A1. Site/Study ID #:___ ___ / **G**___ ___ ___ ___ A2. Visit Date:___ ___ / ___ ___ / ___ ___ ___ ___
Month Day Year

SECTION B: GENERAL INCLUSION/EXCLUSION CRITERIA

B1. Date of Birth: **ELG5B01MM V2(2)/ ELG5B01DD V2(2)/ ELG5B01YY V2(2)/ ELG5B01DT**

Month Day Year

B2. Disease: **ELG5B02 V2(3)**

3. ☐ α1-Antitrysin (α1-AT) Deficiency
4. ☐ Alagille Syndrome (AGS)

GENERAL INCLUSION CRITERIA:

B3. Less than 25 years at enrollment 1. ☐ Yes 2. ☐ No **ELG5B03 V2(3)**

B12. Absence of liver disease 1. ☐ Yes 2. ☐ No **ELG5B12 V2(3)**

GENERAL EXCLUSION CRITERIA:

B5. Inability to comply with follow-up 1. ☐ Yes 2. ☐ No **ELG5B05 V2(3)**

B6. Failure to sign consent or HIPAA medical record release form 1. ☐ Yes 2. ☐ No **ELG5B06 V2(3)**

B7. Date consent form signed: **ELG5B07MM V2(2)/V ELG5B07DD V2(2)/ ELG5B07YY V2(4)/ ELG5B07DT**
Month Day Year

SECTION E: a1-ANTITRYPSIN (a 1-AT) DEFICIENCY

INCLUSION CRITERIA:

E2. Diagnosis of α 1-AT deficiency by:

a. Low serum α 1-AT level (_**ELG5E2AMG V2(10)**__mg/dL) **–AND–** **ELG5E2A V2(3)** 1. ☐ Yes 2. ☐ No

b. At least one of the following:

i. PiZZ phenotype **–OR–** **ELG5E2BI V2(3)** 1. ☐ Yes 2. ☐ No

ii. PiSZ phenotype **–OR–** **ELG5E2BII V2(3)** 1. ☐ Yes 2. ☐ No

iii. PiZZ genotype **–OR–** **ELG5E2BIII V2(3)** 1. ☐ Yes 2. ☐ No

iv. PiSZ genotype **ELG5E2BIV V2(3)** 1. ☐ Yes 2. ☐ No

–AND–

E3. Sibling with α 1-AT deficiency enrolled in CLiC Longitudinal Study **ELG5E03 V2(3)** 1. ☐ Yes 2. ☐ No

A1. Site/Study ID #:___ ___ / **G**___ ___ ___ ___ A2. Visit Date:___ ___ / ___ ___ / ___ ___ ___ ___
Month Day Year

SECTION F: AGS

INCLUSION CRITERIA:

F4. Sibling with AGS enrolled in CLiC Longitudinal Study **–AND–** 1. ☐ Yes 2. ☐ No **ELG5F04 V2(3)**

F2. Confirmed diagnosis of AGS 1. ☐ Yes 2. ☐ No **ELG5F02 V2(3)**

a. Diagnostic Characteristics (specify one of the following scenarios): **ELG5F2A**

	Family History Of AGS[a]	Paucity of interlobular bile ducts	*Jagged1*[d] *or Notch2* mutations	Number of clinical criteria
1. ☐	Present or absent	Present	Identified[b]	Any or no features
2. ☐	None (proband)	Present	Not identified[c]	3 or >
3. ☐	None (proband)	Absent or unknown	Not identified	4 or >
4. ☐	None (proband)	Absent or unknown	Identified	1 or >
5. ☐	Present	Present	Not identified	1 or >
6. ☐	Present	Absent or unknown	Not identified	2 or >
7. ☐	Present	Absent or unknown	Identified	Any or no features

[a]Family history = AGS present in a first degree relative
[b]Identified = *Jagged1* mutation may have been identified in clinical or research laboratory
[c]Not identified = Not identified on mutation screening, or not screened for
[d]*Jagged1* mutation = mutation, whole gene deletion or deletion of chromosome 20p which includes *Jagged1* locus

Major clinical criteria include cholestasis, consistent cardiac, renal, ocular disease, butterfly vertebrae, or characteristic “Alagille” facies of childhood or adulthood. The specific clinical criteria are:

Cardiac: Heart murmur (with further studies to clarify), Pulmonary stenosis (valvular, pulmonary artery stenosis), pulmonary atresia, tetralogy of Fallot, ASD or VSD.

Ocular: posterior embryotoxon or other anterior chamber defect, retinal pigmentary anomalies

Vertebral: butterfly vertebrae

Characteristic facial features: broad forehead, deep set eyes, pointed chin in child (preteen) or prognathism in adults, triangular face.

Renal: functional defects (such as tubular acidosis), renal insufficiency, renal vascular hypertension, vesicoureteral reflux, structural defects (agenesis, small kidneys, renal cysts, renal artery stenosis, dysplastic kidneys)

Cholestasis: See question F3 for evidence of cholestasis.

A1. Site/Study ID #:___ ___ / **G**___ ___ ___ ___ A2. Visit Date:___ ___ / ___ ___ / ___ ___ ___ ___
Month Day Year

F3. Evidence of cholestasis: (check all that apply)

a. ☐ Fasting total serum bile acid > 3x ULN for age **ELG5F03A V2(2)**

b. ☐ Direct bilirubin > 2 mg/dl **ELG5F03B V2(2)**

c. ☐ Fat soluble vitamin deficiency otherwise unexplainable **ELG5F03C V2(2)**

d. ☐ yGTP > 3x ULN for age **ELG5F03D V2(2)**

e. ☐ Intractable pruritus explainable only by liver disease **ELG5F03E V2(2)**

SECTION G: SUMMARY

G1. Is the subject eligible? 1. ☐ Yes 2. ☐ No 3. ☐ Eligible by exemption **ELG5G1**

ELG5INSIG V2(3) **ELG5SIGMM V2(2)/ ELG5SIGDD V2(2)/ ELG5SIGYY V2(4)/ ELG5SIGDT**

Investigators Signature Date