

Understanding Disease Background and Hallmark Symptoms of Fibrodysplasia Ossificans Progressiva (FOP)

Clinical Pearls: Hallmark FOP Symptoms

Check the toes

Great toe malformations are always seen at birth in classic FOP (eg, hallux valgus), even if a newborn otherwise appears normal.

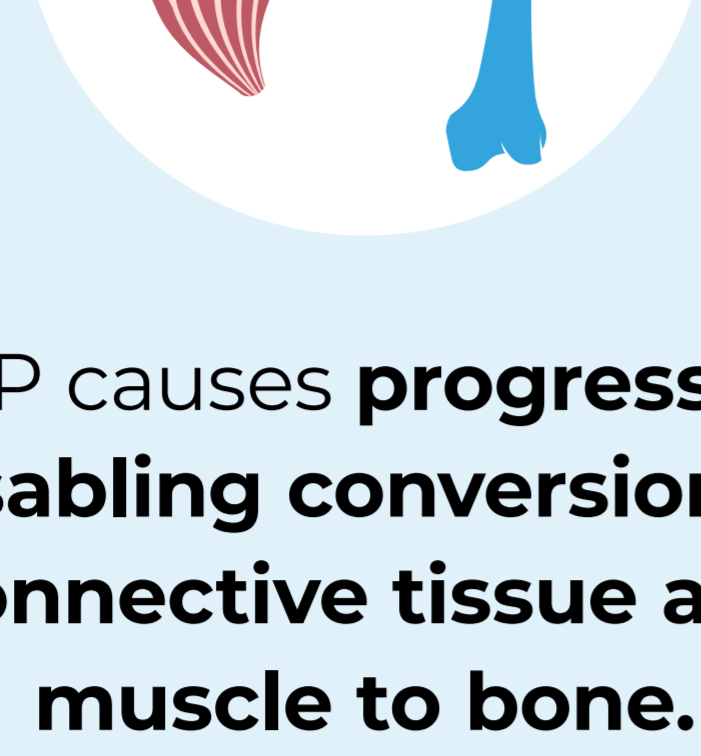
Use caution with invasive procedures

Some diagnostic procedures (eg, biopsy) can cause explosive heterotopic ossification (HO) formation in patients with FOP.

Note unusual neck stiffness

Patients with FOP often develop neck stiffness in early childhood, which can make crawling difficult for them.

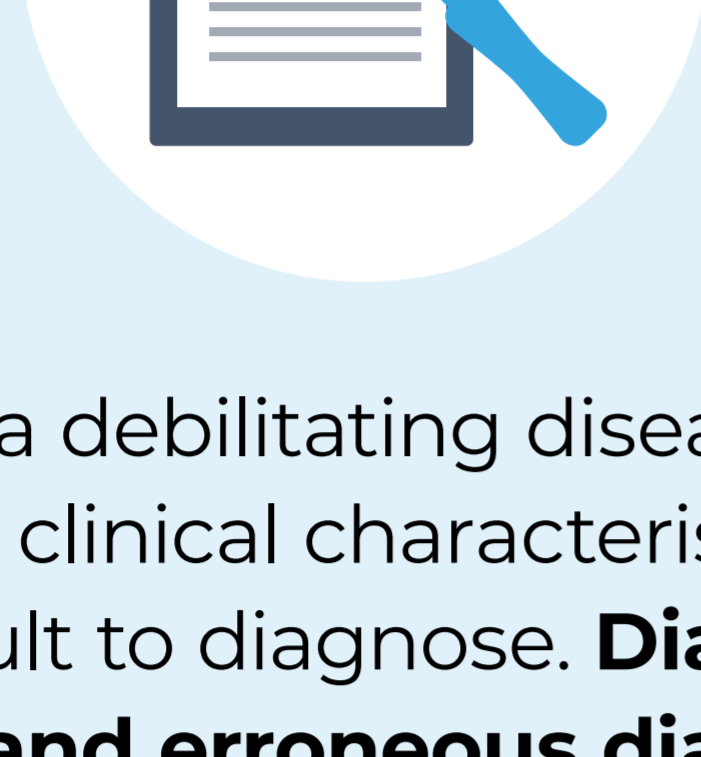
FOP Is an Ultra-Rare Genetic Condition



FOP causes **progressive, disabling conversion of connective tissue and muscle to bone.**



