**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）  
Myhre症候群-[Myhre syndrome]-**

1. □臨床資料 (必要)
2. □影像檢查 (Cardiac echo(必要))
3. □基因檢測報告 (必要)

| 項目 | 填寫部分 |
| --- | --- |
| 1. **病歷資料**   包含病史、身體檢查(必要) | **臉部特徵 (至少符合兩項)**   * 短瞼裂[Short palpebral fissures] * 深陷的眼[Deeply set eyes] * 上頜發育不全[Maxillary underdevelopment] * 人中短[Short philtrum] * 狹窄嘴Narrow mouth] * 上唇紅緣薄[Thin vermilion of the upper lip] * 凸頜[Prognathism] * 唇裂及/或腭裂[Cleft lip and/or palate]   **其他特徵 (至少符合一項)**   * 身材矮小/胎兒生長遲滯[Short stature / IUGR] * 智能不足/學習困難[Intellectual disability / Learning challenges] * 聽力喪失[Hearing loss] * 關節異常[Joint problems] * 呼吸道/呼吸異常[Airway / Respiratory problems] * 心血管異常[Cardiovascular problems] |
| 1. **影像檢查** | □ Cardiac echo(必要)  □ Pulmonary function test |
| 1. **基因檢測 (必要)**   (請附實驗室報告影本) | □ SMAD4基因 |

參考文獻

1. 參考GeneReviews: Starr LJ, Lindor NM, Lin AE. Myhre Syndrome. 2017 Apr 13. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018.Available from: https://www.ncbi.nlm.nih.gov/books/NBK425723/

**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)**

**- Myhre症候群 [Myhre syndrome] -**

|  |
| --- |
| 應檢附資料  □臨床資料 (必要)  □影像檢查  □基因檢測報告 (必要) |

|  |
| --- |
| **臉部特徵 (至少符合兩項)**   * Short palpebral fissures * Deeply set eyes * Maxillary underdevelopment * Short philtrum * Narrow mouth * Thin vermilion of the upper lip * Prognathism * Cleft lip and/or palate   **其他特徵 (至少符合一項)**   * Short stature / IUGR * Intellectual disability / Learning challenges * Hearing loss * Joint problems * Airway / Respiratory problems * Cardiovascular problems |

|  |
| --- |
| 影像檢查  □ Cardiac echo(必要)  □ Pulmonary function test |

|  |
| --- |
| 基因檢測報告結果  □ SMAD4基因 |

找到一個Pathogenic/Likely pathogenic點位

|  |
| --- |
| **Myhre症候群** |

**參考文獻**

1. 參考GeneReviews: Starr LJ, Lindor NM, Lin AE. Myhre Syndrome. 2017 Apr 13. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018.Available from: https://www.ncbi.nlm.nih.gov/books/NBK425723/