# 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 |
|---|---|
| **A. 病歷資料**<br>包含病史、身體檢查(必要) | **須符合四項主要特徵**<br>**或符合三項主要特徵及兩項次要特徵**<br><br>**主要特徵**<br>☐ Hematologic problems<br>☐ Recurrent severe infection<br>☐ Restrictive growth condition<br>☐ Adrenal insufficiency<br>☐ Genital abnormalities<br>☐ Enteropathy<br><br>**次要特徵**<br>☐ Thymic hypoplasia or aplasia<br>☐ Developmental delay<br>☐ Bone and joint problems<br>☐ Esophageal achalasia |
| **B. 常規檢驗** | ☐ Hemogram<br>☐ Survey for adrenal gland insufficiency<br>☐ Microbiological culture<br>☐ Bone marrow |
| **C. 影像學檢查** | ☐ Abdominal or renal sonography / abdominal CT/MRI<br>☐ Chest X-ray / CT / MRI<br>☐ Bone survey<br>☐ UGI series |
| **D. 基因檢測 (必要)**<br>(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

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**常規檢驗**

- ☐ Hemogram
- ☐ Survey for adrenal gland insufficiency
- ☐ Microbiological culture
- ☐ Bone marrow

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**影像學檢查**

- ☐ Abdominal or renal sonography / abdominal CT/MRI
- ☐ 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
2. Orphanet: https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=494433
3. OMIM(# 617053): https://www.omim.org/entry/617053
4. NIH Rare Diseases: https://rarediseases.info.nih.gov/diseases/13108/mirage-syndrome
5. MalaCards: https://www.malacards.org/card/mirage_syndrome
6. Onuma et al. Human Genome Variation (2020) 7:4
7. 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
8. 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