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Development of targeted re-sequencing approach for muscular dystrophy diagnosis

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With the rapid expansion of knowledge regarding monogenic disorders, there are now often many genes associated with particular phenotypes. For example, in the muscular dystrophies, there are over 47 causative genes identified to date, with few if any phenotypic features able to distinguish between the different underlying genetic causes. Many of the muscular dystrophy genes on the panel are quite large (e.g. nebulin–147 exons;dystrophin – 79 exons; dysferlin – 55 exons). As a result, patients and physicians can often pursue an extended molecular genetics odyssey, involving send-outs to different laboratories, with commensurate high costs and long turn-around times. An alternative approach is nextgen sequencing, where all candidate muscular dystrophy genes are sequenced in parallel through either targeted re-sequencing or exome sequencing. We compared three approaches to nextgen sequencing in muscular dystrophy patients; RainDance targeted amplification of 1,841 amplimers covering the exons and exon/intron boundaries of 47 muscular dystrophy genes, and exomic sequencing (with and without a 47 gene filtering step). The RainDance unit fuses individual microbubbles of the amplimer mix with patient DNA. The emulsion is then PCR amplified, and amplimers were sequenced using 2nd(Illumina) or 3rd generation (Pacific Biosciences) sequencers. In parallel, whole exome sequencing was done on muscular dystrophy patients and was analyzed with or without the 47 gene filter. Data was assembled and analyzed using the NextGENe software and variants were called and reported accordingly.The observed advantages and disadvantages of each approach will be described.

Biography

Akanchha Kesari is molecular biologist and geneticist by training; she had pursued a number of complex and interdisciplinary projects, although the focus is always on human genetic disease. Her research project has been focused to define the molecular basis for the muscular dystrophies, both with regards to identification of causative genes, and biochemical pathways immediately downstream of the primary genetic defect. Recently, she has been awarded the T32 grant in Genetics and Genomics of Muscle from NIAMS (NIH) to purse my reasearch in development of next-generation sequencing technology for muscle disease. Along with that, she is also in the fellowship training program of National Human Genome Research Institute (NHGRI) in Clinical and Molecular Genetics. Suceessful completion of the trianing will help her to become the certified molecular geneticist by American Boards of Medical Genetics (ABMG).

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