Research Article

A study of congenital anomalies of human adult cadaveric kidneys

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Abstract

Aim: The present study was undertaken to observe and study the congenital anomalies of the human adult cadaveric kidneys. **Material and Methods**: Forty human adult cadavers over a period of two years from the department of Anatomy, J.N. Medical College, Belagavi, were included in this study. Congenital anomalies of kidneys were observed and studied during routine dissections. **Observations:** Congenital anomaly like unilateral absence of kidney was found in one cadaver. Also we found unilateral lobulated kidney in six cadavers. **Embryological basis:** The metanephric buds begin to develop into kidneys in the fifth week of gestation. Failure of development of metanephric buds leads to renal agenesis. Renal agenesis is closely associated with ipsilateral congenital anomalies of genito-urinary system, cardiac and skeletal systems. The unilateral renal agenesis has also been reported to be associated with X-linked dominant trait, Kallmann's syndrome, chromosomal abnormalities as trisomy 21, trisomy 22, trisomy 7 etc. Persistant fetal lobulations occurs due to incomplete fusion of the developing renal lobules. **Conclusion:** Patients with unilateral renal agenesis and a normal solitary kidney are at increased risk of proteinuria, hypertension, and renal insufficiency. Therefore, it is essential to have prolonged and careful follow-up and to employ strategies that maximize renal preservation. Often, persistant fetal lobulated kidney is a normal variation, but it should be distinguished from inflammatory scarring of the kidney, renal infarcts and tumours, which it can mimic as a pseudotumour. The knowledge of these anomalies will help the clinicians during the procedures of kidney.

Keywords: Renal agenesis, lobulated kidney.

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INTRODUCTION

Kidney is the most common site of congenital abnormalities. Congenital anomalies of the kidney and urinary tract constitute approximately 20 to 30 percent of all anomalies identified in the prenatal period[1]. The present study was undertaken to observe and study the congenital anomalies of the human adult cadaveric kidneys.

Renal Agenesis

Renal agenesis is the absence of one or both the kidneys, more prevalent in male. The incidence of unilateral agenesis of kidney (URA) is 1:1000 births[2]. The incidence of bilateral renal agenesis is reported to be 1 or 2 in every 10000 births [2].URA is the most common congenital abnormality of the urinary tract and this entity has been recognized and known since the time of Aristotle.[3,4] In URA the left kidney is more commonly absent as compared to right side.[3] Unilateral renal agenesis may be asymptomatic and is often incidentally diagnosed by abdominal ultrasound or computerised tomography (CT) scan secondary to another condition, with the contralateral kidney demonstrating compensatory hypertrophy. Bilateral renal agenesis is invariably fatal. Kidneys develop from the metanephric buds in the fifth week of gestation. Failure of development of metanephric buds, lack of induction of metanephric blastema with ureteric bud or maldevelopement of mesonephric duct leads to renal agenesis.The solitary kidney may also be the result of postnatal involution of multicystic kidney. The renal agenesis has also been reported to be due to

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teratogenicity of diabetes mellitus, use of renin angiotensin inhibitors with high doses of vitamin A derivatives, chlorambucil, and cocaine abuse.[4] kidney defects are associated with gene mutations[5]. The mutations occur naturally. However the exact etiology of renal agenesis is still unknown. The incidence of URA is increased in newborns with a single umbilical artery.[6] URA also occurs with the caudal regression syndrome, which is seen with increased frequency in diabetes mellitus.[4] In a study of prenatal and perinatal factors associated with renal agenesis demonstrated that diabetes mellitus, black race, younger pregnancies, and alcohol intake are independent risk factors for development of URA.[3] A mother with pre-existing diabetes mellitus has a 5-fold increased risk of having a fetus with URA.Renal agenesis is closely associated with ipsilateral congenital anomalies of genito-urinary system, cardiac and skeletal systems. Genital anomalies occur in 37-60% of females and 12% of males with congenital URA.[7,8] The unilateral renal agenesis has also been reported to be associated with X-linked dominant trait, Kallmann's syndrome, chromosomal abnormalities as trisomy 21, trisomy 22, trisomy 7, trisomy 10, 45 X mosaicism, and 22q11 microdeletion, as well as congenital hearing loss and double uterus in females.

PERSISTENT FETAL LOBULATION

Normal congenital variant. It is the result of fetal lobulation that persists into adulthood. Typically, the fetal kidneys are subdivided into lobes by grooves that disappear by the end of the fetal period. It occurs due to incomplete fusion of the developing renal lobules. It is discovered incidentally and carries no clinical significance. It is important in imaging to distinguish between lobulation and scarring, which can occur from reflux and/or chronic infection. Lobulation can be seen on CT or ultrasonography as indentations that occur between the medullary pyramids, compared with renal scars, which are located overlying the medullary pyramids.

