Collaboration, Education and Test Translation (CETT) Program
Clinical data diagnostic testing X- Linked Recessive Chondrodysplasia Punctata (CDPX1)
Please complete and return with sample

Patient Name: _____ **ID:** _____

Submitting Physician: _____ **Date:** ____/____/____ (m / d / y)

Current Information: _____
(address, phone #)

Date of Birth: ____/____/____ (m / d / y) **Age (yrs & mos):** _____ **Sex: M F**

Ethnicity: Caucasian Black/African Hispanic Asian
Hawaiian / Pacific Islander American Indian Mixed

Ancestral country of origin for maternal family: _____

Growth

Gestational Age	_____	
Birth weight	_____	
Birth length	_____	
OFC	_____	
Weight	_____ Kg	
Height	_____ cm	
Failure to thrive	**Yes**	**No**
Demise	**Yes**	**No**

Development

Motor	**Normal**	**Delayed**
Cognitive	**Normal**	**Delayed**

Other systems involved

Hearing loss	**Yes**	**No**
If yes, which type? _____		
(conductive, neurosensorial, or mixed)		
Cataracts	**Yes**	**No**
Ichthyosis	**Yes**	**No**
Cardiovascular defect	**Yes**	**No**
Genitourinary defect	**Yes**	**No**
GE reflux	**Yes**	**No**
Respiratory abnormalities	**Yes**	**No**

Additional findings or describe the above
_____

Craniofacial

Nasomaxillary hypoplasia	**Yes**	**No**
Flattened nose	**Yes**	**No**
Alar grooves [1]	**Yes**	**No**

Other _____
_____

Skeletal

Brachytelephalangy [2]	**Yes**	**No**
Limbs shortening	**Yes**	**No**
Cervical spine abnormality	**Yes**	**No**
Chondrodysplasia punctacta		
In feet	**Yes**	**No**
In hands	**Yes**	**No**
Bronchi	**Yes**	**No**
Trachea	**Yes**	**No**

Other bone /cartilage structure involved
_____
_____

Prior Tests

Karyotype: _____		
Deficient plasmalogen synthesis[3]	**Yes**	**No**
FISH for STS gene[4]	**Deleted**	**Normal**
Sterol profile[5]	**Normal**	**Abnormal**

Relevant Family History
Other affected family members:
_____
Medicine used during pregnancy or pregnancy complications: *(Special concern for vitamin K antagonists which are warfarin and phenytoin)*
_____
Maternal illness: *(Special concern for autoimmune diseases, pancreatitis or other condition that may cause vitamin K deficiency)*
_____

[1] Alar groove: a longitudinal groove at the medial side of the nasal alar wing
[2] Brachytelephalangy: shortness of the fingertips
[3] Plasmalogens synthesis is a biochemical test for peroxisomal RCDP
[4] STS: Steroid sulfatase gene is situated close to ARSE on Xp22 and is often involved in interstitial deletion
[5] Sterol profile: Used to diagnose X-Linked Dominant Chondrodysplasia Punctata