Gynécologie Obstétrique & Fertilité 44 (2016) 187–191

Available online at
ScienceDirect
www.sciencedirect.com

Elsevier Masson France
EM|consulte
www.em-consulte.com

Letters to the editor

Prenatal diagnosis of bilateral multicystic dysplastic kidney in three siblings

Diagnostic anténatal d'une dysplasie rénale multikystique bilatérale dans une fratrie

1. Introduction

Anomalies in the development of the urinary tract affect about 3 to 4% of the population and account for about 20% of the defects detected in utero. Multicystic dysplastic kidney (MCDK) is a renal pathology referring to the presence of multiple kidney cysts, surrounded by dysplastic parenchymal tissue, and is one of the most common urinary malformations detected in the prenatal and early neonatal periods (incidence 1 in 4300 live births) [1]. Since the multicystic kidney is almost always non-functional, the prognosis depends entirely on the contralateral kidney. The presence of bilateral MCDK is nearly always lethal. Thus, when diagnosed prenatally, the obstetrician and parents may discuss the possibility to terminate the pregnancy [2,3].

We will be describing the case of a patient, Mrs P, who underwent three medical abortions for recurrent renal disease for which several hypotheses were raised before reaching the diagnosis of bilateral multicystic dysplastic kidney. Notably, a family history of three siblings with MCDK may allow us to raise hypotheses concerning the genetic transmission pattern.

2. Case report

Mrs P., who was born in 1986, gravida five, para four, mother of two healthy children, underwent three medical abortions for fetal kidney disease. After a normal delivery from a first partner, she then became pregnant with another partner. She underwent a genetic amniocentesis test at 22 weeks of gestation due to a unilateral nephromegaly with digestive compression, visible in echography and confirmed by MRI (Fig. 1A) and substantially reduced quantity of amniotic fluid. The karyotype was normal, 46 XX. Fetal biology showed an absence of acetylcholinesterase, normal level of catecholamine, with however high concentrations of alpha-fetoprotein and total proteins in the amniotic fluid. This latter was an indirect sign of renal dysfunction, unfortunately, neither any fetal blood test nor any urine analysis was performed. The medical team was orientated towards a nephrotic syndrome or a tumor, although the latter assumption did not in any way explain the protein leakage. The pregnancy was medically interrupted at 24 weeks and 2 days. A histological examination of the heavier and larger right fetal kidney showed an almost total destruction of the renal parenchyma, which was replaced by a large number of cysts of various sizes (Fig. 1A). Moreover, the atrophic left kidney was almost completely destroyed, and replaced by fibrosis punctuated with cysts. The initial hypothesis of oligomeganephronia was ruled out, because there were no enlarged nephrons in reduced number. With the aim of correlating with a genetic origin of the disease and its potential association with MODY diabetes, the gene *HNF1B* was analyzed, but showed no abnormality. Renal echography was performed in both parents, and was normal.

The third pregnancy started a year later. Amniocentesis, performed at 16 weeks of gestation, showed a normal 46 XY karyotype. The ultrasonography at 17 weeks of gestation showed renal asymmetry, with hyper echogenic parenchyma and many irregular anechogenic areas, suggestive of cystic formations. Furthermore, chest compression was observed with bilateral hydrothorax, anamnios and occurrences of fetal bradycardia. Doppler ultrasound showed normal vascularisation. Bladder remained visible despite the lack of amniotic fluid. The request for medical termination of pregnancy was accepted by the multidisciplinary team of prenatal diagnosis at 21 weeks of gestation. At autopsy, it was seen that the fetus had a particular Potter's facial features and joints fixed flexion with irregularity of the lower limbs. The right kidney was four times the size of the left one, with pulmonary hypoplasia and a pericardial effusion. Histological examination showed atrophic renal cortex in both kidneys with the presence of fibrosis and subcortical cysts. The diagnosis was not clearly defined at that point. Suspecting a renal-coloboma syndrome, the gene *PAX 2* was studied but showed no anomaly.

The fourth pregnancy went well, and the patient gave birth to a healthy 38-week little girl of 3300 g. Finally, for her last pregnancy, although first-trimester ultrasound showed no irregularity, a nephromegaly was detected at 22 weeks. The kidney had a hyperechogenic parenchyma dotted with cysts and a loss of cortico-medullary differentiation, and was associated with a heart compression responsible for a pericardial effusion (6.5 mm), in the context of oligohydramnios. Contralateral kidney was barely visible. Waist circumference was still measurable although the excessive volume of the kidney caused a deformation of the shape of the abdomen. The medical termination of pregnancy was accepted and autopsy samples showed one kidney weighing 7.5 g and the other 4.5 g, both with numerous cystic formations. The hypothesis of medullary cystic kidney diseases type 2 (associated with renal dysplasia and cysts in the collecting ducts or the distal tubules) was raised, but then ruled out since no mutation of the gene *UMOD* was identified. After consulting other expert opinions, the diagnosis of bilateral multicystic dysplastic kidney was retained.