MATERIALS AND METHODS

Forty human adult cadavers over a period of 2 years from the department of Anatomy, J.N. Medical College, Belagavi, were included in this study. Congenital anomalies of kidneys were observed and studied during routine dissections.

OBSERVATION

Congenital anomaly like unilateral absence of kidney was found in one cadaver. Also unilateral lobulated kidney were found in 6 cadavers.

ANOMALY NO.1

During routine dissection, in the Department of Anatomy, J.N. Medical College, Belagavi, in a 63 yr old male cadaver, we found the unilateral absence of left kidney and ureter. (Fig.1).

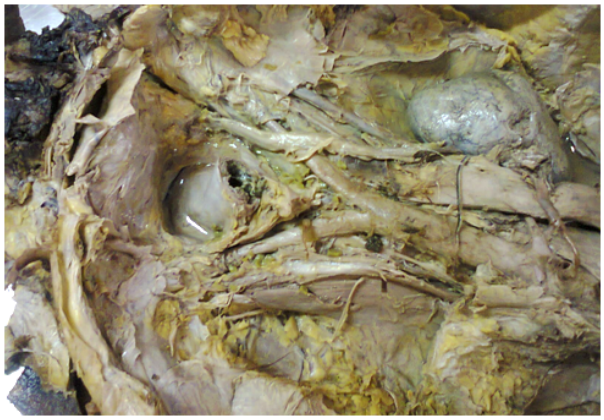

Figure 1: Showing absence of left kidney and ureter

There was no post-operative scar on abdomen. The right kidney was observed in normal anatomical position, slightly hypertrophied. The Right ureteric opening was seen in the urinary bladder. The left kidney was absent (fig. 1) and the left ureteric opening was blunt. The left testis was present. The suprarenal, seminal vesicles and vas deferens on both sides were normal. The thoracic, pelvic & abdominal organs were normal.

Kidney

Right kidney was seen in sub-hepatic region.It was well covered with the renal capsule (dimensions 13X8X4 cm) and hilum showed renal vessels and ureter. The external surface of kidney was smooth without any lobulations.

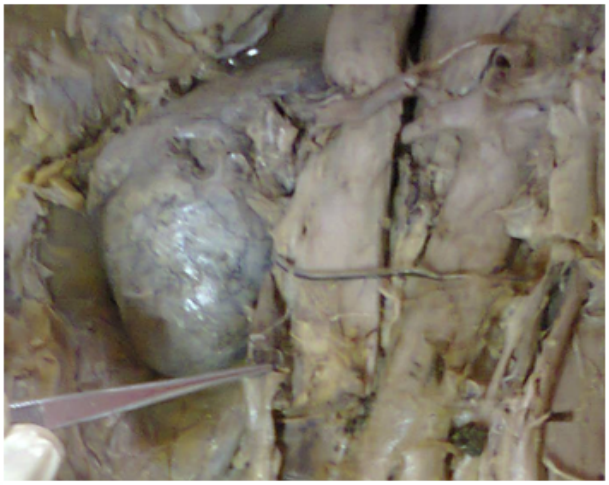

Figure 2: Showing right kidney and ureter

Urinary Bladder

The urinary bladder was normal. Trigone of urinary bladder showed right ureteric orifice and blunt left ureteric orifice.

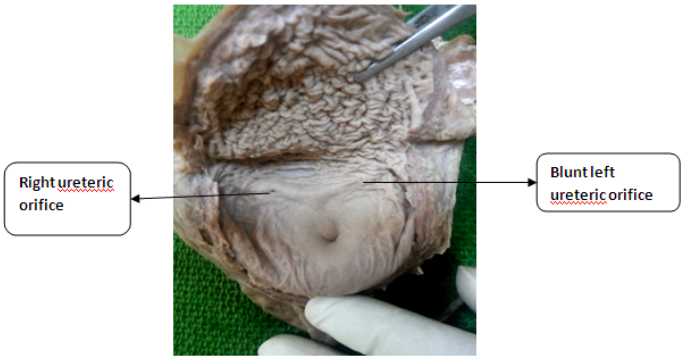

Figure 3: Showing right ureteric orifice and blunt left ureteric orifice

Ureter

The right ureter was normal and vesico -ureteral opening was observed. The left ureter was absent, and vesico-ureteral opening in trigone of urinary bladder was absent.

