UKGTN Testing Criteria

Approved name and symbol of disease/condition(s):	OMIM number(s):
Ichthyosis, Congenital, Autosomal Recessive 6; ARCI6	612281
Ichthyosis, Congenital, Autosomal Recessive 1; ARCI1	242300
Ichthyosis, Congenital, Autosomal Recessive 2; ARCI2	242100
Ichthyosis, Congenital, Autosomal Recessive 5; ARCI5	604777
Approved name and symbol of gene(s):	**OMIM number(s):**
transglutaminase 1; TGM1	190195
arachidonate 12-lipoxygenase, 12R type; ALOX12B	603741
arachidonate lipoxygenase 3; ALOXE3	607206
NIPA-like domain containing 4; NIPAL4	609383
cytochrome P450, family 4, subfamily F, polypeptide 22; CYP4F22	611495

Patient name:	**Date of birth:**
Patient postcode:	**NHS/CHI 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 Dermatologist	
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
At least two of the following are required: • Born with collodion membrane • Erythroderma • Dark plate-like scales or fine white scaling • Ectropium/ eclabium • Hyperkeratosis	
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