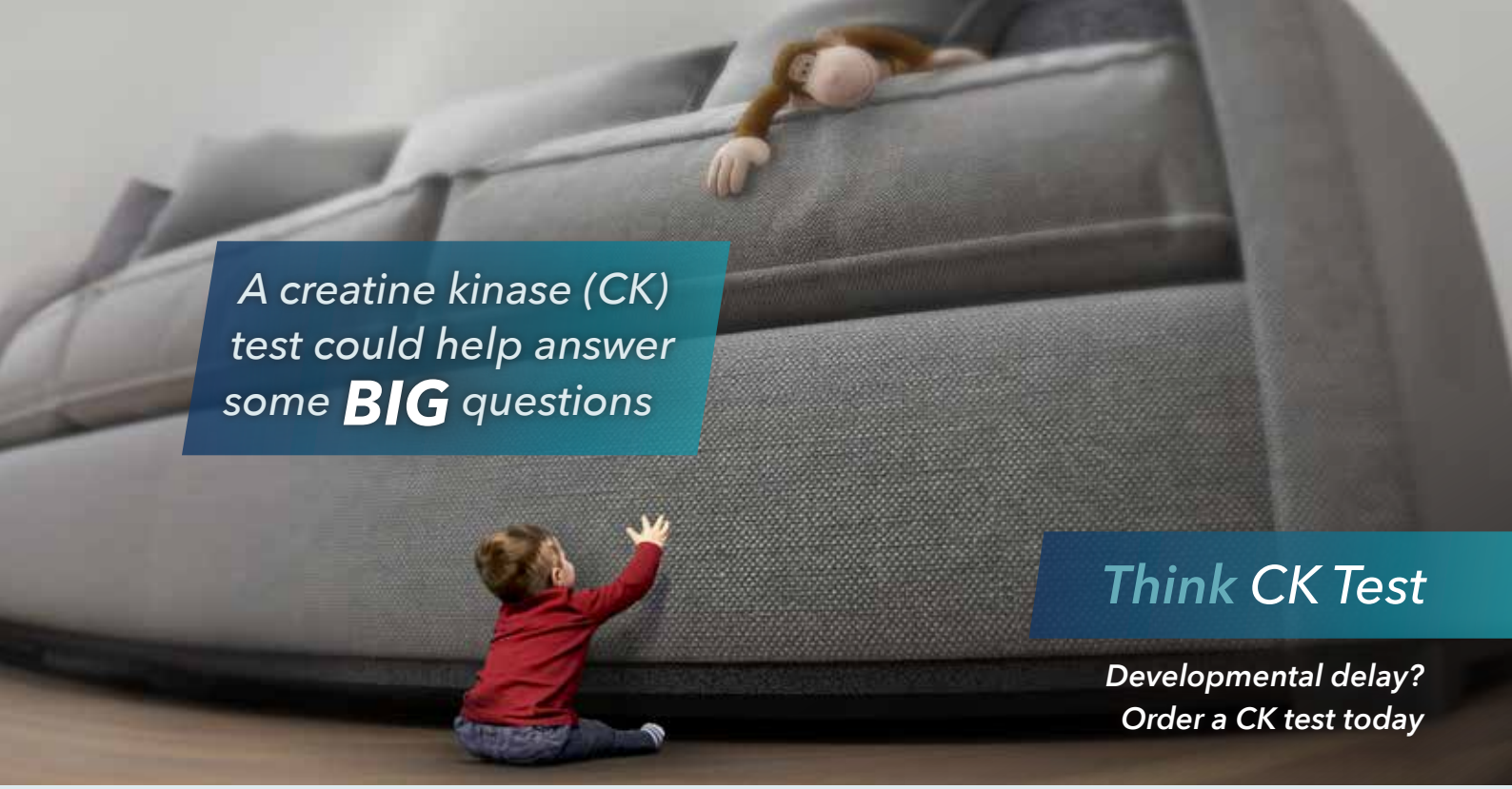

DUCHENNE MUSCULAR DYSTROPHY (DMD) IS A RARE GENETIC DISORDER[1,2]

- DMD affects ~1 in every 3,600–6,000 live male births[1–3]
- DMD is characterised by progressive muscle degeneration leading to loss of ambulation, respiratory and cardiac failure, and subsequent early death[1,2,4,5]

