A mother with VCFS and unilateral dysplastic kidney and her fetus with multicystic dysplastic kidneys: additional evidence to support the association of renal malformations and VCFS

Dreviendt et al. recently described in this journal a female fetus with Potter sequence caused by unilateral renal agenesis and contralateral multicystic dysplasia renal, who was retrospectively found to have a deletion in chromosome 22q11 following identification of the deletion in the father. The father presented with typical VCFS features but no urological anomalies. We describe a patient with a clinical diagnosis of VCFS and a unilateral dysplastic kidney but with negative high resolution cytogenetic and FISH studies, who had a female fetus with bilateral multicystic kidneys. This provides additional evidence to support the conclusion of Dreviendt et al. that in VCFS the renal malformation can dominate the clinical phenotype.

Our patient is a 24 year old female initially referred because of facial dysmorphology and developmental delay. She had a long nose and a long, thin face, a small chin, prominent incisors, a deep philtrum (fig 1), a high palate which had the appearance of a cleft, velopharyngeal insufficiency, and long, thin fingers and toes. She also had a repaired ASD, developmental and speech delay, depression, chemical dependency, and seizures. A renal ultrasound showed a unilateral multicystic dysplastic kidney. Karyotype analysis and FISH using a digoxigenin labelled probe localised to 22q11.2 (Oncor Inc, Gaithersburg, MD) were negative. Her first pregnancy was uncomplicated and she delivered a healthy male with no dysmorphic features. He had a normal renal ultrasound and at the age of 2 years is developmentally appropriate. During her second pregnancy, ultrasound examination of her female fetus at 19 weeks 4 days identified bilateral multicystic kidneys and anhydramnios. The pregnancy was terminated and necropsy confirmed the presence of multicystic dysplastic kidneys, hypoplastic bladder, and low set ears. No other abnormalities were noted. Karyotype analysis was normal.

Of patients diagnosed clinically with VCFS, only 68% to 81% have a deletion of 22q11.12.1 Several recent articles have noted the presence of nephropathological malformations as a component of VCFS syndrome.7 Of the 39 patients reported by Dreviendt et al. with 22q11 deletions, four had nephropathological malformations. Another patient with unilateral renal agenesis and VCFS phenotype features suggestive of DiGeorge sequence had a normal G banded karyotype.8 Driscoll et al. reported a patient with a multicystic kidney and a normal karyotype; however, molecular studies showed the absence of a paternal 22q11 allele. Of 11 patients with DiGeorge syndrome reported by Palacios et al., one had a dysplastic right kidney and left ureteropyonephrosis and one had a right megaloureter; karyotype analysis was not performed on these two patients.

We concur with Dreviendt et al. that renal malformations associated with VCFS can lead to the Potter sequence and can dominate the clinical phenotype. These authors retrospectively investigated 10 additional cases of Potter sequence and no other patient with a 22q11 deletion was found. They performed FISH for 22q11.2 on all fetuses with Potter sequence, along with a thorough evaluation of both parents for physical features of VCFS, needs to be examined further.

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