Kinetics 毅力醫護健康集團

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嬰兒基因篩檢項目
Newborn Genetic Screening Items

Number	Disease / Syndrome / Overview
1	 **CHROMOSOME 1p36 DELETION SYNDROME** Cause: Deletion of chromosome 1p36 Symptom: Typical craniofacial features, developmental delay, intellectual disability, structural brain abnormalities, congenital heart defect, eye/vision problem, hearing loss and seizures
2	 **CHROMOSOME 1p32-p31 DELETION SYNDROME** Cause: Deletion of contiguous gene on chromosome 10p32 - p31 Symptom: Developmental delay, intellectual disability, hydrocephalus, seizures, inguinal hernia, urinary incontinence, cryptorchidism, muscular hypotonia and polydactyly
3	 **OMPHALOCELE, AUTOSOMAL** Cause : Duplication of genes on chromosome 1p31 Symptom: Umbilical hernia, precocious labor, prematurity and mild developmental delay
4	 **CHROMOSOME 1q21.1 DUPLICATION SYNDROME** Cause: Duplication of one or more genes within the chromosome 1q21.1 Symptom: Impaired communication and socialization skills, developmental delay in speech and language, attention deficit /hyperactive disorder, recurrent seizures (epilepsy) and malformations of heart
5	 **CHROMOSOME 1q21.1 DELETION SYNDROME, 1.35-MB** Cause: Deletion of contiguous gene on chromosome 1 Symptom: Developmental delay and intellectual disability, physical abnormalities (microcephaly, short statue), eye problems(cataracts), seizures and weak muscle tone (hypotonia)
6	 **CHROMOSOME 1q41-q42 DELETION SYNDROME** Cause : Deletion of a contiguous gene on chromosome 1q41-q42 Symptom: Developmental delay, lung hypoplasia, diaphragmatic hernia, cryptorchidism, holoprosencephaly, frontal bossing and deep-set eyes, midline cleft lip
7	 **CHROMOSOME 2p16.3 DELETION SYNDROME** Cause: Deletion at 2p16.3 Symptom: Autism, schizophrenia, developmental delay and intellectual disability
8	 **CHROMOSOME 2p16.1-p15 DELETION SYNDROME** Cause: Deletion of contiguous gene on chromosome 2p16.1-p15 Symptom: Neurodevelopmental disorder, dysmorphic feature (microcephaly, bitemporal narrowing, smooth and long philtrum and hypertelorism)
9	 **CHROMOSOME 2q31.1 DUPLICATION SYNDROME** Cause : Duplication of genes at chromosome 2q31.1 Symptom: Short statue and distinctively short forearms
10	 **CHROMOSOME 2q31.2 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 2q31.2 Symptom: Severe mental retardation, absence of speech, sleep disturbances, macrocephaly, micrognathia, strabismus, proptosis, scoliosis and pectus excavatum
11	 **CHROMOSOME 2q35 DUPLICATION SYNDROME** Cause: Duplications on chromosome 2q35 Symptom: Syndactyly and craniosynostosis
12	 **CHROMOSOME 2q37 DELETION SYNDROME** Cause: Deletion of several genes on chromosome 2q37.2 Symptom: Brachydactyly, short stature, intellectual disability and behavioral abnormalities
13	 **SETD5 SYNDROME** Cause : *SETD5* gene mutation on chromosome 3 Symptom: Intellectual disability, autism, poor speech development, brachycephaly, low-set ears, depressed nasal bridge, long philtrum, and thin upper lip
14	 **CHROMOSOME 3q13.31 DELETION SYNDROME** Cause: Deletion of chromosome 3q13.31 Symptom: Developmental delay, language and motor delays, mild to moderate cognitive delays, autism spectrum disorders, macrocephaly, downslanting palpebral fissures, bulbous nose and short philtrum
15	 **DANDY-WALKER MALFORMATION, INCLUDED; DWM, INCLUDED** Cause: Deletions on chromosome 3q Symptom: Cystic dilation of the fourth ventricle, hypoplasia and upward rotation of the cerebellar vermis, delayed motor development, hypotonia, ataxia, mental retardation and hydrocephalus

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16	 **CHROMOSOME 3q29 DUPLICATION SYNDROME** Cause: Microduplication of chromosome 3q29 Symptom: Mild to moderate mental retardation, microcephaly, round face, bulbous nose, short or downslanting palpebral fissures
