**Cornelia de Lange Syndrome Clinical Questionnaire**
***Please complete and return with sample***

**Name:** ______________________ **DOB** __________ **Date** __________

| Features | Present: | Yes | No |
|---|---|---|---|
| **Growth** | | | |
| IUGR | | ☐ | ☐ |
| Failure to Thrive | | ☐ | ☐ |
| Gestational Age | ______ wks | | |
| Birth weight | ______ gms | | |
| Birth length | ______ cm | | |
| OFC | ______ cm | | |
| Age at Exam | ______ yrs & mos | | |
| Height | ______ cm | | |
| Weight | ______ kg | | |
| OFC | ______ cm | | |
| **Craniofacial** | | | |
| Microbrachycephaly | | ☐ | ☐ |
| Synophrys/arched eyebrows | | ☐ | ☐ |
| Long, thick eyelashes | | ☐ | ☐ |
| Low-set ears | | ☐ | ☐ |
| Posteriorly rotated ears | | ☐ | ☐ |
| Thickened helices | | ☐ | ☐ |
| Broad nasal bridge | | ☐ | ☐ |
| Upturned/anteverted nares | | ☐ | ☐ |
| Long, smooth philtrum | | ☐ | ☐ |
| Thin upper lip | | ☐ | ☐ |
| Downturned corners of mouth | | ☐ | ☐ |
| High arched palate | | ☐ | ☐ |
| Cleft palate | | ☐ | ☐ |
| Small teeth | | ☐ | ☐ |
| Widely spaced teeth | | ☐ | ☐ |
| Micrognathia | | ☐ | ☐ |
| Bluish tinge around eyes or mouth | | ☐ | ☐ |
| Low-pitched cry | | ☐ | ☐ |
| Short neck | | ☐ | ☐ |
| Low posterior hairline | | ☐ | ☐ |
| **Developmental Delay** | | ☐ | ☐ |
| Age Sat ______ | | | |
| Age Walked ______ | Verbal IQ ______ | | |
| Age Talked ______ | Performance IQ ______ | | |
| **Neurologic** Seizures | | ☐ | ☐ |
| **Limb Abnormalities** | | | |
| Upper extremity deformity | | ☐ | ☐ |
| Describe ______________________ | | | |
| Small hands | | ☐ | ☐ |
| Proximally placed thumbs | | ☐ | ☐ |
| 5th finger clinodactyly | | ☐ | ☐ |
| Limitation of elbow extension | | ☐ | ☐ |
| Lower extremity involvement | | ☐ | ☐ |
| Small feet | | ☐ | ☐ |
| 2-3 toe syndactyly | | ☐ | ☐ |

| Features | Present: | Yes | No |
|---|---|---|---|
| **Gastrointestinal** | | | |
| GER | | ☐ | ☐ |
| Pyloric stenosis | | ☐ | ☐ |
| Intestinal malrotation | | ☐ | ☐ |
| Congenital diaphragmatic hernia | | ☐ | ☐ |
| Other: ______________________ | | | |
| **Otolaryngologic** | | | |
| Sensorineural hearing loss | | ☐ | ☐ |
| **Ophthalmologic** | | | |
| Ptosis | | ☐ | ☐ |
| Strabismus | | ☐ | ☐ |
| Lacrimal duct obstruction | | ☐ | ☐ |
| Myopia | | ☐ | ☐ |
| **Genitourinary** | | | |
| Cryptorchidism | | ☐ | ☐ |
| Hypoplastic genitalia | | ☐ | ☐ |
| Renal abnormalities | | ☐ | ☐ |
| **Cardiovascular** | | ☐ | ☐ |
| If so, what is the defect? ______________ | | | |
| **Dermatologic** | | | |
| Hirsutism | | ☐ | ☐ |
| Cutis marmorata | | ☐ | ☐ |
| Hypoplastic nipples | | ☐ | ☐ |
| Small umbilicus | | ☐ | ☐ |
| Single palmar crease | | ☐ | ☐ |

**Additional findings:**

**Relevant family history:**

**LAB USE ONLY** (Do not write in this box):

Mutation Detected: