Is he lagging behind?

is it Duchenne?

WHAT TO DO IF YOU SUSPECT DUCHENNE MUSCULAR DYSTROPHY

OBSERVE

KEY SIGNS OF DMD[1]

Early signs may be present before the age of 2 years[2,3]

- *Signs of muscle weakness*
- *Not walking by 16–18 months*
- *Cognitive and speech delay*
- *Unexplained increases in transaminases*
- *Evidence of Gowers' sign or toe walking (any age, especially <5 years old)*

TEST

ORDER CREATINE KINASE (CK) TEST[2–4]

Elevated CK levels reflect muscle damage, therefore testing is recommended in primary care[2–4]

Elevation in CK warrants prompt referral to a neuromuscular specialist[1,3]

REFER

REFER TO A NEUROMUSCULAR SPECIALIST[1]

Diagnosis is confirmed through:

1. CLINICAL ASSESSMENT[1]

Includes neuromuscular and skeletal examinations

2. GENETIC ASSESSMENT[1,5–8]

MLPA detects large dystrophin gene mutations

Dystrophin gene sequencing detects small/single nucleotide mutations

3. MUSCLE BIOPSY[1]

*Detects the presence/absence of dystrophin protein**

*If genetic testing does not confirm a clinical diagnosis of Duchenne, then a muscle biopsy sample should be performed[1]

MLPA, multiplex ligation-dependent probe amplification

CARE

EARLY DIAGNOSIS MAKES A DIFFERENCE:

- *Timely intervention may delay disease progression and complications[1,2,8]*
- *Access to emerging therapies and clinical trials[2]*
- *Access to genetic counselling and family planning[2]*

For more information about Duchenne visit www.**TAKE ON** DUCHENNE.eu*

Make every day count

*Take on Duchenne is developed and funded by PTC Therapeutics and is intended for EU HCPs

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