28 year old G5 P1031, in a consanguineous marriage, referred for sonography at 14w1d (by dates) due to a suspected head abnormality. Composite fetal biometry was consistent with 12w6d. Past history was notable for early second trimester losses of anomalous fetuses.

On ultrasound, the fetus measured 67.9 mm and had an NT of 1.1 mm. A clear butterfly was noted and the orbits, nasal bone, maxilla and mandible were seen. The fetal stomach, bladder, cord insertion and spine were seen as well. However, there was an occipital encephalocele, the kidneys were enlarged and polycystic, and the digits demonstrated polydactyly. On assessing the fetal heart, an atrioventricular septal defect (AVSD) was noted. Differential diagnosis included trisomy 13 and 18, autosomal dominant polycystic kidney disease and Meckel-Gruber Syndrome (MGS).

In our case, the fetus manifested the classic triad of MGS: an occipital encephalocele, polycystic kidneys and polydactyly. In addition there was an AVSD. Though trisomy 13 and 18 and autosomal dominant polycystic kidney disease were in the differential, the fact that this was a consanguineous couple, with a prior history of anomalous fetuses, made MGS the most likely diagnosis. The family elected termination of pregnancy. They declined a postmortem examination or a genetic workup.

Given its lethality, early diagnosis of MGS, as well as other such lethal anomalies, has a tremendous impact on counseling the family particularly when termination of pregnancy may be a consideration.