17	 **CHROMOSOME 3q29 DELETION SYNDROME** Cause: Deletion of contiguous gene on chromosome 3q29 Symptom: Mild to moderate mental retardation, slightly dysmorphic facial features (long and narrow face, short philtrum, cleft lip and palate) and horseshoe kidney
18	 **WOLF-HIRSCHHORN SYNDROME; WHS** Cause: Deletion of chromosome 4p16.3 Symptom: "Greek warrior helmet" appearance, delayed growth and development, intellectual disability, seizures and weak muscle tone (hypotonia)
19	 **CHROMOSOME 4q21 DELETION SYNDROME** Cause: Deletion of genes on chromosome 4q21 Symptom: Neonatal muscularhypotonia, delayed speech, marked progressive growth restriction, and distinctive facial features (broad forehead, frontal bossing, hypertelorism, short philtrum and downturned corners of the mouth)
20	 **AXENFELD-RIEGER SYNDROME, TYPE 1** Cause: Mutation in *PITX2* gene on chromosome 4q25 Symptom: Abnormal development of the anterior segment of the eye, results in blindness from glaucoma and dental hypoplasia
21	 **CHROMOSOME 4q32.1-q32.2 TRIPLICATION SYNDROME** Cause: Triplication of a contiguous gene on chromosome 4q32.1-q32.2 Symptom: Delayed psychomotor development and variable mental retardation, macrocephaly, hypoplastic zygoma, long midface, short nose, wide nasal bridge, underdeveloped columella, downslanting and small palpebral fissures
22	 **FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1; FSHD 1** Cause: Deletion of the D4Z4 macrosatellite repeat on chromosome 4q35 Symptom: Contraction of face, upper arms and shoulder muscles, hearing loss and retinal capillary abnormalities
23	 **CRI-DU-CHAT SYNDROME** Cause: Deletion of short arm of chromosome 5 Symptom: High-pitched cat-like cry (In newborn), microcephaly, hypertelorism, low-set ears, severe psychomotor and mental retardation
24	 **CHROMOSOME 5p13 DUPLICATION SYNDROME** Cause: Duplication of several genes on chromosome 5p13 Symptom: Developmental delay and mental retardation, low birth weight and become overweight in adulthood, frontal bossing, large or broad forehead, bitemporal narrowing, short or slanted palpebral fissures, short philtrum, high-arched palate and low-set ears
25	 **CHROMOSOME 5q12 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 5q12 Symptom: Developmental delay or mental retardation, visual impairment, neonatal hypotonia and seizures, dysmorphic features (esotropia, thin palpebral fissures, large nose and nasal tip, brachycephaly and short arm)
26	 **CHROMOSOME 5q14.3 DELETION SYNDROME (MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE C; MEF2C)** Cause: Deletion of genes on chromosome 5q14.3 Symptom: Mental retardation, absence of speech, seizure, ADHD, hearing loss, dysmorphic facial features (small chin, short nose with anteverted nares and large open mouth)
27	 **CHROMOSOME 5q14.3 DELETION SYNDROME, DISTAL** Cause: Deletion of a contiguous gene on chromosome 5q14.3 Symptom: Mental retardation, epilepsy, no speech acquisition, delayed motor development, minor dysmorphic facial features (high forehead, depressed nasal bridge)
28	 **MENTAL RETARDATION, AUTOSOMAL DOMINANT 31; MRD31** Cause: Mutation in the *PURA* gene on chromosome 5q31 Symptom: Neurodevelopmental disorder, respiratory insufficiency, early-onset seizures, nonverbal, nonambulatory and strabismus
29	 **SOTOS SYNDROME 1; SOTOS1** Cause: Mutation in the *NSD1* gene or deletion in the 5q35 region Symptom: Excessively rapid growth, nonprogressive cerebral disorder with mental retardation, long face and a prominent forehead
30	 **AXENFELD-RIEGER SYNDROME, TYPE 3** Cause: Mutation in the *FOXC1* gene on chromosome 6p25 Symptom: Iris stromal hypoplasia, corectopia (anterior segment dysgenesis), heart anomalies and hearing loss

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31	**CHROMOSOME 6pter-p24 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 6 Symptom: Hypertelorism, anterior eye chamber abnormalities, palatal and dental abnormalities, hearing loss, congenital heart defects, neuronal defects, anomalies of the extremities and structural ear and nose anomalies
