**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 |
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| 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]
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| 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] -**

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| 應檢附資料□臨床資料 (必要)□影像檢查 □基因檢測報告 (必要) |

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

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| 基因檢測報告結果□ SMAD4基因 |

找到一個Pathogenic/Likely pathogenic點位

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| **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/