**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 |
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| 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]-**

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| 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
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**次要特徵*** □ Thymic hypoplasia or aplasia
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* □ Chest X-ray / CT / MRI
* □ Bone survey

□ UGI series |

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| **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