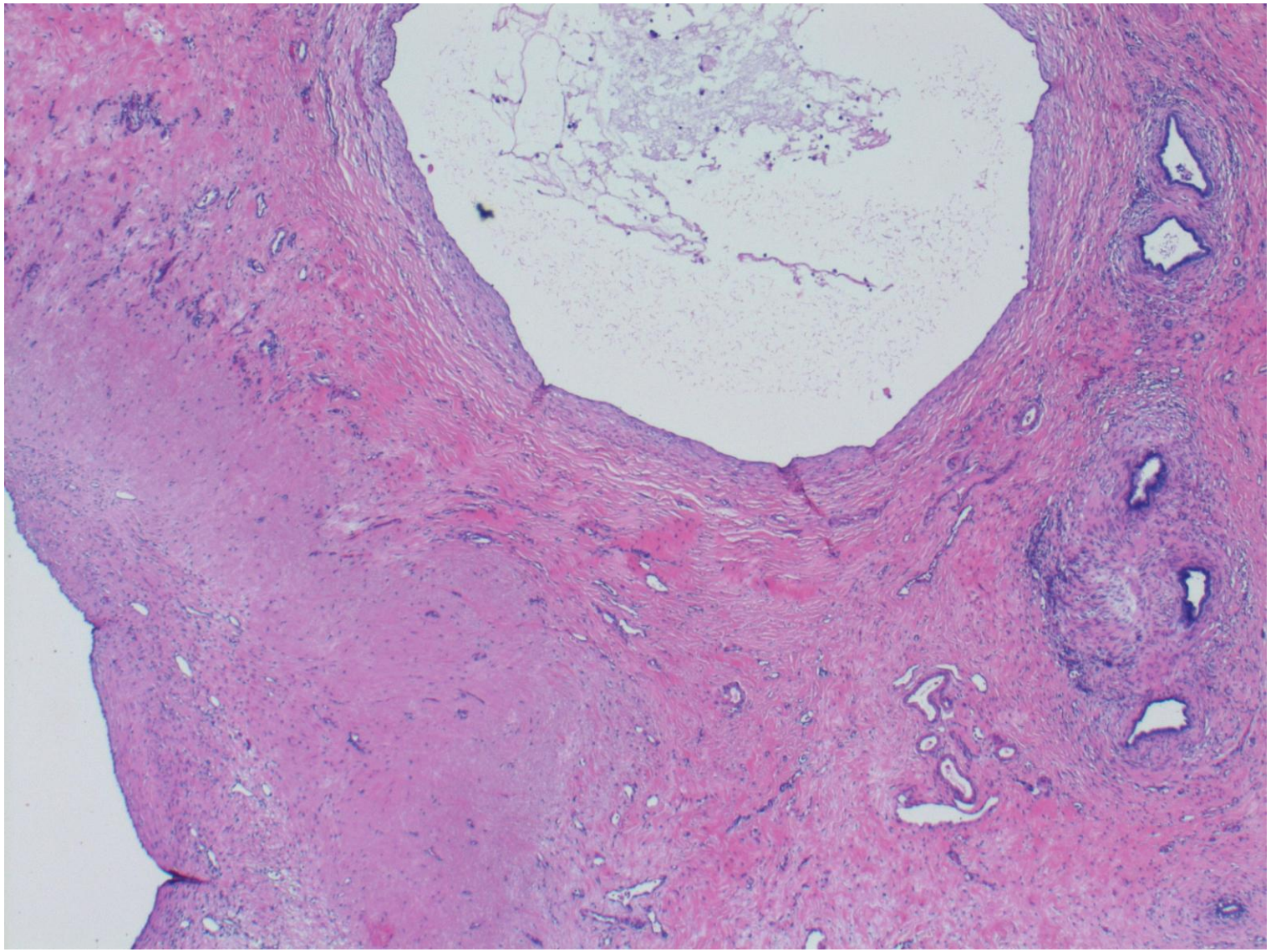
Autosomal recessive polycystic kidney disease

Autosomal recessive polycystic kidney disease (ARPKD) primarily involves the kidney and liver. The gene PKHD1 is mutated. Historically, it was termed as infantile polycystic kidney disease, but the disease can present not only in the neonatal but also infantile, juvenile or even adult populations. Hence, the old nomenclature of adult and infantile polycystic kidney disease is not used anymore. Commonly, the disease is presented in the perinatal or infantile (four weeks to one year) with the classic finding of enlarged kidneys. Less commonly, initial presentation occurs in childhood (after age one year) to young adulthood, and hepatobiliary manifestations may be predominant, characterized by the clinical consequences of developmental anomalies of biliary ductal plate remodeling (known as Caroli disease). In cases with childhood presentation (age >1 year), the enlarged kidneys show multiple micro- and macrocysts, increased echogenicity, and reduced or absent corticomedullary differentiation. The liver disease may be predominant, including hepatomegaly, bile duct dilatation and liver fibrosis. Signs of portal hypertension and Caroli disease may be associated. Here, a 2 years-old boy case is presented. An intrapelvic kidney was surgically removed and microscopically showed features of ARPKD.

