End stage renal disease in a child with oral facial digital syndrome

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ABSTRACT

Background: Oral facial digital syndrome (OFDS) is the name of a group of congenital diseases with involvement of face, oral cavity, digits and other organs. Renal involvement in OFD may be in form of polycystic kidney disease.

Case Presentation: This case report presents a patient with OFDS who developed end-stage renal disease (ESRD) at age of six without evidence of renal cyst in sonography.

Conclusions: It may be recommended to evaluate and follow up renal function in children with cleft lip and cleft palate especially when they have other anomalies such as polydactyly and oral lesions considering possibility of OFDS.

Implication for health policy/practice/research/medical education:
Renal involvement should be considered in all patients with multiple organ anomalies, especially in patients with cleft lip, cleft palate and polydactyly. Absence of renal cysts does not exclude kidney involvement and further follow up of patient’s growth, urine specific gravity, renal function and other sonographic findings such as renal echogenicity may be helpful.


1. Background
Oral facial digital syndrome (OFDS) is the name of a group of rare congenital diseases characterized by typical malformations of face, oral cavity and digits. Renal involvement in OFDS has been described mainly as polycystic kidney disease that usually results in end-stage renal disease (ESRD) in adulthood. This patient with OFDS developed ESRD at age of six without evidence of renal cyst in sonography.

2. Case Presentation
The patient is a 6-year-old boy with history of several hospital admissions for aspiration pneumonia because of cleft lip and cleft palate since early infancy, this time he was scheduled to be operated for multiple tongue masses. On physical examination he had short stature (height = 93 cm/< 3 percentile/-4.4SD), flat nasal bridge, multiple masses of tongue (Figure 1), prominent skull (due to hydrocephaly) and polydactyly of hands. Blood pressure was normal (Figure 2). Pre-operative evaluation showed; hemoglobin = 7.3 g/dL, blood urea nitrogen (BUN) = 36 mg/dL, creatinine = 3 mg/dL, K = 5.7, mEq/L, pH = 7.3 ad HCO3 = 17.4 mmol/L. Urinalysis showed specific gravity of 1.005, +1 proteinuria and +1 glycosuria. In renal sonography patient had bilateral echogenic kidneys. Estimated glomerular filtration rate (GFR) (calculated by modified Schwartz’s formula) was 12.8 cc/min/1.73 m². Due to significant rise of serum creatinine, continuous ambulatory peritoneal dialysis (CAPD) was started for patient and he is candidate for renal transplantation now.

3. Discussion
OFDS is the name of a group of rare congenital diseases
characterized by typical malformations of face, oral cavity and digits. The first description of this syndrome was conducted by “Mohr” in 1941 who reported patients with distinct features of lobulated tongue, hypertelorism, high arched palate and broad nasal bridge (1). Since then 11 types of OFDS have been described. Two of those types (OFDS I and OFDS IV) are associated with miscellaneous phenotypes and organ involvements that could overlap with each other in some presentations. OFDS type I is the most frequent type with estimated incidence of 1:50,000 – 1:250,000 live births (2). It may occur in 1:5,1000 patients with cleft lip, cleft palate or both. Seventy-five percent of OFDS1 is sporadic. X linked dominant inheritance pattern is lethal in males (3). Other types of OFDS except type VIII (X linked) are transmitted as autosomal recessive trait. OFDS I is caused by mutation of OFD I gene located on XP22.2-22.3 (4). This gene is recognizable in 85% of patients with OFD1 phenotype. OFDS I gene is a component of distal centriole (5), and is responsible for production of a protein present in structure of both primary cilium and nucleus. Hence, we can consider OFD1 as a member of “ciliopathies” family (6).

Renal involvement in OFD has been described in types I, III, IV, VI and VII (7). Major form of renal involvement in OFDS is polycystic kidney disease (8). Renal Cysts may be detected from neonatal period and childhood but more commonly are discovered in early adulthood. In one study frequency of polycystic kidney disease was around 35% (9). Another study showed that, 16% of cysts presented below 18 years of age and 63% after 18 years of age(10). In spite of autosomal dominant polycystic kidney disease (ADPKD), cysts in OFDS I originate from glomeruli rather than renal tubules. Additionally, size of cysts are not so large and overall renal size is not enlarged as in ADPKD (2).

Renal insufficiency has been reported in OFDS primarily due to polycystic kidney disease. Renal insufficiency usually presents after 18 years of age (11). In one study ESRD in two patients with 28 and 35 years of age was regarded as early ESRD and authors recommended monitoring of renal function in OFDS patients (12). In one report renal failure was detected in an 11 year old girl with OFDS type I (13). In our case despite characteristic clinical findings of OFDS, chronic kidney disease was not associated with polycystic kidney disease but rather with increased renal echogenicity and low urine specific gravity without hypertension or hematuria. These findings are more consistent with renal involvement in ciliopathies such as nephronophthisis.

4. Conclusions
It may be recommended to evaluate and follow up renal function in children with cleft lip and cleft palate especially when they have other anomalies such as polydactyly and oral lesions considering possibility of OFDS.

Author’s contribution
The author passed all criteria for authorship contribution based on recommendations of the International Committee of Medical Journal Editors. HEM handled the case and managed him, supervised the treatment, prepared the primary draft, edited and finalized the manuscript. The author read and signed the final paper.

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The authors declared no potential conflicts of interest with respect to the case, authorship, and/or publication of this article.

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References

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