敏兒安 safe T21 express

Kinetics® 毅力醫護健康集團

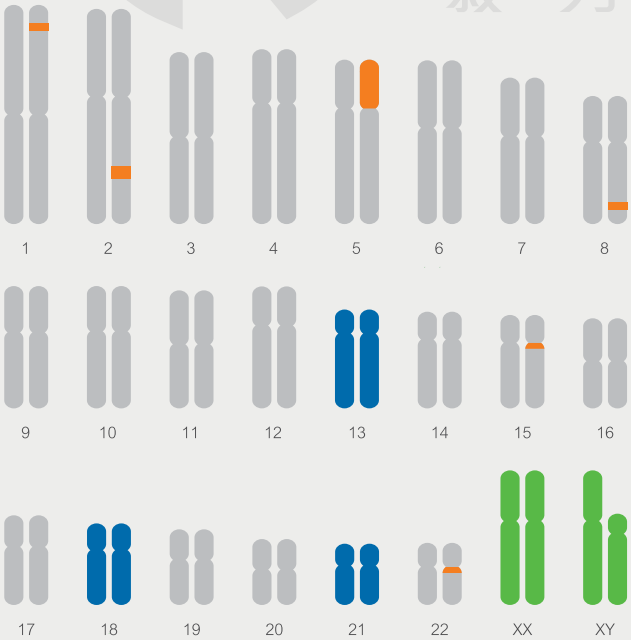
無創性胎兒DNA產前篩檢測試項目

Non-Invasive Analysis of Fetal DNA for Prenatal Screening - Standard Panel

標準版
Standard Panel

為了使服務達到國際標準[3]，我們建議媽媽選擇敏兒安 safe T21 express 標準版，其所報的14項常見異常有較多數據支持。

safe T21 express Standard Panel screens for 14 validated conditions. In alignment with international guidelines[3], this panel is the recommended choice for most mothers.



藍色為染色體三體症
Blue represents Trisomies

橙色為微缺失症候群
Orange represents Microdeletion Syndromes

綠色為性染色體相關疾病
Green represents Sex Chromosome Aneuploidies

*敏兒安 safe T21 express 只能檢測微小至3Mb長度的微缺失及微重複，然而上述的微缺失卻有機會小於3Mb。由於部分微缺失的發生率較低，因此未獲大型數據驗證。

*Since microdeletions are rare, limited data is available for validating the detection rate of most microdeletions. Microdeletions may occur in less than 3Mb in size, safe T21 express only searches for microdeletions with a minimum size of 3Mb.

測試項目包括 Testing items include

3項 染色體三體症 Trisomies	7項 微缺失症候群 Microdeletion Syndromes	4項 性染色體相關疾病 Sex Chromosome Aneuploidies

染色體三體症 Trisomies

- T21唐氏綜合症 T21 Down Syndrome
- T18愛德華氏綜合症 T18 Edwards Syndrome
- T13巴陶氏綜合症 T13 Patau Syndrome

微缺失症候群 Microdeletion Syndromes

- 1p36缺失綜合症 1p36 Deletion Syndrome
- 2q33.1缺失綜合症 2q33.1 Deletion Syndrome
- 15q11.2缺失綜合症/天使綜合症 15q11.2 Deletion/ Angelman Syndrome
- 5p缺失綜合症/貓哭綜合症 5p Deletion/ Cri-du-chat Syndrome
- 22q11.2缺失綜合症/迪喬治綜合症 22q11.2 Deletion/ DiGeorge Syndrome
- 8q24.1缺失綜合症 毛髮-鼻-指骨綜合症 8q24.1 Deletion/ Langer-Giedion Syndrome
- 15q11.2 缺失綜合症/普瑞德威利綜合症/小胖威利綜合症 15q11.2 Deletion/ Prader-Willi Syndrome

性染色體相關疾病 Sex Chromosome Aneuploidies

- X0 X染色體單體症（特納綜合症） X0 Monosomy X (Turner Syndrome)
- XYY 三體綜合症（XYY 超雄綜合症/雅各氏綜合症） XYY Syndrome (Jacob's Syndrome)
- XXY 柯林菲特氏綜合症 XXY Klinefelter Syndrome
- XXX 三體綜合症（XXX 超雌綜合症） XXX Triple X Syndrome

