Collection of disease

单基因遗传疾病

Xeroderma pigmentosum

week 1

- DNA repair disorder, can not repair the damaged DNA
- increasing risk of cancer
- no primary therapy

Alkaptonuria

Week 2

- A kind of Autosomal recessive inheritance
- 尿黑症

Albinism

Week 2

- Autosomal recessive inheritance
- can't produce melanin
- 白化病

Marfan syndrome

Week 2

- Autosomal dominant disorder
- caused by mutations in the FBN1 gene on chromosome 15, which encodes fibrillin-1
- 患有这种疾病的人往往又高又瘦，胳膊、腿、手指和脚趾都很长。 它们通常还具有异常灵活的关节和异常弯曲的脊柱。

Rett syndrome

Week 2

- Mutation located on X chromosome
- Mutated gene is Mecp 2 gene
  - can inhibit the expression of some gene
  - Mecp2 has been implicated plays important roles in dendritic arborization, synaptic strength, excitatory-inhibitory balance and long-term potentiation
- Neurological disorder
- Partial rescue of Mecp 2 deficiency by postnatal activation

Red-green colour blindness

Week 2

- X-linked Recessive genetic disorder
- mainly in male

Fragile X syndrome

Week 2

- Triplet repeat disorder
- single gene disorder associated with autism
- cause tremor and ataxia
  an expansion of the CGG repeats within the fragile X mental retardation 1 gene (FMR1) on the X chromosome
- female more more often to be affected
- Symptoms
  - Low intelligence
  - cause of autism
  - long and narrow face
  - long ears, flexible fingers
  - seizures

Tay-Sachs disease

Week 12 Genetic Testing

- A rare autosomal disorder
- caused by mutation in HEXA gene on chromosome 15
  - which results in the buildup of the molecule GM2 ganglioside within cells, progressively destroying neurons in the brain and spinal cord.
- Three types
  - Infantile Tay-Sachs disease
  - juvenile Tay-Sachs disease
  - Adult Tay-Sachs disease
- Carrier Detection
  - Serum may be used to test affect hormones
  - Leukocytes are used to test

Cystic fibrosis

Week 12 Genetic Testing

- Most common genetic disorder for white
- Mutation in the gene CFTR on chromosome 7
- causes problems with **digestion** and **breathing**
- Treatment: daily respiratory therapy, digestive enzymes, medication to promote lung function
- Carrier screening
  - DNA mutation analysis
  - potential risk should be aware after negative test

Thalassemia

Week 12 Genetic Testing

- Autosomal recessive genetic disorder
- abnormal hemoglobin production
- a high frequency in tropical and sub-tropical areas

- Symptoms depend on the type and can vary from none to severe
- Two main type : alpha thalassemia and beta thalassemia
- Symptom
  - Anemia
  - feeling tired and pale skin
  - bone problems
  - enlarged spleen
- Treatment
  - blood transfusion
  - bone marrow transplant
  - If the spleen becomes overly enlarged, surgical removal may be required.
  - Gene therapy

Spinal muscular atrophy

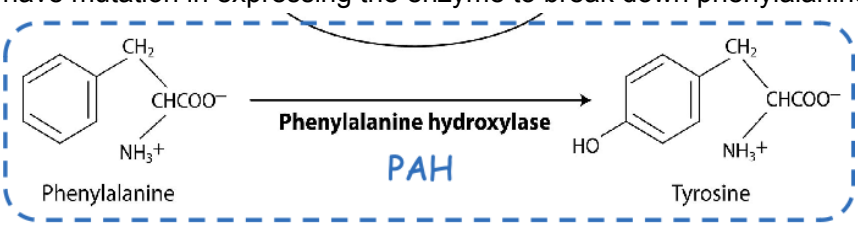
Week 12 Genetic Testing

- autosomal recessive
- Gene defection on SMN1 gene
- Another gene SMN2, more SMN2 copies, the disease is more mild
- Neuromuscular disease
- Detection
  - A blood test
  - electromyography
  - muscle biospy
  - Carrier testing needs a measurement of SMN1 copy number, by qPCR
  - general population have two SMN1 copies on one chromosome and no copies on the other and will not be identified as being a carrier of spinal muscular atrophy using this approach.
- Treatment
  - Nutrition through feeding tube
  - Antisense oligonucleotide
    - Nusinersen, 一种治疗这个的药物

Phenylketonuria

Week 12 Genetic Testing

- Also called PKU
- autosomal recessive disorder
- have mutation in expressing the enzyme to break down phenylalanine



- Cause phenylalanine accumulation, which can be toxic in high concentration
  - can harm nervous system
  - cause intellectual disability
  - fair hair and skin, delayed cognitive development

- Criteria for PKU
    - classic PKU
    - Atypical or mild PKU
    - Non-PKU benign hyperpheylalanine
-
    - Treatment
    - Restriction of dietary intake of Phe
        - low protein food , without Phe or with supplemental tyr(because Tyr is turned forn Phe but here they can't do that)
- Methods of PKU diagnosis
    - Bacterial inhibition assay
        - agar plate 上存在analog of phenylalanine
        - The size of the halo was related to the concentration of phenylalanine in the sample
        - 血液样本中的phenylalanine会影响细菌生长。通过细菌生长来反映是否PKU

Medium-chain acyl-CoA dehydrogenase deficiency(MCADD)

Week 12 Genetic Testing

- Autosomal recessive
- Medium-chain acyl-CoA dehydrogenase deficiency
- fatty acid oxidation disorders
- 患者不能通过脂肪酸氧化的途径获得能量
- Newborn Screening for MCADD and other fatty acid oxidation disorders became available with the introduction of TMS in NS programs.

