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Developmental outcome of isolated fetal macrocephaly

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Objective: Macrocephaly is defined as a head circumference greater than 2 standard deviations. The counseling regarding the prognosis of macrocephaly diagnosed in-utero is difficult due to lack of available studies. The neuropsychological outcome of children with macrocephaly is not clear due to contradictory results in the literature. This study was design to evaluate the neurodevelopmental outcome of children with prenatally diagnosed isolated macrocephaly.

Methods: We evaluated the performance of 17 children, aged 2-7 years diagnosed in utero as macrocephalic and compared them to 17 normocephalic children, utilizing a standardized neuropsychological battery, which included attribution to cognitive, emotional and behavioral domains.

Results: All the fetuses in the study group had a head circumference between two and three standard deviation (SD). No significant differences were found between the groups on the cognitive, language and motor domains. The study group scored significantly lower than the control on three parameters reflecting executive functioning, behavior and social-emotional development. Children with familial macrocephaly showed significantly better executive functioning compared to children with non-familial macrocephaly. Multiple linear regression analysis found paternal head circumference as the only significant variable in positively predicting the cognitive functioning of the child.

Conclusions: Our results indicate that the prenatal diagnosis of isolated macrocephaly is not a risk factor for abnormal long-term neuropsychological development. Paternal macrocephaly was found to be most important in positively predicting cognitive performance of the child.