Available online at www.elixirpublishers.com (Elixir International Journal)

Embryology

Elixir Embryology 102 (2017) 44464-44468

ISSN: 2229-712X

"RENAL AGENESIS" EMBRYOLOGICAL BASIS AND ITS CLINICAL IMPORTANCE

Ganesh Elumalai and Moganelwa Sharline Mampa

Department of Embryology, College of Medicine, Texila American University, South America.

ARTICLE INFO

Article history:

Received: 9 December 2016;

Received in revised form:

13 January 2017;

Accepted: 26 January 2017;

Keywords

Renal agenesis,

Unilateral agenesis ,

Bilateral agenesis,

Kidney tubule,

Collecting ducts.

ABSTRACT

Renal agenesis is a congenital birth defect in which an infant is born missing one or both kidneys. The cause of renal agenesis is not known, though some cases result from inherited mutated genes. Babies with one kidney are often able to live and lead normal lives with ongoing tests and treatment. Those with no kidneys need long-term dialysis to survive.

Introduction

Renal agenesis is a congenital malformation in which one, unilateral, or both, bilateral, fetal kidneys fail to develop. Unilateral renal agenesis (URA) is defined as the one-sided congenital absence of renal tissue resulting from failure of embryonic kidney formation. Human renal development is initiated at the fifth gestational week and is characterized by highly orchestrated interactions between the outgrowing ureteric bud of the mesonephric duct and the metanephric mesenchyme. As a consequence, renal agenesis occurs when the ureteric bud fails to form the ureter, the renal pelvis and the collecting ducts and the renal mesenchyme to form nephrons .URA should be discriminated from abnormal or incomplete renal development leading to a non-functioning kidney, as can be identified in a multicystic dysplastic kidney (MCDK) or renal aplasia. With fetal ultrasonography screening routinely performed, clinicians are more often confronted with an apparent diagnosis of URA. Bilateral renal agenesis (Potter's syndrome) is a rare and always fatal malformation with bilateral absent kidneys

The kidneys carry out functions that are essential for life. In healthy persons, the kidneys: produce urine, which eliminates urea, or fluid waste, from the blood. Kidneys maintain a stability of sodium, potassium, and other electrolytes in the blood. They Supply the hormone erythropoietin, helping red blood cell growth. They produce the hormone renin to assist regulate blood pressure. They produce calcitriol, also known as Vitamin D, which assists the body in absorbing calcium and phosphate from the GI tract. Everybody needs at least one kidney to survive. Without either one of the kidneys, the body is unable to remove waste or water appropriately. The accumulation of waste and fluid can counteract the balance of important chemicals in the blood, and leads to death without treatment.

Incidence

Unilateral agenesis is much more common, but is not usually of any major health consequence, as long as the other kidney is healthy.

It is more related with an increased occurrence of Müllerian duct abnormalities, which are abnormalities concerning the development of the female reproductive tract and can be result into infertility, blocked menstrual flow, increased necessity for Caesarean sections, or other problems. Herlyn-Werner-Wunderlich syndrome is a type of a syndrome in which unilateral renal agenesis is collective with a blind hemivagina and uterus didelphys. About 40% of women with a urogenital tract anomaly also have a related renal tract anomaly. Adults living with unilateral renal agenesis have noticeably higher chances of hypertension . People with this condition are advised to approach contact sports with caution. The probability of a person being born with this condition is roughly 1 in 750.

Bilateral renal agenesis is a medical condition in which both kidneys of a fetus fail to develop during gestational period. It is one contributory agent of Potter sequence. This absence of kidneys causes Oligohydramnios, an insufficiency of amniotic fluid in a pregnant woman, which can place extra pressure on the developing baby and cause further malformations. The condition is commonly, but not always the result of a genetic disorder, and is more common in infants born to one or more parents with a malformed or absent kidney. Males are more commonly affected and most infants that are born alive do not live beyond four hours.

Both types of renal agenesis occur in less than 1 percent of births annually, according to the March of Dimes

March of Dimes. Less than 1 in every 1,000 newborns has URA. Unilateral agenesis should be suspected in infants with a single umbilical artery. BRA is much rarer, occurring in about 1 in every 3,000 births. Risk factors for renal agenesis in newborns appear to be multi-factorial. This means that genetic, environmental, and lifestyle factors combine to create

Tele: +91-9989069546

E-mail address: coolncandy@gmail.com

a person's risk. For example, some early studies have linked maternal diabetes, young maternal age, and alcohol use during pregnancy to renal agenesis. More recently, studies have shown pre-pregnancy obesity, alcohol use, and smoking to be linked to renal agenesis.

