Early Diagnosis:
Fibrodysplasia Ossificans Progressiva (FOP)

Fibrodysplasia Ossificans Progressiva (FOP) is a rare, severely debilitating congenital myopathy characterized by a hallmark great toe malformation, painful and recurrent episodes of soft tissue swelling (flare-ups) and heterotopic bone formation in muscle, tendons, and ligaments. In FOP, disability is cumulative and increases the risk of a shortened lifespan.

Why is Early Diagnosis so Important?

Common diagnostic and medical procedures, such as biopsies, intramuscular injections and surgery, can provoke flare-ups that result in rapid, debilitating heterotopic ossification.

A 2005 global survey of people with FOP found that **87%** of individuals were initially incorrectly diagnosed and that almost **50%** of them had undergone medical interventions that triggered bone formation and caused permanent loss of mobility. Furthermore:

4.1 The average number of years elapsed from initial symptom onset to correct diagnosis

6 The average number of physician specialists consulted before the correct diagnosis was made

Early diagnosis of FOP can help to avoid harmful interventions.
FOP can easily be identified early in life if attention is paid to specific signs and symptoms.

At birth, nearly **100%** of affected individuals have a hallmark, bilateral toe malformation in which the great toes are shortened and bent inwards resembling a bunion.

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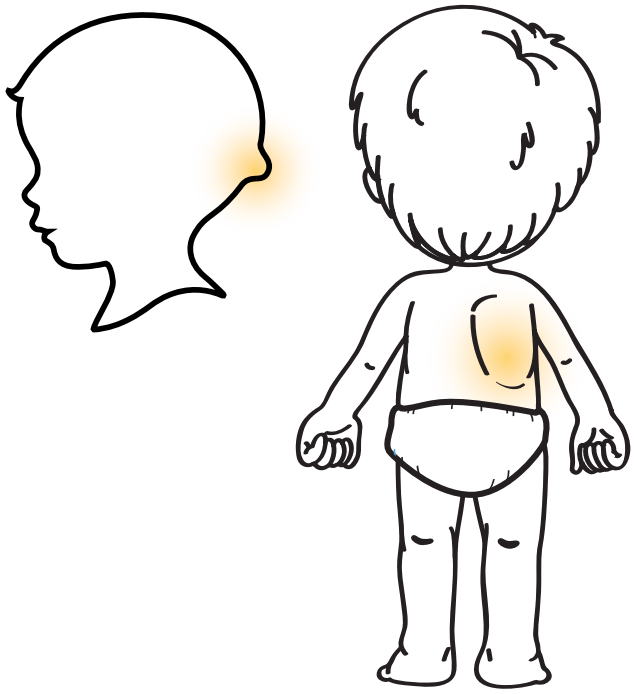
Early FOP
Signs and Symptoms

In infancy or early childhood, additional symptoms may emerge such as:

- ***Neck stiffness*** that causes difficulty crawling
- ***Scalp lumps*** that appear and disappear rapidly, migrate, or change shape
- Painful, ***soft-tissue swellings*** (flare-ups) on the neck, chest, or back



If an infant or young child presents with neck stiffness, scalp lumps, or soft tissue swellings, visual inspection of the toes may help prevent misdiagnosis and inappropriate medical interventions.

In the presence of the hallmark bilateral great toe malformation, prompt referral for genetic consultation is warranted. Confirmation of a clinical diagnosis with genetic testing for ACVR1 gene mutations associated with FOP is now available.

For more information:
www.clementiapharma.com

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[1]Kitterman, J. A., Kantanie, S., Rocke, D. M. and Kaplan, F. S. (2005). Iatrogenic harm caused by diagnostic errors in fibrodysplasia ossificans progressiva. Pediatrics 116, e654-e661.