

**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