Binge drinking, or having more than 4 drinks over 2 hours, during the second month of pregnancy also increases risk. Environmental factors may also result in kidney defects like renal agenesis. For example, maternal medication use, illegal drug use, or exposure to toxins or poisons during pregnancy may be factors.

Ontogenesis of the normal development of the Kidney

Human kidney develops from two sources; the secreting part- or the kidney tubules (Nephrons) of kidney develop from metanephros which is the caudal part of nephrogenic cord located in sacral region of embryo. The collecting part (collecting ducts) of kidney develops from ureteric bud- a diverticulum-arising from lower part of mesonephric duct.

The metanephros forms the kidney tubules. Each newly formed collecting tubule is covered at free end by metanephric tissue cap. The cells of metanephros form small renal vesicles which later form S-shaped kidney tubules. The proximal end of these tubules from a cup-shaped Bowman's capsule, inside this capsule blood capillaries form the glomeruli (tufts of arterial capillaries). As the kidney tubules grow they get transformed into proximal convoluted tubule (PCT), loop of henle and distal convoluted tubule (DCT). There are about one million nephrons in each kidney in adult. The collecting ducts develop from ureteric bud of mesonephric duct. The ureteric bud grows towards metanephric blastema. The bud divides repeatedly.

The first five generations form pelvis of kidney. Subsequently further division form calyces major, calyces minor and collecting ducts. The terminal ends of the branches that become slightly dilated after repeated divisions come in close contact with metanephric blastema that develops into kidney tubules thus the ureteric bud gives rise to; ureter, calyces major and minor and collecting ducts. The secreting part (kidney tubules) and collecting part (collecting ducts) unite and get canalized; tubules so that the urine formed by the kidney tubules passes down the ducts. Ascent of Kidneys- the kidney develops in sacral region of embryo from two sources, as mentioned above. As the embryo and fetus grow in length, there is differential growth of body wall.

The result is that kidneys gradually ascend up and finally reach their adult position in lumbar region of posterior abdominal wall. As kidney ascends it receives its blood supply from higher branches of abdominal aorta. Below is the Fig-1, showing the development of pronephros, mesonephros, metanephros and mesonephric duct opens into the cloaca. The Fig -2 (A, B, C and D) shows the development of the collecting system of the kidney. The Fig-3, shows the stages in the development of the nephron.

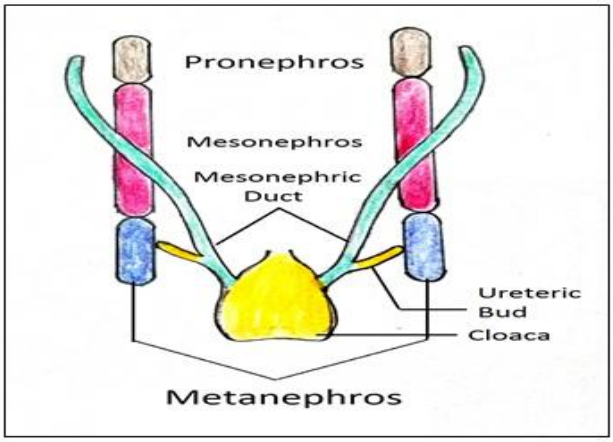

Fig 1.The mesonephric duct opens into the cloaca; and gives off the ureteric bud.

