**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)  
-面肩胛肱肌失養症[Facioscapulohumeral muscular dystrophy, FSHD]-**

1. □臨床症狀及徵兆的病歷紀錄(必要)
2. □實驗室檢查報告:□CK值(必要); □肌電圖/神經傳導報告(必要);□肌肉病理報告(選擇)
3. □基因檢測報告(必要)

| 項目 | 填寫部分 |
| --- | --- |
| 1. **病歷資料(必要)** |  |
| A1臨床病史(必要) | 發病年齡 [Age at disease onset] \_\_\_\_\_\_\_ 歲  病程描述: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| 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臨床症狀及徵兆 (必要) | 1. □臉部肌肉無力(必要) 2. □肩胛骨翼狀突起(必要) □左=右；□左>右；□左<右 3. □無力或肌肉萎縮(必要)   □上肢近端:□左 □右；□上肢遠端:□左 □右；  □下肢近端:□左 □右；□下肢遠端:□左 □右   1. □視網膜血管病變 2. □聽力減退 3. □脊柱側彎或前彎 |
| 1. **實驗室檢驗報告**   (請附相關檢驗資料) | □CK值(必要)  □肌電圖/神經傳導報告(必要): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  □肌肉病理報告(選擇):\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| 1. **基因檢測報告(必要)**   (請附實驗室報告) | □ FSHD1: contraction of the D4Z4 macrosatellite repeat in chromosome 4q35 □正常 □異常＿＿＿＿＿＿＿＿＿  □ FSHD2: *SMCHD1*基因檢測結果：□正常 □異常＿＿＿＿＿＿  □ FSHD3: *LRIF1*基因檢測結果：□正常 □異常＿＿＿＿＿＿  □ FSHD4: *DMNT3B*基因檢測結果：□正常 □異常＿＿＿＿＿＿  □ \_\_\_\_\_\_\_\_\_\_\_基因檢測結果：□正常 □異常＿＿＿＿＿＿ |

參考文獻：

1. Schatzl, T., Kaiser, L., Deigner, H.-P. faciscapulohumeral **muscular dystrophy: genetics, gene activation and downstream signalling with regard to recent therapeutic approaches: an update.**Orphanet. J. Rare Dis. 16: 129, 2021.
2. Richards, M., Coppee, F., Thomas, N., Belayew, A., Upadhyaya, M. **Facioscapulohumeral muscular dystrophy (FSHD): an enigma unravelled?** Hum. Genet. 131: 325-340, 2012.
3. Facioscapulohumeral muscular dystrophy 1; FSHD1 on OMIM (2021, September 4), https://www.omim.org/entry/158900?search=facioscapulohumeral&highlight=facioscapulohumeral
4. Facioscapulohumeral muscular dystrophy on MDA (2021, September 4), https://www.mda.org/disease/facioscapulohumeral-muscular-dystrophy/signs-and-symptoms

