March 1999 (Volume 40, Number 3)

Prenatal Diagnosis of Spinal Muscular Atrophy Type I (Werdnig- Hoffmann) by DNA Deletion Analysis of Cultivated Amniocytes

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Aim. Presentation of a prenatally diagnosed case of Werdnig-Hoffmann disease, the most severe type of spinal muscular atrophy.
Methods. DNA obtained from cultivated amniocytes was analyzed for deletions in the survival motor neuron gene and neuronal apoptosis inhibitory protein gene.
Results. The fetus was diagnosed as an affected homozygote for deletions in exon 7 and exon 8 of the survival motor neuron gene. No deletions of exon 5 in the neuronal apoptosis inhibitory protein gene were found.
Conclusion. Direct DNA deletion analysis of the survival motor neuron gene and neuronal apoptosis inhibitory protein gene in affected families represents a highly reliable and fast method for prenatal diagnosis of Werdnig-Hoffmann disease.

Key words: amniotic fluid; gene deletion; muscular atrophy, spinal; polymerase chain reaction; Werdnig-Hoffmann disease