衛生福利部國民健康署「罕見疾病個案通報審查標準機制」(送審資料表)
-多發性硬化症/泛視神經脊髓炎 [MS/NMOSD]

-表 1.多發性硬化症[Multiple sclerosis, MS]

應檢附文件(必要)

□病歷資料 (包括臨床表徵、發病年齡、家族史、發病次數、病程發展過程、神經學身體診察等)

□相關科會診病歷紀錄(必要)：

□眼科會診_____ □免疫科會診_____ □血液腫瘤科會診_____

□影像學報告(包括腦及脊髓等)

□實驗室檢驗(包括排除疾病相關的檢驗等)

□符合 DIT 及 DIS 的準據

項目	填寫部分
A.病歷資料	
主要病史(必要)	□病歷資料 (包括臨床表徵、發病年齡、家族史、病程發展過程、神經學身體診察等) □發病次數一次(具臨床客觀徵兆)：□是 □否 □發病次數二次(含)以上(具臨床客觀徵兆)：□是 □否 □相關科會診病歷紀錄(必要)： □眼科會診_____ □免疫科會診_____ □血液腫瘤科會診_____ □排除腦中風/腦梗塞 □排除 Sarcoidosis □排除中樞神經系統 Lymphoma □排除 Paraneoplastic Syndrome □排除系統性身體免疫等疾病侵入中樞神經系統之疾病 □排除感染性腦脊髓炎 □排除其他原因不明發炎去髓鞘疾病(Other idiopathic demyelinating disorders (IIDD)) □排除神經代謝疾病
主要表徵(必要)	□視力減退或喪失 □動眼功能異常 □顱神經麻痺_____ □肢體無力_____ □兩側肢體麻痺或癱瘓 □膀胱功能異常 □脊髓病灶以下的體感覺消失 □腦幹病灶側顱神經麻痺伴有對側肢體半側無力或半側麻痺或半側感覺喪失 □症狀性大腦症候群(客觀徵兆)_____ □其他客觀徵兆_____

<table>
<tr><td>項目</td><td>填寫部分</td></tr>
<tr><td>實驗室檢驗報告(選擇)</td><td>□血液檢查(WBC/DC, Hgb 等) □ESR:_____ □CRP:_____
□VDRL: □正常 □異常_____
□ANA:□正常 □異常_____
□C3/C4: □正常 □異常_____
□脊髓液病毒或細菌培養：□正常 □異常_____
□其他_____</td></tr>
<tr><td>脊髓檢驗報告(必要)</td><td>□正常
□異常 _____</td></tr>
<tr><td>影像學檢查報告(必要)</td><td>1. 胸部 X 光：□正常 □異常_____
2. 腹部影像學：□正常 □異常_____
3. 脊髓 MRI：□正常 □異常_____
4. 腦部及視神經 MRI：□正常 □異常_____</td></tr>
<tr><td>符合 DIT 及 DIS 準據</td><td>
<table>
<tr><td></td><td>臨床表現
Clinical Presentation</td><td>MS 診斷所需之附加資料
Additional Data Needed for MS Diagnosis</td></tr>
<tr><td>□</td><td>≧2 attacks；objective clinical evidence of ≧2 lesion or objective clinical evidence of 1 lesion with reasonable historical evidence of a prior attack</td><td>None</td></tr>
<tr><td>□</td><td>≧2 attacks；objective clinical evidence of 1 lesion</td><td>Dissemination in space, demonstrated by: ≧1 T2 lesion in at least 2 to 4 MS-typical regions of the CNS (periventricular, juxtacortical, infratentorial, or spinal cord)；or Await a further clinical attack implicating a different CNS site</td></tr>
<tr><td>□</td><td>1 attack；objective clinical evidence of ≧ 2 lesions</td><td>Dissemination in time, demonstrated by: Simultaneous presence of asymptomatic gadolinium-enhancing and nonenhancing lesions at any time；or A new T2 and/or gadolinium-enhancing lesion(s) on follow-up MRI, irrespective of its timing with reference to a baseline scan；or Await a second clinical attack</td></tr>
<tr><td>□</td><td>1 attack；objective clinical evidence of 1 lesion
(clinically isolated syndrome)</td><td>Dissemination in space and time, demonstrated by: For DIS:≧1 T2 lesion in at least 2 of 4 MS-typical regions of the CNS (periventricular, juxtacortical, infratentorial, or spinal cord)；or Await a second clinical attack implicating a different CNS site；and For DIT: Simultaneous presence of asymptomatic gadolinium-enhancing and nonenhancing lesions at any time; or A new T2 and/or gadolinium-enhancing lesion(s) on follow-up MRI, irrespective of its timing with reference to a baseline scan；or Await a second clinical attack</td></tr>
<tr><td>□</td><td>Insidious neurological progression suggestive of MS (PPMS)</td><td>1 year of disease progression (retrospectively or prospectively determined) plus 2 of 3 of the following criteria:
1. Evidence for DIS in the brain based on ≧1 T2 lesions in the MS-characteristic (periventricular, juxtacortical, or infratentorial) regions
2. Evidence for DIS in the spinal cord based on ≧2 T2 lesions in the cord
3. Positive CSF (isoelectric focusing evidence of oligoclonal bands and/or elevated IgG index)</td></tr>
</table>
</td></tr>
</table>

