Essential genetics 5

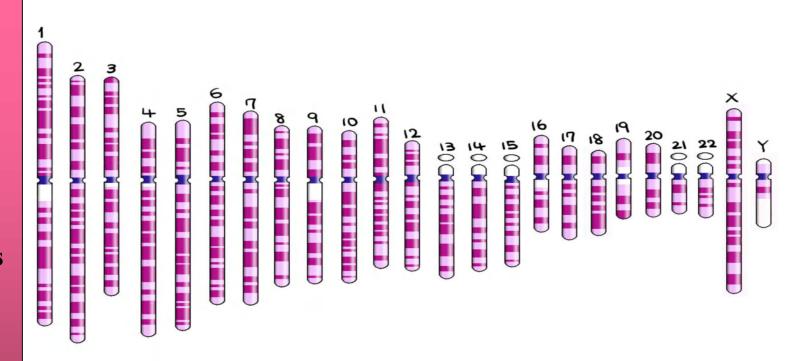
Disease map on chromosomes

天使病院

外木秀文

挿画 Koromo

臨床遺伝診療室



例

Gaucher disease 単一遺伝子病

Prader-Willi syndrome 隣接遺伝子症候群、欠失が主因となる疾患

Trisomy 13 複数の遺伝子の重複によって起こる疾患

遺伝子の座位あるいは欠失等の範囲を示す

1p36 deletion syndrome

Adrenoleukodystrophy, neonatal Zellweger syndrome

Hypercholesterolemia, familial

Ehlers-Danlos syndrome, type VI

Congenital disorder of glycosylation, type Ic

Diamond-Blackfan anemia 6

Marshall syndrome Stickler syndrome, type II

Alagille syndrome 2

Gaucher disease

Chromosome 1q21.1 deletion syndrome

Cardiomyopathy, dilated, 1A Charcot-Marie-Tooth disease Emery-Dreifuss muscular dystrophy Hutchinson-Gilford progeria Muscular dystrophy, limb-girdle type

Insensitivity to pain, congenital, with anhidrosis

Charcot-Marie-Tooth disease Dejerine-Sottas syndrome

Chronic granulomatous disease due to deficiency of NCF-2

Epiphyseal dysplasia, multiple Spondyloepimetaphyseal dysplasia

> Noonan syndrome Peters anomaly

Parkinson disease, familial

Seizures, benign familial neonatal-infantile

Ehlers-Danlos syndrome, type I, III, IV

Rhizomelic chondrodysplasia punctata, type 3

Split-hand/foot malformation

Brachydactyly, type D-E, Brachydactyly-syndactyly syndrome Synpolydactyly, type II and V

Leigh syndrome

Multiple pterygium syndrome Escobar syndrome

Brachydactyly, type A1

Waardenburg syndrome

Alport syndrome, autosomal recessive

Crigler-Najjar syndrome

GM1-gangliosidosis, type I-III Morquio syndrome B

Larsen syndrome Spondylocarpotarsal synostosis syndrome Atelosteogenesis

Tietz syndrome Waardenburg syndrome, type IIA

Bardet-Biedl syndrome

Glycogen storage disease IV

Seckel syndrome 1,

Blepharophimosis, epicanthus inversus, and ptosis

Dandy-Walker malformation

Epilepsy

Split-hand/foot malformation, type 4

Chromosome 3q29 microdeletion syndrome

Achondroplasia Hypochondroplasia Thanatophoric dysplasia, types I and II

Wolfram syndrome

Ellis-van Creveld syndrome

Parkinson disease

Hirschsprung disease, short-segment

Wolf-Hirschhorn syndrome

Mucopolysaccharidosis Ih-Is

Huntington disease

Fraser syndrome

Brachydactyly, type A2 Chrondrodysplasia, acromesomelic

Axenfeld-Rieger syndrome, type 1 Iridogoniodysgenesis, type 2 Peters anomaly

Hypercalciuria, absorptive

Craniometaphyseal dysplasia Adenomatous polyposis coli Cri-du-chat syndrome Cutis laxa, recessive, type I Brachydactyly, type A1 Clubfoot, congenital **Achondrogenesis Ib-II** Laron dwarfism Diastrophic dysplasia Epiphyseal dysplasia Hirschsprung disease **Treacher Collins** Cornelia de Lange syndrome mandibulofacial dysostosis **Maroteaux-Lamy syndrome** Parietal foramina with cleidocranial dysplasia **Cockayne syndrome Sotos syndrome** Weaver syndrome