Worldwide prevalence is estimated to be between 0.5 and 1.4 per million with **no ethnic, racial, gender, or geographic predilection.**



FOP is a debilitating disease with distinct clinical characteristics, yet is difficult to diagnose. **Diagnostic delay and erroneous diagnosis are common.**



During the diagnostic journey, **patients with FOP often have dangerous and unnecessary diagnostic procedures that lead to permanent harm and lifelong disability.**

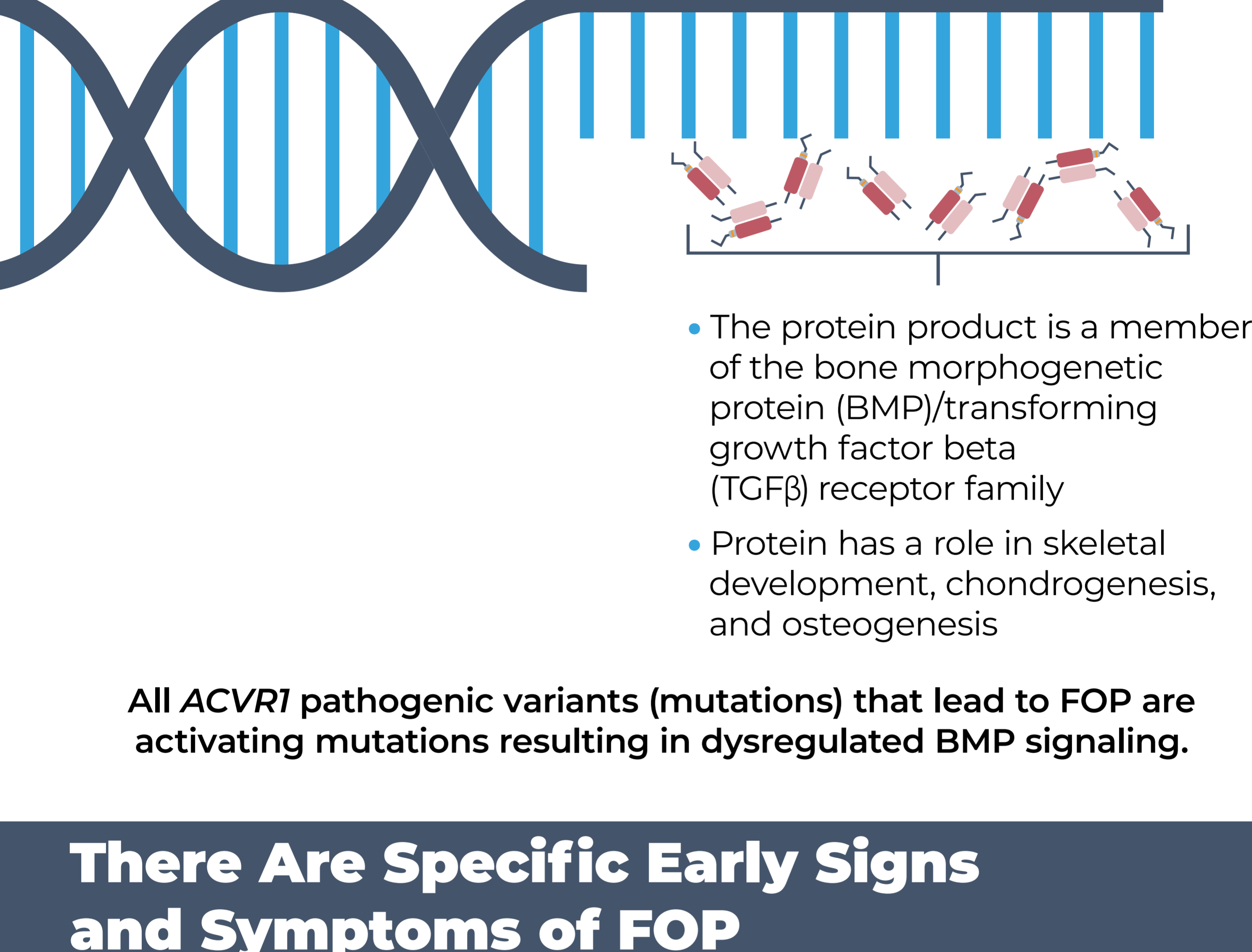


Heightened awareness of FOP and its hallmark symptoms can **hasten an accurate diagnosis for patients.**

