Proximal femoral focal deficiency (PFDD) represents a rare and complex deformity manifested by hypoplasia of a variable portion of the femur with shortening of the entire limb.

We report a case of fetal non familial PFFD diagnosed prenatally using two-dimensional and three-dimensional images.

A 22 year old woman with no medical history and unremarkable ongoing pregnancy. The ultrasound showed a left femur at 8 mm, the right one at 50 percentiles with no other fetal abnormalities.

PFFD is a rare and complex congenital anomaly (1:50,000-200,000 population) that results in varying degrees of femoral hypoplasia with limb shortening and pelvic abnormalities. It may be present bilaterally in association with other malformations/deficiencies of the lower limbs, and the upper limbs may also be involved. Other anomalies may also be present such as cleft palate, congenital heart defects, and spinal anomalies. The etiology is unknown.

The early diagnosis of Prenatal diagnosis of PFFD could provide useful information to both parents and physicians concerning management and therapeutic planning.