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Medical Genetics for the Pediatrician: "How much do you really need to know?"

The Aim of this presentation is to give an overview of the rapidly growing field of clinical genetics as it relates to Pediatrics. It aims to equip doctors with some basic clinical skills to tackle their encounter with a suspected genetic disorder. This is achieved by giving an overview of the types of genetic disorders encountered in everyday practice. By introducing the clues to suspect a genetic disease and a guide of what to do afterwards. By explaining the underlying rationale of why we need to diagnose genetic syndromes, and how we go about doing that. Practical examples from the clinic will be given throughout the presentation. With these examples, some sophisticated molecular and cytogenetic investigative tools will be introduced- including when they are or are not indicated, and what to do with the intriguing results.

Speaker Biography

Jumana Y Al-Aama is a Professor and Consultant of Medical Genetics, Chairman of the Department of Genetic Medicine and Director of the Princess Al-Jawhara Al Brahim Center of Excellence in Research of Hereditary Disorders at King Abdulaziz University (KAU), Jeddah, Saudi Arabia. She is Certified by the Canadian College of Medical Genetics and by the Royal College of Pediatrics and Child Health. She is a member of the board of Directors of the Princess Aljawhara Center of Molecular Medicine and Genetic Disorders at the Gulf University in Bahrain. She is also the Founding Specialty Chief Editor of (Frontiers in Genetic Disorders and Frontiers in Pediatric Genetic Disorders) based in Geneva, and an academic editor for the PLOS One journal of Science. She was Visiting Professor at Beijing Genomic Institute (BGI) College in China from December 2012 – 2016. She has 116 publications in international journals with 3932 citations and H-index of 26. She has won several research awards including Best Health Sciences Researcher at KAU for 2018. She has given invited talks in international conferences in several countries including France, Spain, China, Malaysia and others.

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