

FOXG1 clinical phenotype guideline:

Core phenotype (need to be met for PP4)

- Microcephaly
- Severe intellectual disability
- Dyskinesia
- No period of normal development
- Neonatal hypotonia

Additional features (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)

- Abnormal brain imaging (e.g. partial agenesis of the corpus callosum, simplified gyral pattern, reduced white matter volume)
- Delayed motor development
- Impairment of postnatal growth
- Stereotypies
- Generalized seizures
- GE reflux
- Poor sleep pattern
- Unexplained episodes of crying
- Recurrent aspiration