32	**COFFIN-SIRIS SYNDROME 1: CSS1** Cause: Mutation in the *ARID1B* gene on chromosome 6q25 Symptom: Mental retardation, craniofacial abnormalities, hypoplastic or absent fifth fingernails or toenails
33	**CHORDOMA, SUSCEPTIBILITY TO; CHDM** Cause: Duplication in the gene encoding brachyury Symptom: A chordoma is a rare type of cancerous tumor that can occur anywhere along the spine. Headache, double vision (diplopia)
34	**GREIG CEPHALOPOLYSYN DACTYLY SYNDROME; GCPS** Cause: Mutation in the *GLI3* gene on chromosome 7p14 Symptom: Frontal bossing, scaphocephaly, polydactyly and variable syndactyly and hypertelorism
35	**CHROMOSOME 7q11.23 DELETION SYNDROME, DISTAL, 1.2-MB** Cause: Deletion of a contiguous gene on chromosome 7q11.23 Symptom: Increased risk of epilepsy, learning difficulties, intellectual disabilities, neurobehavioral abnormalities, developmental delay, autism and heart defects
36	**WILLIAMS-BEUREN SYNDROME; WBS** Cause: Deletion of some part of chromosome 7q11.23 Symptom: Heart and blood vessel problems, abnormalities of connective tissue, increased calcium level (hypercalcemia) in infancy, distinctive facial feature (a broad forehead and wide mouth with full lips)
37	**WILLIAMS-BEUREN REGION DUPLICATION SYNDROME** Cause: Duplication of genes on chromosome 7q11.23 Symptom: Multisystem developmental delay (speech delay, motor skills delay), diaphragmatic hernia, cryptorchidism, heart defects, cognitive defects(mental retardation, autism and ADHD)
38	**MONOSOMY 7 OF BONE MARROW** Cause : Loss of chromosome 7 or deletion of genes on chromosome 7q Symptom: Myelodysplasia, acute myelogenous leukemia, thrombocytopenia and erythrocyte macrocytosis
39	**CURRARINO SYNDROME** Cause: Mutation in the *HLXB9* homeobox on chromosome 7q36 Symptom: Sickle-shaped sacrum by partial sacral agenesis, presacral mass, anorectal malformation, perianal sepsis and genitourinary anomalies
40	**SCHIZOPHRENIA 16; SCZD16** Cause: Duplication of a region on chromosome 7q36.3 Symptom: Hallucinations (hearing voices), delusions (often bizarre or persecutory in nature), lacking of motivation and disorganized thinking and speech
41	**CHROMOSOME 8q12.1-q21.2 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 8q12.1-q21.2 Symptom: Ear and renal defects, impairment in horizontal eye movement, hydrocephalus and aplasia of the trapezius muscle
42	**MESOMELIA-SYNOSTOSES SYNDROME** Cause: Microdeletion on chromosome 8q13 Symptom: Shortening of mesomelic limb, ptosis, hypertelorism, palatal abnormality, congenital heart disease and ureteral anomalies
43	**CHROMOSOME 8q21.11 DELETION SYNDROME** Cause: Deletion on chromosome 8q21.11 Symptom: Intellectual disability, facial dysmorphic features (round face with full cheeks, high forehead, corneal opacities, wide nasal bridge, a short philtrum, downturned corners of the mouth), short neck, hearing loss and syndactyly
44	**NABLUS MASK-LIKE FACIAL SYNDROME; NMLFS** Cause : Deletion of a contiguous gene at chromosome 8q22.1 Symptom: Abnormal hair pattern with an upswept frontal hairline, sparse arched eyebrows, flat and broad nose, long philtrum, small chin, developmental delay and cryptorchidism
45	**CHROMOSOME 8q22.1 DUPLICATION SYNDROME** Cause: Microduplication of chromosome 8q22.1 Symptom: Skeletal disorder, short stature, spinal cord compression, chronic joints pain and scleroderma-like skin change
46	**TRICHORHINOPHALANGEAL SYNDROME, TYPE II; TRPS2** Cause: Deletion of a contiguous gene on chromosome 8q24.1 Symptom: Multiple dysmorphic features (spare scalp hair, bulbous nose, defects on fingers), mental retardation, cognitive impairment, delayed speech, hearing loss and seizures
47	**MENTAL RETARDATION, AUTOSOMAL DOMINANT 2; MRD2** Cause: Disruption of the DOCK8 gene on chromosome 9p24 Symptom: Mental retardation, developmental disability, mild dysmorphic features, delayed speech and psychomotor development and seizures.
