Case of the week: #1
Fong Disease (Nail-Patella Syndrome)

Case History:
None provided.

Diagnosis:
Fong Disease.

Findings:
A frontal view of the pelvis demonstrates iliac horns.

Discussion:
Fong disease, also known as Nail-Patella Syndrome, is an autosomal dominant disease which manifests with nail dysplasia, patellar aplasia/hypoplasia, iliac horns, nephropathy, and arthrodysplasia of the elbows. The most serious complication of the disease is its effect on the kidneys which can lead to asymptomatic proteinuria for several years to end stage renal disease.

Fong disease has a prevalence of 1 in 50,000 live births and has no race or sex predilection. This condition is caused by mutations in the \( \text{LMX1B} \) gene. Some studies have shown that the location of this mutation may predict the frequency and severity of the proteinuria.

The pathognomonic feature of Fong disease is the iliac horn. It has been demonstrated with MRI imaging that the iliac horn, usually bilateral, is the site of attachment of the gluteus medius muscle.\(^1\) The horns actually consist of cortex and medulla that are continuous with the iliac wings. Other radiographic findings include patellar absence or hypoplasia with associated complications such as joint effusions and/or osteoarthritis. The elbows may demonstrate radial head dysplasia with posterior displacement. Clinically, this manifests with limited supination and pronation.

The other clinical symptoms include absent or dysplastic nails in addition to nonspecific changes such as discoloration or longitudinal ridging. Open angle glaucoma may also occur with a variable age of presentation. This manifestation is highly associated with
the NPSI gene. Some may experience irritable bowel syndrome or constipation. Other may have neurological associations which include Raynaud’s phenomenon, parasthesias, and epilepsy. The renal pathology in Fong disease is the leading cause of death in these patients. The associated nephropathy resembles glomerulonephritis in 30-55% of patients and can progress to end stage renal failure.

References:

