# 2024년도
# 국가통합 바이오 빅데이터 구축 사업

## ( 301개 희귀질환군 목록)

2024. 5.

국가통합바이오빅데이터구축사업단

| No | 소분류 |
|---|---|
| 1 | Familial breast and or ovarian cancer |
| 2 | Intellectual disability |
| 3 | Hereditary neuropathy (including Charcot-Marie-Tooth disease) |
| 4 | Congenital muscular dystrophy |
| 5 | Dilated Cardiomyopathy |
| 6 | Hypertrophic Cardiomyopathy |
| 7 | Retinal disorders |
| 8 | Neurofibromatosis Type 1 |
| 9 | Early onset or syndromic epilepsy |
| 10 | Hereditary ataxia and cerebellar anomalies |
| 11 | Hereditary spastic paraplegia |
| 12 | Congenital malformation and dysmorphism syndromes - microarray and sequencing |
| 13 | Congenital myopathy |
| 14 | Epilepsy plus other features |
| 15 | Limb girdle muscular dystrophy |
| 16 | Epileptic encephalopathy |
| 17 | Unexplained skeletal dysplasia |
| 18 | Growth failure in early childhood |
| 19 | Syndromic congenital heart disease |
| 20 | Malformations of cortical development |
| 21 | Thoracic aortic aneurysm or dissection |
| 22 | Primary immunodeficiency |
| 23 | Classical tuberous sclerosis |
| 24 | Familial Hypercholesterolaemia |
| 25 | Cystic renal disease |
| 26 | Severe microcephaly |
| 27 | Autism |
| 28 | Mitochondrial disorders |
| 29 | Primary lymphoedema |
| 30 | Infantile enterocolitis and monogenic inflammatory bowel disease |
| 31 | Multiple Tumours |
| 32 | Proteinuric renal disease |
| 33 | Disorders of sex development |
| 34 | Childhood onset dystonia, chorea or related movement disorder |
| 35 | Other rare neuromuscular disorders |
| 36 | Moyamoya disease |
| 37 | VACTERL-like phenotypes |
| 38 | Cytopenia and pancytopenia |
| 39 | Inherited bleeding and or platelet disorders |
| 40 | Colorectal cancer pertinent cancer susceptibility |
| 41 | Pulmonary arterial hypertension |
| 42 | Classical Ehlers-Danlos Syndrome |
| 43 | Arthrogryposis |
| 44 | Undiagnosed neurocutaneous disorders |
| 45 | Undescribed renal disorders |
| 46 | Early onset dystonia |
| 47 | Familial pulmonary fibrosis |
| 48 | Inherited optic neuropathies |
| 49 | Brain cancer pertinent cancer susceptibility |
| 50 | Early onset and familial Parkinson's Disease |

