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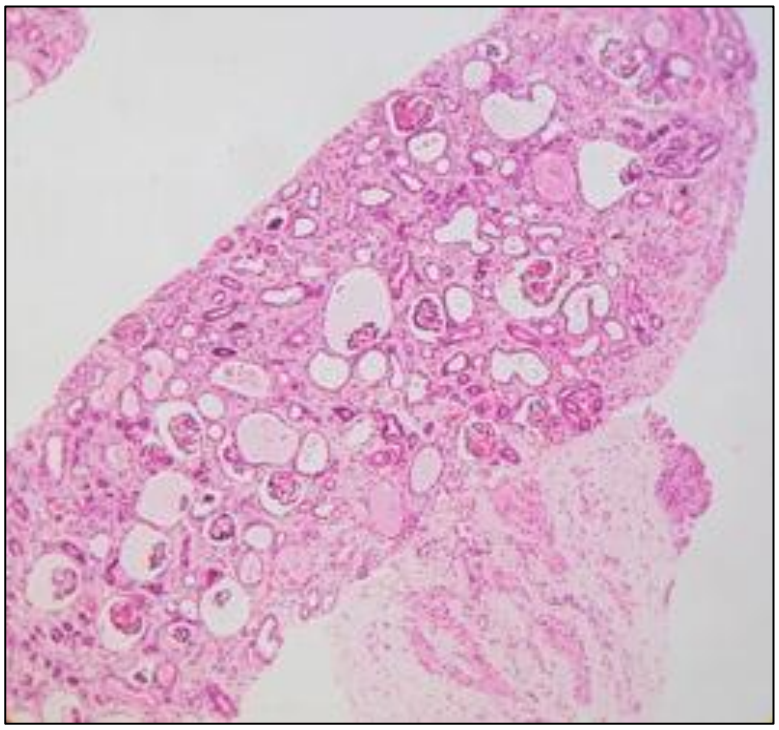
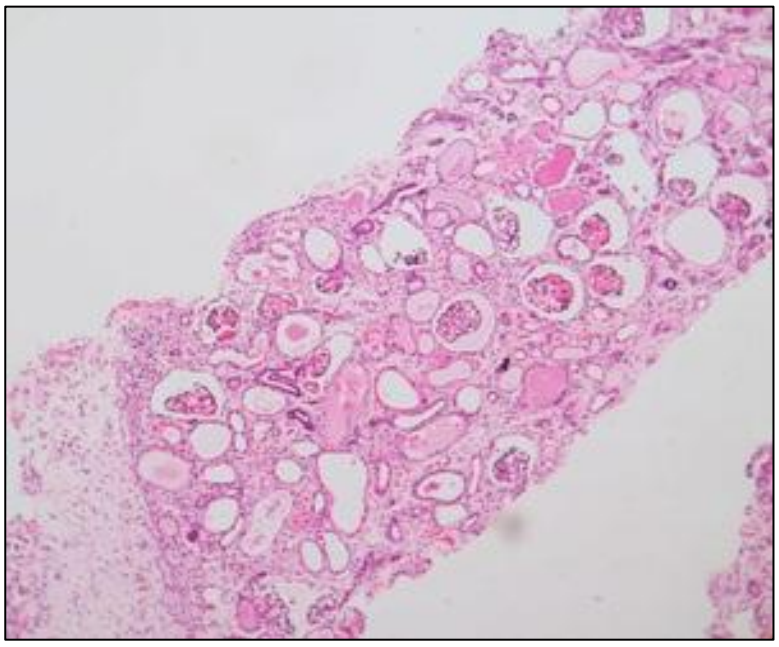
Image Quiz 10