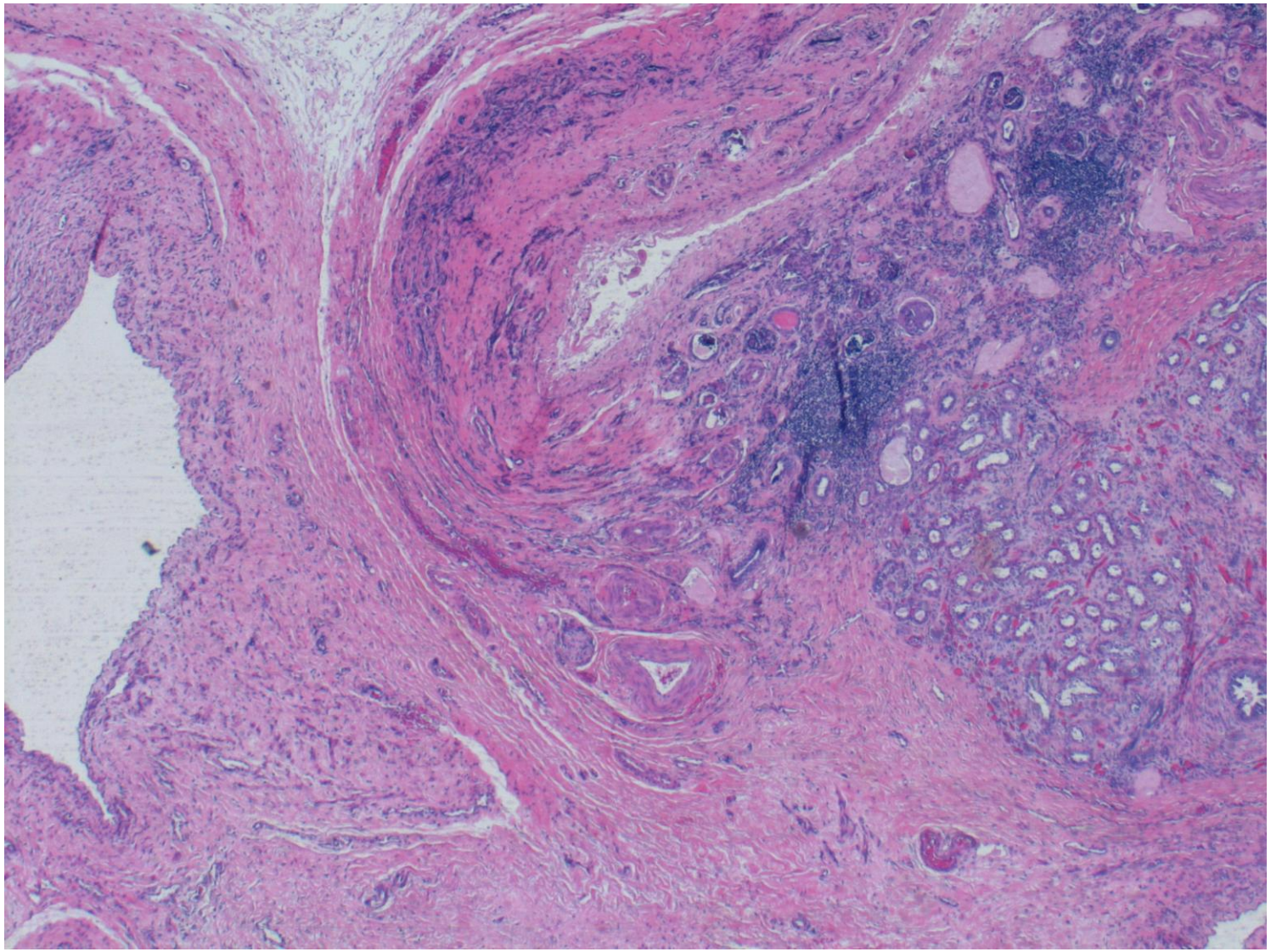
Ref.: Burgmaier K, et al. Autosomal recessive polycystic kidney disease – PKHD1. 2001 Jul 19 [Updated 2024 Apr 4]. In: Adam MP, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1326/

Features of the polycystic kidney

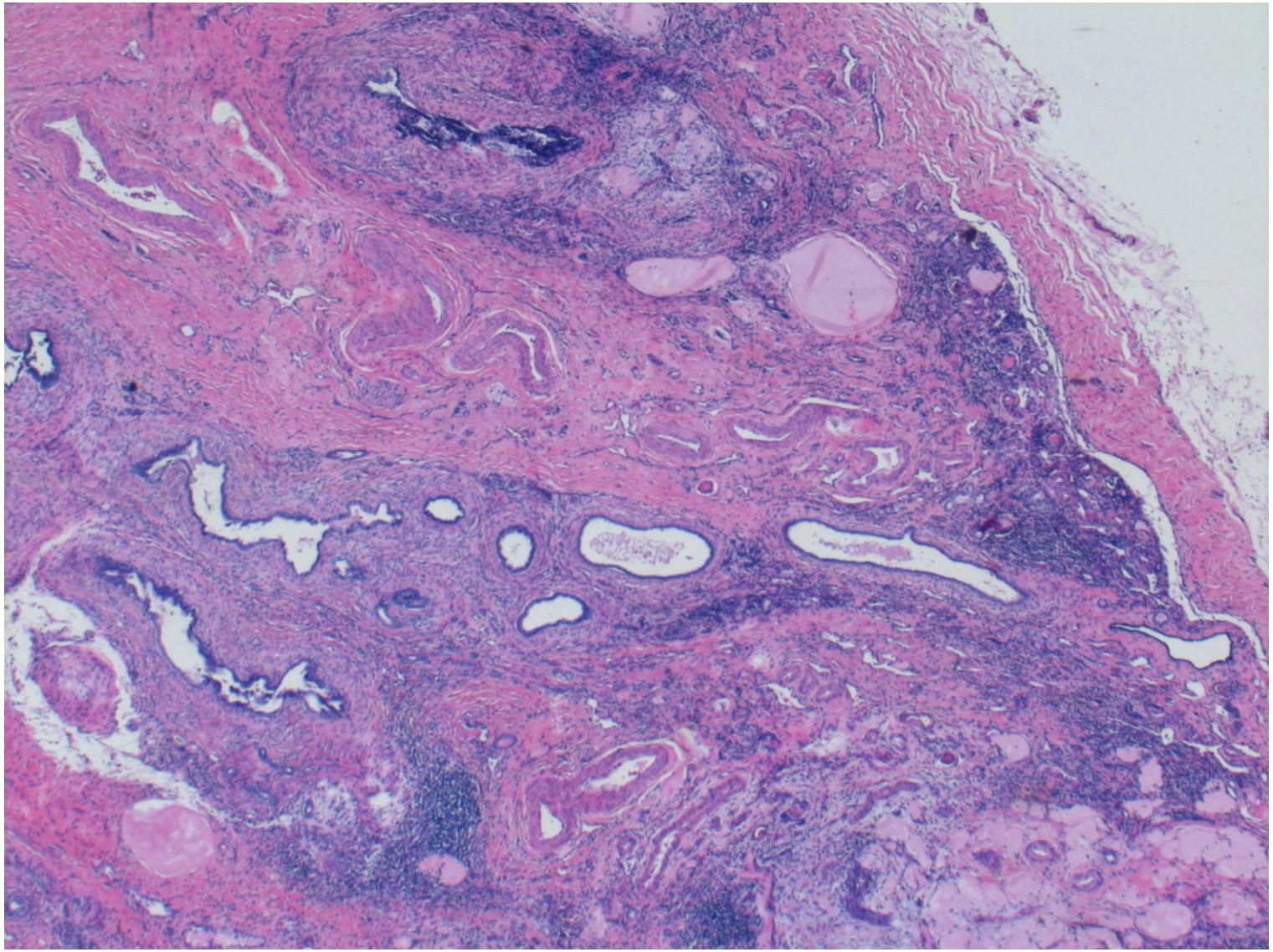
The intrapelvic kidney was removed from a 2 y-o boy, who was also complicated with retention testis. The underdeveloped kidney measures 32 x 19 mm and contains multiple cysts. The maximal size of the cysts is 15 mm. Microscopically, the renal parenchyma consists of multiple islands of inflamed renal cortex (glomeruli and proximal renal tubules) and collecting tubules in the fibrotic stroma. Macro- and microcysts are dispersed.



