Genetic Evaluation, Diagnosis and Treatment

--Start from Knowing

遺傳諮詢診治

--從瞭解開始

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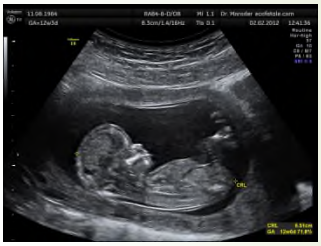
How is genetics visit relevant for my child?

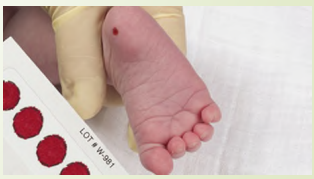
Start from knowing

Genetics is actually not far from you…

- Prenatal
- Ultrasound
- Newborn screening
- Health screening

- Referring for common indications:
  - recurrent infections
  - developmental delay
  - short stature
  - low muscle tone
  - Etc.





Overview

- When to have a genetic evaluation?
- What are the benefits of having a genetic diagnosis?
- What to expect in a typical appointment with genetics?
- The basics of genetics and genetic testing – understand your child's testing report
- Case examples (3 brief cases)
- Dispelling the common myths about genetics
- Q & A

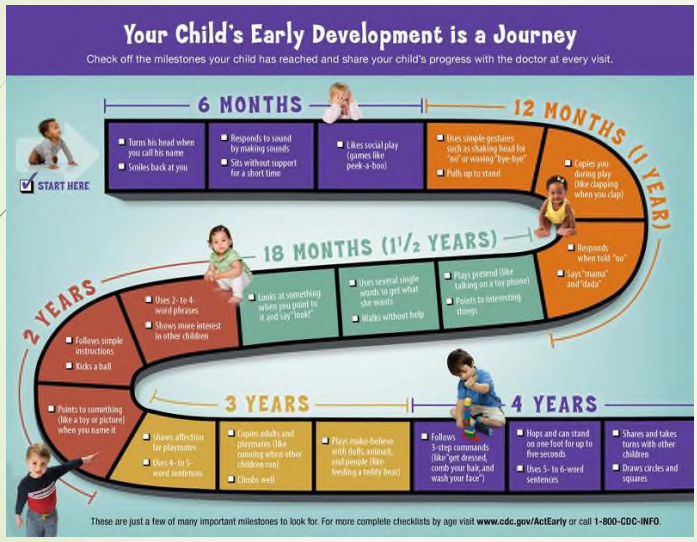
要述

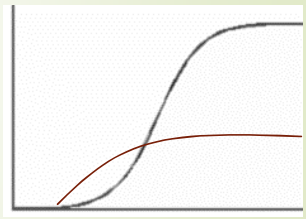
- 什麼時候需要醫學遺傳門診服務？
- 遺傳診斷能對您和家人有什麼幫助？
- 遺傳門診通常做些什麼？
- 基礎遺傳學和遺傳檢測小知識 – 讀懂孩子的檢查報告
- 一些關於遺傳的常見誤解
- 典型病例 （3例）
- 問答環節

When to see genetics

- Multiple congenital abnormalities (such as a neural tube defect, heart defect, cleft lip or palate)
- Rare or unusual symptoms (eg. heterochromia)
- *Developmental delay/Intellectual Disability/Autism*
- Family history of traits/diseases
- Previous child with a genetic disorder
- Early onset of a condition that usually starts much later (such as heart muscle disease in a child)

Developmental Delay





Intellectual Disability

- Characterized by intelligence below normal (IQ<70)
- Lack of needed skills for daily functioning
- People with intellectual disabilities can learn new skills and tasks but will do so more slowly

Autism Spectrum Disorder

Characterized by:

- Difficulties with social interaction and communication
- Stereotypical and repetitive behaviors
- Delay in language acquisition
- Begins before 3 years of age

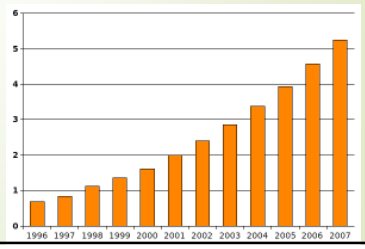
- Associated conditions:
  - Intellectual disability
  - Epilepsy
  - Anxiety

Autism Spectrum Disorder (ASD)

• ASD affects 14.7 per 1,000 children (up to 8 years of age)

• It is estimated that of the 4 million children born in the USA each year 36.500 will be diagnosed with autism. It is estimated that 730.000 people between 0 and 21 years have ASD.