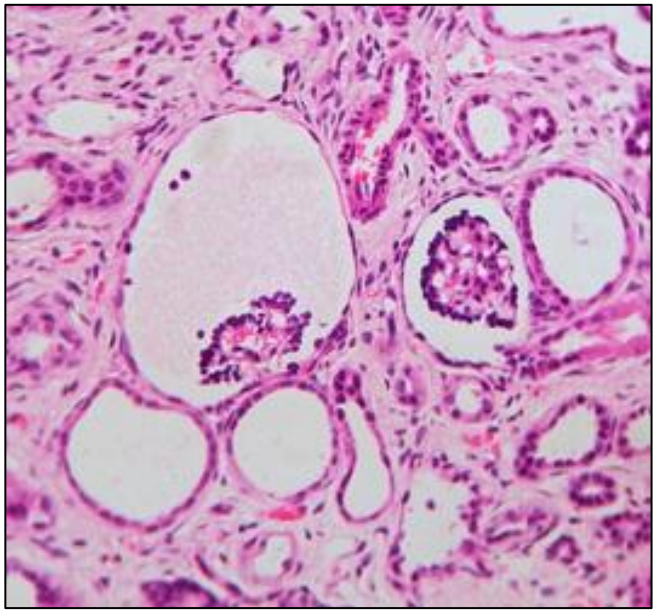
CLINICAL HISTORY:

Renal biopsy from a 3 year old boy with renal dysfunction (creatinine 1.9 mg%/167.9 μ mol/L), central obesity, hypogonadism, astigmatism and polydactyly.

