Commentary

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Renal Dysplasia - Limb Defects Syndrome

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Renal dysplasia-limb defects syndrome (RL syndrome), also known as Ulbright–Hodes syndrome, is a very rare autosomal recessive congenital disorder. It has been described in three infants, all of whom died shortly after birth. RL syndrome is inherited in an autosomal recessive manner. This means the defective gene responsible for the disorder is located on an autosome, and two copies of the defective gene (one inherited from each parent) are required in order to be born with the disorder. The parents of an individual with an autosomal recessive disorder both carry one copy of the defective gene, but usually do not experience any signs or symptoms of the disorder.

Ulbright-Hodes syndrome is characterised by renal dysplasia, growth retardation, phocomelia or mesomelia, radiohumeral fusion, rib abnormalities, anomalies of the external genitalia and a potter-like facies. The syndrome has been described in three infants (one pair of sibs and an unrelated case), all of whom died shortly after birth from respiratory distress resulting from pulmonary hypoplasia and oligohydramnios caused by renal dysplasia. The mode of transmission appears to be autosomal recessive. Renal dysplasia-limb reduction defects syndrome (RL syndrome), is a rare lethal congenital anomaly with only few cases reported in the literature. It is characterized by mesomelia, radiohumeral fusion, bilateral talipes equinovarus, Potter's facies, and renal dysplasia.

The syndrome is characterised by renal dysplasia, growth retardation, phocomelia or mesomelia, radiohumeral fusion (joining of radius and humerus, rib abnormalities, anomalies of the external genitalia and potter-like facies among many others. Congenital heart disease (CHD) is one of the most common birth defects, and recent studies indicate cilia-related mutations play a central role in the genetic etiology of CHD. As cilia are also known to have important roles in kidney development and disease, it is not surprising that renal anomalies were found to be enriched among CHD mutant mice recovered in a large-scale mouse forward genetic screen. The common limb defects include oligodactyly, ectrodactyly, syndactyly or brachydactyly anomalies of the carpal and tarsal bones and the common renal anomalies observed are unilateral renal agenesis (URA), bilateral renal hypoplasia, ureteric hypoplasia, hydroureteronephrosis and duplication abnormalities.

Limb and urinary tract anomalies have frequently been reported to occur together as components of a single acro-renal defect or multiple malformation syndromes. The acro-renal syndrome has a restrictive definition with limb defects usually bilateral, like cleft hands or feet and longitudinal defects involving radius or ulna, tibia or fibula. Renal anomalies include agenesis (unilateral or bilateral), hypoplasia and rarely polycystic kidneys. Additional malformations may involve the oro-mandibular region, the trachea and lungs, skin derivatives including sweat glands, mammary glands, the uterus, vas deferens, the nasal placodes and the eyes.