敏兒安 safe T21 express

Kinetics® 毅力醫護健康集團

無創性胎兒DNA產前篩檢測試項目

Non-Invasive Analysis of Fetal DNA for Prenatal Screening - Advanced Panel

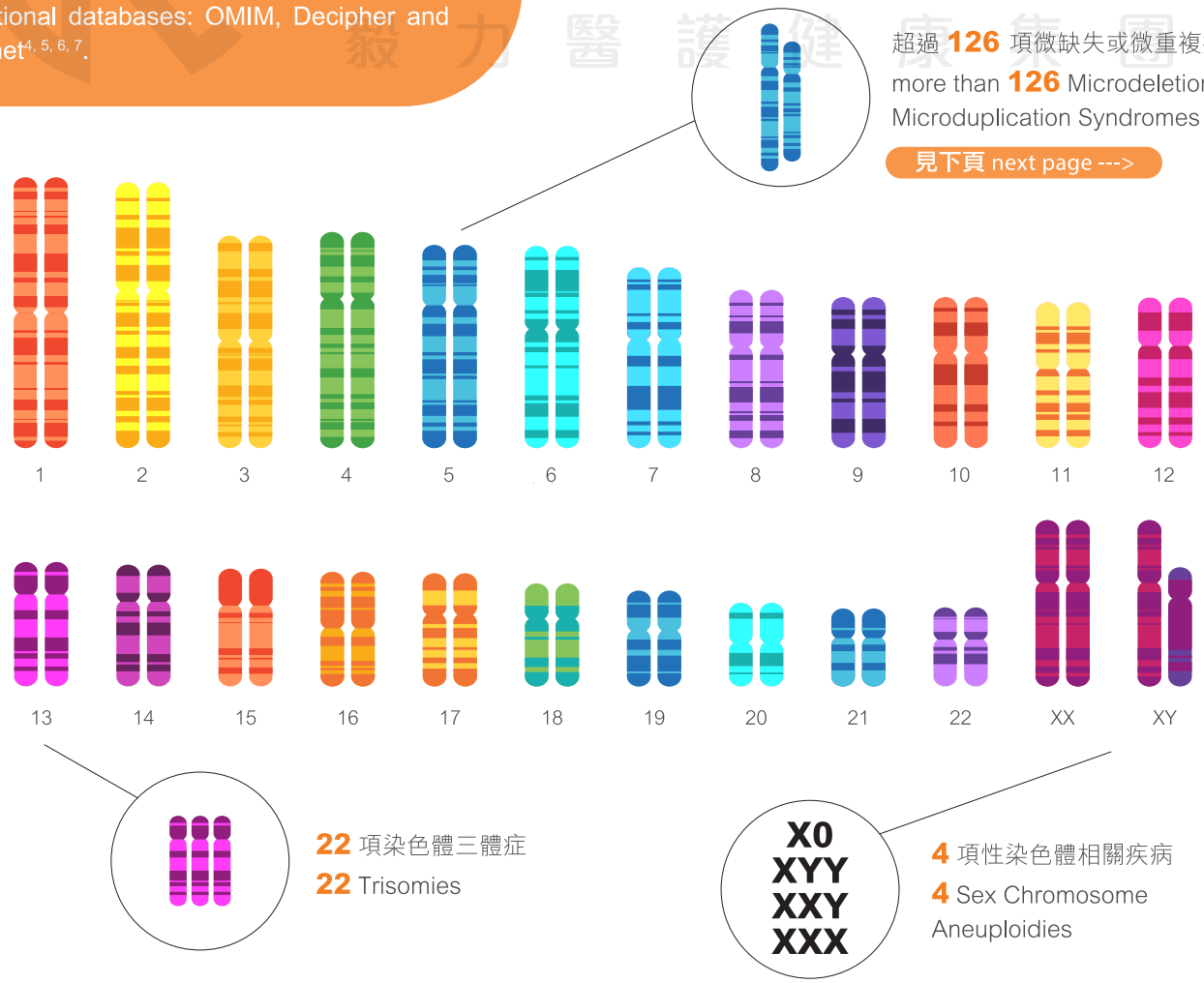
進階版 Advanced Panel

敏兒安 safe T21 express 進階版可以全面檢測到23對染色體相關疾病，包括染色體三體症及目前已在國際數據庫(OMIM, Decipher和Orphanet)中記錄的126項微小至3Mb的微缺失或微重複[4, 5, 6, 7]。