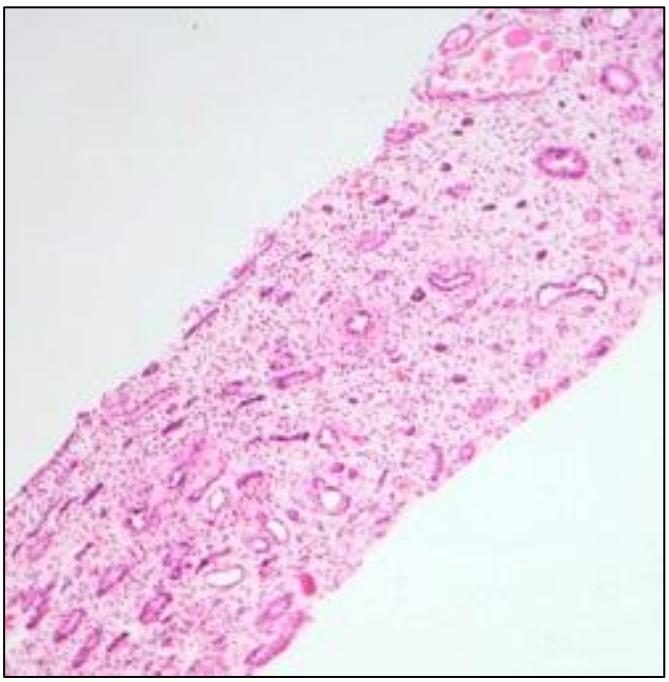
RENAL BIOPSY

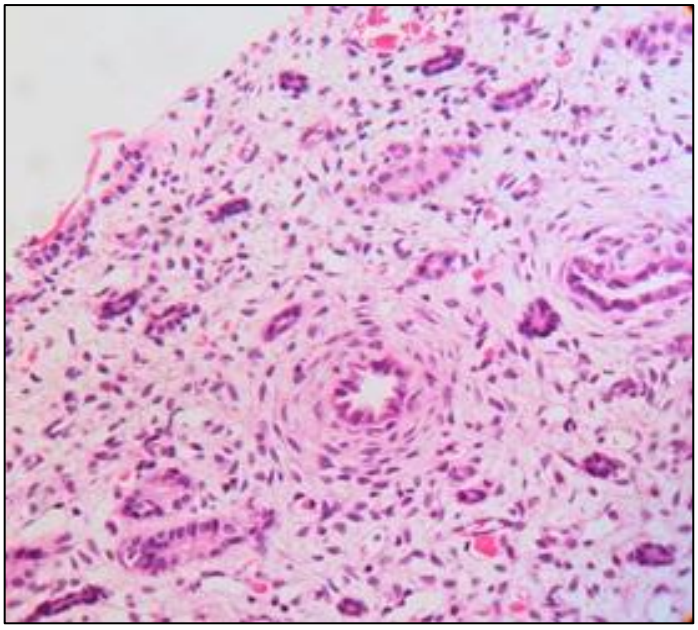


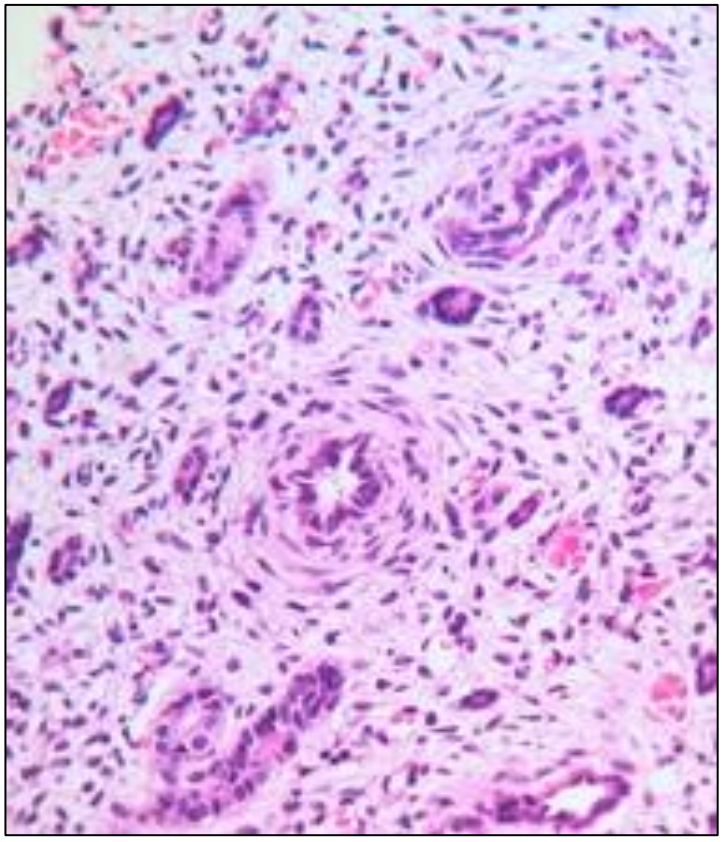




Low and higher magnification photomicrographs showing tubulointerstitial scarring with inspissated tubular casts and prominent dilatation of Bowman’s space in amy glomeruli (glomerulocystic change).







Renal medulla shows variably sized, primitive appearing tubules showing "collarets" of spindly mesenchymal cells.

DIAGNOSIS:

Renal dysplasia in a child with Bardet-Biedl syndrome

DISCUSSION:

Bardet-Biedl Syndrome (BBS) is a rare, hereditary (autosomal recessive) disease, now classified as a ciliopathy characterized by juvenile obesity, hypogonadism, polydactyly, retinal dystrophy, mental retardation, and renal abnormalities. Hypertension and/or diabetes are commonly encountered in these patients.

BBS is distinguished from the much rarer Laurence- Moon syndrome, in which retinal pigmentary degeneration, mental retardation, and hypogonadism

occur in conjunction with progressive spastic paraparesis and distal muscle weakness, but without polydactyly.

Till date, 14 BBS-causing genes have been identified (BBS1 through BBS14; for review see BBS. Seven of the most conserved BBS proteins (BBS1, BBS2, BBS4, BBS5, BBS7, BBS8, and BBS9) form a stable complex called the "BBSome" , excluding three other proteins, BBS6, BBS10, and BBS12, that are vertebrate-specific chaperonin-like proteins.

Diagnosis of BBS (Diagnostic criteria)

Primary features:

Rod-cone dystrophy

Polydactyly

Obesity

Learning disabilities

Hypogonadism in males

Renal anomalies

Secondary features

Speech disorder/delay

Strabismus/cataracts/astigmatism

Brachydactyly/syndactyly

Developmental delay

Polyuria/polydipsia (nephrogenic diabetes insipidus)

Ataxia/poor coordination/imbalance

Mild spasticity (especially lower limbs)

Diabetes mellitus

Dental crowding/ hypodontia/small roots/high arched palate

Left ventricular hypertrophy/congenital heart disease

Hepatic fibrosis

Diagnosis is established when four primary features or three primary plus two secondary features are present

Renal abnormalities are a cardinal and important features of BBS and are the prime determinants of clinical outcome in these patients.

The various renal abnormalities/anomalies reported include parenchymal cysts/communicating calyceal cysts, Calyceal clubbing & blunting), Fetal

lobulation , Scarring, Unilateral agenesis, Dysplastic kidneys, Renal calculi, & Vesicoureteric reflux (VUR). Renal dysplasia has been reported in about 5% of cases of BBS. Histological hallmarks of renal dysplasia include the disorganized appearance of renal parenchyma, primitive tubules with collar like condensation of mesenchyme and metaplastic cartilage (seen in 1/3 rd of cases).

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