**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)
-嚴重複合型免疫缺乏症[Severe Combined ImmunoDeficiency, SCID]-**

□ 病歷資料，包含臨床病史、身體診察、特殊外觀及發展里程碑等 (必要)

□ 實驗室檢驗報告 (必要)

□ 影像學或特殊檢查檢查報告 (選擇)

□ 基因檢測報告 (必要)

**一、新生兒篩檢陽性個案，請填寫此表格。**

| 項目 | 填寫部分 |
| --- | --- |
| 1. **病歷資料(必要)**
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| A1臨床病史(必要) | □ 發病年紀在3個月內者，必須**與孕程母親狀況無關** □母親孕程期間無自體免疫疾病 (SLE, RA or others: \_\_\_\_\_\_\_) □母親孕程期間未服用免疫抑制劑 (藥物\_\_\_\_\_\_\_\_\_\_\_\_)□已排除其他原因及排除特定疾病(如：DiGeorge症候群等) |
| A2家族病史(請附上家族中其他病患之詳細病歷記錄或個案病歷資料) (必要) | □ 家族史 (Family history) □ 有 □ 無  |
| A3臨床症狀 | □ 有 □ 無 |
| 1. **實驗室檢驗報告** (必要)(請附相關檢驗資料)
 | **淋巴球表面標記分析(Lymphocyte subset analysis ) (全必填)**□總淋巴球數\_\_\_\_\_\_\_/mm3□CD3\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)□CD4\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in CD3)□CD8\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in CD3)□Naïve \_\_\_\_\_\_% in CD4; Memory \_\_\_\_\_\_% in CD4□CD19\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)□CD16CD56 NK \_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)**淋巴增生功能 (Lymphocyte proliferation function) (選擇)**檢測方法 □Thymidine □CFSE □Ki67 or □others: \_\_\_\_\_\_\_ 檢測結果 □正常 □低下，為正常的\_\_\_\_\_\_% |
| 1. **影像學檢查報告**

(選擇) (請附相關報告資料) | 胸部X光：Thymus shadow □ 有 □ 無其他異常影像檢查: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| 1. **基因檢測報告(必要)**

(請附實驗室報告) | □找到符合遺傳模式之致病基因\*變異：基因名\_\_\_\_\_\_\_\_\_\_\_\_，　　　　　　　　　　　　　　　　　變異 □尚未找到基因變異，但淋巴增生功能 (Lymphocyte proliferation function) 低於正常10% |

**二、非新生兒篩檢陽性個案，請填寫此表格。**

| 項目 | 填寫部分 |
| --- | --- |
| 1. **病歷資料(必要)**
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| A1臨床病史(必填) | □ 發病年齡 (Age at disease onset) \_\_\_\_\_\_\_ 歲□ 發病年紀在3個月內者，必須**與孕程母親狀況無關** □母親孕程期間無自體免疫疾病 (SLE, RA or others: \_\_\_\_\_\_\_) □母親孕程期間未服用免疫抑制劑 (藥物\_\_\_\_\_\_\_\_\_\_\_\_)□已排除其他原因及排除特定疾病(如：DiGeorge症候群等) |
| A2家族病史(請附上家族中其他病患之詳細病歷記錄或個案病歷資料) (必要) | □ 家族史 (Family history) □ 有 □ 無  |
| A3臨床症狀及徵兆(必要，至少出現右欄一項) | **感染相關：**□肺炎 (Pneumonia) \_\_\_次 (pathogens\_\_\_\_\_)□中耳炎 (Otitis media) \_\_\_\_\_ 次□鼻竇炎 (Sinusitis) \_\_\_\_\_ 次□敗血症 (Sepsis) \_\_\_\_\_次 (Pathogens\_\_\_\_)□伺機性感染(Opportunistic infection, pathogens\_\_\_\_\_)□鵝口瘡 (Oral thrush)□皮膚念珠菌感染 (Cutaneous candidiasis)□其他 (請儘量陳述 )**非感染直接相關：**□肝腫大 (Hepatomegaly)□脾腫大 (Splenomegaly)□紅皮症 (似嚴重異位性皮膚炎，Erythroderma)□不易控制的腹瀉 (Refractory diarrhea)□先天心臟異常 (Congenital heart diseases,)□腦神經病變 (Cerebro-neuropathy: □ seizure，□other: \_\_\_\_\_\_\_\_)□外觀異常 (Abnormal appearance，請儘量陳述 )□生長發育遲滯 (Failure to thrive) |
| 1. **實驗室檢驗報告**(請附相關檢驗資料)**(必要)**
 | **□淋巴球表面標記分析(Lymphocyte subset analysis ) (全必填)**□總淋巴球數\_\_\_\_\_\_\_/mm3□CD3\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)□CD4\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in CD3)□CD8\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in CD3)□Naïve \_\_\_\_\_\_% in CD4; Memory \_\_\_\_\_\_% in CD4□CD19\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)□CD16CD56 NK \_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)**□淋巴增生功能(Lymphocyte proliferation function)異常(選擇)**檢測方法：□Thymidine □CFSE □Ki67 or □others: \_\_\_\_ 檢測結果：□正常 □低下，為正常的＿＿＿% |
| 1. **基因檢測報告(必要)** (請附實驗室報告)
 | □找到符合遺傳模式之致病基因\*變異：基因名\_\_\_\_\_\_\_\_\_\_\_\_，　　　　　　　　　　　　　　　　　變異 □尚未找到基因變異，但淋巴增生功能 (Lymphocyte proliferation function) 低於正常10% |

