

**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