safe T21 express Advanced Panel screens for chromosomal aneuploidies of all 23 pairs of chromosomes, including trisomies and the 126 microdeletions/microduplications with a minimum size of 3Mb that has been recorded on the international databases: OMIM, Decipher and Orphanet[4, 5, 6, 7].

測試項目包括 Testing items include

22 項	**126** 項	**4** 項
染色體三體症 Trisomies	微缺失或微重複症候群 Microdeletion/ Microduplication Syndromes	性染色體相關疾病 Sex Chromosome Aneuploidies



超過 **126** 項微缺失或微重複症候群
more than **126** Microdeletion or Microduplication Syndromes

見下頁 next page --->

22 項染色體三體症
22 Trisomies

X0
XYY
XXY
XXX

4 項性染色體相關疾病
4 Sex Chromosome Aneuploidies

*敏兒安 safe T21 express 只能檢測微小至3Mb長度的微缺失及微重複，然而上述的微缺失卻有機會小於3Mb。由於部分微缺失的發生率較低，因此未獲大型數據驗證。

*由於進階版的檢測項目罕見及複雜，未獲大型數據驗證，因此準確率會降低。

*Since microdeletions are rare, limited data is available for validating the detection rate of most microdeletions. Microdeletions may occur in less than 3Mb in size, safe T21 express only searches for microdeletions with a minimum size of 3Mb.

*Since advanced findings are rare and complex, insufficient data for validation may lower the accuracy.

Kinetics® 毅力醫護健康集團

敏兒安 safe21 express 進階版

* 根據美國醫學遺傳學及基因組學學會（ACMG）的陳述及美國婦產科醫學會（ACOG）的指引#163，不建議透過無創性產前篩查檢測拷貝數變異。Screening CNVs by non-invasive prenatal test (NIPT) is NOT recommended according to ACMG statement and ACOG practic e bulletin #163.

* 以下微缺失/微重複症候群有機會小於 3Mb 長度，敏兒安 safeT21express™ 只能檢測微小至 3Mb 長度的微缺失/微重複症候群。 Below Microdeletion/ Microduplication Syndromes may occur in less than 3Mb in size. safeT21express ™ only searches for Microdeletion/Microduplication Syndr omes with a minimum size of 3Mb.

微缺失/微重複症候群不小於 3Mb 列表 List of Microdeletion/Microduplication Syndromes with size greater than 3Mb

