

# CHKB-Related Myopathy

Multicentre Clinical Data Collection Form (Case Report Form)

*One form per patient · Fields marked \* are required*

**1. Centre / Hospital \***

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**2. Country \***

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**3. Clinician's name \***

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**4. Patient ID \***

*Format: ERN Centre code/Number — e.g. FR01-001*

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**5. Sex \***

Female

Male

**6. Consanguinity (parents) \***

Yes

No

Unknown

**7. Family history of similar disease**

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**8. Current patient status \***

Alive

Deceased

Unknown/Not available

**9. Age at death (if deceased)**

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**NEONATAL**

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**10. Pregnancy complications \***

Yes

No

Unknown

**11. Gestational age at birth (weeks)**

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**12. Birth weight (kg)**

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**13. Neonatal hypotonia \***

- Yes
- No
- Unknown

**14. Respiratory problems at birth \***

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**15. Neonatal ICU admission \***

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**ONSET & MILESTONES**

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**16. Age at first symptoms \***

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**17. Age at diagnosis \***

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**18. First symptoms**

*Select all that apply:*

- Hypotonia
- Delayed motor milestones
- Frequent falls
- Difficulty running
- Difficulty climbing stairs
- Gower's sign
- Waddling gait
- Rhabdomyolysis/myoglobinuria
- Other

**19. Independent sitting acquired \***

- Yes
- No
- Unknown

**20. Age at sitting (months)**

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**21. Independent walking acquired \***

- Yes
- No
- Unknown

**22. Age at walking (months) \***

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**23. Walking delayed \***

- Yes
- No
- Unknown

**24. Lost independent ambulation \***

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**25. Age at loss of ambulation (years)**

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**26. Current ambulatory status \***

- Ambulant unaided
- Ambulant with aid
- Wheelchair full-time
- Unknown

**CLINICAL**

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**27. Predominant pattern \***

- Limb-girdle/proximal
- Distal
- Scapulooperoneal
- Axial
- Generalized
- Unknown
- Other

→ *Limb-girdle/proximal | Distal | Scapulooperoneal | Axial | Generalized | Unknown | Other*

**28. Symmetry \***

- Symmetrical
- Asymmetrical
- Unknown

**29. Skeletal deformities \***

- No
- Unknown
- Yes — Scoliosis
- Yes — Kyphosis
- Yes — Pes cavus
- Yes — Pectus excavatum
- Yes — Other

→ *Select No/Unknown or choose the deformity type. Can type multiple, e.g.: Yes — Scoliosis + Pes cavus*

**30. Contractures \***

- No

- Unknown
- Yes — Achilles
- Yes — Hip
- Yes — Elbow
- Yes — Multiple
- Yes — Other

→ Select No/Unknown or choose the location. Can type multiple locations.

**31. Rhabdomyolysis \***

- Yes
- No
- Unknown

**32. Intellectual disability \***

- No
- Yes mild
- Yes moderate
- Yes severe
- Suspected
- Unknown

**33. Developmental delay \***

- Yes
- No
- Unknown

**34. Speech / language delay \***

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**35. Epilepsy / seizures \***

- Yes focal
- Yes generalised TC
- Yes absence
- Yes myoclonic
- No
- Unknown

**36. Respiratory involvement \***

- Yes
- No
- Unknown

**37. Ventilatory support \***

- Spontaneous breathing
- Nocturnal NIV

- Continuous NIV
- Tracheostomy
- Unknown

→ *If Tracheostomy: add age after selecting — e.g. Tracheostomy (age 12 yrs)*

**38. Cardiac involvement \***

- Yes
- No
- Unknown

**39. Cardiac device \***

- None
- Unknown
- Yes — ICD
- Yes — Pacemaker

→ *If device: add age after selecting — e.g. Yes — ICD (age 35 yrs)*

**40. Skin abnormalities \***

- No
- Yes — ichthyosis
- Yes — hyperkeratosis
- Yes — other
- Unknown

→ *If other: specify after selecting*

**41. Hearing loss \***

- No
- Yes — sensorineural
- Yes — conductive
- Yes — mixed
- Suspected
- Unknown