| No | 소분류 |
|---|---|
| 51 | Cardiac arrhythmias |
| 52 | Cytopenia (NOT Fanconi anaemia) |
| 53 | Noonan syndrome |
| 54 | Genetic Epilepsies with Febrile Seizures Plus |
| 55 | Primary ciliary dyskinesia |
| 56 | Rare anaemia |
| 57 | Pancreatitis |
| 58 | Pigmentary skin disorders |
| 59 | Adult onset dystonia, chorea or related movement disorder |
| 60 | Neonatal cholestasis |
| 61 | Long QT syndrome |
| 62 | Renal cancer pertinent cancer susceptibility |
| 63 | Renal tract calcification (or Nephrolithiasis or nephrocalcinosis) |
| 64 | Significant early-onset obesity with or without other endocrine features and short stature |
| 65 | Syndromic cleft lip and or cleft palate |
| 66 | Rhabdomyolysis and metabolic muscle disorders |
| 67 | Silver Russell syndrome |
| 68 | Neuroendocrine cancer pertinent cancer susceptibility |
| 69 | Familial young-onset non-insulin-dependent diabetes |
| 70 | Coarse facial features including Coffin-Siris-like disorders |
| 71 | Adult onset leukodystrophy |
| 72 | Hypnotic infant with a likely central cause/Floppy infant with a likely central cause |
| 73 | Early onset or familial intestinal pseudo obstruction |
| 74 | Craniosynostosis syndromes |
| 75 | Cerebellar hypoplasia |
| 76 | Congenital anaemias |
| 77 | Familial Focal Epilepsies |
| 78 | Myotonia congenita |
| 79 | Monogenic venous thrombosis |
| 80 | Noonan syndrome plus other features |
| 81 | Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy |
| 82 | Congenital hearing impairment |
| 83 | Amyotrophic lateral sclerosis or motor neuron disease |
| 84 | Congenital hypothyroidism |
| 85 | Tubulointerstitial kidney disease |
| 86 | Inborn errors of metabolism |
| 87 | Cerebral malformation |
| 88 | Familial haematuria |
| 89 | Unexplained paediatric onset end-stage renal disease |
| 90 | Osteogenesis imperfecta |
| 91 | Cerebral vascular malformations |
| 92 | Wilson disease |
| 93 | Isolated hemihypertrophy or macroglossia |
| 94 | Kabuki syndrome |
| 95 | Familial and multiple pulmonary arteriovenous malformations |
| 96 | Hereditary haemorrhagic telangiectasia |
| 97 | Vascular skin disorders |
| 98 | Familial exudative vitreoretinopathy |
| 99 | Familial Genetic Generalised Epilepsies |
| 100 | Ectodermal dysplasia |

| No | 소분류 |
|---|---|
| 101 | Non-syndromic familial congenital anorectal malformations |
| 102 | Sotos syndrome |
| 103 | Endometrial cancer pertinent cancer susceptibility |
| 104 | Multiple Epiphyseal Dysplasia |
| 105 | Congenital adrenal hypoplasia |
| 106 | Corneal dystrophy and abnormalities |
| 107 | Joubert syndrome |
| 108 | Glycogen storage disease |
| 109 | Skeletal Muscle Channelopathies |
| 110 | Structural eye disease (including Anophthalmia or microphthamia) |
| 111 | Childhood solid tumours cancer susceptibility |
| 112 | Cataracts |
| 113 | Pituitary hormone deficiency |
| 114 | Pain channelopathies |
| 115 | Intestinal failure |
| 116 | Lysosomal storage disorder |
| 117 | Currarino triad |
| 118 | Familial or syndromic hypoparathyroidism |
| 119 | Inherited susceptibility to acute lymphoblastoid leukaemia (ALL) |
| 120 | Albright hereditary osteodystrophy, pseudohypoparathyroidism and pseudopseudohypoparathyroidism |
| 121 | Complex Parkinsonism (including pallido-pyramidal syndromes) |
| 122 | IUGR and IGF abnormalities |
| 123 | Hypogonadotropic hypogonadism |
| 124 | Adult onset neurodegenerative disorder |
| 125 | Childhood onset leukodystrophy |
| 126 | Diabetes with additional phenotypes suggestive of a monogenic aetiology |
| 127 | Inherited macular dystrophy |
| 128 | Congenital myaesthenia |
| 129 | Renal tubular acidosis |
| 130 | Atypical haemolytic uraemic syndrome |
| 131 | Thyroid cancer pertinent cancer susceptibility |
| 132 | Hydrocephalus |
| 133 | Hypophosphataemia or rickets |
| 134 | Distal myopathies |
| 135 | Haematological malignancies cancer susceptibility |
| 136 | Inherited white matter disorders |
| 137 | Brugada syndrome |
| 138 | Classical Beckwith-Wiedemann syndrome |
| 139 | Hyperthyroidism |
| 140 | Catecholaminergic Polymorphic Ventricular Tachycardia |
| 141 | Neuro-endocrine tumours- PCC and PGL |
| 142 | Ear malformations with hearing impairment |
| 143 | Polycystic liver disease |
| 144 | Arrhythmogenic Right Ventricular Cardiomyopathy |
| 145 | Atypical Beckwith-Wiedemann syndrome |
| 146 | Iron metabolism disorders (NOT common HFE mutations) |
| 147 | Ketotic hypoglycaemia |
| 148 | Brain channelopathy |
| 149 | Hyperinsulinism |
| 150 | Periodic fever syndromes and amyloidosis |