MMWR, 2010 & http://www.cdc.gov)



Characteristics of genetic condition

Family as a unit of receiving service

Lifelong diagnosis

Benefits of genetic evaluation for the child

- Early diagnosis and recognition may influence long-term outcome
- Find appropriate medical & non-medical therapies
- Screen for associated complications or disabilities before symptoms seen
- Avoid unnecessary testing & evaluations
- Help with educational planning (immediate & long-term)
- Easier to obtain therapy and services if the child has a diagnosis

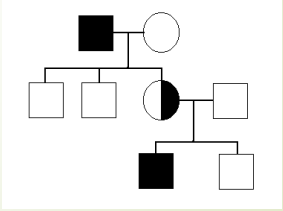
Benefits of genetic evaluation for the family

- Provide information about the cause, treatment and dispel misinformation
- Relieve the guilt some parents feel
- Offer support, guidance and an outline of what to expect
- Reduce the sense of helplessness when the diagnosis/prognosis is not known
- Learn about opportunities for education, advocacy and research
- Provide reproductive options
- Help with decisions about long-term care planning (insurance, education, medical, contraception, adult living)

What happens in a typical appointment with genetics?

Before the visit – What to prepare in advance

- Gather family health history up to three generations
- Share with your doctor without holding back information that you think is irrelevant
- Prepare questions for your genetics doctor



Before the visit – Five questions for the genetics doctor

- What's the diagnosis?
- What's the cause?
- What's the future hold?
- What's the treatment?
- What's the chance of the condition occurring again?

During the visit

- Family Health history
- Medical history
- Physical exam
- Discuss clinical impression and test recommendations: blood and urine tests, imaging exams, etc.

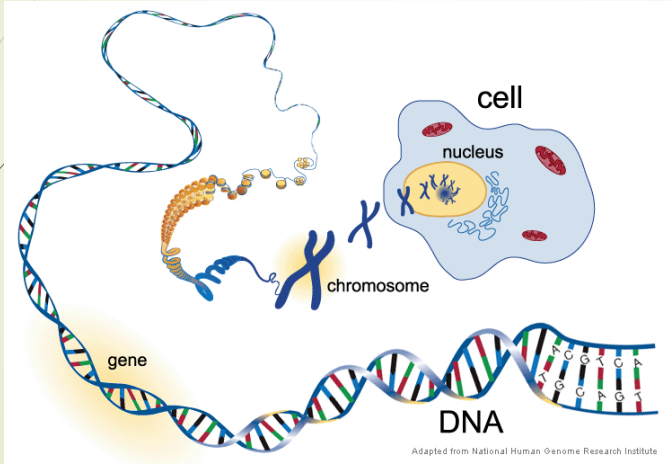
How is genetics diagnosis made?

- Analyzing the family history
- Looking for a combination of clinical features
- Looking at the progression of medical problems
- Genetics testing on blood, saliva, urine – get to the bottom

Testing

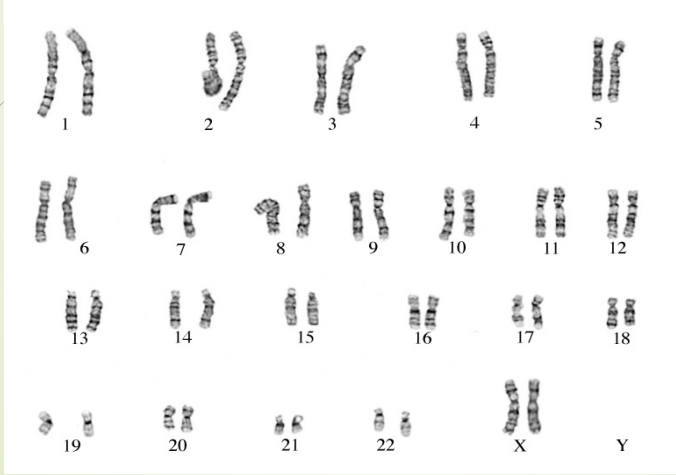
- Results available in days to months
- When diagnosis made, family contacted by phone with results and schedule follow-up visit as soon as possible.

