**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）**

**–Kabuki症候群[Kabuki syndrome]–**

* + - 1. □病歷資料(包含主要臨床表徵、次要性臨床表徵等)(必要)
			2. □基因檢測報告(必要)
			3. □臉部照片(選擇)

| **項目** | **填寫部分** |
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| 1. **臨床病歷(必要)**
 | 家族史[Family history]□ 有＿＿＿＿ □ 無 |
| 主要臨床表徵 | □ A. 嬰兒期肌張力低下，發展遲緩或智能障礙[Infantile hypotonia, developmental delay and/ or intellectual disability]□ B. 典型的異常表徵[Typical dysmorphic features at some point of life] 1. 長眼瞼並合併下眼瞼外三分之一外翻[Long palpebral fissures with eversion of the lateral third of the lower eyelid]
2. 弧型彎眉，且外三分之一部份稀疏或有缺口[Arched and broad eyebrows with the lateral third displaying notching or sparseness]
3. 鼻小柱短而鼻尖扁平[Short columella with depressed nasal tip]
4. 大而發育異常的耳朵[Large, dysplastic ears]
5. 斜視[Strabismus]
6. 藍鞏膜[Blue sclera]
7. 眼瞼下垂[Ptosis]
8. 小下巴[Micrognathia]
9. 口腔、嘴唇異常：顎裂[Cleft palate]、缺牙及/或門齒異常[Oligodontia and/or abnormal incisors]、嘴唇凹窩[Lip pits]、上唇薄下唇厚[Thin vermillion of upper lip and full lower lip]
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| 次要臨床表徵 | □小頭症[Microcephaly]□先天性心臟病[Congenital heart defect]□四肢異常[Limb/extremity features]：持續性指尖肉墊[Persistent fingertip pads)、短指(趾)畸形[Brachydactyly]、關節鬆弛[Lax joints]、先天性臗關節脫位[Congenital hip dislocation]□泌尿系統異常[Genitourinary anomalies]：腎臟異位[Malpositioned kidneys]、男性病人尿道下裂[Hypospadias in males]□身材矮小[Short stature] □其他[Others]\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| **2. 臉部照片(選擇)****(請附電子檔)** | □有□無 |
| **3. 基因檢測報告** **(必要)**（請附實驗室報告影本） | □ *KMT2D* 基因檢測 □異常 □正常 □ *KDM6A* 基因檢測 □異常 □正常 □其他致病基因變異：\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |

**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）**

**–Kabuki症候群[Kabuki syndrome] –**

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| **應檢附文件**1. □病歷資料(包含主要臨床表徵、次要性臨床表徵等)(必要)
2. □基因檢測報告(必要)
3. □臉部照片(選擇)
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| 家族史[Family history](**必填**)□ 有＿＿＿ □ 無 |

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| **臨床症狀 (A及B項為必要條件，且B項中至少4點符合；C項必須至少3點符合)** □A. 嬰兒期肌張力低下，發展遲緩或智能障礙[Infantile hypotonia, developmental delay and/ or intellectual disability] □B. 典型的臉部異常表徵[Typical facial dysmorphic features (defined below) at some point of life. 1. 長眼瞼並合併下眼瞼外三分之一外翻[Long palpebral fissures with eversion of the lateral third of the lower eyelid]
2. 弧型彎眉，且外三分之一部份稀疏或有缺口 [Arched and broad eyebrows with the lateral third displaying notching or sparseness]
3. 鼻小柱短而鼻尖扁平[Short columella with depressed nasal tip]
4. 大而發育異常的耳朵[Large, dysplastic ears]
5. 斜視[Strabismus]
6. 藍鞏膜[Blue sclera]
7. 眼瞼下垂[Ptosis]
8. 小下巴[Micrognathia]
9. 口腔、嘴唇異常：顎裂[Cleft palate]、缺牙及/或門齒異常[Oligodontia and/or abnormal incisors]、嘴唇凹窩[Lip pits]、上唇薄下唇厚[Thin vermillion of upper lip and full lower lip]

□Ｃ.次要臨床表徵1. 小頭症[Microcephaly]
2. 先天性心臟病[Congenital heart defect]
3. 四肢異常[Limb/extremity features]：持續性指尖肉墊[Persistent fingertip pads]、短指(趾)畸形[Brachydactyly]、關節鬆弛[Lax joints]、先天性臗關節脫位[Congenital hip dislocation]
4. 泌尿系統異常[Genitourinary anomalies]：腎臟異位[Malpositioned kidneys]、男性病人尿道下裂[Hypospadias in males]
5. 身材矮小[Short stature]
6. 其他[Others]\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
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| **基因檢測報告(必要條件)**□ *KMT2D*基因檢測異常、或□ *KDM6A*基因檢測異常、或□其他致病基因變異： |

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| □符合臨床必要表徵及具致病性基因變異之**Kabuki症候群[Kabuki syndrome]** |