48	**46,XY SEX REVERSAL 4; SRXY4** Cause: Deletion on chromosome 9p Symptom: Showed female external genitalia and uterus, immature testicular tissue
49	**CHROMOSOME 9p DELETION SYNDROME** Cause: Partial deletion of the short arm of chromosome 9 Symptom: Trigonocephaly, prominent forehead, broad flat nasal bridge, anteverted nares, malformed external ears, mental retardation and cardiac murmurs
50	**EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY-4: EIEE4** Cause: Mutation in the *STXBP1* gene on chromosome 9q34.1 Symptom: Tonic seizures, spastic quadriplegia and delayed psychomotor development
51	**KLEEFSTRA SYNDROME** Cause: Mutation in the *EHMT1* gene on chromosome 9q34.3 Symptom: Severe mental retardation, hypotonia, microcephaly, epileptic seizures, flat face with hypertelorism, anteverted nares, everted lower lip, carp mouth with macroglossia and heart defects
52	**DIGEORGE SYNDROME / VELOCARDIOFACIAL SYNDROME COMPLEX 2; DGS2** Cause: Deletions of chromosome 10p Symptom: Heart defect, T-cell deficiency, hypocalcemia, hypoparathyroidism, deafness and renal dysplasia, immune deficiency, cleft palate, facial dysmorphia, developmental delay and microcephaly
53	**CHROMOSOME 10q22.3-q23.2 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 10q22.3-q23.2 Symptom: Developmental delay, speech delay, multiple congenital anomalies, scoliosis, hypertelorism, low-set ears and flat nasal bridge
54	**SPLIT-HAND/FOOT MALFORMATION 3; SHFM3** Cause: Duplication of a contiguous gene on chromosome 10q24 Symptom: Limb malformation, syndactyly, aplasia or hypoplasia of the phalanges and mental retardation
55	**CHROMOSOME 10q26 DELETION SYNDROME** Cause: Partial deletion of chromosome 10q Symptom: Developmental delay, moderate mental retardation, short stature, microcephaly, long philtrum, small pointed jaw, strabismus and cryptorchidism
56	**WILMS TUMOR, ANIRIDIA, GENITOURINARY ANOMALIES, MENTAL RETARDATION, AND OBESITY SYNDROME; WAGRO** Cause: Microscopic or submicroscopic deletion in chromosome 11p13-p12 Symptom:Child obesity, Wilms' tumor, aniridia, genitourinary anomalies and intellectual disability
57	**CHROMOSOME 11p13 DELETION SYNDROME, DISTAL** Cause: Deletion of a contiguous gene on chromosome 11p13 Symptom: Neurodevelopmental disorders, developmental delay with intellectual disability, speech and language disorder, congenital eye malformations, autism or autistic features
58	**WILMS TUMOR, ANIRIDIA, GENITOURINARY ANOMALIES. AND MENTAL RETARDATION SYNDROME; WAGR** Cause: Deletions on chromosome 11 Symptom: Wilms' tumor (a rare form of kidney cancer), aniridia (an absence of the colored part of the eye), anomalies of the genitalia and urinary tract and intellectual disability
59	**POTOCKI-SHAFFER SYNDROME** Cause: Deletion of a contiguous gene on chromosome 11p11.2 Symptom: Disorder on development of the bones, nerve cells in the brain and other tissues. Distinctive facial features, developmental delay, intellectual disability, multiple exostoses, enlarged parietal foramina, brachycephaly and short philtrum
60	**OSTEOGENESIS IMPERFECTA, TYPE XVI; 0116** Cause: Deletion of a contiguous gene on chromosome 11p11 Symptom: Brittle bone disorder, osteogenesis imperfecta, fractures in utero, short gestational age, constipation, bronchopneumonia, liver hypertrophy

