

MECP2 clinical phenotype guidelines:

Core phenotype (need to be met for PP4)

- Regression of developmental progress and loss of at least 2 of 4 of following
- Loss, partial or complete of fine motor skills (hand use)
- Loss, partial or complete of spoken communication
- Abnormal (dyspraxic) or absent gait
- Stereotypies

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)

- Periodic breathing (breath-holding/hyperventilation) when awake
- Bruxism when awake
- Impaired sleep pattern
- Abnormal muscle tone
- Peripheral vasomotor disturbances
- Scoliosis/kyphosis
- Growth retardation (small stature)
- Small, cold hands and feet
- Inappropriate laughing/screaming spells
- Diminished response to pain
- Intense eye communication (“eye pointing”)

Additional notes: If information is provided such that a phenotype of Rett syndrome is suspected, with specific minimal features used for the diagnosis, then this can be used for PP4 in lieu of the specific clinical features listed.