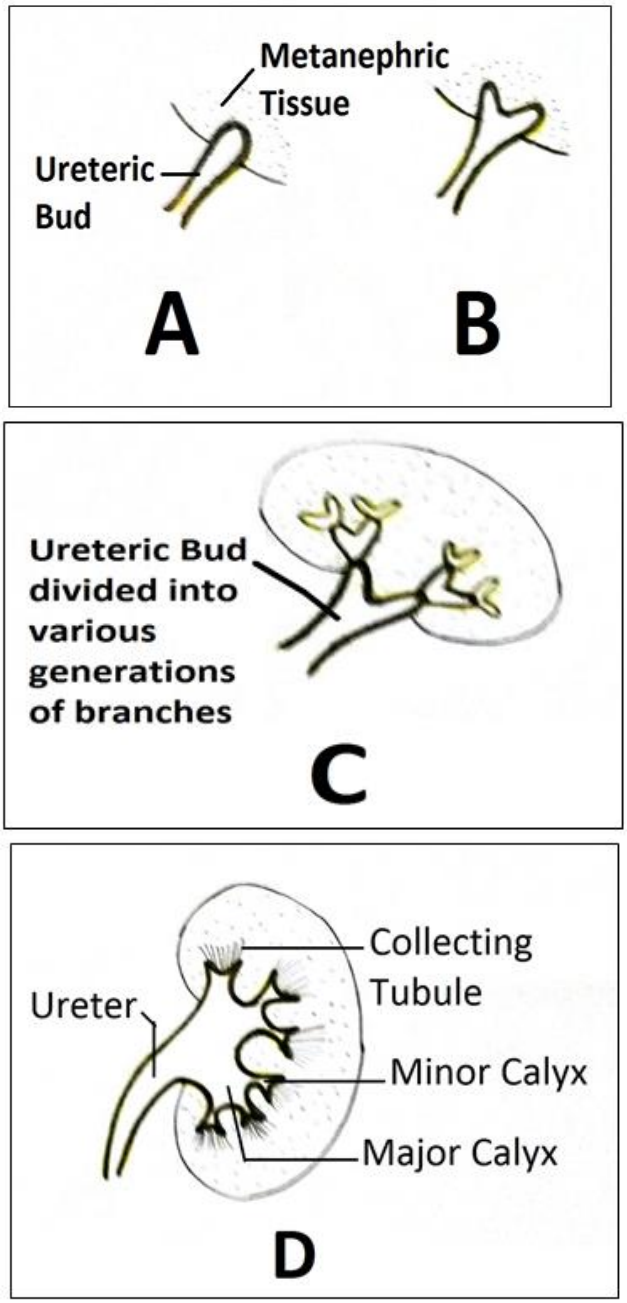

Fig 2. Formation of collecting system of the kidney, from ramifications of the ureteric bud. (a) Development of metanephric tissue and ureteric bud. (b) A well-developed ureteric bud. (c) Ureteric bud develops into various

branches. (d) Shows a developed collecting system of kidney.

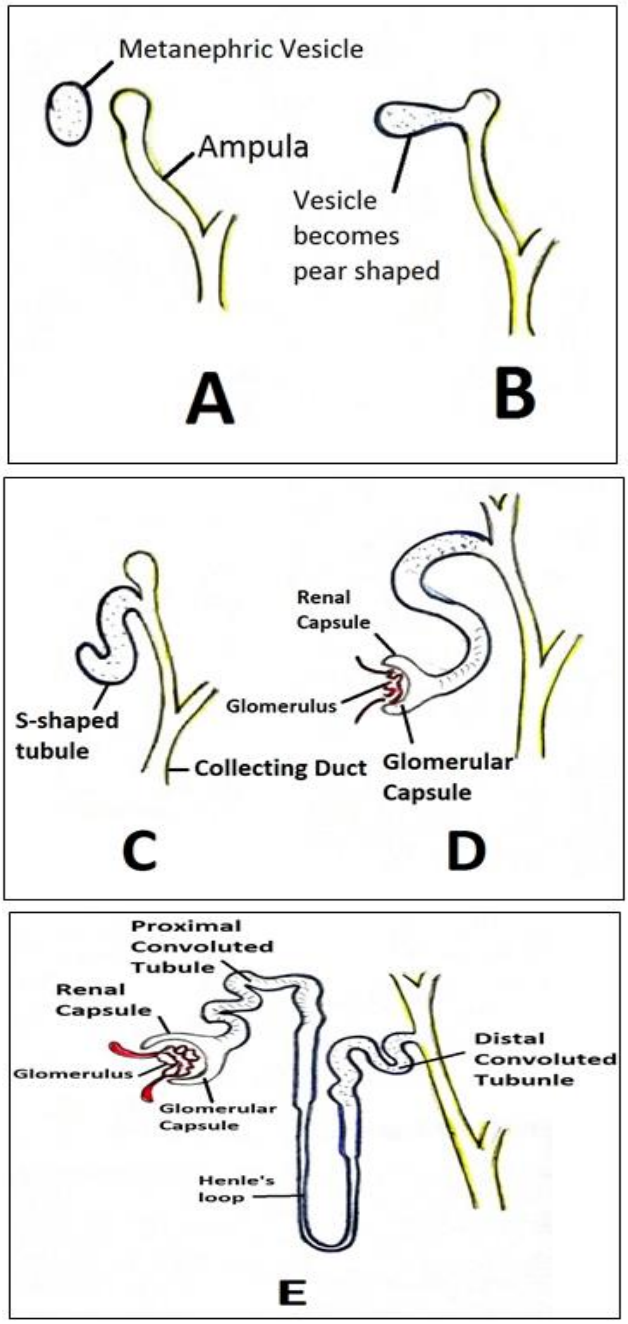

Fig 3. A scheme to show stages in the development of the nephrons. (a) Metanephric vesicle and ampulla develops. (b) Vesicle becomes pear-shaped. (c) S-shaped tubule begins to appear and collecting duct develops. (d) Development of glomerular capsule, renal capsule and glomerulus. (e) a well-developed nephron.