The family tree is drawn in Fig. 1B.

3. Discussion

The pathogenesis of MCDK remains unclear, and most cases are sporadic [2]. To our knowledge, this is the first family report of three cases (two female and one male) of bilateral MCDK among five siblings, raising hypotheses concerning the transmission pattern of the disease.

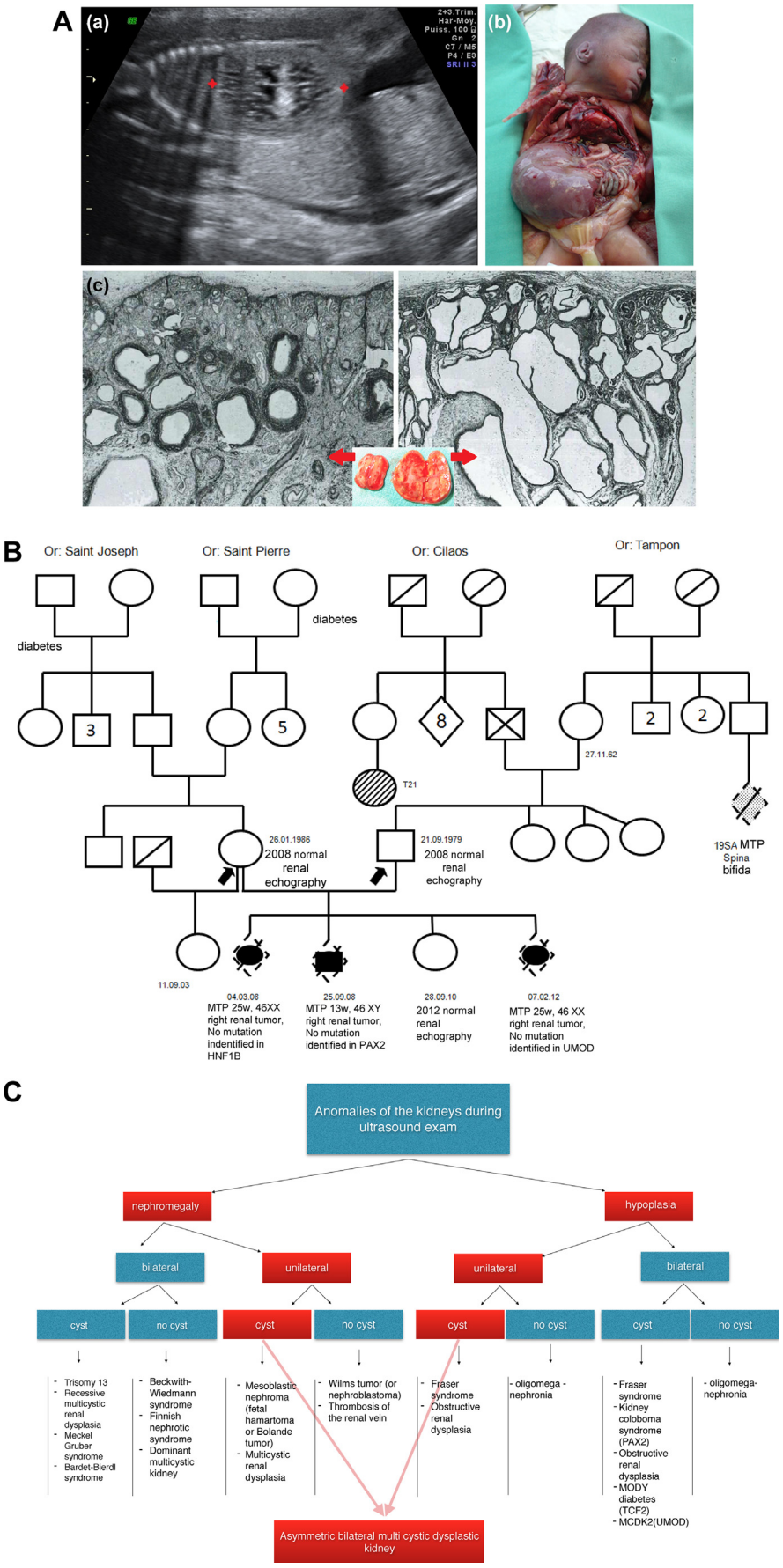

Fig. 1. A. Correlation of nephromegaly between ultrasound (a), fetopathology (b) and histology (c). B. Family tree. MTP: medical termination of pregnancy. C. Diagnostic diagram.