

SLC9A6 clinical phenotype guidelines:

Core phenotype (need to be met for PP4)

- Global developmental delay
- Intellectual disability
- Epilepsy
- Autistic spectrum disorder
- Ataxia
- Craniofacial dysmorphism

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)

- Happy, excitable, frequent smiling, laughter
- Angelman-like features
- Microcephaly