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Stuve-Wiedemann syndrome in one family: diagnosis and outcome

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Objective

This is a case report of one family with two live children affected by the autosomal recessive skeletal dysplasia Stuve-Wiedemann Syndrome (SWS). As the majority of SWS cases did not survive beyond infancy as reported in older cases, a poor outlook was predicted. However, both children did well in infancy with the eldest child now surviving beyond three years.

Methods

This is a case report.

Results

Our patient had a normal first trimester scan at 13 weeks in her fourth pregnancy then had minimal antenatal input until her return at 38 weeks gestation, after moving abroad. Her first two pregnancies sadly resulted in early neonatal deaths: abroad with no medical details known. Fetal medicine review in her third pregnancy revealed severe short long bones with possible fractures. Appearances suggestive of skeletal dysplasia, but invasive testing was declined. Neonatal genetic testing confirmed Stuve-Wiedemann Syndrome. Karyotype confirmed the parents as carriers for the LIFR gene variant c.756dup, p. Lys253Ter, putting offspring at 25% risk of inheritance. In the index pregnancy, the first fetal medicine scan at 38 weeks revealed short long bones with a curved appearance of both femora: suspicious of Stuve-Wiedemann Syndrome, like her previous child. Invasive testing was declined again. Post-natal skeletal survey showed characteristic bowing of tibia bilaterally consistent with SWS; confirmed by genetic analysis. The child is now 16 months old and doing well.

Conclusion

Stuve-Wiedemann Syndrome is a rare autosomal recessive skeletal dysplasia caused by a biallelic loss-of-function variant in the leukaemia inhibitory factor receptor (LIFR) gene. First described by Stuve and Wiedemann in 1971, it is considered to have significant mortality; few survive the first year. Cases are characterized by bowed long bones, joint restrictions, dysautonomia, respiratory and feeding issues.