Myhre syndrome, caused by a heterozygous mutation in the SMAD4 gene, 18q21, is characterized by mental retardation, dysmorphic facial features, microcephaly, midface hypoplasia, prognathism, blepharophimosis, skeletal anomalies and CV defects. Currently, 60 cases were reported, all post-natal.

36 y.o. patient, G3P2. NT = 3.1 mm. CMA = normal. US scans & low dose 3D CT: SGA male fetus, short long bones, abnormal skull shape, dysmorphic facial features, small hands and thickened edematous subcutaneous tissues. Trio WES: **SMAD4**: c.1498A>G; p. Ile500Val (het, de novo) NM_005359.5, rs281875322 = defect in SMAD4 ubiquitination, and a decreased expression of the downstream TGF-b target genes. PM of the abortus = typical facial dysmorphism of Myhre syndrome.