\*致病基因可參照參考文獻1,2, 3，包括IL2RG, RAG1, ADA, IL7R等，不包括WASP, ATM, PIK3CG, TBX1, RMRP, STAT3, IKBKG等其他類會造成免疫缺陷之致病基因。

參考文獻：

1. Dvorak CC, Haddad E, Heimall J, Dunn E, Buckley RH, Kohn DB, Cowan MJ, Pai SY, Griffith LM, Cuvelier GDE, Eissa H, Shah AJ, O'Reilly RJ, Pulsipher MA, Wright NAM, Abraham RS, Satter LF, Notarangelo LD, Puck JM. The diagnosis of severe combined immunodeficiency (SCID): The Primary Immune Deficiency Treatment Consortium (PIDTC) 2022 Definitions. J Allergy Clin Immunol. 2023 Feb;151(2):539-546.
2. Bousfiha A, Moundir A, Tangye SG, Picard C, Jeddane L, Al-Herz W, Rundles CC, Franco JL, Holland SM, Klein C, Morio T, Oksenhendler E, Puel A, Puck J, Seppänen MRJ, Somech R, Su HC, Sullivan KE, Torgerson TR, Meyts I. The 2022 Update of IUIS Phenotypical Classification for Human Inborn Errors of Immunity. J Clin Immunol. 2022 Oct;42(7):1508-1520.
3. Tangye SG, Al-Herz W, Bousfiha A, Cunningham-Rundles C, Franco JL, Holland SM, Klein C, Morio T, Oksenhendler E, Picard C, Puel A, Puck J, Seppänen MRJ, Somech R, Su HC, Sullivan KE, Torgerson TR, Meyts I. Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. J Clin Immunol. 2022 Oct;42(7):1473-1507.
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**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)
-嚴重複合型免疫缺乏症[Severe Combined ImmunoDeficiency, SCID]-**

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| **應檢附文件**□ 病歷資料，包含臨床病史、身體診察、特殊外觀及發展里程碑等 (必要) □ 實驗室檢驗報告 (必要) □ 影像學或特殊檢查檢查報告 (選擇) □ 基因檢測報告 (必要) |

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| □ **新生兒篩檢陽性個案****(含其家屬) (必要)*** 家族史 □ 有 □ 無
* 臨床症狀 □ 有 □ 無
 |
| □ **臨床個案病史 (必要)*** 發病年齡 (Age at disease onset) \_\_\_\_\_ 歲
* 家族史 (Family history) □ 有 □ 無
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**臨床症狀及徵兆 (必要，至少出現下列一項)**

非感染直接相關

□肝腫大 (Hepatomegaly)

□脾腫大 (Splenomegaly)

