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AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE, PREAURICULAR TAG AND EPICANTHUS: A VARIANT OF BOR SYNDROME?

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A female baby was born after an uneventful pregnancy. The prenatal ultrasound scan of the fetus revealed cystic dilatation in the both poles of the left kidney. Postnatal two cysts were seen in the left kidney measuring 16 and 17mm. There were no abnormalities in the right kidney. The physical examination was normal except for an epicanthus and a preauriclar tag on the left side. Ultrasound examination of the relatives revealed bilateral polycystic kidneys in the mother; no cysts were found in her male sibling and father. At the age of 8 months the ultrasound scan of the index patient revealed two cysts in the upper pole of the right kidney measuring 8 and 10 mm. Our search trough literature could not identify reports on association of polycystic kidney disease with a preauricular tag. BOR syndrome may present with phenotypic variety including mild stigmata, but polycystic kidneys have not yet been reported in this syndrome. In conclusion: autosomal dominant polycystic kidney disease may present very early, in utero, as unilateral disease and maybe misdiagnosed as hydronephrosis. US screening of family members may lead to correct diagnosis and avoid invasive radiological investigations.