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The role of clinical genomic testing in treatment discovery for rare neurodevelopmental diseases

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Genomic testing by high resolution chromosomal microarray (CMA) is the guideline-recommended first tier test for neurodevelopmental disorders. Widely used in the clinical setting, accurate and informative interpretation of CMA results can enhance not only the diagnostic understanding of, but also the medical management of, these often rare genetic conditions. We will present the results of our efforts to bring the power of ultra-high resolution microarray analysis, combined with newly developed tools and relational databases, to bear on the complex challenges of interpretation of genomic data. Using our custom microarray optimized for the detection of known critical genomic changes associated with neurodevelopmental disorders, we have performed over 10,000 consecutive CMAs on a US-based, neurodevelopmentally-affected pediatric population. We detected relevant copy number variants (CNV) in approximately 30% of this population, a rate which depends on patient age and indication for testing. A significant proportion (~20%) of these finding were classified as variants of unknown significance (VOUS). We have developed novel technologies and approaches in partnership with patient support groups and members of the medical and academic research communities to bring additional interpretative power to bear on these VOUS. As an example of the clinical utility of ultra-high resolution CMA to map critical genes, we recently reported the identification of a seizure susceptibility candidate region/gene for Wolf-Hirschhorn Syndrome (WHS). Subsequent work using novel analysis techniques has led to identification of additional genes potentially related to congenital heart defects and other conditions associated with WHS. Using these strategies, we have correlated fine-resolution genetic mapping with other rare conditions and predicted potential molecular mechanisms connecting various rare diseases to one another. This in turn impacts the potential for common pharmacotherapeutic development strategies for previously unrelated orphan disorders.

Biography

Karen S Ho is a Principal Scientist of Translational Research Initiatives at Lineagen, Inc., where she is working since five years. She holds MSc degree in Genetics from Cambridge University where she was a Marshall Scholar after graduating summa cum laude from Washington University with a BSc in Biochemistry. She holds a PhD in Developmental Biology from Stanford University and completed her Postdoctoral training as a Howard Hughes Medical Institute Fellow and National Sleep Foundation Fellow in the Department of Neuroscience at the University of Pennsylvania. She is also an Assistant Adjunct Professor in the School of Medicine, Department of Pediatrics at the University of Utah and serves on the Board of two non-profit foundations, NGLY1.org and Rare and Undiagnosed Network, both of which are dedicated to rare disease.

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Notes:

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Page 54