Duchenne muscular dystrophy: Advances in molecular appraoch

Sir,

Duchenne muscular dystrophy (DMD) is the most common muscular dystrophy affecting one in 3,500 live male births.[1] DMD is an X-linked recessive disease that affects boys. The disease results from degeneration and loss of muscle fibers.[1] The natural history of DMD typically affected boys is becoming wheelchair dependant about age 12.[2] The DMD gene (DMD) is the biggest human gene (2.5 Mbp). Once the gene was identified, it was established that affected boys were lacking dystrophin, the protein product of the gene.[2] The most common molecular defect in the DMD gene, accounting for approximately 65% of cases of DMD is deletion of one or more exons. Duplication accounts for 6-10% of cases.[2] The minimum level of diagnosis testing that should be undertaken is a screening that detects the majority of exonic deletions. The most basic method still in regular use involves multiplex PCR (Polymerase Chain Reaction) of the exons known to be most commonly deleted (Chamberlain *et al.* 1988).[3] The multiplex PCR is relatively simple; However, it does not detect duplications, does not characterize all deletion breakpoints, and cannot be used for carrier testing of females. Quantitative analysis of all exons of the gene has brought an improvement in mutation detection rate, as it may detect all exon scale deletions as well as duplications. It also fully delineate the exon boundaries of detected mutations and detect mutations in carrier females. Among the quantitative methods available, multiplex ligation-dependent probe amplification is now the most widely used[4]. An important point to emphasize on is that with both methods that we've just described, any apparent mutation indicated by an abnormal reading from a single probe must be confirmed by an alternative method, since there is a possibility of a single nucleotide polymorphism under a probe or primer binding site. Finally, oligonucleotide-based array comparative genomic accounts for the last generation of quantitative analysis technics. Also known as Chromosomal Microarray Analysis (CMA) is increasingly used in DMD prenatal diagnosis throughout the world. However, routine practices DMD diagnosis are very different among centers.[5]

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Quick Response Code:	Website: www.ijhg.com
	DOI: 10.4103/0971-6866.112931