項目	填寫部分
確診為多發性硬化症 [Multiple sclerosis, MS/NMOSD]	□符合 DIT 及 DIS 準據

參考文獻:
1. Polman CH, Reingold SC, Banwell B, et al. (2011). Diagnostic criteria for multiple sclerosis: 2010 revisions to the McDonald criteria. Ann Neurol, 69(2), 292-302.
2. Swanton JK, Rovira A, Tintore'M, et al. (2007). MRI criteria for multiple sclerosis in patients presenting with clinically isolated syndromes: a multicentre retrospective study. Lancet Neurol, 6, 677-686.
3. Swanton JK, Fernando K, Dalton CM, et al. (2006).Modification of MRI criteria for multiple sclerosis in patients with clinically isolated syndromes. J Neurol Neurosurg Psychiatry, 77, 830-833.
4. Montalban X, Tintore'M, Swanton J, et al. (2010). MRI criteria for MS in patients with clinically isolated syndromes. Neurology, 74, 427-434.
5. Thompson A.J. et al.Diagnosis of multiple sclerosis: 2017 revisions of the McDonald criteria. Lancet Neurol. 2018 Feb;17(2):162-173.
6. Ömerhoca S. et al. Multiple Sclerosis: Diagnosis and Differential Diagnosis. Arch Neuropsychiatry 2018;55: (Suppl 1):S1-S9.

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-多發性硬化症/泛視神經脊髓炎 [MS/NMOSD]

-表 2.泛視神經脊髓炎 [Neuromyelitis optica spectrum disorders, NMOSD]

應檢附文件(必要)

□病歷資料 (包括臨床表徵、發病年齡、家族史、發病次數、病程發展過程、神經學身體診察等)

□相關科會診病歷紀錄(必要)：

□眼科會診_____ □免疫科會診_____ □血液腫瘤科會診_____

□影像學報告(包括腦及脊髓等)

□實驗室檢驗(包括排除疾病相關的檢驗等)

□AQP4 抗體檢驗報告

項目	填寫部分
A.病歷資料	
1. 主要病史(必要)	□病歷資料 (包括臨床表徵、發病年齡、家族史、病程發展過程、神經學身體診察等) □發病次數二次(含)以上：□是 □否 □相關科會診病歷紀錄(必要)： □眼科會診_____ □免疫科會診_____ □血液腫瘤科會診_____ □排除 Sarcoidosis □排除中樞神經系統 Lymphoma □排除 Paraneoplastic Syndrome □排除系統性身體免疫等疾病侵入中樞神經系統之疾病 □排除感染性腦脊髓炎 □排除瀰漫性腦脊髓炎(ADEM)及特發性橫截式脊髓炎(Idiopathic Transverse Myelitis)
2. 主要表徵(必要)	1. □視神經炎(Optic Neuritis) □視力減退或喪失 □其他客觀徵兆_____ 2. □急性脊髓炎(Autle Myelitis) □兩側肢體麻痺或癱瘓 □膀胱功能異常 □脊髓病灶以下的體感覺消失 □其他客觀徵兆_____ 3. □急性菱形窩最後區症候群(Area Postrema Syndrome) □無理由的打嗝或噁心及嘔吐，並持續 24 小時以上 □其他客觀徵兆_____ 4. □急性腦幹症候群(Acute Brainstem Syndrome) □Benedikt syndrome : _____ □Claude syndrome : _____ □Nothnagel syndrome :_____ □Weber syndrome : _____ □Wernekink commissure syndrome : _____

