Cystic kidney disease refers to a group of conditions characterized by the presence of fluid-filled sacs, called cysts, in the kidneys. These cysts can interfere with kidney function and can lead to various complications, depending on the type and severity of the condition. The most common forms of cystic kidney disease are polycystic kidney disease (PKD) and simple kidney cysts.

Key Forms of Cystic Kidney Disease: Polycystic Kidney Disease (PKD):

Autosomal Dominant PKD (ADPKD): This is the most common inherited form, typically manifesting in adulthood. It is characterized by the gradual development of multiple cysts in both kidneys, which can lead to kidney enlargement, hypertension, pain, and eventually kidney failure. Autosomal Recessive PKD (ARPKD): A rarer, more severe form that presents in infancy or early childhood. It can cause significant kidney and liver issues, and is associated with a higher risk of early kidney failure. Simple Kidney Cysts:

These are solitary cysts that typically do not cause symptoms or harm kidney function. They are usually found incidentally during imaging tests for other conditions and are more common as people age. Medullary Sponge Kidney (MSK):

A congenital disorder where cysts form in the inner part (medulla) of the kidneys, leading to a spongy appearance. MSK is often associated with kidney stones, urinary tract infections, and sometimes blood in the urine. Acquired Cystic Kidney Disease (ACKD):

This occurs in people with long-term kidney disease, particularly those on dialysis. The kidneys develop multiple cysts, and although they often do not cause symptoms, there is an increased risk of kidney cancer in this condition. Symptoms and Complications: Pain: Often in the back or sides due to enlarged kidneys or cyst rupture. Hypertension: High blood pressure is common, especially in PKD. Kidney Stones: Increased risk in conditions like MSK. Hematuria: Blood in the urine, which can occur if cysts bleed. Infections: Such as recurrent urinary tract infections (UTIs). Kidney Failure: Particularly in severe forms like ADPKD or ARPKD. Diagnosis and Treatment: Diagnosis: Typically involves imaging studies like ultrasound, CT scan, or MRI to identify cysts and assess kidney function. Genetic testing may be done for inherited forms like PKD. Treatment: Depends on the type and severity. It may include managing symptoms, controlling blood pressure, treating infections, or addressing complications like kidney stones. In severe cases, especially with PKD, treatment may involve dialysis or kidney transplantation if kidney failure occurs. Cystic kidney disease can range from benign, asymptomatic conditions to severe, life-threatening diseases, depending on the specific type and progression of the condition.

Intellectual and developmental disability result from abnormal nervous system development. Over a 1,000genes have been associated with intellectual and developmental disabiliy, driving continued efforts toward dissecting variant functionality to enhance our understanding of the disease mechanism.

A report identified two novel variants in CC2D1A in a cohort of four patients from two unrelated families. They used multiple model systems for functional analysis, including Xenopus, Drosophila, and patient-derived fibroblasts. The experiments revealed that cc2d1a is expressed explicitly in a spectrum of ciliated tissues, including the left-right organizer, epidermis, pronephric duct, nephrostomes, and ventricular zone of the brain. In line with this expression pattern, loss of cc2d1a led to cardiac heterotaxy, cystic kidney disease, and abnormal CSF circulation via defective ciliogenesis. Interestingly, when we analyzed brain development, mutant tadpoles showed abnormal CSF circulation only in the midbrain region, suggesting abnormal local CSF flow. Furthermore, the

analysis of the patient-derived fibroblasts confirmed defective ciliogenesis, further supporting the observations [1].

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Kim AH, Sakin I, Viviano S, Tuncel G, Aguilera SM, Goles G, Jeffries L, Ji W, Lakhani SA, Kose CC, Silan F, Oner SS, Kaplan OI; MarmaRare Group; Ergoren MC, Mishra-Gorur K, Gunel M, Sag SO, Temel SG, Deniz E. CC2D1A causes ciliopathy, intellectual disability, heterotaxy, renal dysplasia, and abnormal CSF flow. Life Sci Alliance. 2024 Aug 21;7(10):e202402708. doi: 10.26508/lsa.202402708. PMID: 39168639.