Chromosome 6pter-p24 deletion syndrome

Iridogoniodysgenesis, type 1 Iris hypoplasia and glaucoma Rieger or Axenfeld anomalies

Branchiooculofacial syndrome

Stickler syndrome, type III

Glycogen storage disease Ic

Cleidocranial dysplasia

Polycystic kidney and hepatic disease

Char syndrome

Carpenter syndrome

Maple syrup urine disease, type Ib,

Split-hand/foot malformation with long deficiency 2

Microphthalmia, syndromic 8

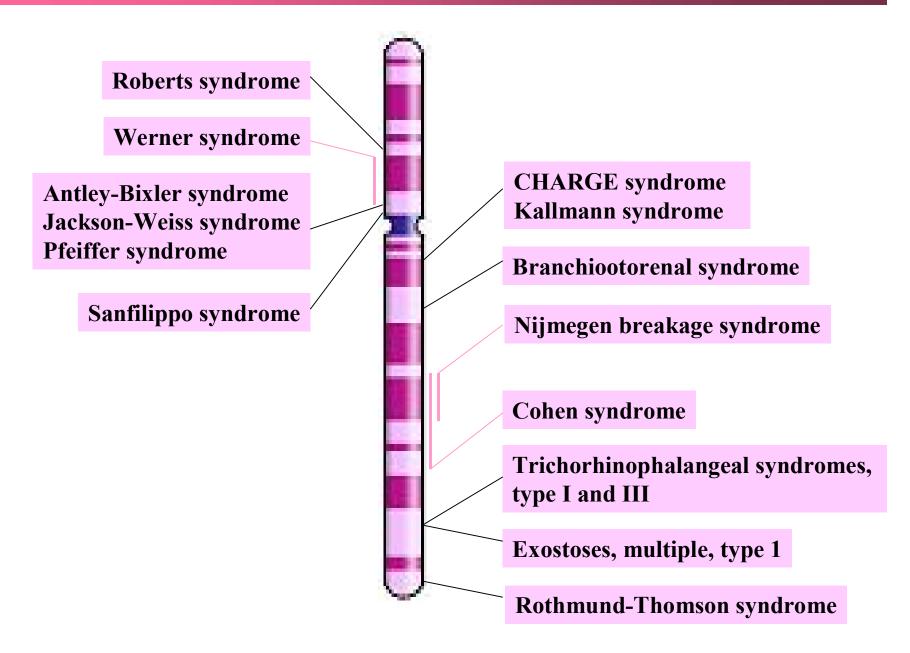
Polycystic liver disease

Metaphyseal chondrodysplasia, Schmid type

Atrioventricular septal defect Hypoplastic left heart syndrome Oculodentodigital dysplasia Syndactyly, type III

Refsum disease Rhizomelic chondrodysplasia punctata, type 1

EEC syndrome-1 Saethre-Chotzen syndrome Split hand/foot malformation Growth hormone deficient Citrullinemia, type II dwarfism Acrocallosal syndrome Refsum disease, infantile Greig cephalopolysyndactyly Zellweger syndrome syndromePallister-Hall syndrome Adrenoleukodystrophy **Silver-Russell syndrome Ehlers-Danlos syndrome,** cardiac valvular form Osteogenesis imperfecta **Cystic fibrosis** Supravalvar aortic stenosis Leigh syndrome Williams-Beuren syndrome **Wolff-Parkinson-White syndrome CHARGE syndrome**



