabetes.

KFSHRC's research breakthroughs continue to elevate its global standing. The hospital successfully diagnosed 38 individuals across three generations of a single family with hereditary osteomalacia, laying a foundation for improved therapies. It also contributes nearly 10% of all global entries in the Online Mendelian Inheritance in Man (OMIM) database, underscoring its role as one of the world's most influential rare disease research hubs.



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Further highlighting its innovation, KFSHRC discovered a novel antibiotic-resistant bacterial species, *Stenotrophomonas riyadhensis*, through WGS, reinforcing its contribution to global efforts against antimicrobial resistance and aligning with Saudi Vision 2030's drive to place the Kingdom at the forefront of healthcare innovation.

Building on this leadership, KFSHRC continues to share its expertise in global forums such as the C3 Davos of Healthcare™ Summit in Japan, where the hospital underscored the role of genomics and precision medicine as catalysts for future healthcare systems, emphasizing the need for international collaboration to address rising global health challenges.

KFSHRC has been ranked first in the Middle East and North Africa and 15th globally among the world's top 250 Academic Medical Centres for the third consecutive year, and was recognized as the most valuable healthcare brand in the Kingdom and the Middle East, according to Brand Finance 2025. The hospital was also named among the World's Best Smart Hospitals for 2025 by Newsweek.

Riyadh

King Faisal Specialist Hospital & Research Centre

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