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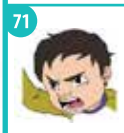
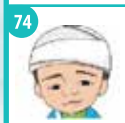
61	 **THROMBOCYTOPENIA, PARIS-TROUSSEAU TYPE; TCPT** Cause: Deletion of a contiguous gene on chromosome 11 Symptom: Abnormality of the cardiovascular system, cognitive impairment, abnormal bleeding, clinodactyly, intellectual disability and finger syndactyly
62	 **JACOBSEN SYNDROME; JBS** Cause: Deletion at the end of chromosome 11q Symptom: Delayed development (speech and motor skills), cognitive impairment and learning difficulties, compulsive behavior, short attention span and easy distractibility, bleeding disorder and heart defects
63	 **CHROMOSOME 13q14 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 13q14 Symptom: Retinoblastoma, mentalimpairment, hypotonia, diplegia, characteristic facial features (high forehead, hypertelorism, bulbous tip of the nose, large mouth with thin upper lip and long philtrum)
64	 **MICROCORIA, CONGENITAL** Cause: Deletion of a contiguous gene at chromosome 13q32 Symptom: Bilateral small pupils that result from an underdevelopment of the dilator pupillae muscle of the iris. Glaucoma and myopia
65	 **PATAU SYNDROME (TRISOMY 13)** Cause: Trisomy 13 having three copies of chromosome 13 Symptom: Severe intellectual disability and physical abnormalities in many parts of the body, heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes, extra fingers or toes, a cleft lip, a cleft palate and weak muscle tone
66	 **CHROMOSOME 14q11-q22 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 14q11-q22 Symptom: Severe psychomotor retardation, microcephaly, poor growth, hypoplasia, dysmorphic features (widely-spaced eyes, broad flat nasal bridge, short nose, a long philtrum and small mouth)
67	 **FRIAS SYNDROME** Cause: Deletion of a contiguous gene on chromosome 14q22.1-q22.3 Symptom: Hypertelorism, proptosis, exophthalmia and short stature
68	 **CHROMOSOME 15q11.2 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 15q11.2 Symptom: Neuropsychiatric or neurodevelopmental problems, dysmorphic features (small face, cleft palate, strabismus, bulbous nose), autism and ADHD
69	 **PRADER-WILLI SYNDROME; PWS** Cause: Deletion of a contiguous gene on chromosome 15q11-q13 Symptom : Diminished fetal activity, obesity, muscular hypotonia, mental retardation, short stature, hypogonadotropic hypogonadism and small hands and feet
70	 **ANGELMAN SYNDROME** Cause: Deletions on chromosome 15q11.2-q13 or mutations Symptom: Neurodevelopmental disorder (mental retardation, movement or balance disorder), typical abnormal behaviors(bursts of laughter, happy disposition) and severe limitations in speech and language
71	 **CHROMOSOME 15q13.3 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 15 Symptom: Behavioral problems (poor attention span, hyperactivity, mood disorder, and aggressive and/or impulsive behavior), intellectual disability, seizures, psychiatric disorders, heart defects, minor abnormalities involving hands and arms
72	 **15q24 MICRODELETION SYNDROME (WITTEVEEN- KOLK SYNDROME)** Cause: Microdeletion in the SIN3A gene on chromosome 15q24 Symptom: Delayed development, speech delay, intellectual disability, autistic behavior and dysmorphic facial features (microcephaly, flat or depressed nasal bridge and long and smooth philtrum, etc.)
