

Case report of prenatal diagnosis of Stüve-Wiedemann Syndrome in a woman with another child affected too.

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ABSTRACT

OBJECTIVE: Stüve-Wiedemann Syndrome (SWS; MIM 601 559) is an autosomal-recessive syndrome characterized by myotonia with mask-like face, skeletal dysplasia and intrauterine growth restriction. Other clinical findings are pursed mouth, hypoplastic midface, congenital contractures and muscular hypotonia. We discuss about the importance of prenatal diagnosis in SWS and the possibility of survival after the first year of life in patients suffering from this disease.

METHODS: we report a case of Stüve-Wiedemann Syndrome detected by morphological examination in our Operative Unit. Prenatal presumptive diagnosis was given with two-dimensional and 3-D probe, during the second trimester of pregnancy. Caesarean section was performed at 38(th) week of gestation. Then diagnosis was genetically performed.

RESULTS: at birth, clinical examination was concordant with the ultrasound findings. Genetic analysis also confirmed the presumptive diagnosis. Episodes of respiratory distress and hyperthermia decreased until it disappeared altogether at 1 year of age.

CONCLUSION: we underline the usefulness of ultrasound study of fetal skeleton in the prenatal diagnosis. It allowed us to do an early detection of birth defects and their appropriate management.