Chromosome 9p deletion syndrome

Arthrogryposis multiplex congenita, distal
Nemaline myopathy

Melkersson-Rosenthal syndrome

Friedreich ataxia

Brachydactyly, type B1

Robinow syndrome

Walker-Warburg syndrome

Tuberous sclerosis-1

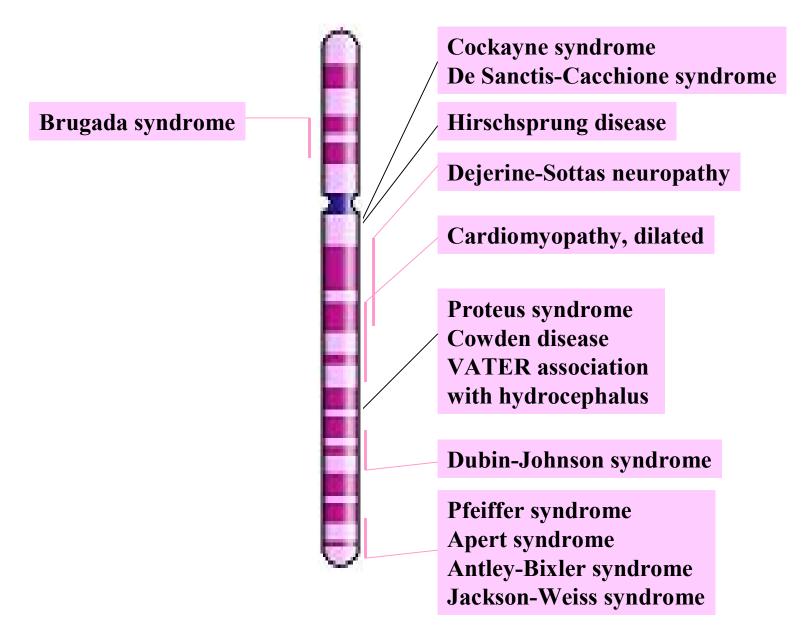
Citrullinemia