DMD IS CAUSED BY MUTATIONS IN THE DYSTROPHIN GENE ON THE X CHROMOSOME[2,5]

- DMD is caused by deletion and duplication mutations in the gene that encodes dystrophin, an important component of the muscle cell membrane[5,6]
- Mutations in the dystrophin gene lead to the absence of, or defects in, dystrophin[2,5]
- This results in ongoing muscle damage and replacement of muscle fibres by scar tissue and fat[6,7]

Within the first 3 years of life, affected infants and young boys with DMD show measurable deficits in gross and fine motor function[8,9]

By aged 5, prominent muscle weakness is observed[8]

By age 10–12, >70% of boys lose ambulation[8]

Many patients require noninvasive ventilator support at night by age 15[8]

Advanced respiratory and/or cardiac complications lead to mortality[1,2,8]

EARLY INTERVENTION MAY IMPROVE PATIENT OUTCOMES[1,2]

- Once muscle is lost it cannot be restored[7,10]
- Early diagnosis is critical to gain access to the right treatments and services[1,2,11]
- The role of GPs is vital as they are in an ideal position to spot early signs of neuromuscular disease[1,11,12]

TIMELY AND ACCURATE DIAGNOSIS CAN ENABLE THE PATIENT AND FAMILY TO RECEIVE THE CARE AND SUPPORT THEY NEED[1,2,13]

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GPs ARE A FIRST POINT OF CONTACT FOR CONCERNED PARENTS AND CAN HELP DRIVE A SUCCESSFUL DIAGNOSTIC JOURNEY[1,2]

OBSERVE

RECOGNISING RED FLAG SIGNS AND SYMPTOMS

0–6 months

- No head control at 2 months[2,3]
- Not making sounds at 4 months[3]
- Not reaching or grasping by 6 months[2,3]
- Not rolling over by 6 months[2,3]

6–18 months

- Not sitting independently at 9 months[3,4]
- Not crawling by 9–15 months[2,5]
- Not speaking first words by 12 months[3]
- Difficulty rising to stand by 18 months[6,7]
- Not walking well by 16–18 months[3,4,6,8]

2–3 years

- Gowers' sign from 2 years old[9]
- Not walking smoothly at 2 years old (tip-toe walking)[9]
- Not jumping at 2 years old[2]
- Difficulty running or climbing at 3 years old[2,3]
- Not speaking in sentence at 3 years old[3]

Other signs and symptoms

- Elevated serum CK or transaminases[10,11]
- Cognitive delay[8]
- Calf hypertrophy[8,12]
- Abnormal gait[8]
- Frequent falls[8,12]

SCHEDULED HEALTH CHECKS ARE A GOOD OPPORTUNITY TO CHECK NEUROMUSCULAR DEVELOPMENT[2,13]

TEST AND REFER

DEVELOPMENTAL DELAY SHOULD TRIGGER A CK TEST

POSITIVE FAMILY HISTORY WITH SUSPICION OF ABNORMAL MUSCLE FUNCTION[8]

DEVELOPMENTAL DELAY, SUCH AS DIFFICULTY RISING TO STAND OR NOT WALKING BY 18 MONTHS[4,6,8,14]

UNEXPLAINED INCREASES IN TRANSAMINASES[8]

PERFORM A CK TEST

ELEVATED CK* (>250 U/L)

PROMPTLY REFER TO A NEUROMUSCULAR SPECIALIST OR GENETICIST[6,8,12]

NORMAL CK*

PATIENTS WITH MISSED MOTOR MILESTONES SHOULD BE REFERRED TO A NEUROMUSCULAR SPECIALIST[6]

Normal or mildly elevated CK levels do not rule out neuromuscular disease[6]

CARE

EARLY DIAGNOSIS MAKES A DIFFERENCE:

- *Timely intervention may delay disease progression and complications[8,13,15]*
- *Access to emerging therapies and clinical trials[13]*
- *Access to genetic counselling and family planning[13]*

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

*The normal CK range is generally up to 250 U/L. Absolute values may differ between laboratories.[6] †Take on Duchenne is developed and funded by PTC Therapeutics and is intended for EU HCPs

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PTC THERAPEUTICS

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