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Use of carrier exome screening for prevention of childhood malignancies and other severe congenital abnormalities

Our work with the Columbia University Medical Center Precision Medicine for Kids with cancer (PIP-seq) project has shown that a significant percentage of pediatric malignancies arise because of genetic predisposition. We can consider cancer predisposition as a specialized case of the parents being carriers for a specific genetic disorder. MNG laboratories has developed a whole exome carrier test designed to fulfill the preconception genetic counseling needs of couples who suspect, based on shared ethnicity or family history, that they are potentially at risk of having children with debilitating health problems, including development of pediatric malignancies. In my presentation, I will discuss the technical, practical and ethical considerations to make such testing widely available for risk couples.

Biography

Peter L Nagy is Board Certified in Anatomic and Molecular Genetic Pathology. He was a Principal Investigator on multiple NIH funded studies at University of Iowa and Columbia University and served as an Associate Director of Personalized Genomic Medicine Laboratory at Columbia University Medical Center. He has established an array of clinical next-generation sequencing tests including whole exome and transcriptome testing for constitutional and somatic (cancer) disorders. At Medical Neurogenetics Laboratories, he is continuing his groundbreaking work on developing computational tools to improve the clinical specificity and sensitivity of next generation sequencing data analysis.

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