

# Multicystic Dysplastic Kidney and Incontinentia Pigmenti Coexistence of 2 Rare Diseases

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Multicystic dysplastic kidney is a congenital kidney malformation consisting of multiple cysts of various sizes without a normal kidney morphology. Incontinentia pigmenti is a rare X-linked dominant genodermatosis, which is usually lethal in males, that presents clinically in 4 stages. Here, we report a case of multicystic dysplastic kidney with ureterovesical junction obstruction and incontinentia pigmenti. Coexistence of these two rare diseases may be a coincidental phenomenon or an association between the two may exist.

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## INTRODUCTION

Multicystic dysplastic kidney (MCDK) is a congenital renal disease presenting with multiple cysts of various sizes in the dysplastic kidneys. Incontinentia pigmenti (IP, Bloch-Sulzberger syndrome, MIM #308300) is an X-linked dominant genodermatosis that is usually lethal in males before birth.<sup>3-5</sup> Incontinentia pigmenti occurs in approximately 1:40 000 to 1:50 000 births. Although greater than 80% of IP cases are caused by *IKBKG/NEMO* (inhibitor of kappa polypeptide gene enhancer in B-cells, kinase gamma/nuclear factor-kappa B essential modulator) mutations, sporadic cases have been reported.<sup>6</sup>

Four characteristic stages occur in IP. In stage 1 (vesicular), vesicles, pustules or both, usually overlying an erythematous base, appear along Blaschko lines. Eosinophilia is often seen at this stage. In stage 2 (verrucous), vesicular lesions become papular or crusted ("wart-like"). In stage 3 (hyper-pigmented), brown or grey-brown linear and/or swirling macules develop by 6 to 12 months of age. Stage 4 (atrophic/hypopigmented) is characterized by hypopigmentation and slightly atrophic linear macules or patches.<sup>7</sup> We report a patient with right MCDK and left ureterovesical junction obstruction accompanied by IP without an *IKBKG/NEMO* mutation.

## CASE REPORT

A 12-day-old girl was referred to our hospital due to renal impairment and vesicular skin lesions. She was born at 32 weeks of gestation by Cesarean due to oligohydramnios weighing 1600 g. She was the first living daughter of a 20-year-old mother's 7th pregnancy. Prenatal ultrasonography revealed a right multicystic dysplastic kidney and left pelvicaliectasis. Her parents were not related and had no history of kidney disease. Her weight at presentation was 1335 g (3rd percentile), length 40 cm (10th to 25th percentile), and blood pressure 95/73 mm Hg. She had linear vesiculobullous lesions on her limbs. Laboratory results included urea, 110 mg/dL; creatinine, 2.75 mg/dL; hemoglobin, 16.1 g/dL; leukocytes,  $23.76 \times 10^9/L$ ; and platelets,  $632 \times 10^9/L$ . Eosinophilia was detected. During the follow-up, creatinine and urea levels were ameliorated after intravenous fluid was begun amlodipine was initiated for hypertension.

Abdominopelvic ultrasonography revealed a right atrophic dysplastic kidney and left hypertrophic kidney. Renal pelvis anteroposterior diameters of the right and left kidneys were 7 mm and 15 mm, respectively. The right ureter was enlarged and twisted. Voiding cystourethrography, which was performed at 18 days of age, was normal. The patient's skin biopsy revealed superficial

perivascular dermatitis and spongiosis with eosinophils and leucocytes as the dominant cells. She was diagnosed with IP based on these findings. She was discharged on the 18th day of hospitalization. When she was 45 days old, she admitted to the hospital presenting with vomiting. She had hypertension. A physical examination revealed dehydration and verrucous and keratotic lesions on her arms and distal limbs (Figures 1 and 2). Diuretic renography using radioisotope renography showed the right nonfunctional kidney had no perfusion, concentration, or excretion, and the hydronephrotic and diuretic-responsive left kidney had normal perfusion and concentration (Figure 3). Right multicystic dysplastic kidney and left ureterovesical junction obstruction were diagnosed based on these findings. At 6 months of age, she had scattered linear hyper-pigmented lesions (Figure 4). Antihypertension drug doses were increased due to persistent hypertension. The *NEMO* gene mutation was negative. She continued receiving amlodipin, captopril, and bicarbonate therapies.

## DISCUSSION

Multicystic dysplastic kidney is the most common cystic malformation of the kidney among infants.<sup>1</sup> It commonly occurs with other congenital anomalies of the kidney and urinary tract (CAKUT), as in our case. However, MCDK with skin involvement is rarely reported.<sup>8,9</sup> To our knowledge, this is the first report of MCDK with IP.

Incontinentia pigmenti (Bloch-Sulzberger syndrome), is an inherited X-linked dominant neurocutaneous disorder that is usually lethal in males before birth.<sup>3-5</sup> Our patient's mother had a history of six miscarriages with unknown etiology. Therefore, we considered that the previous miscarriages might have been of affected male fetuses with IP. We observed three stages of IP in our patient. In the newborn period, she had vesicular lesions. Eosinophilia is often seen at this stage in IP, as in our case. When she was 45 days of age, the vesicular lesions changed into keratotic and verrucous lesions, as seen in stage 2. At 6 months, she had linear hyper-pigmentations on her whole body, as in stage 3 of IP.



**Figure 1.** Verrucous and erythematous vesicular skin lesions of the patient when she was 45 days old.



**Figure 2.** Linear keratotic and verrucous lesions (Stage 2 of incontinentia pigmenti).



Figure 3. Radioisotope renography scintigraphy demonstrates right nonfunctional kidney.



Figure 4. Linear hyper-pigmented lesions of the patient at 6 months of age (stage 3 of incontinentia pigmenti).

In conclusion, we present a case with MCDK and IP. It might be a coincidence or due to an association between MCDK and IP. Even if speculative a contiguous alignment of the gene responsible for IP with other genes involved in kidney development might lead to this picture. Despite this uncertainty we reported this case since MCDK and IP in the same patient has not been reported before.

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#### CONFLICT OF INTEREST

None declared.

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