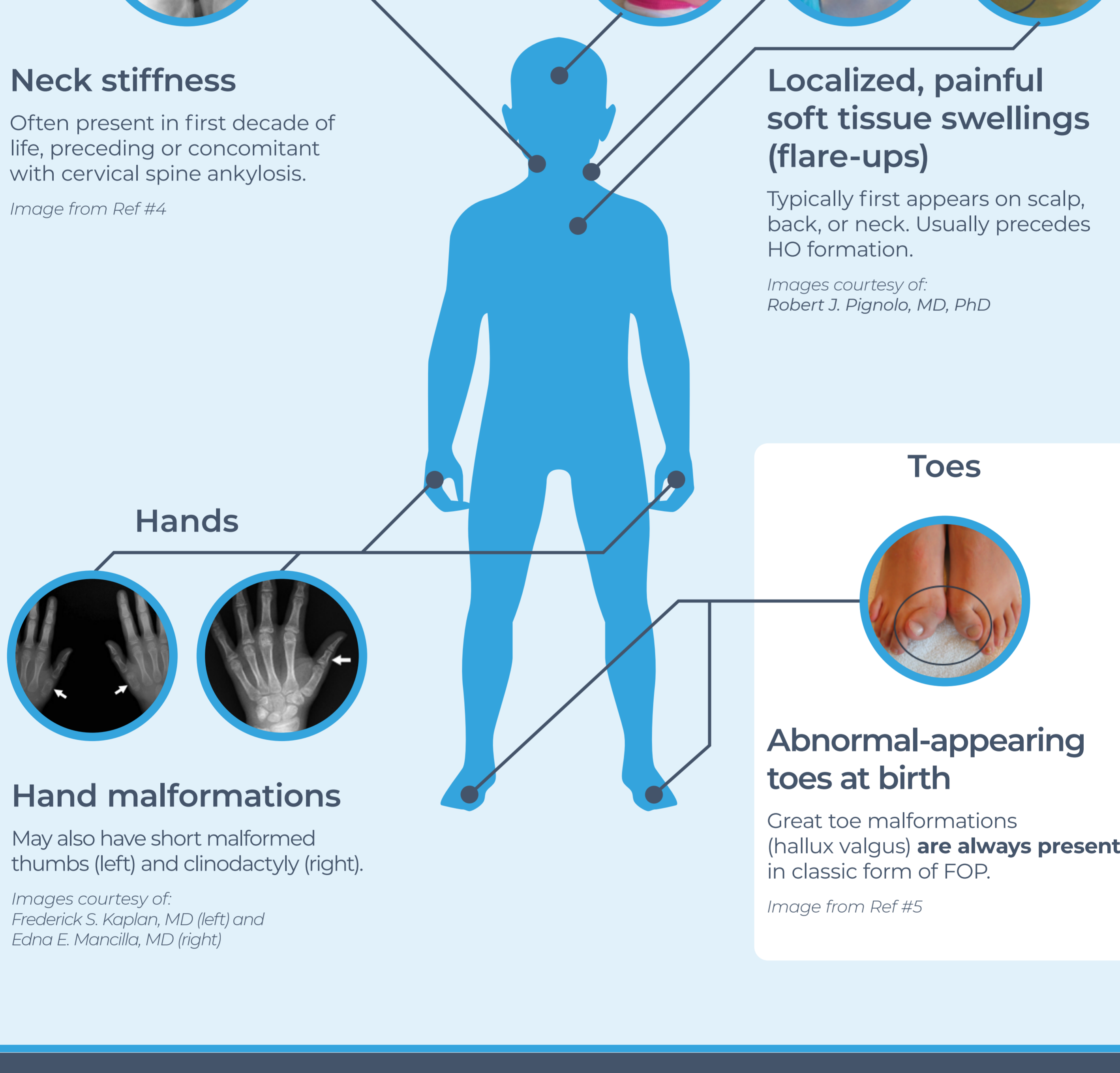


Heightened awareness of FOP opens the door to expert **resources and treatments needed to provide appropriate and individualized care.**