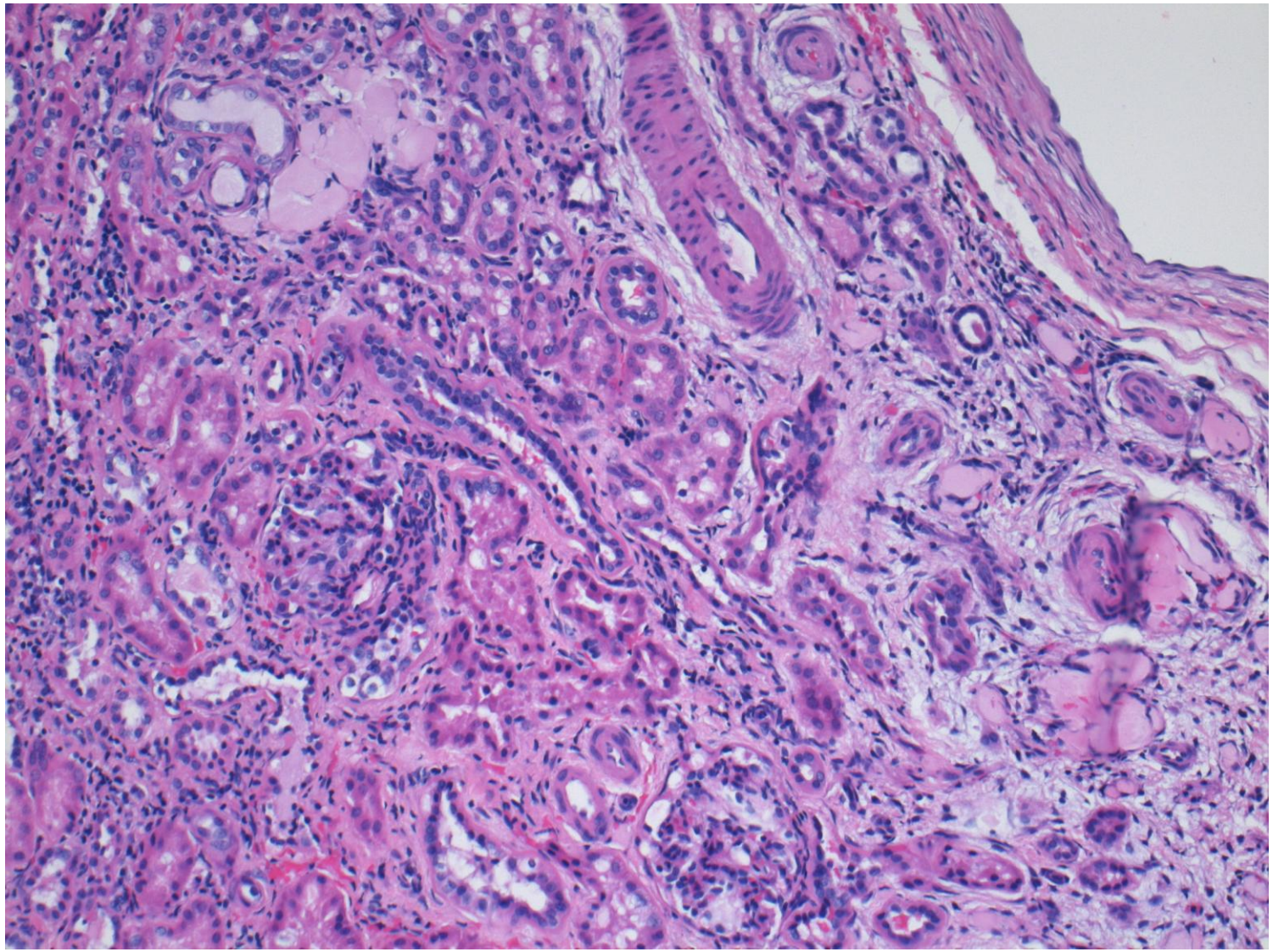
Autosomal recessive polycystic kidney disease seen in a 2 y-o boy. An intrapelvic kidney was surgically removed. Micro- and macrocysts are dispersed in the fibrotic renal tissue (H&E).