Ontogenesis of abnormal development of kidney –Renal agenesis

Events leading to renal agenesis begin when ureteric bud fails to induce metanephric blastema or the primordial of the ureters degenerate. Failure of the metanephric diverticulum to penetrate the metanephrogenic blastema results in failure of kidney development because no nephrons are induced by the collecting tubules to develop from the metanephrogenic blastema. Renal agenesis probably has a multifactorial etiology. There is clinical evidence that complete in utero involution of kidneys could lead to renal agenesis with a blind ending ureter on the same side (Fig-4). below shows the absence of the metanephric diverticulum leading to unilateral agenesis.

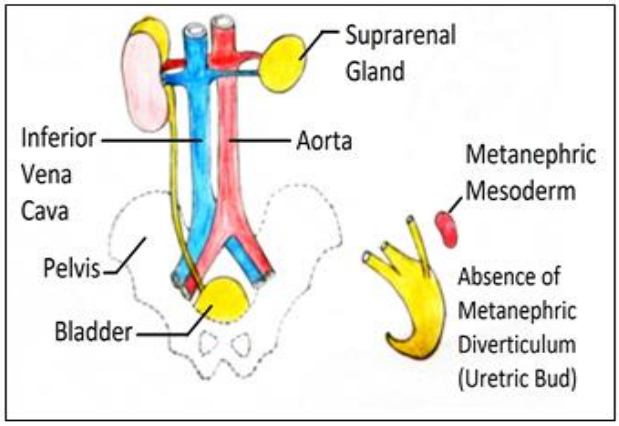

Fig 4. Shows the absence of metanephric diverticulum (Uteric bud). Unilateral renal agenesis.

Discussion

Both Unilateral Renal Agenesis and Bilateral Renal Agenesis occur when the uretic bud, also called the kidney bud, fails to develop at an early stage of fetal growth. The exact source of renal agenesis in newborns is not yet recognized. Most cases of renal agenesis are not inherited from the parents, nor do they result from any behavior by the mother. Certain cases, however, are the result of genetic mutations. These mutations are inherited on from parents who either have the disorder or are carriers of the mutated gene. Prenatal testing can often help determine if these mutations are present. Most newborns with URA have few limitations and live normally. The outlook depends on the health of the remaining kidney and the presence of other abnormalities. To avoid wounding or damaging the remaining kidney, they may need to avoid contact sports when they're older. Once diagnosed, patients of any age with URA need to have their blood pressure, urine, and blood tested annually to check kidney function.

Bilateral renal agenesis is typically fatal within the first few days of a newborn's life. Newborns usually die from underdeveloped lungs shortly after birth. However, some newborns survive. Those who survive will need to have long-term dialysis to do the work of their absent kidneys. Dialysis is a treatment that filters and purifies the blood using a machine. This treatment helps keep the body in balance when the kidneys can't function properly by themselves. Factors such as lung development and general health determine the success of this treatment. The main objective is to keep these infants alive with dialysis and other treatments until they grow strong enough to have kidney transplants.

Conclusion

Since the exact cause of renal agenesis is not known, prevention is not possible. Genetic factors can't be changed. 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 exposure to possible environmental factors before and during pregnancy. These include use of alcohol and certain medications that can affect kidney development. This birth defect is sometimes caused by mutated genes passed from the parents to the baby. If patient have a family history of renal agenesis, they should consider prenatal genetic testing to determine their baby's risk. Babies born with one kidney usually survive and live a relatively normal life, with medical attention and treatment.

Babies born without kidneys usually don't survive. Those who do survive will need long-term dialysis.

References

[1] Bauer SB. Anomalies of the kidney and ureteropelvic junction. In: Walsh PW, Retik Ab, Vaughan Ed Jr, editors.Campbell's Urology. 7th ed. Philadelphia: Wb Saunders Company; 1998. pp. 1709–55.