FOP Has a Genetic Cause



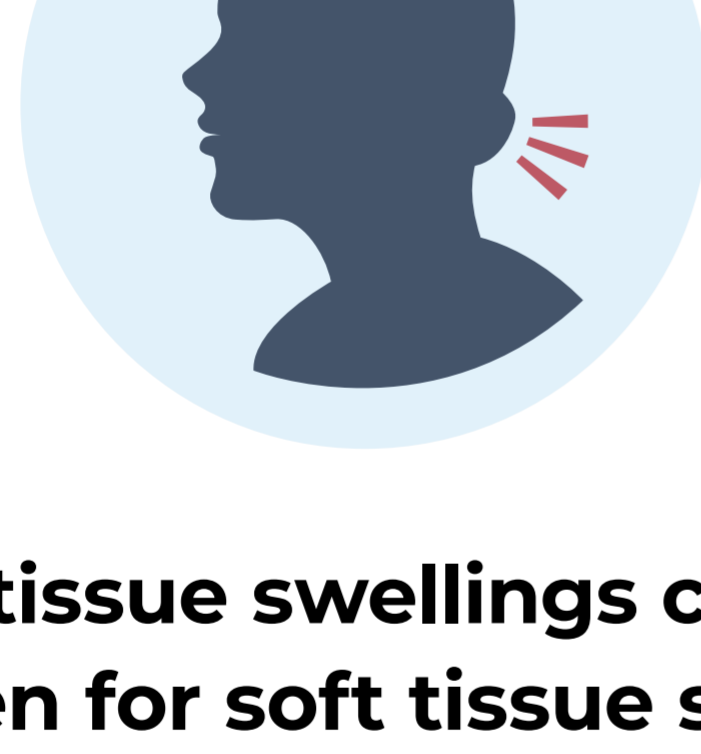
There Are Specific Early Signs and Symptoms of FOP



Possible Additional Clinical Findings

- Cervical spine malformations
Intra-articular ankylosis of facet joints and early cervical spine degenerative changes
- Short broad femoral necks
- Proximal medial tibial osteochondromas
- Hearing loss
- Thumb malformations
Short 1st metacarpal +/- monophalangism

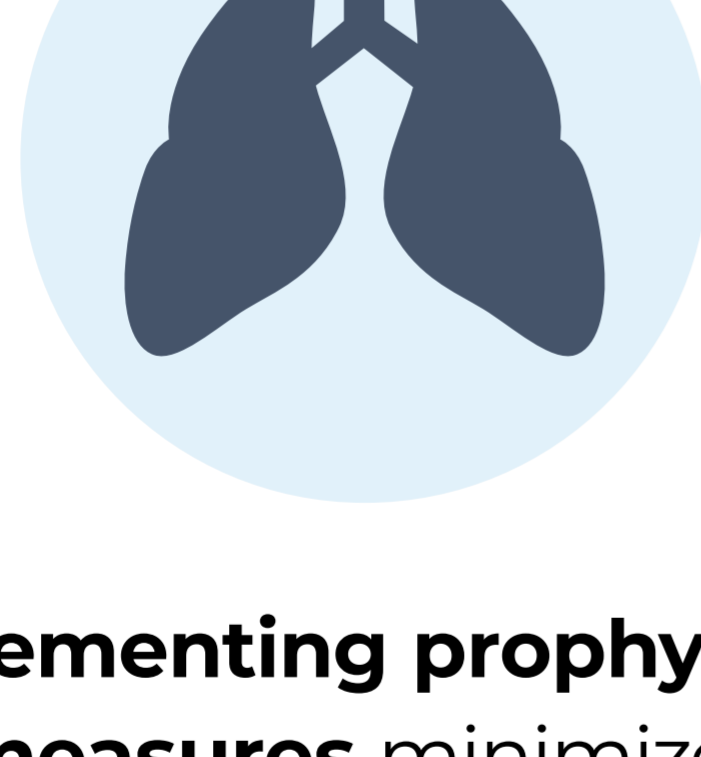
A Timely Diagnosis Prevents or Reduces Harm



Soft tissue swellings can be mistaken for soft tissue sarcoma. Correct diagnosis can reduce need for surgical excision, which can cause explosive HO formation.



