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] -

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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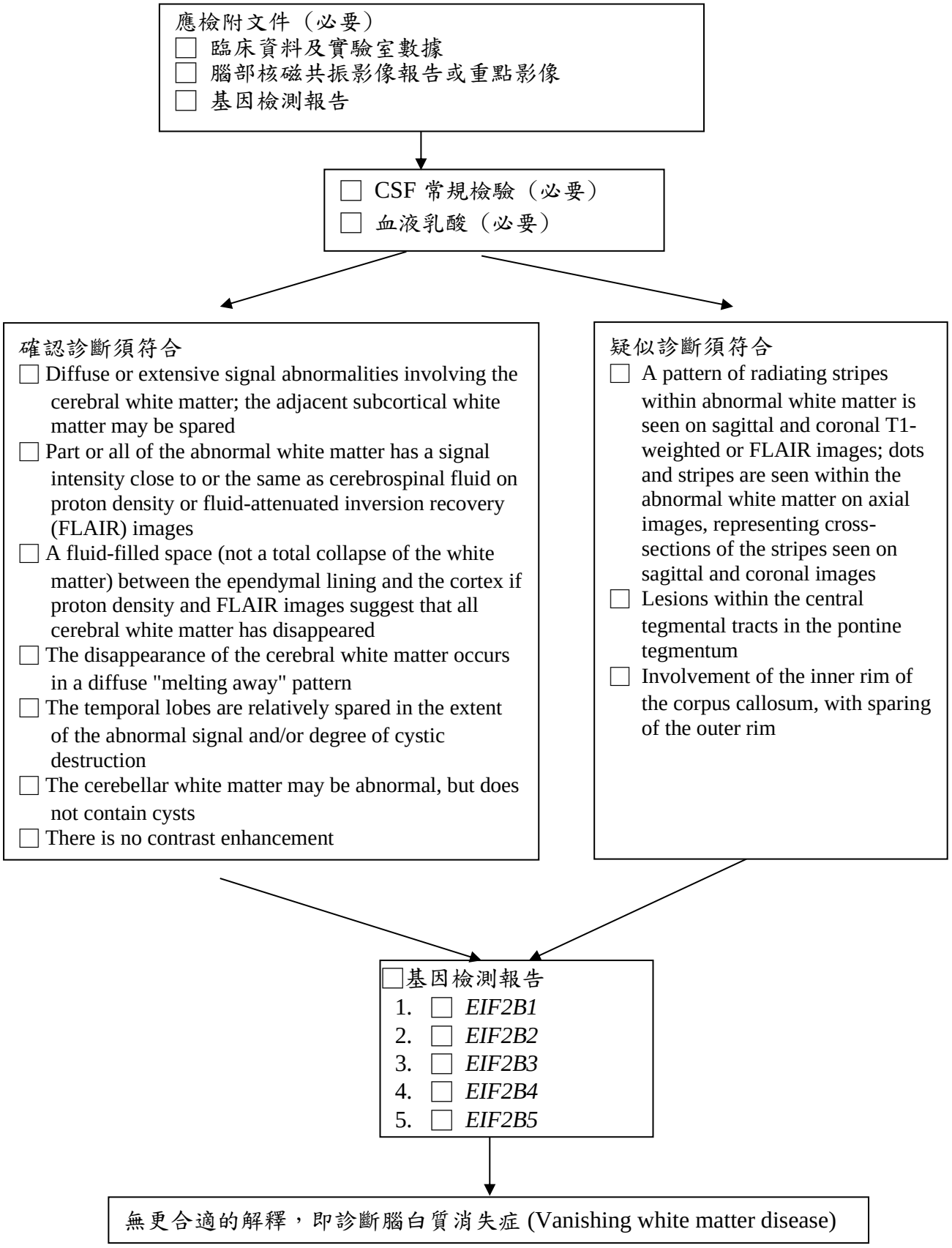

請補說明：_____

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