

Diagnosis of an embryo affected with spinal muscular atrophy - SMA

On 04.04.2013 a prenatal amniotic fluid sample was referred to our center in order to perform routine prenatal chromosomal diagnosis. The referral reason was advanced maternal age.

In parallel with the chromosomal analysis, a portion of the sample was used for genetic testing of spinal muscular atrophy - SMA, which we had just started to apply as a routine supplementary test in all prenatal chromosomal diagnosis cases.

Genetic testing revealed that the fetus was affected with spinal muscular atrophy - SMA, being homozygous for the known pathological deletion of exons 7 and 8 of the SMN1 gene.

Subsequent genetic testing of the parents immediately revealed that both were (unsuspecting) carriers of the disease, having a 25% risk for affected children.

This case clearly highlights the particular value of the routine prenatal screening we introduced for spinal muscular atrophy-SMA, preventing the unexpected birth of even one, up to now, affected child while also providing the knowledge of the risk and the prevention in subsequent pregnancies in the family.