**衛生福利部國民健康署「罕見疾病個案通報審查基準機制」(送審資料表)**

**-Cornelia de Lange氏症候群 [Cornelia de Lange syndrome]-**

1. □臨床資料：包含臨床病史(含臨床表徵、家族史) (必要)
2. □基因檢測報告 (必要)

| 項目 | 填寫部分 |
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| 1. **病歷資料 (必要)**   包含病史(含臨床表徵、家族史)、身體檢查 | **就診年齡** [Age at disease onset] \_\_\_\_\_\_\_ 歲  **家族史** [Family history]  □有 □無 |
| **主要特徵（一項2分，須至少二項）**  □聯眉及/或粗眉[Synophrys and/or thick eyebrows]  □短鼻，凹鼻樑及/或鼻尖及鼻孔朝上[Short nose, concave nasal ridge and/or upturned nasal tip]  □長及/或平的人中[Long and/or smooth philtrum]  □薄上唇及/或口角下彎[Thin upper lip vermilion and/or downturned corners of mouth]  □少指及/或無指[Hand oligodactyly and/or adactyly]  □先天性橫膈膜疝氣[Congenital diaphragmatic hernia]  **次要特徵（一項1分）**  □全面發展遲緩及/或智能不足[Global developmental delay and/or intellectual disability]  □產前生長遲滯[Prenatal growth retardation] (<2 SD)  □產後生長遲滯[Postnatal growth retardation] (<2 SD)  □小頭(產前或產後)[Microcephaly (prenatally or postnatally)]  □小手及/或小腳[Small hands and/or feet]  □短小拇指[Short fifth finger]  □多毛症[Hirsutism] |
| **總分：(註:總分至少≥9，且包含至少二項主要特徵)**  □≥11典型  □9-10 非典型 |
| 1. **基因檢測 (必要)**   (請附實驗室報告影本) | □已知致病基因出現致病變異  □*NIPBL*基因  □*SMC1A*基因  □*SMC3*基因  □*RAD21*基因  □*BRD4*基因  □*HDAC8*基因  □*ANKRD11*基因  □其他：\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |

**衛生福利部國民健康署「罕見疾病個案通報審查基準機制」(審查基準表)**

**-Cornelia de Lange氏症候群 [Cornelia de Lange syndrome]-**

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| 應檢附資料   1. □臨床資料：包含臨床病史(含臨床表徵、家族史) (必要) 2. □基因檢測報告 (必要) |

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| **臨床病史(必要)**  □ 就診年齡 (Age at disease onset) \_\_\_\_\_\_\_ 歲  □ 家族史 (Family history) □ 有 □ 無 |

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| **病歷資料** 包含病史(含臨床表徵、發病年齡、家族史)、身體檢查(必要)  **總分≥9，且包含至少一項主要特徵** | |
| **主要特徵（一項2分，須至少二項）**  □Synophrys and/or thick eyebrows  □Short nose, concave nasal ridge and/or upturned nasal tip  □Long and/or smooth philtrum  □Thin upper lip vermilion and/or downturned corners of mouth  □Hand oligodactyly and/or adactyly  □Congenital diaphragmatic hernia | **次要特徵（一項1分）**  □Global developmental delay and/or intellectual disability  □Prenatal growth retardation (<2 SD)  □Postnatal growth retardation (<2 SD)  □Microcephaly (prenatally or postnatally)  □Small hands and/or feet  □Short fifth finger  □Hirsutism |
| **總分：**  □≥11典型  □9-10 非典型 | |

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| **基因檢測報告結果** (必要)  □已知致病基因出現致病變異  □*NIPBL*基因  □*SMC1A*基因  □*SMC3*基因  □*RAD21*基因  □*BRD4*基因  □*HDAC8*基因  □*ANKRD11*基因  □其他：\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |

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| **確診** |

**參考文獻**

1. Kline, A. D., Moss, J. F., Selicorni, A., Bisgaard, A. M., Deardorff, M. A., Gillett, P. M., ... & Hennekam, R. C. (2018). Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. *Nature Reviews Genetics*, *19*(10), 649-666.
2. Selicorni, A., Mariani, M., Lettieri, A., & Massa, V. (2021). Cornelia de Lange Syndrome: From a Disease to a Broader Spectrum. *Genes*, *12*(7), 1075.