**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)**

* **克片-魯賓斯基症候群[Keppen-Lubinsky syndrome] -**

1. □病歷資料 (包括家族史、生長曲線等) (必要)
2. □智力測驗或發展評估 (必要)
3. □實驗室檢查 (選擇)
4. □影像檢查 (選擇)
5. □基因檢測報告 (必要)

| 項目 | 填寫部分 |
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| 1. **病歷資料(必)** |  |
| 1. 主要病史及家族史 |  |
| 1. 臨床表徵→主要(必要) | 主要條件須至少二項  □全身性脂肪失養症(Generalized lipodystrophy)  □類早老症外觀(Progeroid features)  □嚴重智能不足(Severe intellectual disability) |
| 1. 臨床表徵→次要(必要) | 次要條件 (至少一項)  □神經學表現  □肌張力增強  □張力不全(Dystonia)  □深部肌腱反射增強  □痙攣性四肢麻痺(Spastic tetraparesis)  □抽搐或癲癇  □揮舞肢動症(Ballimus)  □自殘(Self-mutilation)  □骨骼表現  □關節攣縮(Joint contractures)  □脊柱側變(Scoliosis)  □生長遲緩(Growth retardation) |
| 1. **智力測驗或發展評估(必要)** | 智力測驗 □正常 □異常  或發展評估 □正常 □異常 |
| 1. **實驗室檢查 (選擇)** | □白血球CBC/DC  □ IGF-1  □其他 |
| 1. **影像檢查報告 (選擇)** | □脊柱X-ray (Spine X-ray) □正常 □異常  □腦部核磁共振(Brain MRI) □正常 □異常 |
| 1. **基因檢測 (必要)**   (請附實驗室報告影本) | □ *KCNJ6*基因致病性變異 |

**參考文獻**

1. Neuroscience . 2018 August 01; 384: 152–164. doi:10.1016/j.neuroscience.2018.05.031. **Gain-of-function KCNJ6 mutation in a severe hyperkinetic movement disorder phenotype** Gabriella A. Horvath, MD, PhD, Yulin Zhao, PhD, Maja Tarailo-Graovac, PhD, Cyrus Boelman, MD, Harinder Gill, MD, Casper Shyr, PhD, James Lee, MD, Ingrid BlydtHansen, Britt I. Drögemöller, PhD, Jacqueline Moreland, Colin J. Ross, PhD, Wyeth W. Wasserman, PhD, Andrea Masotti, PhD, Paul A. Slesinger, PhD, Clara D.M. van Karnebeek, MD, PhD
2. Am J Med Genet Part A 149A:1827–1829 2009. **Keppen–Lubinsky syndrome: Expanding the phenotype.** Basel-Vanagaite L, Shaffer L, Chitayat D.
3. Am J Hum Genet. 2015 Feb 5;96(2):295-300. doi: 10.1016/j.ajhg.2014.12.011. **Keppen-Lubinsky syndrome is caused by mutations in the inwardly rectifying K+ channel encoded by KCNJ6.** Masotti A1, Uva P2, Davis-Keppen L3, Basel-Vanagaite L4, Cohen L5, Pisaneschi E6, Celluzzi A6, Bencivenga P6, Fang M7, Tian M8, Xu X9, Cappa M6, Dallapiccola B6.

**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)**

**- 克片-魯賓斯基症候群 [Keppen-Lubinsky syndrome] -**

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| **應檢附資料**  1.□病歷資料(包括家族史、生長曲線等) (必要)  2.□智力測驗或發展評估 (必要)  3.□實驗室檢查 (選擇)  4.□影像檢查 (選擇)  5.□基因檢測報告 (必要) |

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| **智力測驗或發展評估(必要)**  智力測驗□正常 □異常 或 發展評估□正常 □異常 |

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| **臨床表徵(必要)**  1.主要條件須至少二項  □全身性脂肪失養症(Generalized lipodystrophy)  □類早老症外觀(Progeroid features)  □嚴重智能不足(Severe intellectual disability)  2.次要條件 (至少一項)  □神經學表現  □肌張力增強  □張力不全(Dystonia)  □深部肌腱反射增強  □痙攣性四肢麻痺(Spastic tetraparesis)  □抽搐或癲癇  □揮舞肢動症(Ballimus)  □自殘(Self-mutilation)  □骨骼表現  □關節攣縮(Joint contractures)  □脊柱側變(Scoliosis)  □生長遲緩(Growth retardation) |

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| **實驗室檢查 (選擇)**  □白血球CBC/DC □ IGF-1 □其他 |

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| **影像檢查報告 (選擇)**  □脊柱X-ray (Spine X-ray) □正常 □異常  □腦部核磁共振(Brain MRI) □正常 □異常 |

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| **基因檢測 (必要)**(請附實驗室報告影本)  □ *KCNJ6*基因致病性變異 |

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| **確定診斷**  □克片-魯賓斯基症候群(Keppen-Lubinsky syndrome) |

**參考文獻**

1.Neuroscience . 2018 August 01; 384: 152–164. doi:10.1016/j.neuroscience.2018.05.031. **Gain-of-function KCNJ6 mutation in a severe hyperkinetic movement disorder phenotype** Gabriella A. Horvath, MD, PhD, Yulin Zhao, PhD, Maja Tarailo-Graovac, PhD, Cyrus Boelman, MD, Harinder Gill, MD, Casper Shyr, PhD, James Lee, MD, Ingrid BlydtHansen, Britt I. Drögemöller, PhD, Jacqueline Moreland, Colin J. Ross, PhD, Wyeth W. Wasserman, PhD, Andrea Masotti, PhD, Paul A. Slesinger, PhD, Clara D.M. van Karnebeek, MD, PhD

2.Am J Med Genet Part A 149A:1827–1829 2009. **Keppen–Lubinsky syndrome: Expanding the phenotype.** Basel-Vanagaite L, Shaffer L, Chitayat D.

3.Am J Hum Genet. 2015 Feb 5;96(2):295-300. doi: 10.1016/j.ajhg.2014.12.011. **Keppen-Lubinsky syndrome is caused by mutations in the inwardly rectifying K+ channel encoded by KCNJ6.** Masotti A1, Uva P2, Davis-Keppen L3, Basel-Vanagaite L4, Cohen L5, Pisaneschi E6, Celluzzi A6, Bencivenga P6, Fang M7, Tian M8, Xu X9, Cappa M6, Dallapiccola B6.