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Commentary

Mechanism and Diagnosis of a Genetic Disorder: Polycystic Kidney Disease

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DESCRIPTION

Polycystic Kidney Disease (PKD or PCKD, also known as polycystic kidney syndrome) is a hereditary disorder characterized by structural abnormalities in the renal tubules, resulting in the development and proliferation of numerous cysts within the kidney. These cysts may begin to develop in utero, in infancy, in childhood, or in maturity. Cysts are non-functioning tubules filled with fluid pumped into them, which range in size from microscopic to huge, damaging surrounding regular tubules and eventually turning them non-functional as well. PKD is caused by defective genes that create a specific aberrant protein that interferes with tubule formation. Autosomal Dominant Polycystic Kidney Disease (ADPKD) and Autosomal Recessive Polycystic Kidney Disease (ARPKD) are the two kinds of PKD that fall under the umbrella term (ARPKD). All of the body's cells contain the defective gene, and as a result, cysts could develop in the liver, seminal vesicles, and pancreas. A diagnosis may be suspected if any, some, or all of the following occur: new-onset flank pain or red urine; a favourable family history; palpation of enlarged kidneys during a physical examination; an incidental discovery on an abdominal ultrasonography; or an incidental finding of abnormal kidney function on regular lab work. Abdominal Computerized Tomography (ACT) imaging is used to provide a definitive diagnosis.

Risks include hypertension brought on by Renin-Angiotensin-Aldosterone System (RAAS) activation, recurrent cyst infections, urination bleeding, and deteriorating renal function. Angiotensin Converting Enzyme Inhibitors (ACEIs) or angiotensin receptor blockers are used to treat hypertension Angiotensin Receptor Blockers (ARBs). Antibiotics are used to treat infections. Decreasing renal function is treated with Renal Replacement Treatment (RRT) dialysis and/or transplantation. Cyst formation in polycystic kidney disease caused by autosomal dominant and autosomal recessive genes is associated with aberrant cilia-mediated signalling. Due to flaws in both proteins, the polycystin-1 and polycystin-2 proteins seem to be involved in both autosomal dominant and recessive polycystic kidney disease. Both proteins interact with calcium channel proteins, which results in a decrease in endoplasmic reticulum storage of calcium as well as resting (intracellular) calcium. The primary cilium, an immobile, hair-like cellular organelle found on the surface of the majority of body cells and anchored in the cell body by the basal body, is the cause of PKD. In the kidney, primary cilia have been found to be present on most cells of the nephron, protruding from the apical surface of the renal epithelium into the tubular lumen. It was once thought that the cilia would bend with the flow of urine, altering signalling, but this has since been proven to be an error in the experiment and cilia bending does not affect Ca flux.

Cyst development is likely to be connected to disruption of one of the numerous signalling channels controlled by the primary cilium, such as intracellular calcium, Wnt/-catenin, Cyclic Adenosine Monophosphate (cAMP), or planar cell polarity, although the exact mechanism by which primary cilium deficiencies cause cyst development is unknown. The malfunction of the main cilium disturbs a number of intracellular signalling pathways that result in the development of cystic epithelium, accelerated cell division, increased apoptosis, and loss of resorptive capacity. Patients with the following symptoms may have polycystic kidney disease, according to the diagnosis, a good familial background, standard symptoms or indications accidental discovery of cysts during imaging studies.

The kidneys are frequently enlarged and appear moth-eaten due to cysts that displace functioning tissue. Imaging typically reveals widespread and bilateral cystic alterations throughout the kidneys. These changes become more noticeable during aging and are less frequent or visible in patients who are younger.

Typically, ultrasonography comes first. Cyst and kidney volume measurements using MRI are very useful. Complete Blood Counts (CBC), renal function tests, and urinalyses are performed, although the results are not specific. Urinalysis finds microscopic or macroscopic hematuria and moderate proteinuria. Gross hematuria might result from a burst cyst or a calculus that has become dislodged. Pyuria is typical even in the absence of bacterial infection, thus the diagnosis of infection should be made using urine results, clinical symptoms, and culture

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results. Blood Urea Nitrogen (BUN) and creatinine are initially normal or just marginally raised, but they gradually rise, particularly in the presence of hypertension. CBC occasionally finds polycythemia. High-resolution CT or magnetic resonance angiography is required for patients who have cerebral aneurysm symptoms. Nonetheless, the majority of experts do not advocate routinely checking asymptomatic people for cerebral aneurysms. Screening patients with autosomal dominant polycystic kidney disease who have a family history of hemorrhagic stroke or cerebral aneurysm is a feasible course of action.