**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）  
–Schaaf-Yang 症候群 [Schaaf-Yang syndrome] –**

□ 病歷資料(必要)：包含臨床病史、臨床表徵、身體診察、神經學檢查及發展里程碑等

□ 基因檢測報告(必要)

□ 實驗室檢查報告(選擇)

| 項目 | 填寫部分 |
| --- | --- |
| 1. **病歷資料(必要)** |  |
| 1. 臨床病史**(必要)** | * 發病年齡 (Age at disease onset) \_\_\_\_\_\_ 歲 |
| 1. 家族史**(必要)** | * 家族史 (Family history) □ 有 □ 無 |
| 1. **臨床表徵** | * 新生兒期全身肌張力低下 (Generalized muscular hypotonia of infancy) (必要) * 新生兒期遠端關節攣縮 (Distal joint contractures in infancy) (必要) * 新生兒期呼吸窘迫 (Respiratory distress in infancy) (必要) * 發展遲緩/認知障礙(Developmental delay/ intellectual disability) (必要) * 嬰幼兒期餵食困難併滋養不良 (Feeding difficulties with failure to thrive at infantile stage) * 兒童及青少年過度攝食及肥胖 (Hyperphagia and obesity in childhood or adolescence) * 自閉症類群障礙或自閉症 (Autistic spectrum disorder or autistic features) * 臉部畸形特徵 (Dysmorphic facial features)   □ 尖下巴 (Pointed chin)  □ 額頭突出 (Frontal bossing)  □ 低耳位 (Low-set ears)   * 身材矮小 (Short stature) * 內分泌異常 (endocrinopathy)   □ 腦下垂體功能低下 (Hypopituitarism)  □ 生長激素缺乏 (Growth hormone deficiency)  □ 性腺功能低下(Hypogonadism)   * 眼睛異常 (Ocular anomalies) * 抽搐 (Seizures) * 骨骼異常，如脊椎側彎/脊柱後凸 (Skeletal abnormalities, such as scoliosis/ kyphosis) |
| 1. **實驗室檢查(選擇)** | * 賀爾蒙檢查報告 |
| 1. **基因檢測報告(必要)** (請附實驗室報告) | *MAGEL2*基因檢測結果：□正常 □異常\_\_\_\_\_\_ |

參考文獻:

1. Schaaf CP, Marbach F. Schaaf-Yang Syndrome. 2021 Feb 11 [Updated 2021 Nov 4]. In Adam MP, Mirzaa GM, Pagon RA, etal., editors. GeneReviews®[Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023.
2. Negishi Y, Ieda D, Tada H, et al. Schaaf-Yang syndrome shows a Prader-Willi syndrome-like phenotype during infancy. Orphanet J Rare Dis 2019;14:277.
3. Patak J, Gilfert J, Byler M, et al. MAGEL2-related disorders: A study and case series. Clin Genet 2019;96:493-505.
4. Marbach F, Elgizouli M, Rech M, et al. The adult phenotype of Schaaf-Yang syndrome. Orphanet J Rare Dis 2020;15:294.

**衛生福利部國民健康署「罕見疾病個案通報審查標準機制」(審查基準表)**

**–Schaaf-Yang 症候群 [Schaaf-Yang syndrome] –**

**應檢附文件**

□ 病歷資料，包含臨床病史、臨床表徵、身體診察、神經學檢查及發展里程碑等(必要)

□ 基因檢測報告 (必要)

□ 實驗室檢查報告 (選擇)

**臨床病史 (必要)**

□ 發病年齡 (Age at disease onset) \_\_\_\_\_\_ 歲

□ 家族史 (Family history) □ 有 □ 無

**臨床表徵**

□ 新生兒期全身肌張力低下 (Generalized muscular hypotonia of infancy) (必要)

□ 新生兒期遠端關節攣縮 (Distal joint contractures in infancy) (必要)

□ 新生兒期呼吸窘迫 (Respiratory distress in infancy) (必要)

□ 發展遲緩/認知障礙(Developmental delay/ intellectual disability) (必要)

□ 嬰幼兒期餵食困難併滋養不良 (Feeding difficulties with failure to thrive at infantile stage)

□ 兒童及青少年過度攝食及肥胖 (Hyperphagia and obesity in childhood or adolescence)

□ 自閉症類群障礙或自閉症 (Autistic spectrum disorder or autistic features)

□ 臉部畸形特徵 (Dysmorphic facial features)

□ 尖下巴 (Pointed chin)

□ 額頭突出 (Frontal bossing)

□ 低耳位 (Low-set ears)

□ 身材矮小 (Short stature)

□ 內分泌異常 (Endocrinopathy)

□ 腦下垂體功能低下 (Hypopituitarism)

□ 生長激素缺乏 (Growth hormone deficiency)

□ 性腺功能低下(Hypogonadism)

□ 眼睛異常 (Ocular anomalies)

□ 抽搐 (Seizures)

□ 骨骼異常，如脊椎側彎/脊柱後凸 (Skeletal abnormalities, such as scoliosis/ kyphosis)

**參考文獻**

1. Schaaf CP, Marbach F. Schaaf-Yang Syndrome. 2021 Feb 11 [Updated 2021 Nov 4]. In Adam MP, Mirzaa GM, Pagon RA, etal., editors. GeneReviews®[Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023.
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**基因檢測報告 (必要)**

□ 具*MAGEL2*基因致病變異

符合四項必要臨床表徵，且具*MAGEL2*致病基因變異

**符合罕見疾病Schaaf-Yang 症候群通報標準**

**實驗室檢查報告 (選擇)**

□ 賀爾蒙檢查報告