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
A.病歷資料(必要)	
1. 臨床病史**(必要)**	☐發病年齡 (Age at disease onset) _____ 歲
2. 家族史**(必要)**	☐家族史 (Family history) ☐ 有 ☐ 無
3. 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)
B.實驗室檢查(選擇)	☐賀爾蒙檢查報告
C.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）

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臨床病史 (必要)

☐ 發病年齡 (Age at disease onset) _____ 歲
☐ 家族史 (Family history) ☐ 有 ☐ 無

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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)

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實驗室檢查報告 (選擇)

☐ 賀爾蒙檢查報告

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基因檢測報告 (必要)

☐ 具 *MAGEL2* 基因致病變異

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符合四項必要臨床表徵，且具 *MAGEL2* 致病基因變異

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符合罕見疾病 Schaaf-Yang 症候群通報標準

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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4. Marbach F, Elgizouli M, Rech M, et al. The adult phenotype of Schaaf-Yang syndrome. Orphanet J Rare Dis 2020;15:294