1. 1p21.3 微缺失綜合症
1p21.3 Microdeletion Syndrome
2. 1p31 重複綜合症
1p31 Duplication Syndrome
3. 1p32-p31 微缺失綜合症
1p32-p31 Microdeletion Syndrome
4. 1p36 缺失綜合症
1p36 Deletion Syndrome
5. 1q21.1 微缺失綜合症[1,3]
1q21.1 Microdeletion Syndrome[1,3]
6. 1q21.1 微重複綜合症[2]
1q21.1 Microduplication Syndrome[2]
7. TAR 綜合症 /血小板減少 - 橈骨缺失綜合症[3]
Thrombocytopenia Absent Radius Syndrome[3]
8. 1q41-q42 微缺失綜合症
1q41-q42 Microdeletion Syndrome
9. 1q44 微缺失綜合症
1q44 Microdeletion Syndrome
10. 2p12-p11.2 缺失綜合症
2p12-p11.2 Deletion Syndrome
11. 2p16.1-p15 微缺失綜合症
2p16.1-p15 Microdeletion Syndrome
12. 2q23.1 微缺失綜合症
2q23.1 Microdeletion Syndrome
13. 2q24 微缺失綜合症
2q24 Microdeletion Syndrome
14. 2q31.1 微缺失綜合症 /裂手裂足症 5 型
2q31.1 Microdeletion Syndrome/ Split Hand/ Foot Malformation 5
15. 2q31.1 微重複綜合症
2q31.1 Microduplication Syndrome
16. 2q32-q33 微缺失綜合症/ 2q33.1 微缺失綜合症
2q32-q33 Microdeletion Syndrome/ 2q33.1 Microdeletion Syndrome
17. 2q35 重複綜合症
2q35 Duplication Syndrome
18. 2q37 微缺失綜合症/全前腦症、前腦無裂畸形 6 型[4]
2q37 Microdeletion Syndrome/ Holoprosencephaly 6[4]
19. 3p-綜合症/3pter-p25 缺失綜合症
3p-Syndrome/ 3pter-p25 Deletion Syndrome
20. 3q13.31 缺失綜合症
3q13.31 Deletion Syndrome
21. Dandy-Walker 畸形、Dandy-Walker 綜合症、第四腦室孔閉塞綜合症
Isolated Dandy-Walker Malformation
22. 3q26-q27 微缺失綜合症
3q26-q27 Microdeletion Syndrome
23. 3q27.3 微缺失綜合症
3q27.3 Microdeletion Syndrome
24. 3q29 微缺失綜合症/3q 亞端粒缺失綜合症
3q29 Microdeletion Syndrome/ 3q Subtelomere Deletion Syndrome/3qter Deletion
25. 3q29 微重複綜合症
3q29 Microduplication Syndrome
26. 4p-綜合症/沃夫-許宏綜合症
4p-Syndrome/ Wolf-Hirschhorn Syndrome
27. 4q21 微缺失綜合症
4q21 Microdeletion Syndrome
28. 4q25 近端缺失綜合症[5]
4q25 Proximal Deletion Syndrome[5]
29. 4q32.1-q32.2 三倍重複綜合症
4q32.1-q32.2 Triplication Syndrome
30. 5p 缺失綜合症 / 5p 單體症 / 貓哭綜合症
5p Deletion Syndrome / Monosomy 5p / Cri-du-chat Syndrome
31. 5p13 微重複綜合症
5p13 Microduplication Syndrome
32. 5q12 缺失綜合症/PDE4D單倍體不足綜合症
5q12 Deletion Syndrome / PDE4D Haploinsufficiency Syndrome
33. 5q14.3 微缺失綜合症
5q14.3 Microdeletion Syndrome
34. 5q23 微缺失綜合症
5q23 Microdeletion Syndrome
35. Sotos 綜合症1 型、大腦巨大症、小兒腦畸形綜合症[6]
Sotos Syndrome 1[6]
36. 6p25微缺失綜合症 / 6p亞端粒缺失綜合症
6p25 Microdeletion Syndrome/ 6p Subtelomeric Deletion Syndrome / 6pterp24 Deletion Syndrome
37. 6q11-q14 缺失
6q11-q14 Deletion
38. 6q16 缺失綜合症
6q16 Deletion Syndrome
39. 6q25 微缺失綜合症 / 6q24-q25 缺失綜合症
6q25 Microdeletion Syndrome / 6q24-q25 Deletion Syndrome
40. 脊索瘤
CHDM/ Chordoma
41. 6q 終端缺失綜合症
6q Terminal Deletion Syndrome
42. 7p22.1 微重複綜合症[7]
7p22.1 Microduplication Syndrome[7]
43. 7q 缺失
7q Deletion
44. 7q11.23 缺失綜合症
7q11.23 Deletion Syndrome
45. 7q11.23重複綜合症
7q11.23 Duplication Syndrome
46. 7q31 微缺失綜合症
7q31 Microdeletion Syndrome
47. 8p23.1 微缺失綜合症
