**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)
– CHARGE症候群 [CHARGE syndrome] –**

1. □病歷資料，包含臨床病史、身體診察、神經學檢查、發展里程碑及眼科/耳鼻喉科/代謝科就診紀錄等(必要)

2. □實驗室檢查報告(選擇)

3. □腦部核磁照影及顳骨核磁照影/電腦斷層檢查報告(選擇)

4. □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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| 1. 臨床病史(必要)
 | □ 發病年齡 [Age at disease onset] \_\_\_\_\_\_ 歲□ 家族史 [Family history] □ 有 □ 無 |
| 1. 臨床表徵(必要)
 | **主要臨床表徵 (必填)**，**共四大項**□ 眼部缺損: 從小視網膜缺損到無眼症 [Ocular coloboma,  ranging from small retinal coloboma to anophthalmia]□ 鼻後孔閉鎖或狹窄: 單側或雙側/骨性或膜性 [Choanal atresia or stenosis：unilateral or bilateral, bony or membranous]□ 顱神經功能障礙或異常 [Cranial nerve dysfunction or anomaly] □ 第一對顱神經: 嗅覺減退或嗅覺喪失  [Cranial nerve I: Hyposmia or anosmia] □ 第七對顱神經: 面癱 (單側或雙側) [Cranial nerve VII: Facial palsy (unilateral or bilateral) □ 第八對顱神經: 感音神經性聽力損失及/或平衡問題  [Cranial nerve VIII: Sensorineural hearing loss and/or balance Problems] □ 第九和十對顱神經: 吸吮/吞嚥困難和吸入性嗆傷，腸道蠕 動問題 [Cranial nerve IX/X: Difficulty with sucking/swallowing and aspiration, gut motility problems]□ 耳朵畸形 [Ear malformations] □ 耳廓異常 [Abnormal auricle] □ 聽骨畸形，因混合感音神經和傳導性聽力損失致典型的楔 形聽力圖 [Ossicular malformations resulting in a typical wedge-shaped audiogram due to mixed sensorineural and conductive hearing loss] □ 顳骨異常，經顳骨斷層掃描確定 [Temporal bone abnormalitiesdetermined by temporal bone CT scan]**次要臨床表徵 (必填) ，共五大項**□ 心血管畸形 [Cardiovascular malformation] □ 錐體幹缺損 [Conotruncal defects] □ 房室中隔缺損 [AV canal defects] □ 主動脈弓異常 [Aortic arch anomalies] □ 單獨出現心房中隔缺損、心室中隔缺損、開放性動脈導管 [Isolated ASD, VSD, PDA] □ 顎裂伴有或未伴有唇裂 (腭裂時鼻後孔閉鎖很少見)  [Cleft palate with or without cleft lip (Note: Choanal atresia is rare  in the presence of a cleft palate)]□ 發展遲緩/認知障礙 [Developmental delay / intellectual disability]□ 身材矮小 [Short stature]□ 性腺功能減退症 [Hypogonadotropic hypogonadism] |
| 1. 實驗室檢查報告 (**選擇)**
 | □ 眼科檢查 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ □ 耳鼻喉科檢查 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_□ 視力檢查 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ □ 聽力檢查 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_□ 代謝檢查 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ □ 心臟超音波檢查 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| 1. **影像報告(選擇)**

(請附相關影像資料) | □ 顳骨核磁共振/斷層掃描: 耳蝸 Mondini 缺損(耳蝸發育不全)，半規管缺失或發育不全 [MRI/CT temporal bone: Mondini defect of the cochlea (cochlear hypoplasia), absent or hypoplastic semicircular canals]□ 腦部核磁共振: 斜坡發育不全，小腦蚓部發育不全 [Brain MRI: Clivus hypoplasia, hypoplasia of cerebellar vermis] |
| 1. **基因檢測報告(必要)**

(請附實驗室報告) | □ *CHD7*基因具異型合子致病基因變異(體染色體顯性遺傳)□ *SEMA3E*基因具異型合子致病基因變異(體染色體顯性遺傳) |
| 1. **確定診斷CHARGE症候群**
 | □ 符合3大項主要臨床表徵，且*CHD7*或*SEMA3E*基因具異型合子致病基因變異□ 符合2大項主要臨床表徵加1大項次要臨床表徵，且*CHD7*或*SEMA3E*基因具異型合子致病基因變異□ 符合1大項主要臨床表徵加3大項次要臨床表徵，且*CHD7*或*SEMA3E*基因具異型合子致病基因變異 |

