

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]-

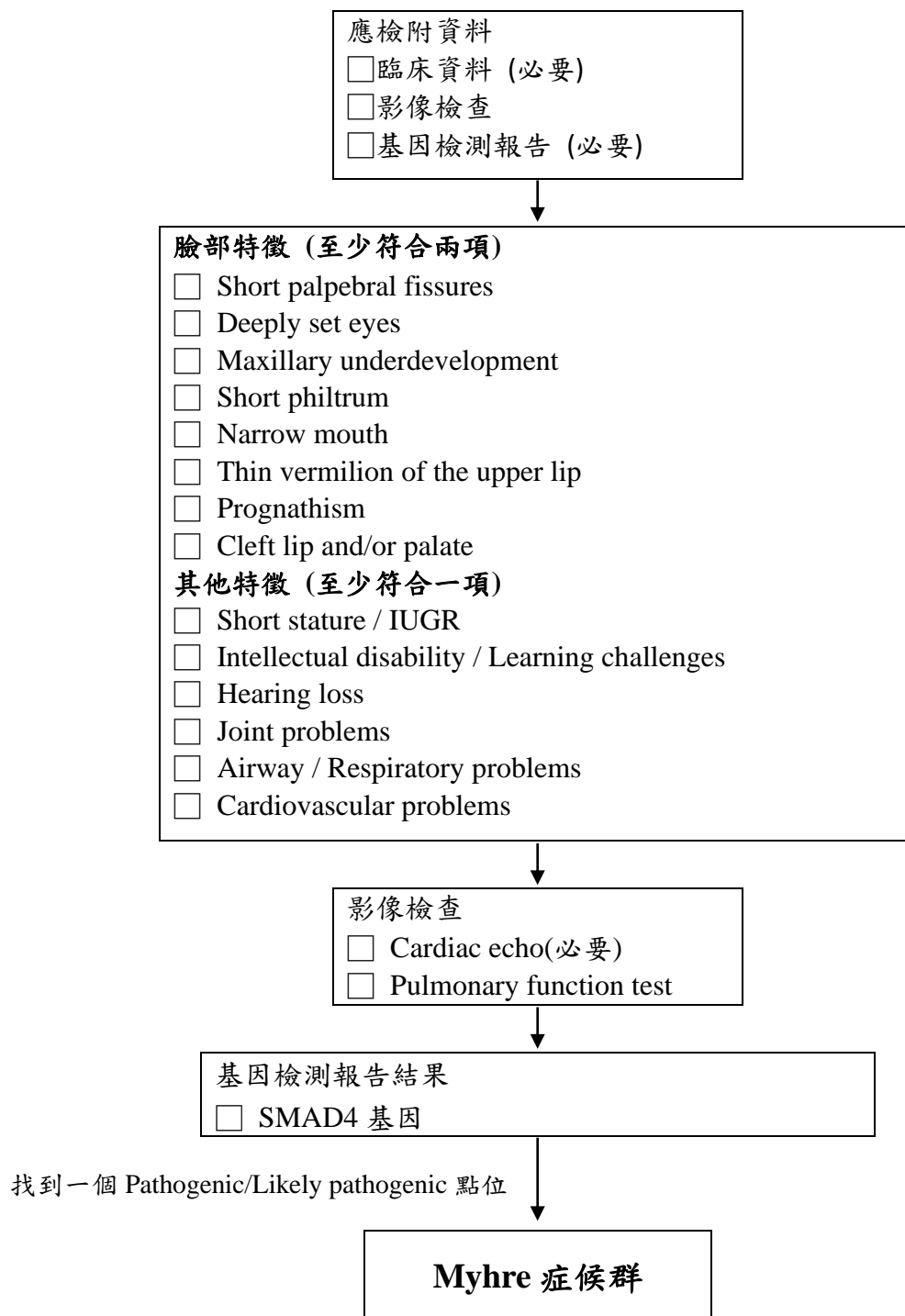
1. 臨床資料 (必要)
2. 影像檢查 (Cardiac echo(必要))
3. 基因檢測報告 (必要)

項目	填寫部分
<p>A. 病歷資料 包含病史、身體檢查(必要)</p>	<p>臉部特徵 (至少符合兩項)</p> <ul style="list-style-type: none"> <input type="checkbox"/> 短瞼裂[Short palpebral fissures] <input type="checkbox"/> 深陷的眼[Deeply set eyes] <input type="checkbox"/> 上頷發育不全[Maxillary underdevelopment] <input type="checkbox"/> 人中短[Short philtrum] <input type="checkbox"/> 狹窄嘴 Narrow mouth] <input type="checkbox"/> 上唇紅緣薄[Thin vermilion of the upper lip] <input type="checkbox"/> 凸頷[Prognathism] <input type="checkbox"/> 唇裂及/或腭裂[Cleft lip and/or palate] <p>其他特徵 (至少符合一項)</p> <ul style="list-style-type: none"> <input type="checkbox"/> 身材矮小/胎兒生長遲滯[Short stature / IUGR] <input type="checkbox"/> 智能不足/學習困難 [Intellectual disability / Learning challenges] <input type="checkbox"/> 聽力喪失[Hearing loss] <input type="checkbox"/> 關節異常[Joint problems] <input type="checkbox"/> 呼吸道/呼吸異常[Airway / Respiratory problems] <input type="checkbox"/> 心血管異常[Cardiovascular problems]
<p>B. 影像檢查</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Cardiac echo(必要) <input type="checkbox"/> Pulmonary function test
<p>C. 基因檢測 (必要) (請附實驗室報告影本)</p>	<ul style="list-style-type: none"> <input type="checkbox"/> 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/>

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