

CDKL5 clinical phenotype guidelines:

Core phenotype (need to be met for PP4)

- Seizures, including infantile spasms, beginning in infancy
- Global developmental delay
- Intellectual disability
- Hypotonia
- Severely impaired gross motor function
- Cortical visual impairment in the first 12 months

Supportive criteria (do not need to be met for PP4, however in the absence of one core phenotype, two or more supportive phenotypes can be used in its place)

- Sleep disturbances
- Gastrointestinal dysfunction
- Subtle dysmorphic features (broad forehead, large, deep-set eyes, tapered fingers, full lips, anteverted nostrils in males)
- Bruxism
- Hand stereotypies
- Periodic breathing
- Laughing, screaming spells
- Cold hands and feet
- Peripheral vasomotor dysfunction