| No | 소분류 |
|---|---|
| 151 | Infantile nystagmus |
| 152 | Familial Hirschsprung Disease |
| 153 | Familial hyperparathyroidism |
| 154 | Epidermolysis bullosa and congenital skin fragility |
| 155 | Fetal hydrops |
| 156 | Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT) |
| 157 | Gastrointestinal epithelial barrier disorders |
| 158 | Palmoplantar keratodermas |
| 159 | Juvenile dermatomyositis |
| 160 | Inherited pancreatic cancer |
| 161 | Familial congenital heart disease |
| 162 | Generalised pustular psoriasis |
| 163 | Bardet-Biedl Syndrome |
| 164 | Bilateral microtia |
| 165 | Non-Fanconi anaemia |
| 166 | Stickler syndrome |
| 167 | Familial cerebral small vessel disease |
| 168 | Familial prostate cancer |
| 169 | Paediatric motor neuronopathies |
| 170 | Familial tumour syndromes of the central and peripheral nervous system |
| 171 | Multiple monogenic benign skin tumours |
| 172 | Exceptionally young adult onset cancer |
| 173 | Developmental macular and foveal dystrophy |
| 174 | Lipoedema disease |
| 175 | Cardio-facio-cutaneous syndrome |
| 176 | Hyperammonaemia |
| 177 | Alstrom syndrome |
| 178 | Osteopetrosis |
| 179 | Segmental overgrowth disorders |
| 180 | Other peroxisomal disorders |
| 181 | Dilated Cardiomyopathy and conduction defects |
| 182 | Familial IgA nephropathy and IgA vasculitis |
| 183 | Unexplained sudden death in the young |
| 184 | Autosomal recessive congenital ichthyosis |
| 185 | Sporadic aniridia |
| 186 | Amyloidosis with no identifiable cause |
| 187 | Head and neck cancer pertinent cancer susceptibility |
| 188 | Kyphoscoliotic Ehlers-Danlos syndrome |
| 189 | Primary Microcephaly - Microcephalic Dwarfism Spectrum |
| 190 | Glaucoma (developmental) |
| 191 | Albinism or congenital nystagmus |
| 192 | Primary membranoproliferative glomerulonephritis |
| 193 | Early onset dementia |
| 194 | Multiple endocrine tumours |
| 195 | Lactic acidosis |
| 196 | Left Ventricular Noncompaction Cardiomyopathy |
| 197 | Rare genetic inflammatory skin disorders |
| 198 | Paroxysmal central nervous system disorders |
| 199 | Severe multi-system atopic disease with high IgE |
| 200 | Hypocalciuric hypercalcaemia |