73	 **CHROMOSOME 15q25 DELETION SYNDROME** Cause: Deletion on chromosome 15q25.2-q25.3 Symptom: Poor growth, developmental delay, cleft palate, congenital diaphragmatic hernia, pectus excavatum, scoliosis and cryptorchidism
74	 **TETRASOMY 15q26** Cause: Tetrasomy of chromosome 15q26 - qter Symptom: Hydrocephalus, craniosynostosis, developmental delay, renal abnormalities and coarse asymmetric facies
75	 **CHROMOSOME 15q26-qter DELETION SYNDROM** Cause: Deletion of a contiguous gene on chromosome 15q26 - qter Symptom: Intrauterine growth retardation, microcephaly, micrognathia, renal anomalies, lung hypoplasia and delayed growth and development

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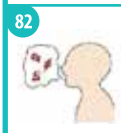
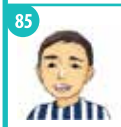
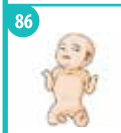
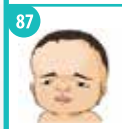
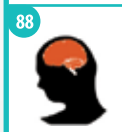
76	 **ALPHA-THALASSEMIA/MENTAL RETARDATION SYNDROME, CHROMOSOME 16-RELATED** Cause: Deletion of a contiguous gene in chromosome 16p Symptom: Anemia, mental retardation, developmental delay and stellate pattern of the iris
77	 **POLYCYSTIC KIDNEY DISEASE, INFANTILE SEVERE, WITH TUBEROUS SCLEROSIS; PKDTS** Cause: Deletion of a contiguous gene on chromosome 16p13.3 Symptom: Infantile polycystic kidneys, a angiomyolipomata and neuro tuberous sclerosis
78	 **CHROMOSOME 16p13.3 DELETION SYNDROME, PROXIMAL** Cause: Deletion of chromosome 16q13.3 Symptom: Mental retardation, a typical facies, broad thumbs and short stature, hypoplastic left heart, abnormal pulmonary lobulation, renal agenesis, neonatal seizures
79	 **CHROMOSOME 16p13.3 DUPLICATION SYNDROME** Cause: Duplication of a contiguous gene on chromosome 16p13.3 Symptom: Mental retardation and/or congenital anomalies, behavioral problems, midfacial hypoplasia in young children and a longer face in older individuals
80	 **CHROMOSOME 16p13.2 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 16p13.2 Symptom: Developmental delay, intellectual disability, autism spectrum disorder, aggressive behavior and absent speech with speech apraxia
81	 **CHROMOSOME 16p12.2-p11.2 DELETION SYNDROME, 7.1- TO 8.7-MB** Cause: Deletion of a contiguous gene on chromosome 16p12.2-p11.2 Symptom: Developmental delay, poor speech, mild dysmorphic features (flat, hypotonic face and low-set, posteriorly rotated ears)
82	 **CHROMOSOME 16p11.2 DELETION SYNDROME, 593-KB** Cause: Deletion of a small piece of chromosome 16 Symptom: Autism spectrum disorder, developmental delay and intellectual disability
83	 **CHROMOSOME 16p11.2 DUPLICATION SYNDROME** Cause : Duplication of a contiguous gene on chromosome 16p11.2 Symptom: Autism spectrum disorder, delayed development of speech and language, behavioral problems and ADHD
84	 **CHROMOSOME 16q22 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 16q22 Symptom: Craniofacial anomalies, a narrow thorax, hydrocephalus, high forehead, broad nasal bridge, hypertelorism, micrognathia, short neck and broad toes
85	 **KBG SYNDROME** Cause: Mutation in the *ANKRD11* gene on chromosome 16q24 Symptom: Developmental delay, intellectual disability, seizures, skeletal anomalies, short stature, macrodontia of the upper central incisors
86	 **CHROMOSOME 17p13.3, TELOMERIC, DUPLICATION SYNDROME** Cause : Duplication one or more genes on chromosome 17p13.3 Symptom: Limb abnormalities
87	 **CHROMOSOME 17p13.3, CENTROMERIC, DUPLICATION SYNDROME** Cause: Duplication of a contiguous gene on chromosome 17p13.3 Symptom: Hypotonia, mild to moderate psychomotor retardation, dysmorphic features (frontal bossing, low-set ears, broad nasal bridge, small nose and hypertelorism)