Duchenne muscular dystrophy

Week 14

- Monogenic, X-linked recessive
- Mutation in dystrophin gene
- two-thirds of which involve exon deletions that disrupt the dystrophin open reading frame, and point mutations or duplications account for the rest.

多基因遗传疾病

Autism

Week 4

- Genetic heterogeneity
    - a single phenotype or genetic disorder may be caused by any one of a multiple number of alleles or non-allele (locus) mutations
- Autism can caused by many gene mutation
- If you have one child with autism, the risk for the next child is only 2-6%. If you have two children with autism, the chances that the third will be autistic are around 35%.

Type I diabetes

Week 13

- Insulin dependent
- can't produce insulin

- more genetic factors than environmental factors

Type II diabetes

Week 13

- Insulin resistance
- polygenic risk
- More environmental factors than genetic factors

CAD

One copy of genetic risk variant for coronary artery disease
Week 13

Breast cancer

Week 13

- Cause
  - gene Mutations
    - Especially BRCA1/2 gene mutation
      - BRCA1/2 can repair the DNA damage, but the mutations make them can't
  - Risk factors include:
    bold means related to **estrogen**
    - **Being female**
    - Increasing age
    - radiation therapy to the chest
    - **Post-menopausal obesity
    - **First menstrual period before age 12
    - **Beginning menopause after age 55
    - **Having first child after 35
    - **Having no child
    - **Postmenopausal hormone replacement therapy
    - Diet

Alzheimer’s disease(AD)

Week 13

- progressive neurodegenerative disease
- Cause
  - Gene cause
    - **APOE variants**
      - subtypes:
        - ε2 : 2 Cys, normal
        - ε3 : 1 Cys, heterozugous
        - ε4: 2 Arg, homozygous, higher chance to get AD
    - Accumulation of amyloid, because can't clear them in the brain

线粒体基因遗传疾病

Primary mitochondrial Disease(PMD)

Week 3

- caused by the mutations in nDNA and mtDNA
- 直接对维持线粒体有作用的

LHON

Week 3

- Caused by the mutation in mtDNA, to mRNA
- Leber's hereditary optic neuropathy
- features
  - Loss of central vision of eyes
  - retinal ganglion cells apoptosis
  - Gender-bias:
  - Males more than female
  - High threshold for pathogenicity
  - Homoplasmic or nearly

Maternal inherited deafness

Week 3

- Caused by the mtDNA mutaion, to rRNA(which is similar to the bacteria)
- Aminoglycosiders can also harm the hearing
  - Aminoglycosides are antibiotics, can harm mitochondria

MERRF

Week 3

- Caused by the mutations in mtDNA, to tRNA

MELAS

Week 3

- Caused by the mutations in mtDNA, to tRNA

未分类疾病

Cleft lip

week 4

- Multifactors
- 裂唇
- example:
  - bilateral cleft lip --> sibling risk is 6%
  - unilateral cleft lip --. sibling risk is 2%
- index patient/ patient zero:
  - the first known case of a infectious disease

Neural tube defect

Week 4

- Multifactors
- a baby with it --> recurrence risk is about 2-4%
- 2 baby with it. --> recurrence risk is about 10%

Down Syndrome

week 5

- Trisomy 21

Turner syndrome

week 5

- Monosomy X, only one X
- lack of secondary sexual characters

22Q11.2 Deletion syndrome

week 5

- Chromosom structure abnormalities
  - Large part of a chromosome has been lost
- Palatal abnormalities
- Congenital heart defects
- Facial anomalies

Translocation Down's syndrome

week 5

- the parents are balanced 21 rebertsonian translocation carrier

Angelman syndrome

week 5

- Two copies comes from the father, no materal chromosome 15
- UBE3A gene in chromosome 15 only expressed form the Materal gene(gene imprinting)
- seizures, unprovoked Smiling and laughter, lack of Speech, dev delay

Maternal PKU

Week 12 Genetic Testing

- The mother with Phe should keep on Phe-free food, or their children might mature in a high Phe environment.

Melanoma

Week 13

- Skin cancer
- UV light damages DNA

Hemochromatosis

Week 14

- hyperactivity to absorb too much iron from the diet

Type I Tyrosinemia

Week 14

- Type 1 Tyrosinemia(Tyrosine的代谢收到影响)
  - deficiency of fumarylacetoacetate hydrolyase（FAH)
  - cause accumulation of Fumarylacetoacetate, which is toxic to liver

Congenital hypothyroidism

Week 14

- defect in thyroid development caused by gene mutation

Heterodoxy Syndrome

- a condition in which the internal organs are abnormally arranged in the chest and abdomen
  - situs inverse
    - 器官颠倒，比如心脏到了右边
  - right isomerism
  - left isomerism