Abstract Title: A de novo paradigm for sporadic genetic disease

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Abstract: De novo germline and somatic mutations can cause disease when they affect functionally relevant bases in the genome. Our studies using exome and genome sequencing have shown for the first time that these mutations may be the major cause of early-onset disorders such as severe intellectual disability. In spite of this we still know very little of the causes and consequences of these important mutations in health and disease. What is the effect of de novo non-coding mutations? How many de novo mutations does a child "inherit" from her 50 year old father, with or without the use of assisted reproductive technology? Can de novo mutations be the cause of other conditions with reduced fecundity such as male infertility? These are the some of the questions which I will address in this presentation, highlighting the importance of well-characterized patient/control cohorts and high quality next generation sequencing technology in combination with state-of-the-art bioinformatics.

References:

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Area of expertise: Genomics, disease gene identification, rare disease, human reproduction