

ANNEX 1 –MINIMAL CLINICAL DATA FOR SUSPICION OF IEPBAS

1. Demographic and familial data

Demographic

- Initials Surname (3 first letters) and First name (2 first letters)
- Sex Male Female
- Date of birth / / yyyy/mm/dd

Family history

- Siblings with the same clinical picture Y N Not Known
 If Yes: Numbers
- Parental consanguinity? Y N Not Known

2. Clinical Examination: Diagnosis/Treatment Initiation OR Visit Follow-up

Clinical Examination Diagnosis/Treatment Initiation

- Date of Clinical Examination / / yyyy/mm/dd
- Or hospitalization date Start date / / End date / /
- Height (cm)
- Weight (kg)
- Icterus (Jaundice) Y N Not Known
- Pruritus Y N Not Known
- Steatorrhoea (loose or fatty stools) Y N Not Known
- Ascites Y N Not Known
- Hepatomegaly Y N Not Known
- Splenomegaly Y N Not Known

Vitamine A deficiency

- Keratitis Y N Not Known
- Decreased night vision Y N Not Known

Vitamine D deficiency

- Rickets Y N Not Known

Vitamine E deficiency

- Hyporeflexia Y N Not Known
- Cerebellar ataxia Y N Not Known

Vitamine K deficiency

- Hemorrhagic signs Y N Not Known

3. Blood Biochemistry

- Date of biochemistry examination / / yyyy/mm/dd
- ASAT UI/L Not Known
- ALAT UI/L Not Known
- Alkaline Phosphatase (AP) UI/L Not Known
- γ-GT UI/L Not Known
- Total bile acids μmol/L ..Not Known
- Bilirubin (total) μmol/L ..Not Known
- Conjugated bilirubin μmol/L ..Not Known

Haemostasis

- Prothrombin time..... Sec Not Known
- Prothrombin time..... %..... Not Known
- Factor II %..... Not Known
- Factor VII + X..... %..... Not Known
- Factor V %..... Not Known

4. Abdominal US

- Date of abdominal US examination ... / / yyyy/mm/dd

Liver

- Increased volume Y N Not Known
- Heterogeneous Y N Not Known

Dilated Bile Ducts

- Intrahepatic Y N Not Known
- Extrahepatic..... Y N Not Known
- Lithiasis or sludges Y N Not Known

Spleen

- Volume Normal Abnormal Not Known

Kidneys

- Size Normal Abnormal Not Known
- Renal cysts..... Y N Not Known

Gallbladder

- Morphology Normal Abnormal Not Known
- Lithiasis or sludges Y N Not Known

Comments:

5. Excrement Investigation

- Date of excrement investigation / / yyyy/mm/dd
- Mean weight (g/24h) Not Known
- Dry weight %..... Not Known
- Lipids..... (g/24h) Not Known
- Ratio Lipids/Dry weight %..... Not Known

6. Treatment (bile acid)

- Indication Text
- Trade Name Name®
- International Nonproprietary Name INN
- Dosage Text
- Posology Text
- Start Date / / yyyy/mm/dd
- End Date / / yyyy/mm/dd