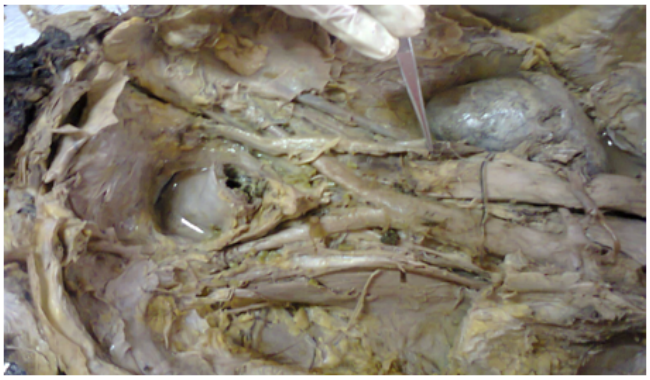

Figure 4: Showing Right uteter and absence of left ureter

ANOMALY NO. 2

Unilateral lobulated kidney was found in 6 cadavers. (fig.5,6) Right side- 2, Left side - 4

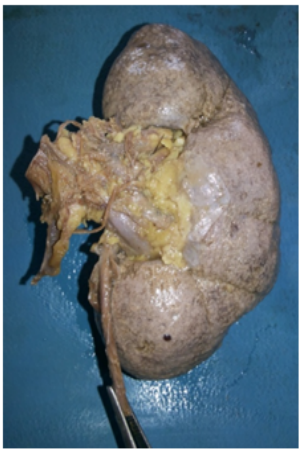

Figure 5: Showing unilateral lobulated left kidney

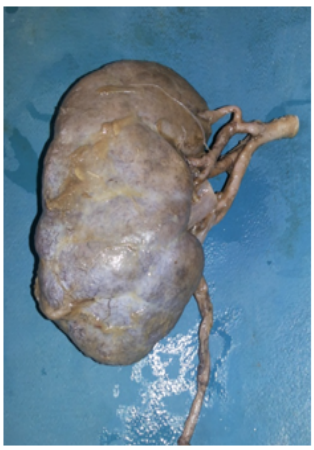

Figure 6: Showing unilateral lobulated right kidney

DISCUSSION

Renal agenesis may be unilateral or bilateral. Unilateral renal agenesis is usually asymptomatic when it occurs as an isolated anomaly[1]. URA is 4 to 8 times more common than bilateral renal agenesis. Bilateral renal agenesis is a rare anomaly incompatible with life, and approximately 20–36% of it presents as familial recurrence. It is 2.5 times more common in males than in females[9]. Renal agenesis is associated with other congenital anomalies like maldevelopment of Mullerian duct, unilateral/bilateral agenesis of vas deferens, seminal vesicle, hemi/complete absence of trigone of bladder, oligohydraminos and single umbilical artery in umbilical cord.[10,11,12]. Associated anomalies are more common in females as Mullerian duct system develops at a later stage in embryogenesis than Wolfian duct.[13] The ureter and triagone are absent in 80 % of cases of renal agenesis.[13] In present study, a male cadaver showed left renal agenesis, with absent ureter and absence of ureteric opening in bladder. The right kidney appeared hypertrophied. Adrian S Woolf *et al*.(2006)[14], observed in his study that if solitary kidney is of normal size, it is either hypoplastic/dysplastic. According to him a solitary functional kidney is always hypertrophied. In present study, our findings are in accordance with Adrian S Woolf *et al*. (2006). In one third to two third of cases of URA, the opposite kidney has been found to be diseased, usually secondary to chronic pyelonephritis.

Embriyological Correlation

During the 5th week of gestation the ureteric bud arises from the mesonephric duct and penetrates the metanephric blastema. Nephrogenesis begins at the 7th week of gestation under the influence of the ureteric bud. By the 20th week the ureteric bud branches into 15 generations and forms the entire collecting duct system. At this time nephrogenesis is only 30% complete. Further maturation proceeds gradually until the 36 th week.[6]

Renal agenesis occurs when there is:

1. Absence of the metanephric blastema;
2. Maldevelopment of ureteric bud ; or
3. Lack of induction of the metanephric blastema by the ureteric bud.

In present study, absence of metanephric blastema and failure of induction of ureteric bud could have resulted in left renal agenesis. The renal agenesis could also be due to the absence of transcription factor WT1 that influences growth factor FGF-2 and BMP-7 to prevent apoptosis of metanephric cells or failure to convert metanephric cells in to nephric epithelium by regulatory genes PAX2 and WNT4 from ureteric bud.[15] Sometimes solitary kidney is the result of post-natal involution of multicystic dysplastic kidney and hydronephrotic kidney. Predictably, the ipsilateral ureter and hemitrigone also fail to develop,

although occasionally a blind-ending ureteric stump may be present, as seen in our study. Ipsilateral adrenal agenesis is seen in 8–10% of cases. Associated ipsilateral urogenital anomalies are common and include absence of the vas deferens, and absence or cysts of the seminal vesicle, which are not seen in our study. Other associated anomalies include skeletal abnormalities, anorectal malformations, cryptorchidism and cardiovascular abnormalities. The classical association of Potter's syndrome is seen in the VATER syndrome where (vertebral and ventricular septal anomalies, anorectal atresia, tracheal and oesophageal lesions and radial bone abnormalities).

