**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)
-假性軟骨發育不全症[Pseudoachondroplasia]-**

1. □病歷資料，包含臨床病史、家族病史、臨床症狀及徵兆(必要)
2. □影像檢查報告 (必要)
3. □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)**
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| A1 臨床病史(必要) | □發病年齡 [Age at disease onset] \_\_\_\_\_\_\_ 歲 |
| A2 家族病史(請附上家族中其他病患之詳細病歷記錄或個案病歷資料) (必要) | □家族史 [Family history] □有 □無  |
| A3 臨床表徵(必要) | **臨床表徵，需符合四項必要表徵：**□面部外觀不具典型軟骨發育不全症之表徵(必要)□出生時身長正常(必要)□生長發育缺陷(必要，至少符合以下其中兩項)□四肢短小併身材矮小(<3%) [Short-limbed short stature(< 3rd  percentile)] □短指[Brachydactyly]□關節鬆弛[Joint laxity]□兒童時期關節疼痛[Joint pain since childhood]□脊椎側彎[Scoliosis]□齒突骨發育不全[Odontoid hypoplasia](選擇)□兒童期早發骨性關節炎[Early osteoarthritis during childhood](選擇)□退化性關節疾病[Degenerative joint disease](選擇)□步態蹣跚[Waddling gait](選擇) |
| 1. **影像學檢查(必填)**

 (請附相關報告資料) | □幹骺端發育不良[Epimetaphyseal dysplasia]□長骨縮短[shortened long bones]□短掌骨/指骨[short metacarpals/phalanges] |
| 1. **基因檢測報告(必要)**

(請附實驗室報告) | □ *COMP*基因檢測結果：符合遺傳模式之致病性基因變異 |

參考文獻：

[1] Hasegawa, K., Futagawa, N., Ago, Y., et al. (2023). Novel and recurrent COMP gene variants in five Japanese patients with pseudoachondroplasia: Skeletal changes from the neonatal to infantile periods. Clinical Pediatric Endocrinology, 32(4), 221–227. https://doi.org/10.1297/cpe.2023-0035

[2] Online Mendelian Inheritance in Man (OMIM). PSEUDOACHONDROPLASIA; PSACH, https://www.omim.org/entry/177170; 2024 [accessed 30 March 2024].

[3] Adam MP, Feldman J, Mirzaa GM, et al., editors. Pseudoachondroplasia (GeneReviews). Seattle (WA): University of Washington, Seattle; 1993-2024.

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