項目	填寫部分
	□腦幹病灶側顱神經麻痺伴有對側肢體半側無力或半側麻痺或半側感覺喪失 □其他客觀徵兆_____ 5. □症狀性猝睡症(Symptomatic Narcolepsy)或急性間腦症後群伴有典型視神經脊髓炎譜系疾病腦部 MRI 影像學間腦病灶(Acute diencephalic clinical syndrome with NMOSD-typical diencephalic MRI lesions) □猝睡症(需附睡眠多項生理檢查,Polysomnography) □急性間腦症之臨床表現(客觀徵兆)_____。 6. □症狀性大腦症候群伴有典型視神經脊髓炎譜系疾病之腦部影像學病灶(Symptomatic cerebral syndrome with NMOSD – typical brain lesion) □症狀性大腦症候群(客觀徵兆)_____
實驗室檢驗報告(選擇)	□血液檢查(WBC/DC, Hgb 等) □ESR: □CRP: □VDRL: □正常 □異常 □ANA:□正常 □異常 □C3/C4: □正常 □異常 □脊髓液病毒或細菌培養：□正常 □異常 □其他
脊髓檢驗報告(必要)	□正常 □異常 _____
影像學檢查報告(必要)	1. 胸部 X 光：□正常 □異常_____ 2. 腹部影像學：□正常 □異常_____ 3. 脊髓 MRI：□正常 □異常_____ 4. 腦部及視神經 MRI：□正常 □異常_____
水通道蛋白 4 自體抗體(AQP4Ab)檢測	□正常 □異常 _____
確診為泛視神經脊髓炎[Neuromyelitis optica spectrum disorders, NMOSD]	□水通道蛋白 4 自體抗體異常，符合下列 1 及 2 1. 具六項主要表徵之一項 2. 排除其他鑑別相關之疾病 □水通道蛋白 4 自體抗體正常，符合下列 1+2+3+4 1. 具主要表徵中第一及二項並伴有第三項或脊髓炎超過(或含)三節連續性病灶(MRI) 2. 具神經影像學(MRI)有 Dissemination in space(DIS)特徵，並對應二項(含)以上之主要表徵 3. 具典型主要表徵相關 MRI 影像病灶*(註解) 4. 排除其他鑑別相關疾病

*註解

Acute optic neuritis : normal findings or only nonspecific white matter lesions, or optic nerve: T2-hyperintense lesion or T1- gadolinium enhancing lesion extending >1/2 optic nerve length or involving optic chiasm ;
Acute myelitis : intramedullary MRI lesion ≥3 contiguous segments (LETM) or ≥3 contiguous segments of focal spinal cord atrophy in patients with history compatible with acute myelitis ;
Area postrema syndrome: dorsal medulla/area postrema lesions ;
Acute brainstem syndrome: periependymal brainstem lesions.

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-多發性硬化症/泛視神經脊髓炎 [MS/NMOSD] -

-表1. 多發性硬化症[Multiple sclerosis, MS]

應檢附文件(必要)
□病歷資料 (包括臨床表徵、發病年齡、家族史、發病次數、病程發展過程、神經學身體診察等)
□相關科會診病歷紀錄(必要):
□眼科會診_____ □免疫科會診_____ □血液腫瘤科會診_____。
□影像學報告(包括腦及脊髓等)
□實驗室檢驗(包括排除疾病相關的檢驗等)
□AQP4抗體檢驗報告
□符合DIT及DIS的準據