[2] Bauer SB, Perlmutter AD, Retik AB. Anomalies of the Upper Urinary Tract. Walsh PC, Retik AB, Vaughan ED Jr, Wein AJ, eds. Campbell's Urology. 6th ed. Philadelphia, Pa: WB Saunders; 1992. Vol 2: 1376-81

[3] Bauer SB, "Anomalies of the upper urinary tract," in Campbell's Urology, P. C. Walsh, A. B. Retik, E. D. Vaughan, and A. J. Wein, Eds., pp. 1898–1906, WB Saunders, Philadelphia, Pa, USA, 8th edition, 2002.

[4] Davis E. M., Peck J. D., Thompson D., Wild R. A., & Langlois P. (2010, September). Maternal diabetes and renal agenesis/dysgenesis. Birth Defects Research Part A: Clinical and Molecular Teratology, 88(9), 722-727.

[5] Ganesh Elumalai, Emad Abdulrahim Ezzeddin. "The sudden soul reaper" - hypertrophic cardiomyopathy – its embryological basis. Elixir Embryology. 2016; 99: 43284-43288.

[6] Ganesh Elumalai, Amal Satheesh Sujitha. "Anomalies origin of left coronary artery" its embryological basis and clinical significance. Elixir Embryology. 2016; 100: 43446-43449.

[7] Ganesh Elumalai, Amodini Dharmalingam. "Left superior vena cava" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43429-43432

[8] Ganesh Elumalai, Anto Sicily Norbert. "APVC - Anomalies Pulmonary Venous Connections" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43450-43453.

[9] Ganesh Elumalai, Sushma Chodisetty, Bridget Omo Usen and Rozminabanu Daud Patel. "Patent Ductus Caroticus" - Embryological Basis and its Clinical significance. Elixir Physio. & Anatomy. 2016; 98: 42439-42442.

[10] Ganesh Elumalai, Ebenezer Asare Sakyi. "Right sided aortic arch" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 43706-43709.

[11] Ganesh Elumalai, Danesha Sanicharan. "Abnormal origin of the right subclavian artery from the right pulmonary artery" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 43714-43718.

[12] Ganesh Elumalai, Enian Senguttuvan. "Double aortic arch" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 43710-43713

[13] Ganesh Elumalai, Hariharan Arjet. "Tracheoesophageal fistula" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43414-43419.

[14] Ganesh Elumalai, Jenefa Princess. "Transposition of Great Vessels" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43442-43444.

[15] Ganesh Elumalai, Logeshwaran Anbazhagan. "Laryngomalacia" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43420-43424.

[16] Ganesh Elumalai, Sushma Chodisetty, Pavan Kumar D.2016. Ganesh Elumalai et al Classification of Type - I and Type - II "Branching Patterns of the Left Arch Aorta". Imperial Journal of Interdisciplinary Research. 2(9): 161-181.

[17] Ganesh E, Sushma C. The deer horn aortic arches" embryological basis and surgical implications. Anatomy Journal of Africa.2016; 5(2): 746 – 759.

[18] Ganesh Elumalai, Sushma Chodisetty. Teratological Effects of High Dose Progesterone on Neural Tube Development in Chick Embryos. Elixir Gynaecology. 2016; 97: 42085-42089.

[19] Ganesh Elumalai, Sushma Chodisetty. "The True Silent Killers" - Bovine and Truncus Bicaroticus Aortic Arches its Embryological Basis and Surgical Implications. Elixir Physio. & Anatomy. 2016; 97: 42246-42252.

[20] Ganesh Elumalai, Sushma Chodisetty, Sanjoy Sanyal. Common Nasal Anomalies and Its Implications on Intubation in Head and Neck Surgeries. Journal of Surgery. 2016; 4 (4): 81-84.

[21] Ganesh Elumalai, Malarvani Thangamani, Sanjoy Sanyal, Palani Kanagarajan. Deficient sacral hiatus cause mechanical low back pain: a radiological study. Int J Anat Res. 2016; 4(1):1758-64.

[22] Ganesh Elumalai, Manoj P Rajarajan. "Type-I vascular rings" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 43700-43705.

[23] Ganesh Elumalai, Muziwandile Bayede Mdletshe. "Arteria lusoria"- aberrant right subclavian artery embryological basis and its clinical significance. Elixir Embryology. 2016; 99: 43289-43292.

[24] Ganesh Elumalai, Nnolika Millington. "Coarctation of Aorta" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43425-43428.

[25] Ganesh Elumalai, Moganelwa Sharline Mampa. "Pulmonary Agenesis" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43439-43441.

[26] Ganesh Elumalai, Mouna Arumugam. "Persistent Left superior vena cava" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43454-43457.