□紅皮症 (似嚴重異位性皮膚炎，Erythroderma)

□不易控制的腹瀉 (Refractory diarrhea)

□先天心臟異常 (Congenital heart diseases,)

□腦神經病變 (Cerebro-neuropathy:

　　□ seizure，□other: \_\_\_\_\_\_\_\_)

□外觀異常 (Abnormal appearance，請儘量陳述 )

□生長發育遲滯 (Failure to thrive)

□其他 (請儘量陳述 )

感染相關

□肺炎 (Pneumonia) \_\_\_次 (pathogens\_\_\_)

□中耳炎 (Otitis media) \_\_\_ 次

□鼻竇炎 (Sinusitis) \_\_\_ 次

□敗血症 (Sepsis) \_\_\_\_次 (Pathogens\_\_\_\_)

□伺機性感染(Opportunistic infection, pathogens\_ \_\_\_)

□鵝口瘡 (Oral thrush)

□皮膚念珠菌感染 (Cutaneous candidiasis)

□其他 (請儘量陳述 )

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| □ 發病年紀在3個月內者，必須**與孕程母親狀況無關 (必全選)** □母親孕程期間無自體免疫疾病 (SLE, RA or others: \_\_\_\_\_\_\_) □母親孕程期間未服用免疫抑制劑□已排除其他原因及排除特定疾病(如：DiGeorge症候群等) |

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| **實驗室檢驗報告(必要)****□淋巴球表面標記分析(Lymphocyte subset analysis ) (全必填)** □總淋巴球數\_\_\_\_\_\_\_/mm3　　　　　　　□CD3\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)□CD4\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in CD3) □CD8\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in CD3)□Naïve \_\_\_\_\_\_% in CD4; Memory \_\_\_\_\_\_% in CD4□CD19\_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)□CD16CD56 NK \_\_\_\_\_\_\_/mm3 (\_\_\_\_\_\_% in total lymphocytes)**□淋巴增生功能(Lymphocyte proliferation function)異常(選擇)**檢測方法：□Thymidine □CFSE □Ki67 or □others: \_\_\_\_ 檢測結果：□正常 □低下，為正常的＿＿＿% |

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| **影像學檢查 (選擇)**胸部X光： Thymus shadow □ 有 □ 無其他異常影像檢查: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |

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| **基因檢測報告 (必要)**□找到符合遺傳模式之致病基因\*變異: 基因名\_\_\_\_\_\_\_\_\_\_\_\_，變異 □尚未找到基因變異，但淋巴增生功能 (Lymphocyte proliferation function) 低於正常10% |

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| **符合罕見疾病之嚴重複合型免疫缺乏症**⬜臨床症狀(含篩檢)，實驗室檢驗報告**顯示有嚴重CD3 T淋巴球低下(< 300 cells/mm3)或CD4 naïve T淋巴球低下(占比<20%)**，與基因檢測找到符合遺傳模式之致病基因變異**三項**皆符合⬜臨床症狀(含篩檢) 符合且實驗室檢驗報告2次以上皆**顯示有嚴重CD3 或CD4 naïve T淋巴球低下**，雖基因檢測報告尚未找到基因變異，但淋巴增生功能 (Lymphocyte proliferation function) 低於正常10% |

\*致病基因可參照參考文獻1,2, 3，包括IL2RG, RAG1, ADA, IL7R等，不包括WASP, ATM, PIK3CG, TBX1, RMRP, STAT3, IKBKG等其他類會造成免疫缺陷之致病基因。

備註：參考文獻

1. Dvorak CC, Haddad E, Heimall J, Dunn E, Buckley RH, Kohn DB, Cowan MJ, Pai SY, Griffith LM, Cuvelier GDE, Eissa H, Shah AJ, O'Reilly RJ, Pulsipher MA, Wright NAM, Abraham RS, Satter LF, Notarangelo LD, Puck JM. The diagnosis of severe combined immunodeficiency (SCID): The Primary Immune Deficiency Treatment Consortium (PIDTC) 2022 Definitions. J Allergy Clin Immunol. 2023 Feb;151(2):539-546.
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