↓

主要病史(必要)
□病歷資料 (包括臨床表徵、發病年齡、家族史、病程發展過程、神經學身體診察等)
□發病次數一次(具臨床客觀徵兆):□是 □否
□發病次數二次(含)以上(具臨床客觀徵兆):□是 □否
□相關科會診病歷紀錄(必要):
□眼科會診_____ □免疫科會診_____ □血液腫瘤科會診_____。
□排除腦中風/腦梗塞
□排除Sarcoidosis
□排除中樞神經系統Lymphoma
□排除Paraneoplastic Syndrome
□排除系統性身體免疫等疾病侵入中樞神經系統之疾病
□排除感染性腦脊髓炎
□排除其他原因不明發炎去髓鞘疾病[others, idiopathic (例如 Idiopathic demyelinating disorders (IIDD)]
□排除神經代謝疾病(選擇)

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主要表徵
□視力減退或喪失
□動眼功能異常
□顱神經麻痺
□肢體無力
□兩側肢體麻痺或癱瘓
□膀胱功能異常
□脊髓病灶以下的體感覺消失
□腦幹病灶側顱神經麻痺伴有對側肢體半側無力或半側麻痺或半側感覺喪失
□症狀性大腦症候群(客觀徵兆)
□其他客觀徵兆_____。

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實驗室檢驗報告(選擇)
□血液檢查(WBC/DC, Hgb 等)□ESR:_____ □CRP:_____□VDRL:□正常 □異常____。
□ANA:□正常 □異常____ □C3/C4:□正常□異常
□脊髓液病毒或細菌培養:□正常 □異常_____ □其他_____。

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脊髓檢驗報告(必要)
□正常　□異常

↓

影像學檢查報告(必要)
1. 胸部X光：□正常 □異常
2. 腹部影像學：□正常 □異常
3. 脊髓MRI：□正常 □異常
4. 腦部及視神經MRI：□正常 □異常

↓

	臨床表現 Clinical Presentation	MS診斷所需之附加資料 Additional Data Needed for MS Diagnosis
□	≧2 attacks；objective clinical evidence of ≧2 lesion or objective clinical evidence of 1 lesion with reasonable historical evidence of a prior attack	None
□	≧2 attacks；objective clinical evidence of 1 lesion	Dissemination in space, demonstrated by: ≧1 T2 lesion in at least 2 to 4 MS-typical regions of the CNS (periventricular, juxtacortical, infratentorial, or spinal cord)；or Await a further clinical attack implicating a different CNS site
□	1 attack；objective clinical evidence of ≧2 lesions	Dissemination in time, demonstrated by: Simultaneous presence of asymptomatic gadolinium-enhancing and nonenhancing lesions at any time；or A new T2 and/or gadolinium-enhancing lesion(s) on follow-up MRI, irrespective of its timing with reference to a baseline scan；or Await a second clinical attack
□	1 attack；objective clinical evidence of 1 lesion (clinically isolated syndrome)	Dissemination in space and time, demonstrated by: For DIS:≧1 T2 lesion in at least 2 of 4 MS-typical regions of the CNS (periventricular, juxtacortical, infratentorial, or spinal cord)；or Await a second clinical attack implicating a different CNS site；and For DIT: Simultaneous presence of asymptomatic gadolinium-enhancing and nonenhancing lesions at any time; or A new T2 and/or gadolinium-enhancing lesion(s) on follow-up MRI, irrespective of its timing with reference to a baseline scan；or Await a second clinical attack
□	Insidious neurological progression suggestive of MS (PPMS)	1 year of disease progression (retrospectively or prospectively determined) plus 2 of 3 of the following criteria: 1. Evidence for DIS in the brain based on ≧1 T2 lesions in the MS-characteristic (periventricular, juxtacortical, or infratentorial) regions 2. Evidence for DIS in the spinal cord based on ≧2 T2 lesions in the cord 3. Positive CSF (isoelectric focusing evidence of oligoclonal bands and/or elevated IgG index)

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（參考文獻1）

符合DIS及DIT準據，確診為多發性硬化症
[Multiple sclerosis, MS]

參考文獻:

