# Flowchart for Clinicians Cure HSPB8

1

### Referral to neuromuscular specialist

- Atrophy of tarsal muscles and tibialis anterior
- Scapular winging
- Waddling gait, lumbar lordosis, or scoliosis
- Decreased ankle reflex

3

#### **Genetic Counselling**

- Single-gene testing in the case of a known familial HSPB8 variant
- Multi-gene panel sequencing in undifferentiated cases\*
- Carrier testing for other family members.

**Suspected HSPB8 Myopathy** 

- Motor weakness
- Balance issues
- Breathing difficulties
- Foot drop

2

## **Testing**

- Nerve conduction studies : motor neuropathy
- Electromyography: mixed myopathic and neurogenic
- Elevated Creatine Kinase
- Muscle MRI with fatty degeneration
- Pulmonary function test: respiratory insufficiency.

4

#### Collaboration

- Geneticist
- Neurologist/neuromuscular specialist
- Cardiology: cardiomyopathy and conduction defects
- Pulmonology
- Supportive: Physical and Occupational therapy
- Nutrition and supplements