Musculoskeletal Best Case
CLINICAL INFORMATION

66-year-old male with bilateral knee and left hip total joint replacements presented with right thigh pain, swelling, and a limp for 4 weeks.
Right knee radiographs 8 years prior
Right knee radiographs 8 years prior
Right knee radiographs 7 years prior
Current right knee radiographs
Current right knee radiographs
Current right knee radiographs
Current right knee radiographs
Current right femur radiographs
DEDIFFERENTIATED CHONDROSARCOMA

Dr. Kapil Wattamwar
Montefiore Diagnostic Radiology Residency
Bronx, NY
Gastrointestinal Best Case
Clinical information

19-year-old previously healthy female with progressive right upper quadrant pain, which persisted following cholecystectomy.
Immunohistochemistry:
(+) vimentin
(-) nuclear INI1 (mutation in tumor suppressor gene hSNF5/INI-1)
CD34, AE1/3, S100, desmin
Malignant Rhabdoid Tumor in the Adult Liver

Andrew McCurry
University of Florida
Gainesville, FL
Neuroradiology Best Case
Clinical Information

58-year-old female with history of Cowden syndrome and 6-month history of progressively worsening ataxia and difficulty with arm and leg movement.
Cerebellar Dysplastic Gangliocytoma (Lhermitte-Duclos Disease)

Adam Orr, M.D.
Spectrum Health/Michigan State University
Grand Rapids, Michigan
Pulmonary and Mediastinal
Best Case
Clinical information

61-year-old male with four-week history of right-sided chest pain and hemoptysis.
Echinococcal Hydatid Cyst

Felipe Belmar, MD
Clínica Dávila
Santiago, Chile
Cardiovascular Best Case
Clinical information

72 year old man with multiple strokes.
Cardiac Myxoma

Thomas Cellini
Rush University Medical Center
Chicago, IL
Pediatric Best Case
Clinical information

20-year-old female with G1P0 who presented enlargement of the fetal abdomen and little amniotic fluid.
### Genetics and Clinical, Pathologic, and Imaging Features of Ciliopathies

<table>
<thead>
<tr>
<th>Ciliopathies</th>
<th>Genetics</th>
<th>Clinical and Pathologic Features</th>
<th>Imaging Features</th>
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<tbody>
<tr>
<td>ARPKD</td>
<td>PKHD1 (6p12): polyductin-fibrocystin</td>
<td>Renal: oliguria, pulmonary hypoplasia, dilated collecting ducts; Liver: portal hypertension due to CHF, biliary duct dilatation</td>
<td>Renal: oligohydramnios sequence, echogenic kidneys with tubular cysts—mostly medul lary; Liver: periporal edema and fibrosis, intra- and extrahepatic biliary dilatation</td>
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<tr>
<td>ADPKD</td>
<td>PKD1 (16p13.3): polyductin 1; PKD2 (4q22): polyductin 2</td>
<td>Renal: cortical and medullary cysts; Liver: hepatic cysts; Cysts of other organs; Vascular defects</td>
<td>Renal: initially normal-sized kidneys with a few round cysts; Liver: hepatic and other abdominal organ cysts in adults</td>
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<tr>
<td>Nephronophthisis, MCKD</td>
<td>Multiple genes, UMOD (16p12.3) (uro-modulin)</td>
<td>Tubulo-interstitial nephropathy leading to renal failure and renal cysts in both entities; Retinal degeneration, hepatic fibrosis, and CNS abnormalities possible in nephronophthisis</td>
<td>Small echogenic kidneys with subcortical and medullary cysts after onset of renal failure</td>
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<tr>
<td>Joubert syndrome-related disorder</td>
<td>Multiple genes</td>
<td>Vermian hypoplasia causing ataxia; developmental delay; may also have CHF, nephronophthisis, ocular or digital defects</td>
<td>Molar tooth sign</td>
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<tr>
<td>Meckel-Gruber syndrome</td>
<td>AR Multiple genes</td>
<td>Perinatal mortality, occipital encephalocoele, polydactyly, cystic renal dysplasia, CHF</td>
<td>Occipital encephalocoele, round renal cysts</td>
</tr>
<tr>
<td>Asphyxiating thoracic dysplasia (jeune syndrome)</td>
<td>AR Multiple genes</td>
<td>Pulmonary hypoplasia causing respiratory distress; nephronophthisis-like nephropathy and renal cysts</td>
<td>Short ribs and small chest; flared iliac wings and narrow sacroiliac notches; cone-shaped epiphyses; renal cysts, intra-hepatic ductal dilatation</td>
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<td>Ellis-van Creveld disease</td>
<td>EVC (4p16.2)</td>
<td>Hair-nail-teeth abnormalities; mildly small chest; congenital heart disease</td>
<td>Barrel-shaped chest with cardiomegaly; similar pelvis to that in Jeune syndrome; cone-shaped epiphyses</td>
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<tr>
<td>Oral-facial-digital syndrome</td>
<td>X-linked dominant type: OFD1 (Xp22) for type 1</td>
<td>Anomalies of oral cavity, face, and digits; renal and hepatic cysts; CNS anomalies (Joubert syndrome)</td>
<td>Renal cysts; some patients have molar tooth sign</td>
</tr>
</tbody>
</table>

Note: AD = autosomal dominant, AR = autosomal recessive, MCKD = medullary cystic kidney disease.
CEP290-related Ciliopathy

Reilly Zenk
Naval Medical Center Portsmouth
Portsmouth, VA
Breast Best Case
Clinical information

31-year-old woman with a lump in the right breast. No family history of breast cancer. No past medical problems.

On clinical exam, there was a firm mobile mass in the upper outer quadrant of the right breast. No palpable adenopathy.
Juvenile Papillomatosiss of the Breast

Massimo Donalisio, MD
CHUV
Lausanne, Switzerland
Genitourinary Best Case
Clinical information

86-year-old female with history of endometrial cancer status post TAHBSO who presented with post-menopausal bleeding for 2 months. Upon further history, the bleeding was present only during urination so a workup for gross hematuria was performed.
Pathology report

- Invasive high-grade papillary urothelial carcinoma within renal pelvis and renal calyces
- Focal invasion into the renal parenchyma
Pathology report

- Invasive high-grade papillary urothelial carcinoma within renal pelvis and renal calyces
- Focal invasion into the renal parenchyma
- Small focus of early noninvasive high-grade papillary urothelial carcinoma at the distal ureter