8p23.1 Microdeletion Syndrome
48. 8p23.1 重複綜合症
8p23.1 Duplication Syndrome
49. 8q12 微重複綜合症[7]
8q12 Microduplication Syndrome[7]
50. 8q12.1-q21.2 缺失綜合症
8q12.1-q21.2 Deletion Syndrome
51. 8q13 微缺失綜合症
8q13 Microdeletion Syndrome
52. 8q21.11 微缺失綜合症
8q21.11 Microdeletion Syndrome
53. 8q22.1 微缺失綜合症
8q22.1 Microdeletion Syndrome
54. 8q22.1重複綜合症/ Leri骨化過早症[7]
8q22.1 Duplication Syndrome/ Leri Pleonosteosis[7]
55. 8q24.1 缺失綜合症 / 毛-鼻-指骨綜合症
8q24.1 Deletion Syndrome/ Langer-Giedion Syndrome/ Trichorhinophalangeal Syndrome Type 2
56. 8q24.3 缺失綜合症[7]
8q24.3 Deletion Syndrome[7]
57. 9p 缺失綜合症 / 9p 單體症
9p Deletion Syndrome/ Monosomy 9p
58. 9p13 微缺失綜合症
9p13 Microdeletion Syndrome
59. 9q21 微缺失綜合症
9q21 Microdeletion Syndrome
60. 10p 遠端單體症/喬治綜合症2 型[4]
Distal Monosomy 10p / DiGeorge Syndrome 2[4]
61. 10q22.3q23.2 微缺失綜合症
10q22.3q23.2 Microdeletion Syndrome
62. 10q22.3q23.3 微重複綜合症
10q22.3q23.3 Microduplication Syndrome
63. 10q26 缺失綜合症 / 10q 端粒缺失綜合症 / 10q 遠端單體症
10q26 Deletion Syndrome / Telomeric Deletion 10q / Distal Monosomy 10q
64. 11p11.2 缺失綜合症/ Potocki-Shaffer 綜合症
11p11.2 Deletion Syndrome/ Potocki-Shaffer Syndrome
65. WAGR 綜合症
WAGR Syndrome
66. WAGRO 綜合症/ WAGR 綜合症[8]
WAGRO Syndrome/ WAGR Syndrome[8]
67. 11q22.2-q22.3 缺失綜合症
11q22.2-q22.3 Deletion Syndrome
68. 11q23 缺失綜合症/雅各布森綜合症、賈可森綜合症
11q23 Deletion Syndrome/ Jacobsen Syndrome
69. 12p12.1 微缺失綜合症
12p12.1 Microdeletion Syndrome
70. 12p13.33 微缺失綜合症/ 12p 遠端單體症[7]
12p13.33 Microdeletion Syndrome/ Distal Monosomy 12p[7]
71. 12q14 微缺失綜合症
12q14 Microdeletion Syndrome
72. 12q15-q21.1 微缺失綜合症
12q15-q21.1 Microdeletion Syndrome
73. 13q12.3 微缺失綜合症[7]
13q12.3 Microdeletion Syndrome[7]
74. 13q14 缺失綜合症
13q14 Deletion Syndrome
75. 13q32 缺失綜合症/ 13q 遠端單體症
13q32 Deletion Syndrome/ Distal Monosomy 13q
76. 14q11.2 微缺失綜合症 / 14q11-q22 缺失綜合症
14q11.2 Microdeletion Syndrome/ 14q11-q22 Deletion Syndrome
77. 14q12 微缺失綜合症
14q12 Microdeletion Syndrome
78. Frias 綜合症
Frias Syndrome
79. 14q32 重複綜合症
14q32 Duplication Syndrome
80. 15q11-q13 重複綜合症
15q11-q13 Duplication Syndrome
81. 15q11.2 缺失綜合症 / 普瑞德威利綜合症、小胖威利綜合症[9]
15q11.2 Deletion Syndrome/ Prader-Willi Syndrome 9
82. 15q11.2 缺失綜合症 / 天使綜合症、天使人綜合症、安格曼綜合症[9]
15q11.2 Deletion Syndrome/ Angelman Syndrome[9]
83. 15q14 微缺失綜合症
15q14 Microdeletion Syndrome
84. 15q24 微缺失綜合症[10]
15q24 Microdeletion Syndrome[10]
85. 15q25 缺失綜合症
15q25 Deletion Syndrome
86. 15q26 缺失綜合症 / 15q 遠端單體症
15q26 Deletion Syndrome / Distal Monosomy 15q
87. Levy-Shanske 綜合症
Levy-Shanske Syndrome
88. 先天性橫隔膜疝氣疾病
HCD/ Hernia, Congenital Diaphragmatic
89. 16p12.2-p11.2 微缺失綜合症[11]
16p12.2-p11.2 Microdeletion Syndrome[11]
90. 16p12.2-p11.2 微重複綜合症[12]
16p12.2-p11.2 Microduplication Syndrome[12]
