UKGTN Testing Criteria

Test name: Familial Paroxysmal Kinesigenic Dyskinesia and Benign Familial Infantile convulsions With or Without Choreoathetosis	
Approved name and symbol of disorder/condition(s): Episodic Kinesigenic Dyskinesia 1; EKD1 Seizures, Benign Familial Infantile, 2; BFIS2 Convulsions, Familial Infantile, with Paroxysmal Choreoathetosis; ICCA	**OMIM numbers:** 128200, 605751, 602066
Approved name and symbol of gene(s): proline-rich transmembrane protein 2; PRRT2	**OMIM number:** 614386

Patient name:	**Date of birth:**
Patient postcode:	**NHS number:**
Name of referrer:	
Title/Position:	**Lab ID:**

Referrals will only be accepted from one of the following:	
Referrer	**Tick if this refers to you.**
Consultant Paediatric Neurologists	
Consultant Neurologists	
Consultant Clinical Geneticists	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Paroxysmal Kinesigenic Dyskinesia (PKD) - Paroxysmal movement disorder characterised by afebrile seizures and dystonia	
AND PKD - No loss of consciousness during episodes	
AND PKD - Normal neurological examination	
Benign Familial Infantile Convulsions(BFIS) - Onset of seizures in infancy (<2 years)	
AND BFIS - Normal neurodevelopment	
OR At risk family members where familial mutation is known.	

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.