



**Arthritis or contractures?**



**Subcutaneous nodules?**



**Hoarse or weak voice?**

**These symptoms may indicate Farber disease, caused by variants in the *ASAH1* gene. Farber disease is commonly misdiagnosed as juvenile idiopathic arthritis (JIA).**

### **Overview**

Farber disease is a form of acid ceramidase deficiency, a rare lysosomal storage disorder.

- Farber disease is progressive.
- Symptoms may take years to appear together.
- The clinical presentation is broad. Some patients die in early childhood while others may live well into adulthood.

### **Other Symptoms**

- Hypotonia/muscle weakness
- Signs of systemic inflammation
- Developmental delay
- Failure to thrive
- Peripheral osteolysis



# No-cost *ASAH1* Genetic Testing

## How to order:



To request a testing kit, contact PerkinElmer Genomics at [Adam.Bennett@perkinelmer.com](mailto:Adam.Bennett@perkinelmer.com)

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Send your patient's dried blood spot or saliva sample

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Test results will be sent via email within 21 days

