# Duchene Muscular Dystrophy

X-linked recessive disorder in a girl child

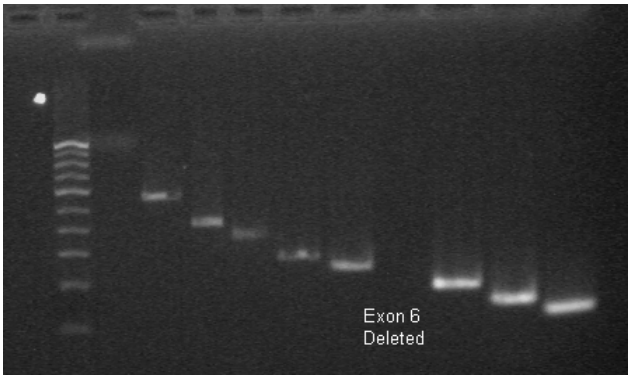
**Case of the Month Series 1004**

## Muscular dystrophy like symptoms in a girl child

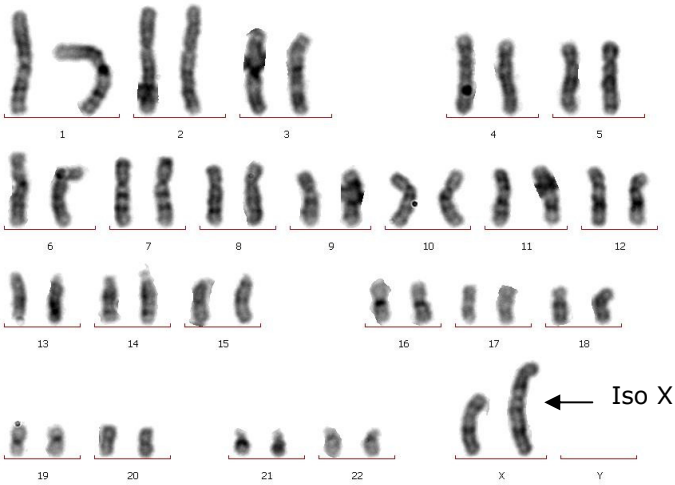
A family with a 3yr old child with growth retardation (?) was referred to GeneTech. The child was normal with no dysmorphology but with reported weakness in muscles and delayed motor skills. Although muscle weakness is a symptom of muscular dystrophies, there was no report to confirm diagnosis. Moreover, Duchene or Becker dystrophies (DMD and BMD) are X-linked recessive disorders affecting mostly boys (1 in 4,000 newborn males). Females are rarely affected by these forms of muscular dystrophy.

## Clues from the Pedigree

During the genetic counseling session we have gathered that atleast 3 males (including mother's brother) were affected with Muscular Dystrophy showing the possibility of X-linked Muscular dystrophy in the family.




Maternal Uncle's DNA analysis: Exon 6 deleted




Additional ISO X anomaly in the child along with Exon 6 deletion

## Recommended Tests and results

1. DNA analysis of maternal uncle confirmed Dystrophin gene mutation (Deletion of Exon 6 on Dystrophin gene)

2. As PCR analysis will not be able to detect Dystrophin gene mutations in female carriers, PCR test for DMD was not recommended. Instead to rule out chromosomal rearrangements, karyotyping of the child was recommended which showed an Iso X condition (One X chromosome is abnormal with duplication of q arm)

## Explanation

With the confirmation of DMD in maternal uncle, it must be assumed that mother is a carrier of the deletion and so is the affected child. All females who are carriers are presumably normal because there is another normal X to take care of normal functioning of Dystrophin gene. But in this case the normal X without deletion is having a chromosomal rearrangement leading to non functioning of dystrophin gene on normal X. With both Dystrophin genes being Non-functional, the female child is affected with Duchene Muscular Dystrophy.

## Recommendations for future pregnancies

Prenatal diagnosis by DNA analysis for Dystrophin Gene Mutations and Fetal karyotyping.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