**INVESTIGATIONS**

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**42. EEG result \***

- Normal
- Abnormal — epileptiform discharges
- Abnormal — slowing
- Abnormal — other
- Unknown/Not done

→ *If abnormal: add detail after selecting — e.g. Abnormal — focal spikes right temporal*

**43. Brain MRI \***

- Yes

- No
- Unknown

**44. Brain MRI result \***

- Normal
- Cerebral atrophy
- White matter abnormalities
- Cerebellar atrophy
- Cortical malformation
- Other
- N/A

→ *Select result; can add free description after option*

**45. Spirometry available \***

- Yes
- No

**46. FVC % predicted \***

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**47. FEV1 % predicted \***

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**48. Echocardiogram \***

- Normal
- Dilated cardiomyopathy
- Hypertrophic cardiomyopathy
- Diastolic dysfunction
- Not done
- Other

→ *Select result*

**49. LVEF % (if reduced)**

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**50. ECG \***

- Normal
- Abnormal — arrhythmias
- Abnormal — other
- Unknown/Not done

→ *If abnormal: add detail after selecting*

**51. EMG \***

- Myopathic
- Myopathic + myotonic
- Neurogenic

- Mixed
- Normal
- Not done
- Other

→ If Other: specify

**52. Nerve conduction studies \***

- Normal
- Polyneuropathy
- Not done
- Other

→ If Other: specify

**53. Muscle MRI performed \***

- Yes
- No

**54. Muscle MRI findings**

*Describe pattern — e.g. rostrocaudal gradient, most affected muscles*

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**55. CPK (U/L) \***

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**MUSCLE BIOPSY**

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**56. Muscle biopsy performed \***

- Yes
- No

**57. Muscle biopsied \***

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**58. Age at biopsy \***

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**59. Histological features**

*Select all that apply:*

- Dystrophic changes
- Fibre size variation
- Necrosis/regeneration
- Endomysial fibrosis
- Fatty replacement
- Lipid accumulation
- Megaconial pattern
- Peripheral accumulation enlarged mitochondria
- Central depletion of mitochondria

- COX/SDH abnormality
- Respiratory chain enzyme abnormality
- Electron microscopy performed
- Other

**60. Biopsy typical for megaconial CMD \***

- Yes
- No
- Uncertain

**61. Biopsy report / images \***

- Yes attached
- Will send separately
- Not available

**62. TEM report / images \***

- Yes attached
- Will send separately
- Not performed

**GENETICS**

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**63. Genetic diagnosis confirmed \***

- Yes
- No
- Pending
- Unknown

**64. Type of genetic analysis \***

- NMD gene panel
- WES
- WGS
- Sanger targeted

**65. Zygosity \***

- Homozygous
- Compound heterozygous
- One variant only
- Unknown

**66. Parental testing \***

- Yes
- No
- Not performed

Unknown

**67. Variant 1 — HGVS cDNA \***

*e.g. c.1119+1G>A*

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**68. Variant 1 — protein effect \***

*e.g. p.Asp373Glyfs\*26*

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**69. Variant 1 — classification \***

- Pathogenic
- Likely pathogenic
- VUS
- Other

**70. Variant 2 — HGVS cDNA**

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**71. Variant 2 — protein effect**

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**72. Variant 2 — classification**

- Pathogenic
- Likely pathogenic
- VUS
- Other

**ADDITIONAL (IF AVAILABLE)**

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**73. MRS performed**

- Yes — normal
  - Yes — reduced choline signal
  - No
  - Unknown/Not done
- *MR Spectroscopy — choline/NAA or choline/Cr ratio if available*

**74. DEXA performed**

- Yes
- No

**75. Lumbar spine T-score / Z-score (paed.)**

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**76. Femoral neck T-score**

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**77. DEXA classification**

- Normal

- Osteopenia
- Osteoporosis

**78. Atypical features / additional notes**

*Any atypical clinical features, unusual disease course, or additional relevant information*

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