2nd International Conference on

Rare Diseases and Orphan Drugs

February 16-17, 2026 | Rome, Italy

Volume: 15

Rapid Genome Sequencing for Critically Ill Children with Rare Diseases

Stephen Kingsmore

Rady Children's Institute for Genomic Medicine, United States

Rapid whole-genome sequencing has emerged as a transformative approach for diagnosing critically ill infants and children with suspected rare genetic disorders. Traditional diagnostic pathways often require weeks or months, delaying targeted treatment and increasing healthcare costs. Recent advances in next-generation sequencing technologies, bioinformatics pipelines, and clinical interpretation have enabled comprehensive genomic analyses to be completed within hours or days. This presentation highlights the integration of rapid genomic diagnostics into neonatal and pediatric intensive care settings, demonstrating their impact on diagnostic yield, clinical decision-making, and patient outcomes. Case studies illustrate how early genetic diagnosis facilitates personalized therapeutic interventions, informs prognosis, and supports family counseling. Furthermore, the presentation discusses challenges associated with variant interpretation, ethical considerations, multidisciplinary collaboration, and equitable access to genomic medicine. Future directions include the incorporation of artificial intelligence for variant prioritization, global genomic data sharing, and the development of precision therapies for rare disorders. These advances are expected to further enhance personalized healthcare for patients with rare genetic diseases.

Biography

Stephen Kingsmore is a pioneer in pediatric genomic medicine and rapid whole-genome sequencing. His work has transformed the diagnosis of critically ill children with suspected genetic disorders by significantly reducing the time required to identify disease-causing variants. His research focuses on implementing precision medicine in neonatal and pediatric intensive care settings, improving clinical outcomes through earlier diagnosis and targeted interventions. He has authored numerous high-impact publications and has received international recognition for advancing genomic medicine and rare disease diagnostics.