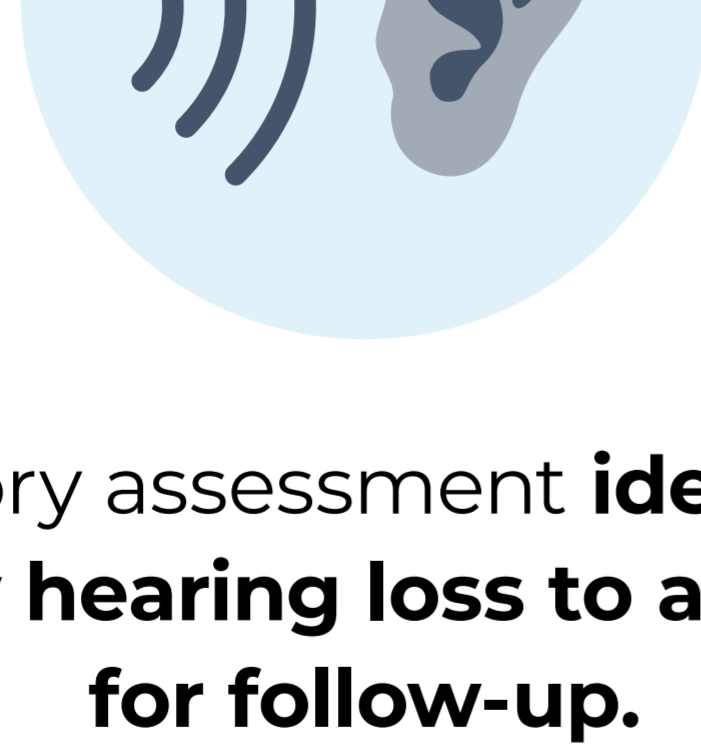
Taking fall reduction precautions minimizes trauma that can lead to flare-ups and HO formation.



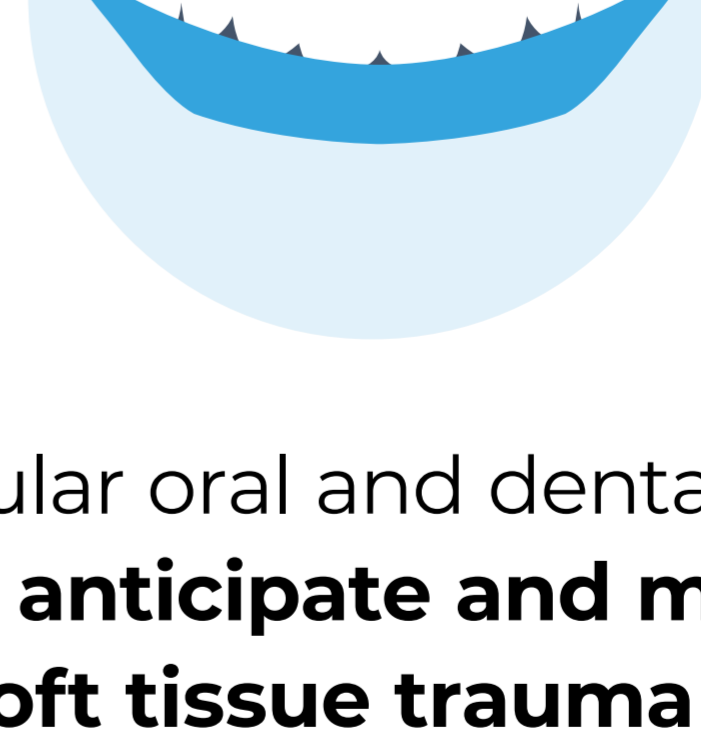
Implementing prophylactic measures minimizes respiratory decline (eg, with incentive spirometry).



Avoid use of early pharmacologics administered in routes that could cause trauma (eg, intramuscular injection).



Auditory assessment **identifies any hearing loss to allow for follow-up.**



Regular oral and dental care **helps anticipate and manage soft tissue trauma or oral HO formation.**

Summary

Understanding the hallmarks of FOP can lead to a timely diagnosis and reduce harm.

Faculty



Matthew T. Drake, MD, PhD

Dr. Matthew Drake is an endocrinologist and associate professor of medicine at Mayo Clinic. He has a particular interest in the mechanisms of bone loss and formation in both healthy and disease states.

References

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