



Diagnostic Imaging of Oligomeganephronia - A Contemporary Case-illustrated Review

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Abstract

Oligomeganephronia is a rare renal anomaly that leads to renal insufficiency in childhood or adolescence. It is morphologically characterized by a decreased number of nephrons with a compensatory hypertrophy of the remaining glomeruli and tubules. The disease belongs to the spectrum of kidney hypoplasia. Oligomeganephronia possesses a rather specific post-contrast computed-tomographic appearance, which paradoxically could present somewhat of a diagnostic challenge due to the rarity and lack of sufficient awareness of the condition. We present a review of the contemporary literature available about this disease, illustrated with a case from our own practice of a 14-year-old patient with oligomeganephronia.

Keywords: Oligomeganephronia; Renal anomalies; Radiology; Nephrology; Renal hypoplasia

Introduction

Oligomeganephronia (OMN) is a rare condition, the awareness of which has been rising in recent years [1-7]. It was first described in 1962 by Royer, Habib et al. [8] as a peculiar form of hypoplastic kidney. This entity is subsequently called (congenital) oligomeganephronia [3,5]. All patients eventually develop progressive renal insufficiency, which leads to complete insufficiency between within a few months from birth to the beginning-middle of adolescence [4-14]. In recent years there are increasing reports of cases with late onset of symptoms – between 20 and 36 years of age [2,10-20]. Final diagnosis is established on the basis of histology – missing corticomedullary borders, reduced number of cortical pyramids and calyces (up to 6), fewer nephrons greatly increased in size (two to three times bigger), frequent hyalinization of said nephrons [4,9,19,20]. The nephron count could be as low as 20% of the norm, with the few available nephrons undergoing functional hypertrophy – the glomeruli become 12-15 times bigger, while the tubules are up to 17 times [9]. Gradually glomerulosclerosis sets [4]. At the time of writing this article there is no known effective treatment or prevention, due to an insufficiently elucidated mechanism of the disease [20].

Epidemiology

Oligomeganephronia has a marked predilection for males with a ratio of 3:1. The exact frequency of the disease is still not established in the literature. Most cases are sporadic, though familial OMN has been described [3,13]. Even though the condition is usually isolated, oligomeganephronia can be associated with genetic syndromes such as renal coloboma syndrome, brachio-oto-renal syndrome, Wolf-Hirschhorn syndrome, and 4p deletion syndrome [7,18]. OMN can also manifest alongside facial anomalies, growth retardation, seizures, extremity deformities, and congenital cardiac defects [3,9]. It is unusual for OMN to be accompanied by other excretory system anomalies [3].

Pathogenesis

At this time the pathogenesis of OMN is not sufficiently clarified. Premature cessation of nephrogenesis is assumed to be the main mechanism [3,9]. Various potential reasons for this are discussed in the literature - genetic defects, association with nuclear transcription

factor PAX2 [19], association with a genetic mutation of the cellular transcription factor TCF2 [20], placental shunting, and intravascular coagulation [9].

The resulting reduced number of glomeruli is faced with filtrating an overwhelming workload - they compensate with hyperfunction, which in turn leads to increased pressure, perfusion, and filtration. In time the glomeruli grow in size as well - again as a manifestation of compensatory response. This enlargement leads to damage to the basal membrane, proteinuria, and renal hypofunction [20]. All these changes gradually lead to glomerular sclerosis and complete loss of renal function. At the same time changes in the interstitium and tubules are also observed, though there the mesangium is spared [20].

Clinical Findings and Evolution

About 30% of all patients are born prematurely or underweight [4]. One or more of the following findings can be observed in the neonatal period: pneumothorax, feeding difficulties, metabolic disorders, and natriuresis [20].

Most patients present with initial manifestations of OMN in the first two years of life - anorexia, vomiting, polyuria, and fever, usually accompanied by azotemia, proteinuria, and metabolic acidosis [8,9]. Other authors report that nearly 63% of patients present clinically before the age of 20 [20]. According to them the most common initial manifestation of OMN is asymptomatic proteinuria [20]. Fuke [6] describes a 23-year-old male patient with oligomeganephronia, but without endstage kidney. All authors are in agreement that OMN leads to total renal failure and that initial presentation after puberty is a rarity [1,8,9,12,16,18,20]. Treatment is symptomatic, with special attention to

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maintaining electrolyte balance and aiding growth [9]. Survival beyond puberty without renal transplantation in OMN patients is considered unusual [8]. In the last decade, however, a series of cases with late onset of OMN have been described [2,6,10,11,18], contrary to the previous statement.

Imaging findings

The diagnostic imaging findings in oligomeganephronia have started becoming the object of scientific papers only in recent years [7]. Radiography and ultrasound offer little value due to unspecific findings - bilateral reduction in renal size, retained shape, and retarded rate of growth [1,3,9]. No literature data was found regarding the application of contrast-enhanced ultrasound in diagnosing oligomeganephronia. Intravenous urography similarly yields unspecific findings - symmetrical kidneys with a normal shape, weak nephrograms, and slow contrast excretion. The same is true for radionuclide study results, namely small and hypofunctioning kidneys which can be observed in many chronic renal diseases [9].

Few accounts of the computed tomographic (CT) imaging findings in OMN exist in the literature, possibly because of the combination of the low frequency of the disease, the accompanying renal disfunction (which is a contraindication to applying intravenous iodinated contrast material), and the relatively recent mass introduction of CT in the routine diagnostic algorithms [9]. Conducting a CT study of the kidneys without intravenous contrast is usually forgone due to its inadequately low contribution of additional information, other than establishing gross morphologic changes and locating radioopaque stones. The potentially insignificant diagnostic gain of non-contrast CT cannot outweigh the detrimental effect of the ionising radiation dose received by the patient, especially in the pediatric population. To fully demonstrate all relevant renal parenchymal pathology, and be justified in the use of ionising radiation with diagnostic purposes, intravenous contrast is mandatory.