1. Polman CH, Reingold SC, Banwell B, et al. (2011). Diagnostic criteria for multiple sclerosis: 2010 revisions to the McDonald criteria. Ann Neurol, 69(2), 292-302.
2. Swanton JK, Rovira A, Tintore'M, et al. (2007). MRI criteria for multiple sclerosis in patients presenting with clinically isolated syndromes: a multicentre retrospective study. Lancet Neurol, 6, 677-686.
3. Swanton JK, Fernando K, Dalton CM, et al. (2006).Modification of MRI criteria for multiple sclerosis in patients with clinically isolated syndromes. J Neurol Neurosurg Psychiatry, 77, 830-833.
4. Montalban X, Tintore'M, Swanton J, et al. (2010). MRI criteria for MS in patients with clinically isolated syndromes. Neurology, 74, 427-434.
5. Thompson A.J. et al.Diagnosis of multiple sclerosis: 2017 revisions of the McDonald criteria. Lancet Neurol. 2018 Feb;17(2):162-173.
6. Ömerhoca S. et al. Multiple Sclerosis: Diagnosis and Differential Diagnosis. Arch Neuropsychiatry 2018;55: (Suppl 1):S1-S9.

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-表2.泛視神經脊髓炎 [Neuromyelitis optica spectrum disorders, NMOSD]

應檢附文件(必要)
☐病歷資料(包括臨床表徵、發病年齡、家族史、發病次數、病程發展過程、神經學身體診察等)
☐相關科會診病歷紀錄(必要)：
☐眼科會診_____ ☐免疫科會診_____ ☐血液腫瘤科會診_____。
☐影像學報告(包括腦及脊髓等)
☐實驗室檢驗(包括排除疾病相關的檢驗等)
☐AQP4抗體檢驗報告

↓

☐排除Sarcoidosis ☐排除中樞神經系統Lymphoma
☐排除Paraneoplastic Syndrome ☐排除系統性身體免疫等疾病侵入中樞神經系統之疾病
☐排除感染性腦脊髓炎
☐排除瀰漫性腦脊髓炎(ADEM)及特發性橫截式脊髓炎(Idiopathic Transverse Myelitis)

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主要表徵
1. ☐視神經炎(Optic Neuritis)
   ☐視力減退或喪失 ☐其他客觀徵兆_____。
2. ☐急性脊髓炎(Autle Myelitis)
   ☐兩側肢體麻痺或癱瘓 ☐膀胱功能異常
   ☐脊髓病灶以下的體感覺消失 ☐其他客觀徵兆_____。
3. ☐急性菱形窩最後區症候群(Area Postrema Syndrome)
   ☐無理由的打嗝或噁心及嘔吐，並持續24小時以上 ☐其他客觀徵兆_____。
4. ☐急性腦幹症候群(Acute Brainstem Syndrome)
   ☐Benedikt syndrome :
   ☐Claude syndrome :
   ☐Nothnagel syndrome :
   ☐Weber syndrome :
   ☐Wernekink commissure syndrome :
   ☐腦幹病灶側顱神經麻痺伴有對側肢體半側無力或半側麻痺或半側感覺喪失
   ☐其他客觀徵兆_____。
5. ☐症狀性猝睡症(Symptomatic Narcolepsy)或急性間腦症後群伴有典型視神經脊髓炎譜系疾病腦部MRI影像學間腦病灶(Acute diencephalic clinical syndrome with NMOSD-typical diencephalic MRI lesions)
   ☐猝睡症(需附睡眠多項生理檢查,Polysomnography)
   ☐急性間腦症之臨床表現(客觀徵兆)_____。
6. ☐症狀性大腦症候群伴有典型視神經脊髓炎譜系疾病之腦部影像學病灶(Symptomatic cerebral syndrome with NMOSD – typical brain lesion)
   ☐症狀性大腦症候群(客觀徵兆)_____。