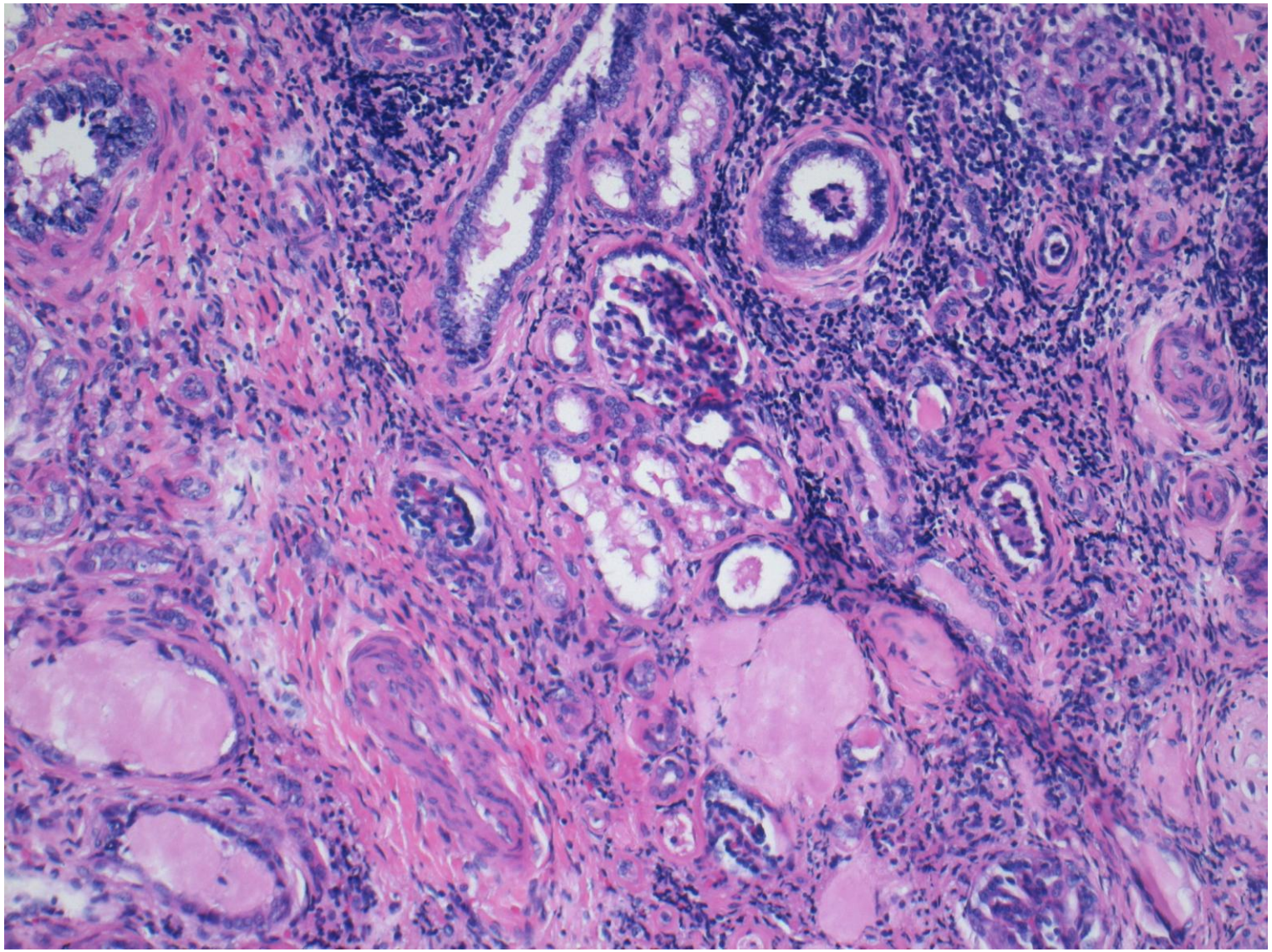
Autosomal recessive polycystic kidney disease seen in a 2 y-o boy. An intrapelvic kidney was surgically removed. A microcyst is formed in the fibrotic renal tissue. Chronic inflammatory change is seen among the clustered renal tubules (H&E-2).