88	 **MILLER-DIEKER LISSENCEPHALY SYNDROME; MDLS** Cause: Deletion of a contiguous gene on chromosome 17p13.3 Symptom: Abnormal brain development, severe intellectual disability, developmental delay, seizures, abnormal muscle stiffness, hypotonia, feeding difficulties and microcephaly
89	 **CHROMOSOME 17p13.1 DELETION SYNDROME** Cause: Deletion of chromosome 17p13.1 Symptom: Severe mental retardation, very poor speech, small head, poor growth, broad and low-set nasal bridge, short philtrum, thin upper lip, pes planus and hypertelorism
90	 **SMITH-MAGENIS SYNDROME; SMS** Cause: Deletion in chromosome 17p11.2 Symptom: Mental retardation, hypotonia, speech delay, small ears, conductive hearing loss, esotropia and dental enamel dysplasia

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91	**POTOCKI-LUPSKI SYNDROME; PTLS** Cause: Duplication of a contiguous gene on chromosome 17p11.2 Symptom: Mental retardation, pervasive developmental disorders, ADHD, autism, short stature and structural cardiovascular abnormalities
92	**YUAN-HAREL-LUPSKI SYNDROME; YUHAL** Cause: Duplication of a contiguous gene on chromosome 17p12-p11.2 Symptom: A complex neurodevelopmental disorder, delayed walking, speech delay, behavioral difficulties, chronic constipation, foot deformities, joint laxity and congenital heart defects
93	**CHROMOSOME 17q11.2 DELETION SYNDROME, 1.4-MB** Cause: Deletion of chromosome 17q11.2 Symptom: Mild facial dysmorphism, hypertelorism, ptosis, mental retardation and/or for learning disabilities, plexiform neurofibromas
94	**CHROMOSOME 17q12 DUPLICATION SYNDROME** Cause: Duplication of a contiguous gene on chromosome 17q12 Symptom: Mild to moderate mental retardation, cognitive impairment, dolichocephaly, large and anteverted ears and micrognathia
95	**KOOLEN-DE VRIES SYNDROME** Cause: Deletion on chromosome 17q21.31 Symptom: Delayed psychomotor development, intellectual disability, hypotonia, characteristic facial features (broad forehead, long face, upslanting palpebral fissures, epicanthal folds, tubular nose with bulbous nasal tip) and friendly personality
96	**HYPERTRICHOSIS, CONGENITAL GENERALIZED, WITH OR WITHOUT GINGIVAL HYPERPLASIA; HTC3** Cause: Microdeletions or microduplications on chromosome 17q24.2-q24.3 Symptom: Hair overgrowth on the back and limbs, a broad flat nose (in adults), bulbous soft nose (in children), large ears with thick, long and hairy lobes and thickened lips
97	**CHROMOSOME 18p DELETION SYNDROME** Cause: Deletion of chromosome 18p Symptom: Mental retardation, growth retardation, craniofacial dysmorphism (round face, dysplastic ears, wide mouth, dental anomalies), and abnormalities of the limbs, genitalia, brain, eyes and heart
98	**TETRASOMY 18p** Cause: Tetrasomy of chromosome 18p Symptom: Neonatal feeding problems, developmental delay, cognitive impairment, microcephaly, seizures, cardiac defects, strabismus, and scoliosis/kyphosis
99	**CHROMOSOME 18q DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 18q Symptom: Short stature, microcephaly, carp-like mouth, external ear anomalies, mental retardation, hearing impairment and cardiac anomalies
100	**EDWARDS SYNDROME (TRISOMY 18)** Cause : Trisomy 18 having three copies of chromosome 18 Symptom:Slow growth before birth, low birth weight, heart defects, abnormalities of other organs, a small, abnormally shaped head (small jaw, and mouth) and clenched fist with overlapping fingers
101	**PIGMENTED NODULAR ADRENOCORTICAL DISEASE, PRIMARY, 4; PPNAD4** Cause: Duplication on chromosome 19p13 Symptom: Hypercortisolism, hypertension, skin fragility and osteoporosis
102	**CHROMOSOME 19q13.11 DELETION SYNDROME** Cause: Deletion of a contiguous gene on chromosome 19q13.11 Symptom: Minor facial anomalies (retrognathia, hypertelorism, broad nasal tip, a broad mouth with thin lips, broad gums, irregularly placed teeth) and cutis aplasia of the posterior frontal head
