**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）  
MIRAGE症候群-[** **MIRAGE syndrome]-**

1. □ 臨床資料 (必要)
2. □ 常規檢驗 (非必要)
3. □ 影像學檢查 (非必要)
4. □ 基因檢測報告 (必要)

| 項目 | 填寫部分 |
| --- | --- |
| 1. **病歷資料**   包含病史、身體檢查(必要) | **須符合四項主要特徵**  **或符合三項主要特徵及兩項次要特徵**  **主要特徵**   * Hematologic problems * Recurrent severe infection * Restrictive growth condition * Adrenal insufficiency * Genital abnormalities * Enteropathy   **次要特徵**   * Thymic hypoplasia or aplasia * Developmental delay * Bone and joint problems * Esophageal achalasia |
| 1. **常規檢驗** | * Hemogram * Survey for adrenal gland insufficiency * Microbiological culture * Bone marrow |
| 1. **影像學檢查** | * Abdominal or renal sonography / abdominal CT/MRI * Chest X-ray / CT / MRI * Bone survey * UGI series |
| 1. **基因檢測 (必要)**   (請附實驗室報告影本) | * SAMD9 gene mutation |

**參考文獻**

* Syndromes: Rapid Recognition and Perioperative Implications, 2e  
   <https://accesspediatrics.mhmedical.com/content.aspx?bookid=2674&sectionid=220538595>
* Orphanet: <https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=494433>
* OMIM(# 617053): <https://www.omim.org/entry/617053>
* NIH Rare Diseases: <https://rarediseases.info.nih.gov/diseases/13108/mirage-syndrome>
* MalaCards: <https://www.malacards.org/card/mirage_syndrome>
* Onuma et al. Human Genome Variation (2020) 7:4

**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）  
MIRAGE症候群-[** **MIRAGE syndrome]-**

|  |
| --- |
| 須符合四項主要特徵  或符合三項主要特徵及兩項次要特徵  **主要特徵**   * □ Hematologic problems * □ Recurrent severe infection * □ Restrictive growth condition * □ Adrenal insufficiency * □ Genital abnormalities * □ Enteropathy   **次要特徵**   * □ Thymic hypoplasia or aplasia * □ Developmental delay * □ Bone and joint problems   □ Esophageal achalasia |

|  |
| --- |
| **常規檢驗**   * □ Hemogram * □ Survey for adrenal gland insufficiency * □ Microbiological culture   □ Bone marrow |

|  |
| --- |
| **影像學檢查**   * □ Abdominal or renal sonography / abdominal CT/MRI * □ Chest X-ray / CT / MRI * □ Bone survey   □ UGI series |

|  |
| --- |
| **基因檢測報告結果**  □ SAMD9 |

**參考文獻**

1. Syndromes: Rapid Recognition and Perioperative Implications, 2e

<https://accesspediatrics.mhmedical.com/content.aspx?bookid=2674&sectionid=220538595>

1. Orphanet: <https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=494433>
2. OMIM(# 617053): <https://www.omim.org/entry/617053>
3. NIH Rare Diseases: <https://rarediseases.info.nih.gov/diseases/13108/mirage-syndrome>
4. MalaCards: <https://www.malacards.org/card/mirage_syndrome>
5. Onuma et al. Human Genome Variation (2020) 7:4
6. Narumi, Satoshi et al. “SAMD9 mutations cause a novel multisystem disorder, MIRAGE syndrome, and are associated with loss of chromosome 7.” Nature genetics vol. 48,7 (2016): 792-7. doi:10.1038/ng.3569
7. Buonocore, Federica et al. “Somatic mutations and progressive monosomy modify SAMD9-related phenotypes in humans.” *The Journal of clinical investigation* vol. 127,5 (2017): 1700-1713. doi:10.1172/JCI91913