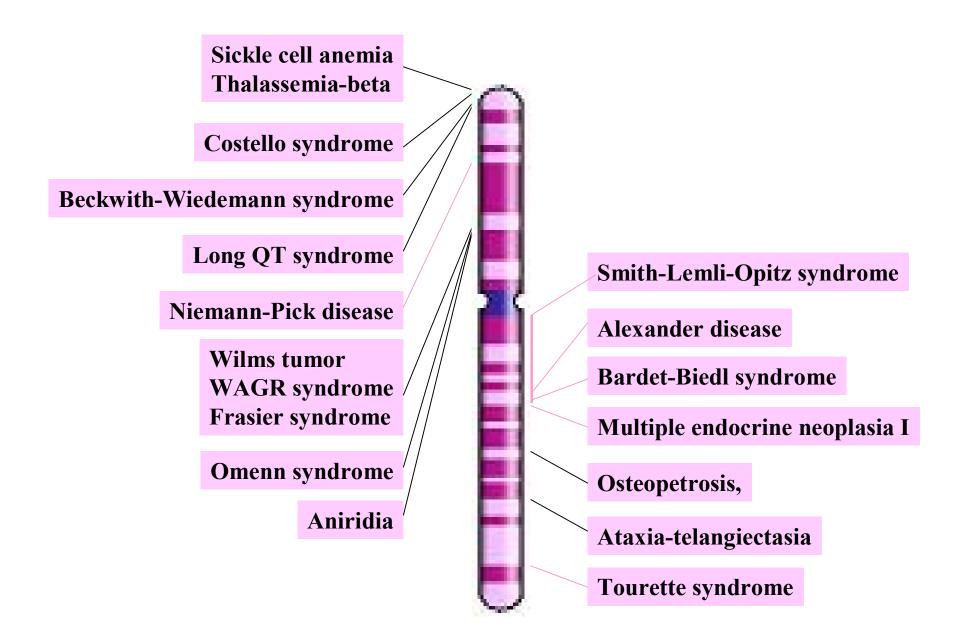
Nail-patella syndrome

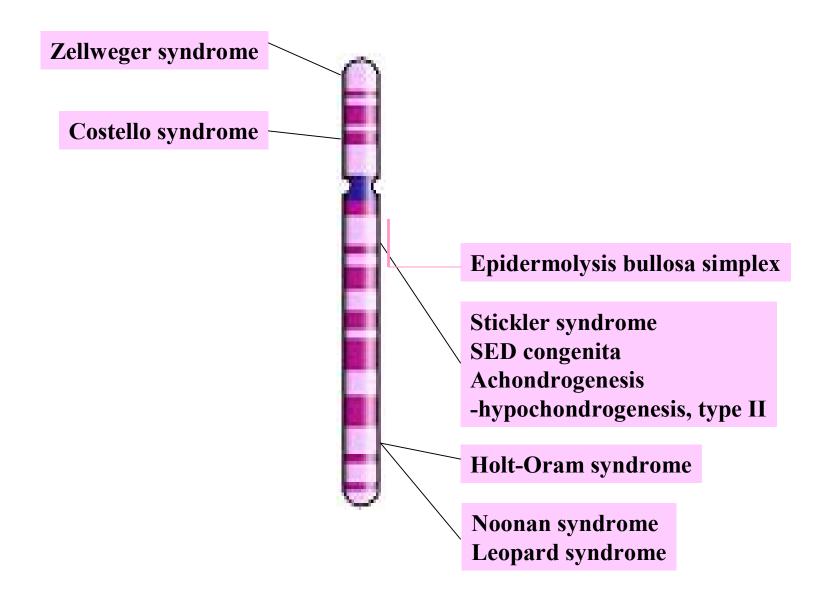
Walker-Warburg syndrome

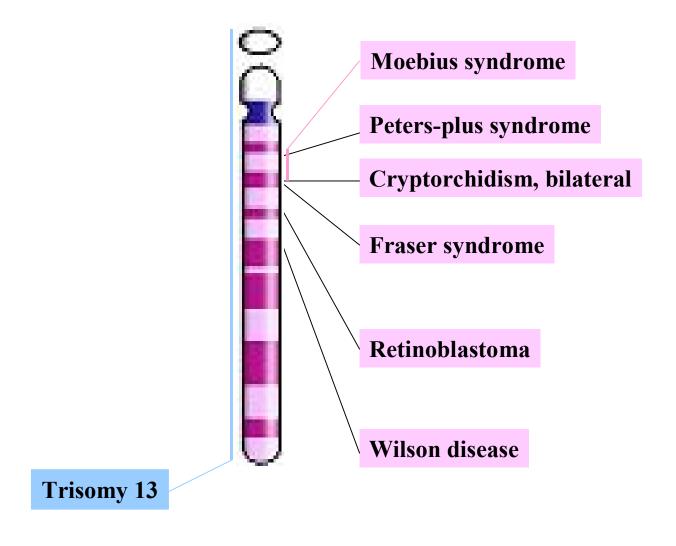
Ehlers-Danlos syndrome, types I and II

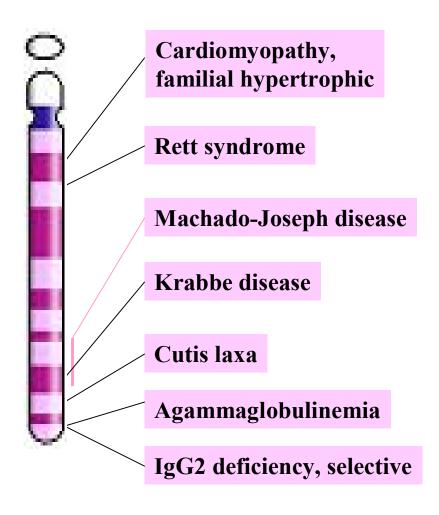
Chromosome 9q subtelomeric deletion syndrome