91. 16p13.3 缺失綜合症
16p13.3 Deletion Syndrome
92. 16q22 缺失綜合症
16q22 Deletion Syndrome
93. 16q24.1 微缺失綜合症
16q24.1 Microdeletion Syndrome
94. 17p11.2 微缺失綜合症 / 史密斯-馬吉利氏綜合症
17p11.2 Microdeletion Syndrome / Smith-Magenis Syndrome
95. 17p11.2 微重複綜合症 / Potocki-Lupski 綜合症
17p11.2 Microduplication Syndrome/ Potocki-Lupski Syndrome
96. 17p12-p11.2 微重複綜合症 / Yuan-Harel-Lupski 綜合症
17p12-p11.2 Microduplication Syndrome/ Yuan-Harel-Lupski Syndrome
97. 17p13.3 缺失綜合症
17p13.3 Deletion Syndrome
98. 17p13.3 微重複綜合症
17p13.3 Microduplication Syndrome
99. 17q11 微缺失綜合症
17q11 Microdeletion Syndrome
100. 17q12 缺失綜合症
17q12 Deletion Syndrome
101. 17q12 微重複綜合症
17q12 Microduplication Syndrome
102. 17q21.31重複綜合症
17q21.31 Duplication Syndrome
103. 17q23.1-23.2 缺失綜合症
17q23.1-23.2 Deletion Syndrome
104. 18p-綜合症 / 18p 缺失綜合症 / 18p 單體症
18p-Syndrome / 18p Deletion Syndrome / Monosomy 18p
105. 18q-綜合症 / 18q 缺失綜合症
18q-Syndrome/ 18q Deletion Syndrome
106. 19q13.11 微缺失綜合症
19q13.11 Microdeletion Syndrome
107. 20p12.3 微缺失綜合症
20p12.3 Microdeletion Syndrome
108. 20q11.2 微缺失綜合症[7]
20q11.2 Microdeletion Syndrome[7]
109. 21q-綜合症 / 21q 缺失綜合症 / 21 單體症
21q-Syndrome/ 21q Deletion Syndrome/ Monosomy 21
110. 21q22.11-q22.12 微缺失綜合症
21q22.11-q22.12 Microdeletion Syndrome
111. 21q22.13-q22.2 微缺失綜合症/因21q22.13-q22.2 微缺失引致的 DYRK1A 相關智力障礙綜合症
21q22.13-q22.2 Microdeletion Syndrome/ DYRK1A-Related Intellectual Disability Syndrome due to 21q22.13-q22.2 Microdeletion
112. 貓眼綜合症
Cat Eye Syndrome
113. 22q11.2 缺失綜合症 / 22q11DS / 迪喬治綜合症 / 顎心臉綜合症
22q11.2 Deletion Syndrome/ 22q11DS/ DiGeorge Syndrome/ Velocardiofacial Syndrome
114. 22q11.2 微重複綜合症[13]
22q11.2 Microduplication Syndrome[13]
115. 22q13 缺失 / 22q13 單體症
22q13 Deletion / Monosomy 22q13
116. 22q22.3 缺失綜合症/全前腦症、前腦無裂畸形 1 型
22q22.3 Deletion Syndrome/ Holoprosencephaly 1
117. Xp11.3 缺失綜合症 / X-性聯智障- 視網膜色素病變綜合症
Xp11.3 Deletion Syndrome/ X-linked Intellectual Disability-Retinitis Pigmentosa Syndrome
118. Xp11.23-p11.22 微重複綜合症
Microduplication Xp11.23-p11.22 Syndrome
119. Xp21 微缺失綜合症 / Xp21 鄰近基因缺失綜合症
Xp21 Microdeletion Syndrome/ Xp21 Contiguous Gene Deletion Syndrome
120. Xp22.2-p22.13重複綜合症
Xp22.2-p22.13 Duplication Syndrome
121. Xp22.3 微缺失綜合症
Xp22.3 Microdeletion Syndrome
122. Xp28 缺失綜合症/ 霧霧症-矮-畸面-性促素高-性腺功能低下綜合症
Xp28 Deletion/ Moyamoya Angiopathy-Short tature-Facial Dysmorphism-Hypergonadotropic Hypogonadism Syndrome
123. Xq21 缺失綜合症
Xq21 Deletion Syndrome
124. Xq22.3 端粒缺失綜合症
Xq22.3 Telomeric Deletion Syndrome
125. Xq27.3-q28重複綜合症[7]
Xq27.3-q28 Duplication Syndrome[7]
126. Xq28 鄰近基因缺失綜合症/ X-性聯肌小管-性器異常綜合症[7]
Xq28 Contiguous Gene Deletion Syndrome/ X-linked Myotubular Myopathy-Abnormal Genitalia Syndrome[7]

