



Prenatal features of neurofibromatosis type 1

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Objective

Neurofibromatosis type 1 (NF1) is one of the most common autosomal dominant disorders affecting approximately 1/3500 individuals in all ethnic groups. Usually the neurofibromatosis type 1 is recognised postnatally.

Methods

This is a case report.

Results

An ultrasound (US) examination was performed in a 26 years old nulliparous woman at 27 weeks' gestation after referral for suspected fetal anomalies. The scan revealed the following associated anomalies: shortening and flexion of the femur, scoliosis, right hydronephrosis, double outlet right ventricle, ocular hypertelorism and dysmorphic facial features. The fetal karyotype was normal. The pregnant woman and her mother were suffering from mild neurofibromatosis (presenting with café-au-lait spots only). At 31 week of pregnancy a C-section had to be performed because of the fetal distress. After birth the defects observed prenatally were confirmed. We found additionally a single café-au-lait spot on the left leg. The newborn died on the third day of life. The additional microarray test confirmed the diagnosis of neurofibromatosis type 1 in the newborn and his mother. We detected a large deletion involving the NF1 gene.

Conclusion

Our report shows that neurofibromatosis type 1 may be a cause of serious fetal defects even in families with only mild symptoms.

