Introduction: Prenatal diagnosis of NEK 9 mutation, lethal congenital contracture syndrome 10 in a WES in the second trimester have been previously reported but is uncommon. It is reported in 2 traveller families in Ireland by Casey et al in 2016 and in diagnosis of lethal or prenatal-onset autosomal recessive disorders by Stals et al 2017.

Case report: A 28-year old, G2 P0 as referred to our tertiary centre with an incomplete structural ultrasound examination as a result from obesity and position of the foetus. First trimester screening has not been done. Detailed scan at 20+3 weeks showed severe fetal abnormalities: bilateral talipes, scoliosis, bilateral clenched hands, short long bones, bowing of the femura, abnormal ribs with a small thorax, small chin, polyhydramnios and very little fetal movements.

The couple underwent amniocentesis. QF-PCR for common trisomies was normal, microarray CNV analysis was normal. WES gave a NEK 9 mutation compound heterozygous as described in fetal akinesis with skeletal dysplasia and long hypoplasia, lethal congenital contracture syndrome 10.

Conclusion: Prenatal diagnosis of NEK 9 mutation, lethal congenital contracture syndrome 10 in a WES in the second trimester have been previously reported but is uncommon. We describe severe skeletal dysplasia, polyhydramnios and fetal akinesia. Our case underscores the value of WES screening in severe fetal abnormalities.