The Basics of Genetics - Understand your child's testing report

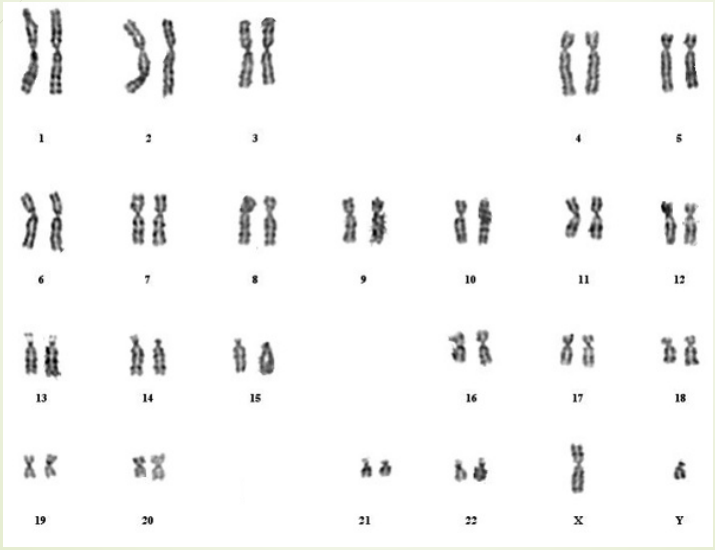


Chromosomes: normal female

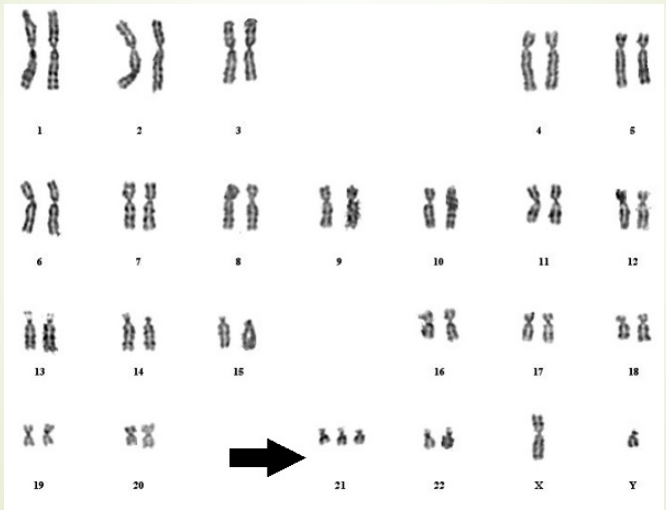
Our ~23,000 genes are on the 23 pairs of chromosomes



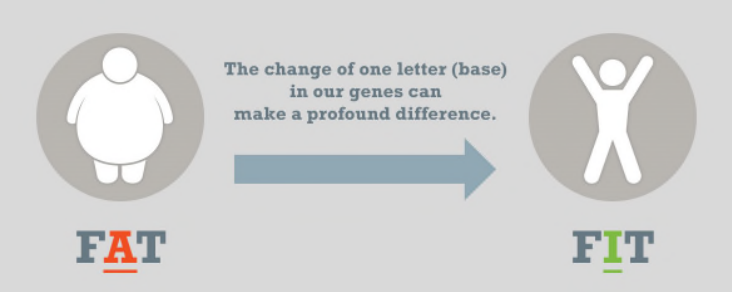
Chromosomes: normal male



Trisomy 21 (Down Syndrome)



Gene Analysis



When Diagnosis is Made:

- Provide information about the cause
- Features of the disease or condition
- What to expect over time
- Plan specific treatment and management
- Arrange for other consults if indicated: Neurology, cardiology, ophthalmology and etc.

When Diagnosis is Made: Counseling Includes

- Inheritance Pattern
- Recurrence risk and reproductive options
- Testing for other family members may be indicated
- Offer information, support, resources

GINA

Genetic Information Nondiscrimination Act passed in 2008.

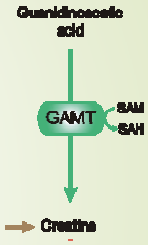
- Prohibits discrimination in health coverage and employment based on genetic information
- Does not extend to life, disability or long-term care insurance
- http://www.genome.gov/24519851

Case 1

- 4 year old girl referred for short stature
- Parents not sure why here
- Turner syndrome
- Kidney and heart check-ups, growth hormone therapy

Case 2

- Developmental delay
- High guanidinoacetate; low creatine -> Genetic testing
- GAMT deficiency
- Treatment is special diet: Arginine-restricted diet and creatine supplementation
- Not fully reversible for early age damage, but can prevent seizures and reduce severity



Case 3

- Family history of aortic rupture
- Asymptomatic siblings 16 yo and 13yo
- Fear in the family; Annual cardiac check-up
- Genetic testing revealed mutation in the FBN1 gene in the elder brother
- Provide diagnosis, while prevent overscreening

Myth 1

- I must have done something wrong/we must have missed something, so that my child has disability/genetic condition.

- There is nothing you did or did not do that lead to the child's disorder.

Myth 2

- Once I have a genetic diagnosis, it can be fully cured like cold.

- There is nothing wrong with taking TCM, but with a genetic diagnosis, it can point to the specific need such as enzyme replacement
- With genetic condition, usually there is no full cure.

Myth 3

- My child already has autism? Why do I still need to test/imaging
- (Genetic disorder cannot be cured, so we don't need any testing.)

- Treat/prevent potential complications; educate the school; reach the maximal potential

Myth 4

- Genetic testing sound fancy. It must be expensive and there is no way I can afford it!

- No! Resources are available.
- Medicaid cover genetic testing
- Private Insurances： United Healthcare, BCBS and etc.
- Financial assistance programs

Myth 5

- Genetic disease are always inherited.

- Some are not (de novo, aneuploidy)
- Some are

Thank you!

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Baylor Adult Genetics: 713-798-7820