URA is associated with malposition of bowel - occurs in empty renal fossa.[13]

- Rt. renal agenesis - malposition of jejunum, deodenum, Rt. Flexure of colon
- Lt. renal agenesis - malposition of distal transverse colon and spenic flexure of colon.

Such bowel position changes should not be mistaken for a form of internal hernia, abnormalility of rotation or displacement by a tumor or organomegaly.

Causes for renal agenesis

Failure of interaction between metanehric mesenchyme & ureteric bud.Mutations in gene regulating GDNF expression. Ex. Gene SALL1- Townes Brock syndrome, PAX2- Renal Coloboma syndrome, EYA1 – Branchio-otorenal syndrome

Failure of expression of WT1, HOX 11, Wnt 11, FGF2,BMP7. Naturally occurring mutations in Pax-2, KAL mutation are associated with renal abnormalities.[16]

Embryological basis of Lobulated kidney

Embryologically, the kidney develops in several distinct lobules that fuse as they develop and grow. Incomplete fusion of these renal lobules can persist postnatally and may be observed in 7% of adults. After 28th week of gestation, varying degrees of assimilation of independent 14 renal lobes occur (8-16). Normally, this lobulated structure of kidney remains apparent at birth and it gradually disappears during infancy as the nephrons increase and grow[17] and fully disappear over the first 5 yrs of life as kidney grows. It is recognized incidentally on imaging studies - intravenous pyelography, ultrasound, computed tomography (CT) or magnetic resonance (MRI) - as smooth regular indentations in the renal contour, without parenchymal thinning or abnormalities in the underlying calyx. Often, this is a normal variation, but it should be distinguished from inflammatory scarring of the kidney, renal infarcts and tumours, which it can mimic as a pseudotumour. At times, fetal lobulation may mimic a solid renal mass and this can be ruled out with CT or MRI. Lobation is frequently symmetrical, often limited to mid and lower part of kidney. These are sharply angulated surface indentations (notches) between the calyces rather than opposite a calyx. Can be distinguished from other causes of irregularity of outline - by its symmetry, shape and absence of any deformity of underlying calices. A cortical defect opposite a calyx represents pathologic loss of lobar tissue. On intravenous urography, fetal lobulation appear as smooth regular indentations in between the renal calyces, without parenchymal thinning or abnormalities in the u0nderlying calyx. Where as in vesicoureteric reflux, the scars occur over calyces, which are abnormally clubbed. Inflammatory scars are deeper and typically associated with an abnormal underlying calyx. Renal infarcts are generally random in distribution and cause a broad flat depression in the renal outline. We observed lobulation in 5% of right kidney specimens and 10% of left kidney specimens. Patil and his associates reported a rare congenital condition of kidney where bilateral lobulation and malrotation were observed in association with the open hilar structure of kidney[18].

CONCLUSION

Although unilateral agenesis is common, its definitive etiology is unknown. Unilateral renal agenesis may or may not be associated with urogenital defects as seen in the present case where the testes, vas deferens seminal vesicle were all normal. As solitary kidney has to compensate for absence of other kidney, patient with unilateral agenesis needs to have life–long nephrological care and follow-up to maximize renal preservation. Most individuals with unilateral renal agenesis lead normal lives although there is an increased risk of renal infections, proteinuria, kidney stones, hypertension, renal insufficiency. and renal failure. If detected, person should have annual surveillance, including a blood pressure measurement, serum creatinine, and urinalysis. Unilateral renal agenesis is inherited as X- linked dominant trait and also associated with Kallmann's syndrome. In such condition, parents and siblings need to be screened for renal agenesis, and proper counselling needs to be given. Prenatal counseling can help prospective parents understand the risks of having a baby with renal agenesis. Women can lower the risk of renal agenesis by reducing known environmental factors before and during pregnancy like - use of alcohol and certain drugs that can affect kidney growth. The characteristic intestinal malposition or displacement on a barium contrast study will help to find out the renal anomaly. This malposition of bowel in empty renal fossa should not be mistaken for a form of internal hernia, abnormality of rotation or dispacement by a tumor or organomegaly. Often, persistant fetal lobulated kidney is a normal variation, but it should be distinguished from inflammatory scarring of

the kidney, renal infarcts and tumours, which it can mimic as a pseudotumour. Also the knowledge of these anomalies will help the clinicians during the procedures of kidney.

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