103	**ALAGILLE SYNDROME 1; ALGS1** Cause: Mutation in the *JAG1* gene on chromosome 20p12 Symptom: Neonatal jaundice, posterior embryotoxon, retinal pigmental changes, heart, pulmonic valvular stenosis, peripheral arterial stenosis, abnormal vertebrae (butterfly vertebrae), prominent forehead, deep-set eyes, and small and anteriorly pointed chin
104	**DOWN SYNDROME** Cause: Trisomy of all or a critical portion of chromosome 21 Symptom: Intellectual disability, characteristic facial appearance and weak muscle tone, heart defect, delayed development and behavioral problems, hypothyroidism, hearing and vision problems and leukemia
105	**CHROMOSOME 22q11.2 DUPLICATION SYNDROME** Cause: Microduplication on chromosome 22q11.2 Symptom: Mental retardation, speech impairment, ADHD, growth retardation, muscular hypotonia, hypertelorism, congenital heart malformation, visual and hearing impairment, microcephaly and ptosis
106	**DIGEORGE SYNDROME; DGS** Cause : Deletion of chromosome 22q11.2 Symptom: Hypocalcemia, thymic hypoplasia, immune deficit, cardiac malformations, upward and downward slanting eyes, short philtrum and relatively small mouth
107	**CAT EYE SYNDROME; CES** Cause : A small supernumerary chromosome 22 Symptom: Cat like eye, combination of coloboma of the iris and anal atresia, downslanting palpebral fissures, malformations of heart and renal, normal or nearnormal mental development
108	**CHROMOSOME 22q11.2 DELETION SYNDROME, DISTAL** Cause : Distal deletion of chromosome 22q11.2 Symptom: Developmental delay, intellectual disability with a significant language delay, congenital cardiovascular defects, diaphragmatic hernia and uterine didelphys
109	**VELOCARDIOFACIAL SYNDROME** Cause: Deletion of chromosome 22q11.2 Symptom: Learning disability, cardiac anomalies, umbilical hernia, hypospadias, cleft palate and slender hands and digits
110	**PHELAN-MCDERMID SYNDROME** Cause : Deletion of contiguous gene at chromosome 22q13 or mutation in the *SHANK3* gene Symptom: Delayed speech, autistic behavior, neonatal hypotonia, increased pain tolerance, sleep disturbance, long eyelashes, bulb nose and large and fleshy hands
111	**SYNPOLYDACTYLY 2; SPD2** Cause : Disruption of the fibulin-1 gene Symptom: Metatarsal fusion, metacarpal fusion and synpolydactyly between the third and fourth fingers
112	**CHROMOSOME 22q13 DUPLICATION SYNDROME** Cause : Heterozygous interstitial duplication in chromosome 22q13 Symptom: Developmental delays, learning problem, kleptomania, destructive behavior, dysmorphic features (upslanting palpebral fissures and epicanthus inversus)
113	**MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD** Cause : Mutation in the gene encoding dystrophin Symptom:X chromosome-linked disease, muscular dystrophy, usually present in early childhood and rapidly progressive and cardiomyopathy
114	**TURNER SYNDROME** Cause: Chromosome number abnormality (45,X) Symptom: Short stature, loss of ovarian function, infertile, lymphedema, webbed neck and heart defect
115	**KLINEFELTER SYNDROME, XXY** Cause : Extra copy of X chromosome (47, XXY) Symptom: One in 600 newborn males, taller than peers, breast enlargement, small penis and microorchidism, hypospadias, and infertility
116	**TRIPLE-X SYNDROME** Cause: Additional X chromosome:XXX Symptom : Female. Delayed development of motor skills, learning disabilities, speech delay, difficulty in social interaction. Mostly, normal sexual development, able to have children. Occasionally early puberty, premature ovarian failure or ovarian abnormality.
117	**JOUBERT SYNDROME** Cause: Mutations in at least ten genes Symptom: One in 2,500 newborn girls worldwide, brain abnormality (molar tooth sign), cerebellarvermian aplasia, hypotonia, breathing abnormalities, developmental delay and eye abnormalities

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