Pena-Shokeir phenotype: A lethal pattern of multiple congenital anomalies.

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Abstract

Pena-Shokeir phenotype is a unique pattern of lethal multiple congenital anomalies with variable expression. The history of a family in which two sibs conform to Pena-Shokeir phenotype is detailed and substantiates the criteria for definition of the phenotype. The addition of a peculiar ultrasound presentation in the antenatal period may indicate the condition.