Introduction - Case report
We present the case of a 24 year old, second gravida. At special ultrasound scan in 21/4 w.o.g. we found multiple abnormalities of the head (fig. 1). Via amniocentesis we identified a normal caryotype, 46 XY. Through next generation sequencing the genetic diagnosis of a MPPH-1-syndrome could be confirmed as a pathogenic change in the PIK3R2-gen. After a difficult counseling of the parents they continued pregnancy until an unexpected intrauterine fetal death at 34+4 w.o.g.

Conclusion
Possible early prenatal diagnosis of extremely rare diseases and syndromes, thanks to the advances with genetic diagnostic tools will increase and confront us with new challenges in the consulting and care of affected expectant mothers.

We did not expect the intrauterine fetal death in this constellation. There are possibly other effects of the disease that may cause unknown molecular changes and further problems not yet explored.

Megalecephaly-polymericyria-polydactyly-hydrocephalus (MPPH) -syndrome
According to orphanet only 6 case reports worldwide refer to a PIK3R2-related MPPH-1-syndrome. The fetal head circumference may be enhanced up to 6 standard deviations; the brain is significantly larger. Typical structural changes are perisylvian polymicrogyria and a ventriculomegaly (in nearly 50% severe). Around 50% of patients show postaxial polydactyly on hands or feet. Main problems are moderate to severe deficiencies in neurological and intellectual development, orofacial muscle dysfunction causing dysphagia, as well as epilepsy in about 50% of cases. Treatment is therefore symptomatic and an intense long-term home care has to be expected.