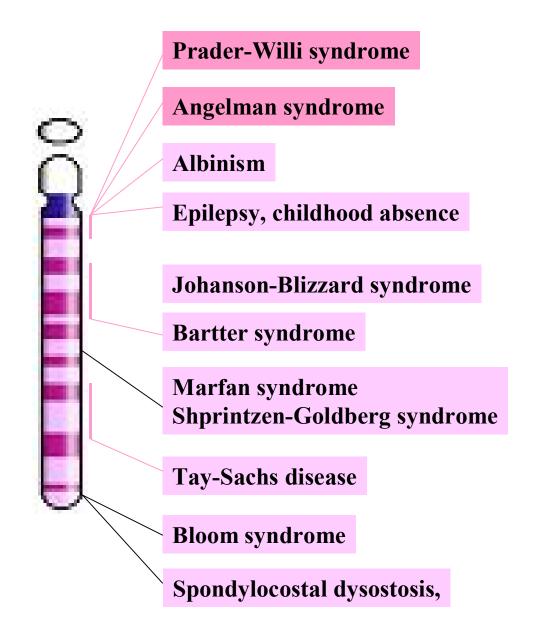


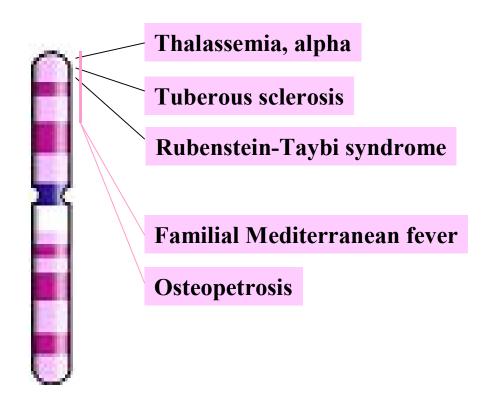




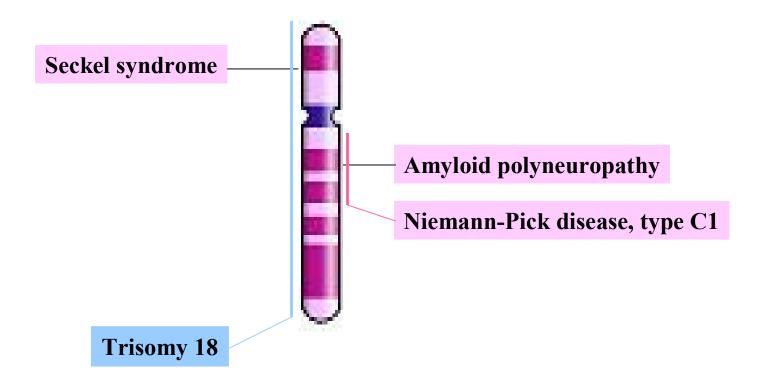


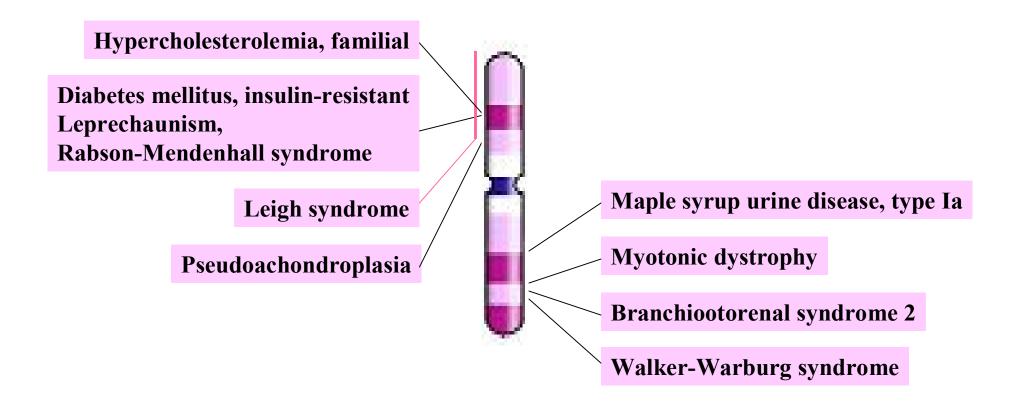


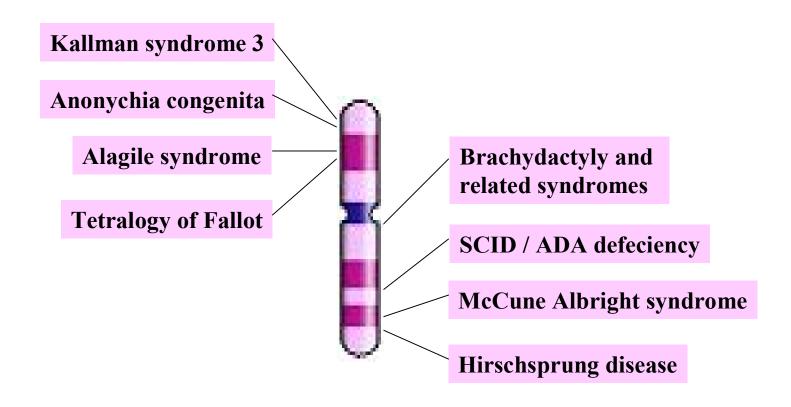


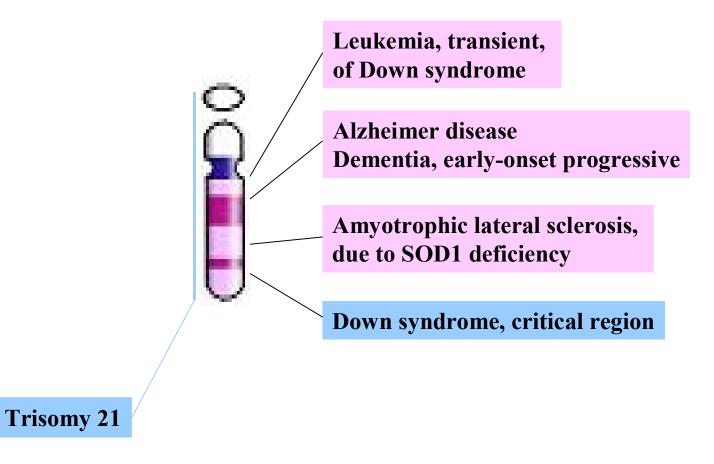


Miller-Dieker syndrome Arthrogryposis, distal, type 2A Li-Fraumeni syndrome Neurofibromatosis, type 1 **Epidermolysis bullosa simplex** Charcot-Marie-Tooth disease, type 1A **Ehlers-Danlos syndrome, type I Dejerine-Sottas disease** Osteogenesis imperfecta, type I Sjogren-Larsson syndrome **Growth hormone deficiency Bardet-Biedl syndrome 13 Smith-Magenis syndrome** Campomelic dysplasia Usher syndrome, type 1G









Cat eye syndrome

Chromosome 22q11.2 deletion syndrome DiGeorge syndrome Velocardiofacial syndrome

Chromosome 22q13.3 deletion syndrome

Adrenoleukodystrophy, neonatal

Zellweger syndrome

Neurofibromatosis, type 2

Rubinstein-Taybi syndrome

Waardenburg syndrome, type IIE

Metachromatic leukodystrophy

Congenital disorder of glycosylation, type Ig

Chondrodysplasia punctata Oral-facial-digital syndrome Kallmann syndrome **Opitz G syndrome Coffin-Lowry syndrome Duchenne muscular dystrophy Becker muscular dystrophy** Ornithine transcarbamylase deficiency **Wiskott-Aldrich syndrome Hypophosphatemic rickets Aarskog-Scott syndrome** Severe combined

immunodeficiency

Menkes disease

Agammaglobulinemia

Fabry disease

Alport syndrome

Lowe syndrome

Simpson-Golabi-Behmel syndrome

Lesch-Nyhan syndrome

Hemophilia B

Fragile X syndrome

Diabetes insipidus, nephrogenic

Frontometaphyseal dysplasia

G6PD deficiency

Hemophilia A

Otopalatodigital syndrome

Rett syndrome