202106

1. 以上所列疾病雖然包含多個臨床表現，但缺乏足夠診斷 TAR 綜合症的臨床表現；2. 雖然外顯率不完整及基因臨床表現參差，但 DECIPHER 數據庫內顯示部分基因突變患者為病原體突變；3. 上表第5項及第7項分別於 RBM8A 基因缺失牽連；4. 相同區域的微缺失，但有不同病徵或臨床表現；5. 文獻指最小為1.2M b (PMC：4988255) 亦發現有患者於 4q24至4q28.3 出現 23Mb 缺失；6. Sotos 綜合症 1 型：5q35 缺失；Sotos 綜合症 2 型 及 3 型：19號染色體單基因突變。以上所列只包括 Sotos 綜合症 1 型 ；7. DECIPHER 只發現一位患；8. WAGRO 綜合症 WAGR 綜合症分別於一個額外的腦源性神經營養因子缺失。一些數據庫會把兩者視為相同的綜合症；9. 基於小胖威利綜合症及天使綜合症的發病獨特性，未能以 DECIPHER 記錄的缺失大小作分；10. 以上所列包含反覆出現的拷貝數變異；11. 大分患缺失約為 550kb；12. 大分患者重複約為 550kb；13. 以上所列疾病為非臨床可識別的臨床疾病。有重複的患者沒有病徵 。

1. Syndrome with variable clinical manifestations but without the clinical picture of Thrombocytopenia Absent Radius (TAR) Syndrome；2. Although penetrance is incomplete and gene expression is variable a number of patients with mutations classified as pathogenic in DECIPHER can be found；3. The difference between item #5 and #7 is the i nvolve ment of deletion in RBM8A gene；4. Same region microdeletion but with variable syndrome or clinical manifestation；5. Smallest size described inliterature is 1.2 Mb (PMC : 4988255) while one patient with 23 Mb deletion from 4q24 to 4q28.3 was also found；6. Sotos Syndrome 1 : 5q35 deletion Sotos Syndrome 2 & 3: single gene mutation in Chr 19. Only Sotos Syndrome 1 is；7. Only one patient was found on DECIPHER；8. WAGRO Syndrome differs from WAGR Syndrome by including the deletion of an additional gene BDNF. Some databases treat them as the same；9. Cannot distinguish PWS and AS from deletion size in DECIPHER due to their unique pathogenesis；10. Involves recurrent CNV；11. Most patients have deletion with size around 550 kb；12. Most patients have duplication with size around 550 kb；13. This is not a clinically recognizable disorder. Some people having the duplication do not have symptoms.