

Recommendations for the management of individuals with CDD with suggested timepoints for completion.

Genetic Testing

Genetic testing should be offered to all individuals with DEE to confirm diagnosis.

Neurological: Clinical Management

At Baseline: Review by a pediatric neurologist and (if not the same professional) an epilepsy specialist. Families should be informed about Sudden Unexpected Death in Epilepsy (SUDEP).

Annually: Review by a pediatric neurologist and (if not the same professional) an epilepsy specialist.

Neurological: Neuroimaging

At Baseline: Individuals should be investigated with a brain MRI scan.

Neurological: EEG

At Baseline: EEG (regardless of clinical seizure status).

As Clinically Indicated: An EEG should be repeated to capture and classify spells of unclear clinical significance.

Neurological: Anti-seizure drugs

As Clinically Indicated: Individuals with seizures should be offered Ztalmy (Ganaxolone), if available. Equally, CBD (Epidiolex) should be offered for epilepsy with CDD, provided this met legal and regulatory requirements.

Neurological: Epilepsy surgery

At Baseline: Individuals should be considered for a VNS insertion if seizures are refractory to medications. Individuals should be considered for corpus callosotomy if seizures are refractory to medication.

Neurological: Stereotypes and movement disorders

At Baseline: Individuals should be screened for movement disorders and have these treated if causing problems.

Annually: Individuals should be screened for movement disorders and have these treated if causing problems.

Neurological: Somnology

At Baseline: Individuals should have their sleep assessed by their clinician.

Annually: Individuals should have their sleep assessed by their clinician.

Systemic: Auxology

At Baseline: Assessment of head circumference, weight and height.

Annually: Assessment of head circumference, weight and height.

As Clinically Indicated: Assessment of head circumference, weight and height.

Systemic: Gastrointestinal management including assessment and management of feeding

At Baseline: Assessment of gastrointestinal complications such as constipation, air swallowing and acid reflux. Individuals should be referred to a Gastrointestinal specialist as well as a Nutrition specialist. Non-specialist feeding and swallowing should be assessed during clinic reviews.

Annually: Assessment of gastrointestinal complications such as constipation, air swallowing and acid reflux. Non-specialist feeding and swallowing should be assessed during clinic reviews.

As Clinically Indicated: A gastrostomy should be considered either when weight plateaus or BMI tails inappropriately or when swallowing is considered unsafe.

Systemic: Respiratory

At Baseline: A non-specialist respiratory assessment to screen for breathing disorders, including hyperventilation, breath-holding or other conditions.

Annually: A non-specialist respiratory assessment to screen for breathing disorders, including hyperventilation, breath-holding or other conditions.

As Clinically Indicated: Referral to a pulmonologist/respiratory clinician.

Systemic: Cardiology

At Baseline: Screening for cardiac issues and this should include an ECG.

Systemic: Dermatology

At Baseline: Individuals should have a routine skin check for pressure ulcers and skin breakdown.

Annually: Individuals should have a routine skin check for pressure ulcers and skin breakdown.

Systemic: Urology

At Baseline: Bladder related issues should be checked regularly (e.g., to assess for urinary retention and urinary tract infections).

Annually: Bladder related issues should be checked regularly (e.g., to assess for urinary retention and urinary tract infections).

Systemic: Audiology

At Baseline: Individuals should have an audiological assessment in the form of auditory brainstem response (AABR) screening.

Systemic: Dental care

At Baseline: Individuals should have a dental check.

Annually: Individuals should have a dental check.

Systemic: Financial

At Baseline: Financial support options should be explored.

Annually: Financial support options should be explored.

Therapy assessments and interventions: Neurorehabilitation

At Baseline: Referral to a neuro-rehabilitation service to assess equipment needs and diagnose problems causing impairment of mobility or hand function and to prevent contractures.

Annually: Referral to a neuro-rehabilitation service to assess equipment needs and diagnose problems causing impairment of mobility or hand function and to prevent contractures.

Therapy assessments and interventions: Development

As Clinically Indicated: Development should be assessed during infancy (0–3 years), preschool age (3–6 years), pre-middle school age (6–9 years), adolescence age (12–16 years), early adulthood (18–25 years) and as needed thereafter.

Therapy assessments and interventions: Ophthalmology

At Baseline: Individuals should have a detailed vision assessment. Individuals should be referred for assessment and management of cortical visual impairment by an ophthalmologist familiar with this condition.

Therapy assessments and interventions: Communication

At Baseline: Individuals should be offered a speech therapy assessment and assessed for augmentative and assistive communication aids such as switches, touch pads or eye gaze aids.

Therapy assessments and interventions: Orthopedics

As Clinically Indicated: Hip and spine X-ray if there is a clinical concern. Screening test for osteopenia (such as wrist X-ray or DEXA scan) if there is a clinical concern.

Therapy assessments and interventions: Physiotherapy (PT)

At Baseline: Individuals should be offered PT assessment.

As Clinically Indicated: Access to PT regularly for any ongoing issues.

Therapy assessments and interventions: Occupational therapy (OT)

At Baseline: Individuals should be offered an OT assessment.

As Clinically Indicated: Access to OT for any ongoing issues.

Therapy assessments and interventions: Educational

At Baseline: Formal educational plans should be reviewed.

Annually: Formal educational plans should be reviewed.

As Clinically Indicated: Educational accommodations should be made if visual impairment is present.

International registry

At Baseline: All individuals with CDD should be offered to be enrolled in an international registry or other research studies.

About the IFCR

Our mission is to treat and cure CDKL5 Deficiency Disorder by funding scientific research, while helping affected individuals and their families to thrive. Please visit our website at www.CDKL5.com to learn more about our work, and let us know who you are by signing up for our “Connect CDKL5” contact registry.

Connect CDKL5



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