Introduction: Kabuki syndrome (incidence of 1 in 32,000 newborns) is a genetic disease caused by mutations in the KMT2D gene. The syndrome features include characteristic facial features (as makeup from traditional Japanese Kabuki Theater), developmental delay, hip, knee joints anomalies, cleft palate, unusual fingerprints with features and fleshy pads.

Case content: We report a case of a 28 years patient, with a previous normal child that presented for the anomaly scan at 24 weeks. The 11-13th weeks scan was reported as normal and combined risk was low. We found bilateral double kidney, single umbilical artery, nonspecific dysmorphic facial aspect and polyhydramnios. Amniotic liquid result was 46xx. On financial reasons the whole genome CG array was not accepted. The pregnancy evolution was uneventful and a 3100 g baby girl was delivered at term. At birth the baby had a posterior palate cleft that was overlooked on ultrasound. After birth many anomalies evolved hydrocephalus, dysmorphic features (cleft palate), delayed motor development, muscle hypertonia and hypotonia, microcephaly, palpebral ptosis, external ear malformation, stenosis of pulmonary artery, abnormal urinary system, and aganglionic megacolon.

Conclusion: Ultrasound is a useful tool to trigger genetic anomalies but without the availability of the extended genetic examination prenatal diagnosis is not possible. Prenatal diagnosis of the Kabuki syndrome cannot be made only on ultrasound signs that are not specific.