| No | 소분류 |
|---|---|
| 201 | Choanal atresia |
| 202 | Auditory Neuropathy Spectrum Disorder |
| 203 | Amelogenesis imperfecta |
| 204 | Structural basal ganglia disorders |
| 205 | Extreme early-onset hypertension |
| 206 | Familial rhabdomyosarcoma or sarcoma |
| 207 | Familial cicatricial alopecia |
| 208 | Immunodysregulation Polyendocrinopathy and Enteropathy, X-Linked |
| 209 | Chondrodysplasia punctata |
| 210 | Idiopathic ventricular fibrillation |
| 211 | Paediatric or syndromic cardiomyopathy |
| 212 | Neonatal diabetes (diagnosed less than 6 months) |
| 213 | Non-CF bronchiectasis |
| 214 | Paediatric congenital malformation-dysmorphism-tumour syndromes |
| 215 | Lipoprotein lipase deficiency |
| 216 | Progressive cardiac conduction disease |
| 217 | Congenital fibrosis of the extraocular muscles |
| 218 | Holoprosencephaly |
| 219 | Epidermodysplasia verruciformis |
| 220 | Xeroderma Pigmentosum-like disorders |
| 221 | Monogenic diabetes |
| 222 | Rare multisystem ciliopathy disorders |
| 223 | PHACE(S) syndrome |
| 224 | Familial tumoral calcinosis |
| 225 | Blepharophimosis ptosis and epicanthus inversus |
| 226 | Pyruvate dehydrogenase (PDH) deficiency |
| 227 | Cone Dysfunction Syndrome |
| 228 | Peroxisomal biogenesis disorders |
| 229 | Ductal plate malformation |
| 230 | Acute intermittent porphyria |
| 231 | Familial primary spontaneous pneumothorax |
| 232 | Elastin-related phenotypes |
| 233 | Intracerebral calcification disorders |
| 234 | Familial disseminated superficial actinic porokeratosis |
| 235 | Multi-organ autoimmune diabetes |
| 236 | Vici Syndrome and other autophagy disorders |
| 237 | Simpson-Golabi-Behmel syndrome |
| 238 | Thoracic dystrophies |
| 239 | Rod-cone dystrophy |
| 240 | Primary pigmented nodular adrenocortical disease |
| 241 | Variegate porphyria |
| 242 | Cerebral folate deficiency |
| 243 | Neuronal ceroid lipofuscinosis |
| 244 | Short QT syndrome |
| 245 | Glucokinase-related fasting hyperglycaemia |
| 246 | Familial dysalbuminaemic hyperthyroxinaemia |
| 247 | Inherited predisposition to GIST |
| 248 | Familial rhabdoid tumours |
| 249 | Severe hypertriglyceridaemia |
| 250 | Parathyroid cancer |

| No | 소분류 |
|---|---|
| 251 | Early onset familial premature ovarian insufficiency |
| 252 | Radial dysplasia |
| 253 | Calcium-sensing receptor phenotypes |
| 254 | Familial hidradenitis suppurativa |
| 255 | Familial Meniere Disease |
| 256 | Ocular coloboma |
| 257 | Insulin resistance (including lipodystrophy) |
| 258 | Carney complex |
| 259 | Non-acute porphyrias |
| 260 | Familial melanoma |
| 261 | Hereditary erythrocytosis |
| 262 | Autosomal dominant deafness |
| 263 | Disseminated non-tuberculous mycobacterial infection |
| 264 | Erythropoietic protoporphyria, mild variant |
| 265 | Hydroa vacciniforme |
| 266 | Legius syndrome |
| 267 | Genomic Advances in Sepsis (GAinS) study |
| 268 | Weaver syndrome |
| 269 | Laterality disorders and isomerism |
| 270 | Generalised arterial calcification in infancy |
| 271 | Barth syndrome |
| 272 | Pontine tegmental cap dysplasia |
| 273 | Schizophrenia plus additional features |
| 274 | Ichthyosis and erythrokeratoderma |
| 275 | Respiratory ciliopathies including non-CF bronchiectasis |
| 276 | Genodermatoses with malignancies |
| 277 | Cutaneous photosensitivity |
| 278 | Familial hemifacial microsomia |
| 279 | Non-syndromic hypotrichosis |
| 280 | Cockayne syndrome |
| 281 | Bladder cancer pertinent cancer susceptibility |
| 282 | Congenital disorders of glycosylation |
| 283 | Rod Dysfunction Syndrome |
| 284 | Inherited predisposition to acute myeloid leukaemia (AML) |
| 285 | Kleine-Levin syndrome and other inherited sleep disorders |
| 286 | Primary hyperaldosteronism |
| 287 | Costello syndrome |
| 288 | Familial non-syndromic cleft lip and or familial cleft palate |
| 289 | Balanced translocations with an unusual phenotype |
| 290 | Milroy disease |
| 291 | Gilbert syndrome |
| 292 | Peeling skin syndrome |
| 293 | Vein of Galen malformation |
| 294 | Rhomboencephalosynapsis |
| 295 | Severe familial anorexia |
| 296 | Surfactant deficiency |
| 297 | Neurotransmitter disorders |
| 298 | Lymphoedema distichiasis |
| 299 | Inherited non-medullary thyroid cancer |
| 300 | LEOPARD syndrome |
| 301 | Meige disease |