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☐具核心表徵之發病兩次(含)以上

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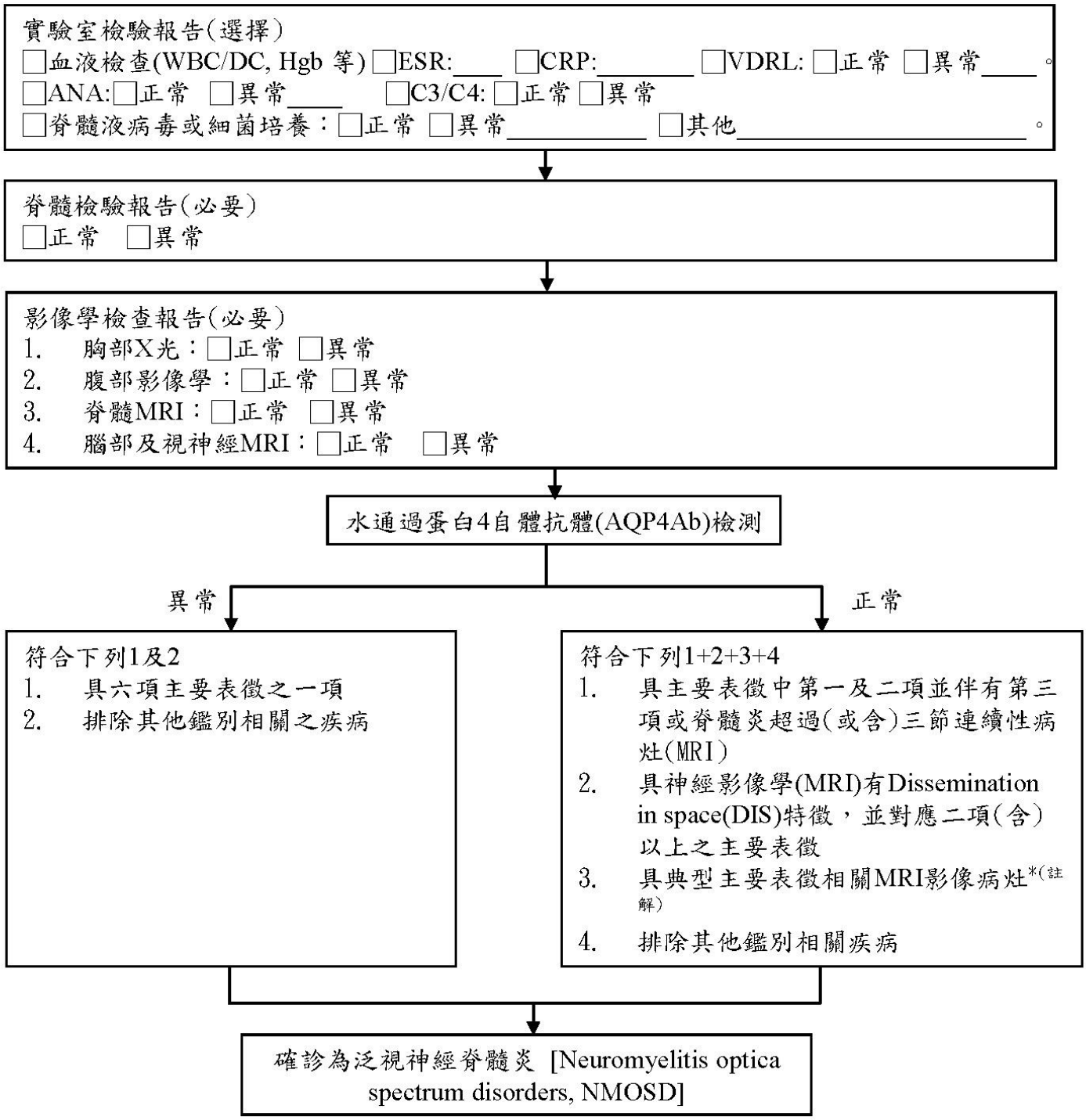

*註解

Acute optic neuritis : normal findings or only nonspecific white matter lesions, or optic nerve: T2-hyperintense lesion or T1- gadolinium enhancing lesion extending >1/2 optic nerve length or involving optic chiasm ;
Acute myelitis : intramedullary MRI lesion ≥3 contiguous segments (LETM) or ≥3 contiguous segments of focal spinal cord atrophy in patients with history compatible with acute myelitis ;
Area postrema syndrome: dorsal medulla/area postrema lesions ;
Acute brainstem syndrome: periependymal brainstem lesions.

參考文獻

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