

FOP: Diagnosis, Imaging Studies, and Common Misdiagnoses

Full abbreviations, accreditation, and disclosure information available at [PeerView.com/NYY40](https://www.PeerView.com/NYY40)

Diagnosis of FOP¹⁻⁵

Malformation of the great toes is visible at birth

- Pediatricians and orthopedic surgeons are often among the first physicians to examine a child with congenital malformation of great toes

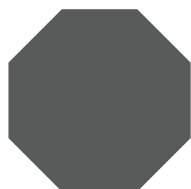
History of disease flare-ups that worsen the condition

Rogue bone growth progressively restricts movement

- Before HO develops, routine physical examinations—including a radiographic skeletal survey—will not provide sufficient evidence to definitively diagnose FOP

Genetic confirmation

- The most authoritative indicator is the detection of the *ACVR1* gene
- Single-gene detection allows rapid and accurate diagnosis of patients with FOP before the onset of HO



If FOP is suspected, all elective procedures—such as surgeries, biopsies, and immunizations—should be deferred until a definitive diagnosis is made

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How Helpful Are Imaging Studies?⁶

In early stages of a flare-up, ultrasound or MRI can evaluate tissue edema

X-rays are the most clinically helpful, accessible, and low-cost imaging modality. Other modalities are typically reserved for clinical trials and not yet standard of care

CT can be helpful for detecting HO

Functional imaging can show increased activity at flare-up sites

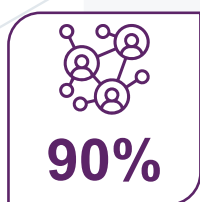
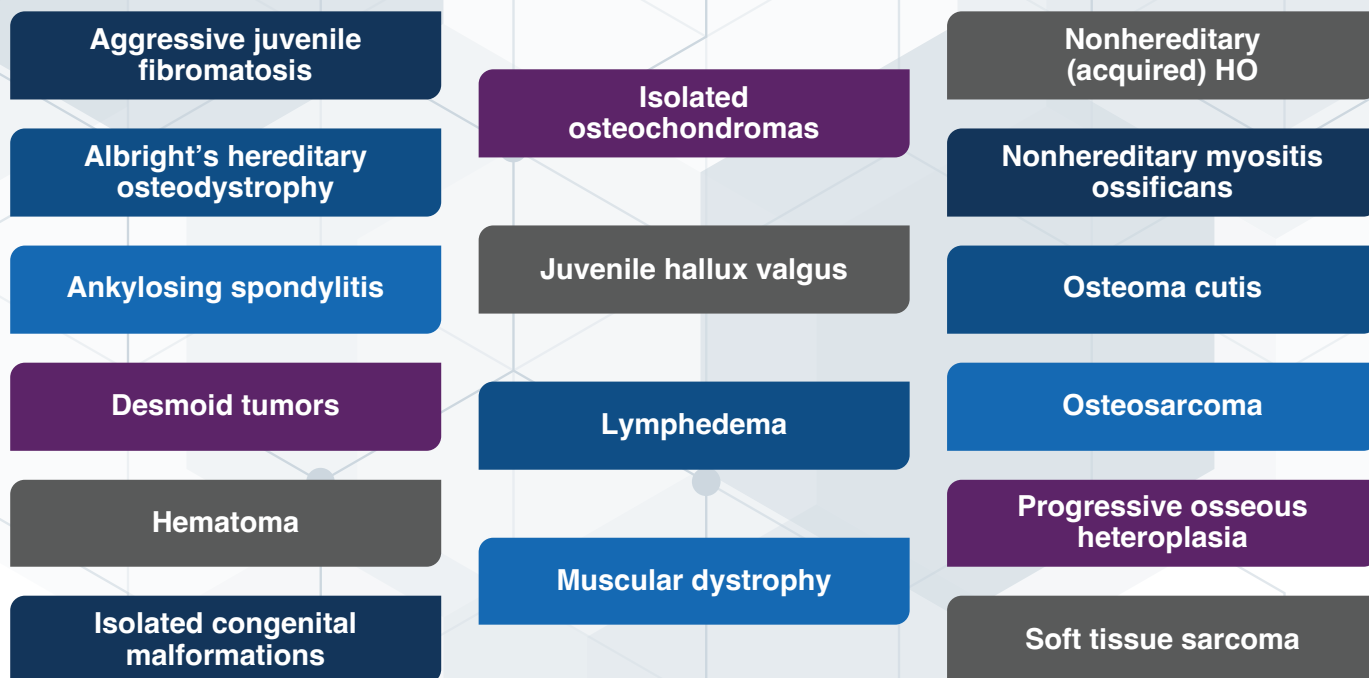
Further studies are needed to determine how well imaging can document disease progression and response to treatment

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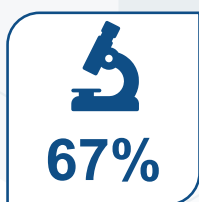
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FOP Differential Diagnosis and Common Misdiagnoses^{2,4,7-11}



of patients are misdiagnosed worldwide



undergo invasive procedures for diagnosis and treatment

Children often undergo unnecessary and harmful diagnostic biopsies that exacerbate the progression of the condition

