THE USE OF CLONED DNA PROBES IN THE DETECTION OF CARRIER STATUS OF X-LINKED DUCHENNE MUSCULAR DYSTROPHY

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Duchenne muscular dystrophy (DMD) is an X-Linked recessive muscular disorder which affects about 1 in 2500 males. The first symptoms appear by the time the affected boy begins to walk. They are wheelchair-bound by the age of 10 and usually death results in the third decade of life. This genetically lethal disorder is transmitted by carrier females who rarely show clinical manifestations[1,2]

The diagnosis of DMD has been previously based on clinical features supported by electromyography, muscle biopsy and enzyme assays[3]. This approach is however unsatisfactory in the diagnosis of carriers, as the results of the above investigations are inconclusive.

In the present preliminary study, we have used recombinant DNA techniques previously not available in Sri Lanka, for the detection of DMD carriers[4]. The inheritance of DMD was studied in 12 families using several X chromosome specific cloned DNA sequences, detecting 'Restriction Fragment Length Polymorphisms' (RFLPs) within and flanking the DMD locus. The results were used to identify RFLPs that were associated with the disease in the respective families. The information was used to identify carrier females and to provide appropriate genetic counselling. (This work is supported by the International Program in the Chemical Sciences and SAREC).

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