The array of post-contrast CT findings in oligomeganephronia creates a rather specific appearance which includes bilaterally reduced kidney size, evenly thickened cortex and medulla with fusion between adjacent pyramids (obliteration of the columns of Bertin). Additionally a typical fine striation of the cortex is observed - easily differentiated from the striated nephrogram in acute pyelonephritis, where the striations are much thicker. Scant cortical scarring (seen as small cortical indentations) could also be present [9]. Most of the described findings are present in our own case, illustrated in Figures 1-3.



Figure 1a: Axial CT image of the right kidney – native phase. No discernible pathology. Left kidney (not shown) is markedly smaller, but also homogeneous. This image neatly illustrates the relatively little diagnostic value of non-contrast CT in evaluating subtle renal pathology.

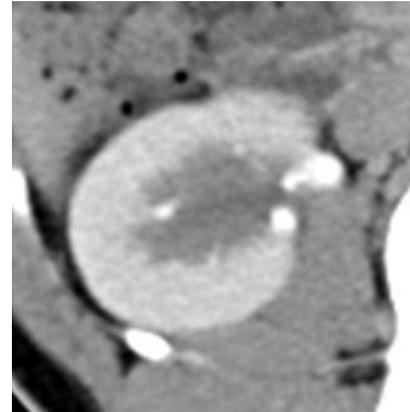


Figure 1b: Axial CT of the right kidney – arterial phase. Demonstration of bright arterial contrast bolus as well as clear demarcation between the contrasting cortex and as of yet non-contrasted medulla. No columns of Bertin are distinguishable.

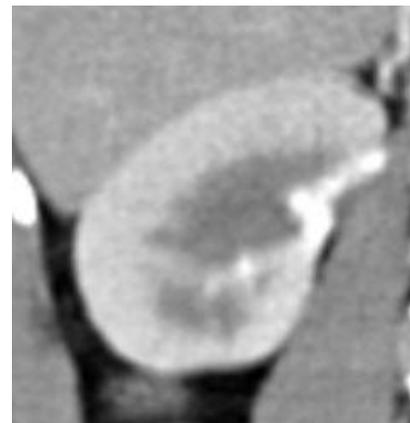


Figure 1c: Coronal CT of the right kidney – arterial phase. Identical findings to 1b. Suspicions of a discrete solitary column of Bertin in the lower half of the kidney.

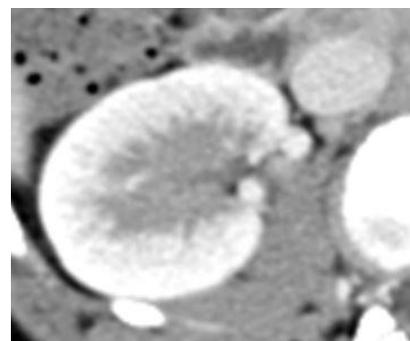


Figure 2a: Axial CT of the right kidney – venous phase. Demonstration of the fine striations between the cortex and the medulla, possibly due to the contrast entering in the enlarged tubules.

We did not find any literature specifically concerning magnetic resonance tomography (MRT) imaging findings of OMN at the time of writing this article.



Figure 2b: Coronal CT of both kidneys – venous phase. Demonstration of the fine striations between the cortex and the medulla bilaterally, more prominent on the right (on the left side of the image). Demonstrable hypoplasia of the left kidney. Suspensions of the discrete solitary column of Bertin on the right, as mentioned in Figure 1c.



Figure 3: Coronal CT of both kidneys, liver, and spleen – venous phase. The spleen (right side of the image) exceeds the craniocaudal dimension of the liver – splenomegaly.

Illustrative Case

We present a 14-year-old girl who is admitted to pediatric nephrology with a classic clinical and laboratory constellation of uroinfection. During a routine abdominal ultrasonography carried out as part of the diagnostic algorithm the examiner establishes renal asymmetry, as well as splenomegaly. It was decided that additional tomographic data was necessary. Blood creatinine, urea, and creatinine clearance levels are normal. Due to technical reasons MRT is unavailable at the time. The decision is made to proceed with a contrast-enhanced abdominal CT with a focus on renal morphology. The asymmetry in the kidneys is confirmed, with the left being severely reduced in size. Cortical hypertrophy as well as the characteristic fusion of the medullary pyramids are evident (Figures 1 and 2). No vascular or drainage anomalies are identified. Splenomegaly is confirmed as an additional finding (Figure 3). The imaging findings are followed up by ultrasound-guided core biopsy, which proves OMN, initially suspected from CT images. The patient is advised to monitor renal function at least once every 3 months after discharge from the hospital.

Conclusion

Oligomeganephronia is a rare congenital disease belonging to the spectrum of renal hypoplasias. It leads to the development of glomerulosclerosis and renal insufficiency. Currently awareness of this entity is starting to increase among radiologists. Unfortunately, due to

the severe impairment of renal function, oligomeganephronia patients rarely undergo CT with intravenous contrast, which could yield the characteristic appearance of the kidneys. Despite that, in patients at an early compensated stage of the disease, whose OMN is yet to be found, a CT performed in search for other abdominal pathology could be diagnostic. Considering the prognosis, a biopsy to confirm the diagnosis early on would potentially improve patient management with frequent monitoring of renal function, careful maintenance of homeostasis, and timely planning of possible transplantation.

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