We present a 35 years old patient, G3P1A1, referred to our high risk center for an abnormal morphologic ultrasound at 25 weeks of pregnancy. The fetal ultrasound in our center demonstrated multiples abnormalities: a craniosynostosis in clover, a protrusion of the eyes balls and thumbs anomalies. Vertebral anomalies were also seen at the ultrasound which is quite unusual in this pathology. A fetal magnetic resonance imaging (MRI) showed a craniosynostosis, a dysmorphia, thick thumbs, a short trunk, a thoraco-abdominal disproportion, a humero-radial synostosis.

An amniocentesis was performed and found a mutation in the FGFR2 gene (c.1019A>G). The patient decided to terminate the pregnancy at 28 weeks of pregnancy. An X-ray of the fetal skeleton was performed and confirmed the ultrasound abnormalities. Pfeiffer syndrome is a common form of acrocephalosyndactyly, characterized by variable degrees of bicoronal craniosynostosis, variable hand and foot malformations and various other associated manifestations. The transmission is variable (autosomal dominant or autosomal recessive or X-linked recessive).