Proforma for estimation of leucocyte cystine

For lab use only:

Lab ID	
Date of receiving sample	
Amount of sample received	_____ ml
Sample received in room temperature	Yes/No

Name	Father's name
Date of birth	Age _____ years, Sex___
Phone number	Address

BASIS OF DIAGNOSIS

Confirmed by genetics	☐	Metabolic alkalosis	☐
Polyuria, polydipsia	☐	Metabolic acidosis	☐
Failure to thrive	☐	Hypophosphatemia	☐
Hyponatremia, salt craving	☐	Hypercalciuria	☐
Mild	☐	Nephrocalcinosis	☐
Severe	☐	Beta-2-microglobinuria	☐
Hypokalemia	☐	Episodes of dehydration	☐
Mild	☐	Hypernatremia	☐
Severe	☐	Water deprivation test positive	☐
Tetany	☐	Cystine crystals (eye)	☐
Rickets	☐	Hepatosplenomegaly	☐
Mild	☐	Hypomagnesemia	☐
Severe	☐	Hypothyroidism	☐
Photophobia	☐	Diabetes	☐
Others			

Genetics

Gene:_____ c._____ p._____

homozygous/heterozygous/compound heterozygous *(Encircle one)*

Classification: variant of unknown significance/likely pathogenic/pathogenic *(Encircle one)*

HISTORY

Symptom	*Duration*	*Symptom*	*Duration*
Polyuria ☐		Intermittent tachypnea ☐	
Polydipsia ☐		Seizures ☐	
Failure to thrive ☐		Visual difficulty ☐	
Salt craving ☐		Photobhobia ☐	
Paralytic episodes ☐		Hearing impairment ☐	
Tetany ☐		Vomiting ☐	
Diarrhoea ☐		Abdominal distension ☐	
Bony deformities ☐		Constipation ☐	
Night blindness ☐		Others	

PERINATAL HISTORY

Parameters	*Details*	*Signs*	*From gestation*
Birth weight		Polyhydramnios ☐	
Gestation		Oligohydramnios ☐	
Adverse perinatal events ☐			

EXAMINATION AT DIAGNOSIS

Weight ____ kg Height ____ cm

Systolic blood pressure _____ mm Hg

Diastolic blood pressure _____ mm Hg

Bony deformities Yes / No

Rickets predominantly involving lower limbs/upper limbs/chest/bossing

Pubertal status Prepubertal / Pubertal /Post pubertal Puberty delayed Yes / No

Systemic examination

Chest: _____

CVS: _____ Abdomen: _____

CNS: _____

FAMILY HISTORY *(draw pedigree)*

Consanguinity: Yes / No , degree (if yes):_____

INVESTIGATIONS:

Time-point	Onset	6-months	12-months	24-months	36-months	Last follow-up
Date						
Hb						
TLC						
DC						
Plt						
Ur						
Cr						
Ca						
PO4						
UA						
Na						
K						
TP						
Alb						
SGOT						
SGPT						
SAP						
PTH						
Vit D						
TSH/T3/T4						
HCO3						
BE						
Serum Chloride						
Mg						
Fasting BS, HbA1c						

Time-point	Onset	6-months	12-months	24-months	36-months	Last follow-up
Date						
Volume, mL/d						
Cr, mg/dl						
Ca/Cr						
24-hr protein, mg/day						
24-hr Ca, mg/kg/day						
Aminoacidogram						

Date
FEPO4
TRP
TmP/GFR
Fasting urine pH
U-B co2
FEHCO3

USG-KUB: Date_____ final interpretation_____

Medullary nephrocalcinosis Yes / No

Vision assessment:

Cystine crystals Yes / No

Other manifestations:_____

Therapy:

Cysteamine bitartrate: Yes / No

Date of initiation of therapy:

Dose (g/1.73m^{2-}/day): _____

If on Cysteamine, date and time of last dose: Time of sample drawn:

Potassium supplements (mEq/kg/day):

Bicarbonate supplements(mEq/kg/day):

Phosphate (mg/kg/day):

Magnesium (mg/kg/day):

Indomethacin: (mg/kg/day): , duration of therapy with indomethacin:

AE-inhibitors Yes / No

Gastrostomy feeding: Yes / No

Recombinant growth hormone Yes / No _____ IU/day

Orthosis: Required/not required

Other therapy/ surgical intervention:

Follow-up:

Date of last follow-up: Height: Weight:

Complications (tick if present)

Hypothyroidism ☐

Diabetes mellitus ☐

Rickets/ osteoporosis ☐

CKD (Stage_____) ☐

Corneal ulcers ☐

Retinopathy ☐

Gastroesophageal reflux ☐

Myopathy ☐

Nephrocalcinosis ☐

Hypercalciuria ☐

Cognitive impairment ☐

Learning disorder ☐

Sleep apnea/aspirations ☐

Others: _____