[27] Ganesh Elumalai, Shubham Jain. "Subglottic stenosis" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43458-43461.

[28]. Ganesh Elumalai, Sushma Chodisetty. Anomalous "Mutilated Common Trunk" Aortic Arch Embryological Basis and its Clinical Significance. Texila International Journal of Basic Medical Science. 2016; 1(1): 1-9.

[29] Ganesh Elumalai, Sushma Chodisetty, Eliza Arineta Oudith and Rozminabanu Daud Patel. Common anomalies origin of left vertebral artery and its embryological basis. Elixir Embryology. 2016; 99: 43225-43229.

[30] Ganesh Elumalai, Thelma U. Ebami. "Patent Ductus Arteriosus" embryological basis and its clinical significance. Elixir Embryology. 2016; 100: 43433-43438.

[31] Ganesh Elumalai, Siva Brinda Jeyapaul. "Choanal Atresia" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 43719-43722.

[32] Ganesh Elumalai, Kelly Deosaran. "Congenital diaphragmatic hernia" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 43723-43728.

[33] Ganesh Elumalai, Basim Arif. "Subclavian Steal Syndrome" Embryological basis and its clinical importance Elixir Embryology. 2016; 100: 4372943733.

[34] Goodyer, P. (2016). Renal dysplasia/hypoplasia. In Avner, E.D., Harmon, W. E., Niaudet, P., Yoshikawa, N., Emma, F., & Goldstein, S. L. (Eds.), Pediatric Nephrology (7th ed.), chap 5, pp. 113-134. New York: Springer Publishing.

[35] Michiel F. Schreuder, Ruud R. Bueters, Marleen C. Huigen, Frans G.M. Russel, Rosalinde Masereeuw, and Lambertus P. van den Heuvel. (2011, January). Effect of drugs

on renal development. Clinical Journal of the American Society of Nephrology, 6(1), 212-217.
[36] Parikh, C. R., McCall, D., Engelman, C., & Schrier, R. W. (2002). Congenital renal agenesis: Case-control analysis of birth characteristics. American Journal of Kidney Diseases, 39(4), 689.
[37] Renal agenesis. Retrieved from http://www.infokid.org.uk/renal-agenesis
[38] Renal agenesis, bilateral. Retrieved from http://rarediseases.org/rare-diseases/renal-agenesis-bilateral/
[39] Slickers, J. E., Olshan, A. F., Siega-Riz, A. M., Honein M. A., Aylsworth, A. S., for the National Birth Defects Prevention Study. (2008). maternal body mass index and lifestyle exposures and the risk of bilateral renal agenesis or hypoplasia: The national birth defects prevention study. American Journal of Epidemiology, 168(11), 1259-1267.
[40] Westland, R., Schreuder, M. F., Ket, J. C. F., & van Wijk, J. A. E. (2013, July 10). Unilateral renal agenesis: A systematic review on associated anomalies and renal injury. Nephrology Dialysis Transplantation.
[41] Ronald W. Dudek. BRS Embryology. Lippincott Williams & Wilkins.2016; 6th ed: 171, 175.
[42] Sadler, T. & Langman, J. Langman's Medical Embryology. Lippincott Williams & Wilkins. 2012; 12th ed: 248-250.
[43] Schoenwolf Gary C. Larsen's Human Embryology. Churchill Livingstone Elsevier. 2008; 4thed: 518-520.
[44] Meizner I, Yitzhak M, Levi A, et al. Fetal pelvic kidney: a challenge in prenatal diagnosis? Ultrasound Obstet Gynecol. 1995;5:391–93
[45] Queisser-Luft A, Stolz G, Wiesel A, Schlaefer K, Spranger J. Malformations in newborn: Results based on 30,940 infants and fetuses from the mainz congenital birth defect monitoring system (1990-1998) Arch Gynecol Obstet. 2002;266:163
[46] Seikaly MG, Ho PL, Emmett L, Fine RN, Tejani A. Chronic renal insufficiency in children: The 2001 Annual Report of the NAPRTCS. PediatrNephrol. 2003;18:796–804.
[47] Scott, J.E., Renwick, M., and Scott, J.E.Antenatal diagnosis of congenital abnormalities in the urinary tract. Result from the Northern Region Fetal Abnormality Survey. Br. J. Urol. 1988; 62:295–300
[48] T. V. Patel and A. K. Singh, "Crossed fused ectopia of the kidneys," Kidney International, vol. 73, no. 5, p. 662, 2008.