PureTarget ：全面解析高挑戰性基因區域，打造靈活擴展的定序方案

PureTarget 的 HiFi 長讀定序流程不需 PCR 步驟，能精準讀取並保留天然 DNA 的甲基化（methylation）資訊，避免 PCR 所造成的偏差。搭配專為「重複序列擴增型（repeat expansion）神經疾病」與「帶因者篩檢（carrier screening）」設計的即用型目標基因 panel，適用於檢測包括 Huntington disease（亨丁頓舞蹈症）、 Fragile X syndrome (X染色體脆折症）、 ALS（肌萎縮性側索硬化症）等多種高挑戰性基因。協助實驗室擺脫繁瑣的傳統分型流程，將所有高難度基因一次整合至現代化的長讀定序平台中。

原廠網頁

PacBio

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Off-the-shelf panels

PureTarget repeat expansion panel 2.0

最新版的 repeat expansion panel 涵蓋 38 個目標，針對多種與神經系統疾病相關、常見的重複序列進行檢測。新版中新增的目標以**粗體**標示。

Disease	Targets
Spinocerebellar ataxia (SCA)	*ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8, ATXN10, CACNA1A, PPP2R2B, TBP* ***BEAN1, DAB1, FGF14, NOP56, ZFHX3***
Fragile-X disease (FXS)	*FMR1*
Fragile X syndrome, FRAXE type	***AFF2***
Intellectual disability associated with fragile site FRA2A	***AFF3***
Frontotemporal dementia (FTD), amyotrophic lateral sclerosis (ALS)	*C9orf72*
Friedreich's ataxia (FRDA)	*FXN*
Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome (CANVAS)	*RFC1*
Neuronal intranuclear inclusion disease, Alzheimer disease and parkinsonism phenotype (NIID)	***NOTCH2NLC***
Myotonic dystrophy (DM)	*DMPK, CNBP*
Huntington disease (HD)	*HTT*
Huntington's disease-like type2 (HDL2)	***JPH3***
Fuchs endothelial corneal dystrophy 3 (FECD3)	*TCF4*
Kennedy Disease, Spinal and bulbar muscular atrophy, (SBMA)	*AR*
Oculopharyngeal muscular dystrophy (OPMD)	*PABPN1*
Oculopharyngodistal myopathy (OPDM)	***ABCD3, GIPC1, LRP12, RILPL1***
Syndactyly (SD5)	***HOXD13***
Congenital central hypoventilation syndrome (CCHS)	***PHOX2B***
Creutzfeldt-Jakob disease (CJD)	***PRNP***
Progressive Myoclonic Epilepsy Type 1 (EPM1) Unverricht-Lundborg Disease (ULD)	***CSTB***
Familial adult myoclonic epilepsy type 1 (FAME)	***SAMD12***

PureTarget carrier panel

這個 carrier screening panel 整合了 12 個包含隱性致病變異的基因，不再需要依賴非 NGS 方法進行輔助分型。PureTarget 是一個流程精簡的檢測方案，可與常規短讀定序的 carrier screening panel 搭配使用，以擴展攜帶者篩檢的基因目標範圍。

Disease	Targets
Hemophilia A	*F8*
Friedreich ataxia (FRDA)	*FXN*
Fragile-X disease (FXS)	*FMR1*
Congenital adrenal hyperplasia	*CYP21A2*
Classical-like Ehlers-Danlos syndrome	*TNXB*
Alpha thalassemia	*HBA1/2*
Gaucher disease	*GBA*
Spinal muscular atrophy	*SMN1/2*
Early-infantile epileptic encephalopathy (EIEE1) and Partington syndrome (PRTS)	*ARX*
Sickle Cell Anemia and Beta thalassemia	*HBB*
X-linked retinitis pigmentosa	*RPGR*
Fragile X syndrome, FRAXE type	*AFF2*

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