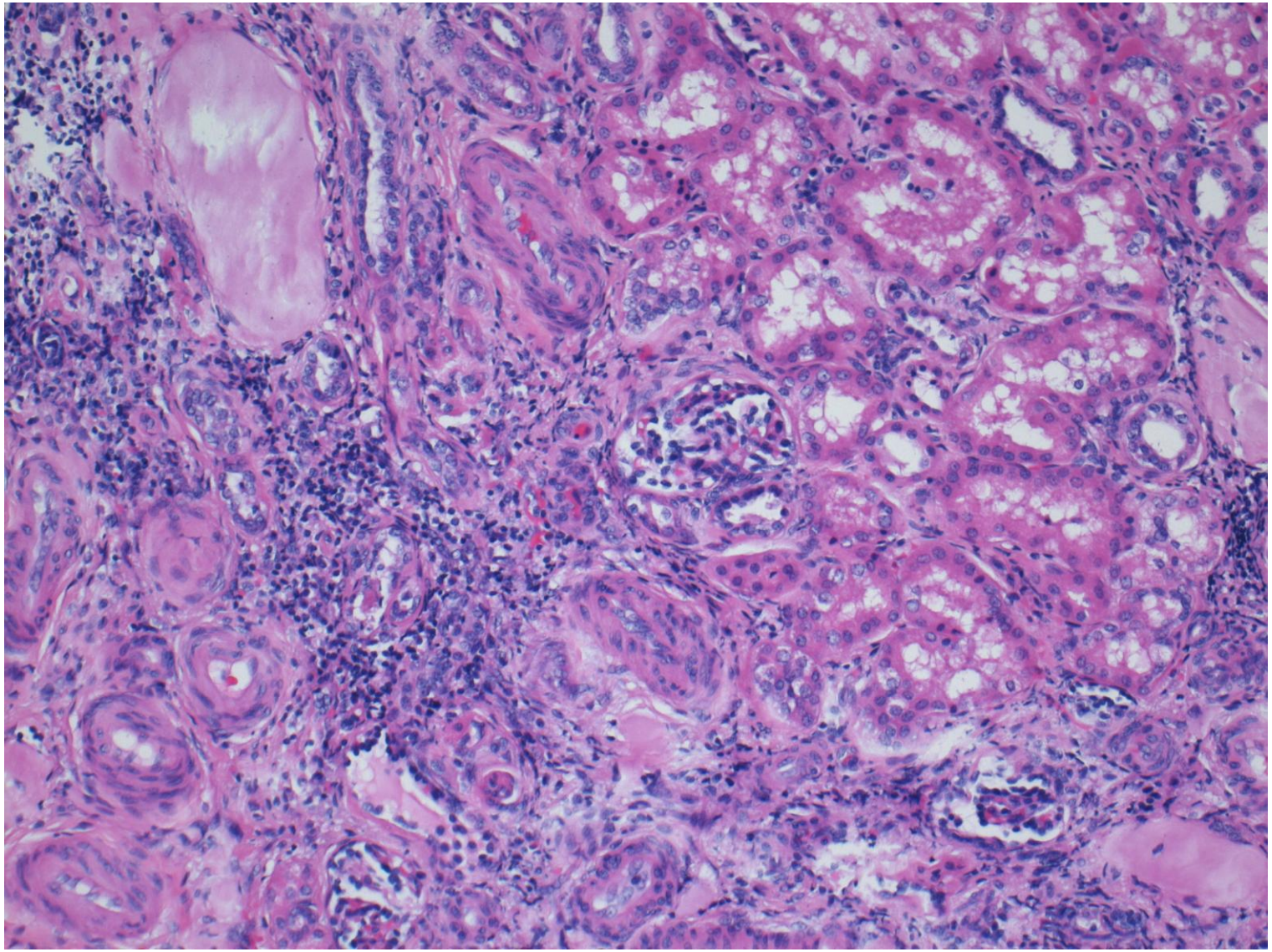
Autosomal recessive polycystic kidney disease seen in a 2 y-o boy. An intrapelvic kidney was surgically removed. Microcysts are formed in the fibrotic renal tissue. Chronic inflammatory change is seen in the stroma (H&E-3).



Autosomal recessive polycystic kidney disease seen in a 2 y-o boy. An intrapelvic kidney was surgically removed. Adjacent to a cystic lesion, renal tubules and glomeruli are observed. Fibrotic stromal change is mildly seen (H&E-4).



Autosomal recessive polycystic kidney disease seen in a 2 y-o boy. An intrapelvic kidney was surgically removed. Microcysts are formed in the fibrotic renal tissue. Chronic inflammatory change is seen in the stroma (H&E-5).



Autosomal recessive polycystic kidney disease seen in a 2 y-o boy. An intrapelvic kidney was surgically removed. Renal tubules and glomeruli are observed, and chronic inflammatory change is associated (H&E-6).