**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)**

**-腦白質消失症 [Vanishing white matter disease] -**

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| **項目** | **勾選** |
| 1.病歷摘要中包含足以佐證之臨床資料及實驗室數據(必要) |  |
| * CSF 常規檢驗 |  |
| * 血液乳酸 |  |
| 2. 腦部核磁共振影像報告或重點影像足以佐證(必要) |  |
| 2-1確認診斷須符合 |  |
| * There are diffuse or extensive signal abnormalities involving the cerebral white matter; the adjacent subcortical white matter may be spared |  |
| * Part or all of the abnormal white matter has a signal intensity close to or the same as cerebrospinal fluid on proton density or fluid-attenuated inversion recovery (FLAIR) images |  |
| * There is a fluid-filled space (not a total collapse of the white matter) between the ependymal lining and the cortex if proton density and FLAIR images suggest that all cerebral white matter has disappeared |  |
| * The disappearance of the cerebral white matter occurs in a diffuse "melting away" pattern |  |
| * The temporal lobes are relatively spared in the extent of the abnormal signal and/or degree of cystic destruction |  |
| * The cerebellar white matter may be abnormal, but does not contain cysts |  |
| * There is no contrast enhancement |  |
| 2-2疑似診斷須符合 |  |
| * A pattern of radiating stripes within abnormal white matter is seen on sagittal and coronal T1-weighted or FLAIR images; dots and stripes are seen within the abnormal white matter on axial images, representing cross-sections of the stripes seen on sagittal and coronal images |  |
| * There are lesions within the central tegmental tracts in the pontine tegmentum |  |
| * There is involvement of the inner rim of the corpus callosum, with sparing of the outer rim |  |
| 3. 基因檢測報告(必要) |  |
| * EIF2B1 |  |
| * EIF2B2 |  |
| * EIF2B3 |  |
| * EIF2B4 |  |
| * EIF2B5 |  |
| 4. 無更適合的解釋  說明： |  |

備註：

1. 檢驗應包括CSF 常規檢驗以及血液乳酸分析，結果應為正常．
2. 參考Lancet Neurol. 2006;5(5):413.
3. 95%的患者應有上述五個基因中之一的異型或同型合子的基因變化．

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- 腦白質消失症[Vanishing white matter disease] -

應檢附文件 (必要)

□ 臨床資料及實驗室數據

□ 腦部核磁共振影像報告或重點影像

□ 基因檢測報告

□ CSF 常規檢驗 (必要)

□ 血液乳酸 (必要)

確認診斷須符合

□ Diffuse or extensive signal abnormalities involving the cerebral white matter; the adjacent subcortical white matter may be spared

□ Part or all of the abnormal white matter has a signal intensity close to or the same as cerebrospinal fluid on proton density or fluid-attenuated inversion recovery (FLAIR) images

□ A fluid-filled space (not a total collapse of the white matter) between the ependymal lining and the cortex if proton density and FLAIR images suggest that all cerebral white matter has disappeared

□ The disappearance of the cerebral white matter occurs in a diffuse "melting away" pattern

□ The temporal lobes are relatively spared in the extent of the abnormal signal and/or degree of cystic destruction

□ The cerebellar white matter may be abnormal, but does not contain cysts

□ There is no contrast enhancement

疑似診斷須符合

□ A pattern of radiating stripes within abnormal white matter is seen on sagittal and coronal T1-weighted or FLAIR images; dots and stripes are seen within the abnormal white matter on axial images, representing cross-sections of the stripes seen on sagittal and coronal images

□ Lesions within the central tegmental tracts in the pontine tegmentum

□ Involvement of the inner rim of the corpus callosum, with sparing of the outer rim

□基因檢測報告

1. □ *EIF2B1*
2. □ *EIF2B2*
3. □ *EIF2B3*
4. □ *EIF2B4*
5. □ *EIF2B5*

無更合適的解釋，即診斷腦白質消失症 (Vanishing white matter disease)

請補說明：