# Medical Genetics, reproduction and eating

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# Menù

- Part 1:
  - Medical Genetics in the 21 century
- Part 2:
  - Medical Genetics and infertility
- Part 3:
  - Genetic, epigenetics, food and reproduction

## Medical Genetics in the 21<sup>st</sup> century

From Mendel to System Biology

# The 4 levels of medical genetics

#### "Classical" genetics

- Mendelian genetics (single gene diseases)
- Cytogenetics (chromosomal diseases)
- Multifactorial inheritance
  - Interaction between genes and environment

#### • Epigenetics

Alteration in DNA function without alteration in DNA structure

#### Post genomics

- Gene expression profiles

#### The "classical genetics"

From genotype to phenotype







# Genotype, phenotype, simple traits, complex traits

# Genotype and Phenotype

- Genotype:
  - Genetic constitution
    of each single
    subject
- Phenotype:
  - Physical, cellular and mental features of a single subject



#### Genotype





#### Genotype-phenotype correlation

P=G

![](_page_11_Picture_1.jpeg)

http://www.bio.davidson.edu/Courses/Immunology/Students/Spring2003/Leese/albino.gif

![](_page_11_Picture_3.jpeg)

![](_page_11_Picture_4.jpeg)

# Simple traits (mendelian inheritance)

- Phenotype directly determined by genes, without environmental influence
- Dichotomous traits
- Inherited according to the Mendel's laws.

![](_page_12_Picture_4.jpeg)

### Single-traits diseases

- Mendelian diseases
  - Autosomic dominant
  - Autosomic recessive
  - X-linked
- Chromosomal disease
  - Abnormalities in chromosome number
  - Abnormalities in chromosome structure

#### Autosomical dominant diseases

![](_page_14_Figure_1.jpeg)

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![](_page_15_Picture_0.jpeg)

#### Examples

![](_page_15_Figure_2.jpeg)

![](_page_15_Picture_3.jpeg)

![](_page_16_Picture_0.jpeg)

#### Autosomal recessive diseases

![](_page_17_Figure_1.jpeg)

## Examples

![](_page_18_Picture_1.jpeg)

![](_page_18_Picture_2.jpeg)

![](_page_18_Picture_3.jpeg)

#### Carriers

DISEASE	AFFECTED	CARRIERS
CF	1:2500	1:25
SMA	1:10000	1:50
Thalassemia (specific regions)	1:400	1:10

#### X-linked inheritance

![](_page_20_Figure_1.jpeg)

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#### Examples

![](_page_21_Picture_1.jpeg)

![](_page_21_Picture_3.jpeg)

![](_page_21_Picture_4.jpeg)

#### **Chromosomal diseases**

![](_page_22_Figure_1.jpeg)

![](_page_23_Picture_0.jpeg)

![](_page_23_Picture_1.jpeg)

![](_page_23_Picture_2.jpeg)

![](_page_23_Picture_3.jpeg)

![](_page_23_Picture_4.jpeg)

#### **Complex Traits**

- Quantitative phenotypes, derived from the interaction between genes and evironment
- Not following Mendel's Law

![](_page_24_Picture_3.jpeg)

![](_page_25_Picture_0.jpeg)

![](_page_25_Picture_1.jpeg)

![](_page_26_Figure_0.jpeg)

#### Phenotype in complex traits

# Susceptibility genes

#### **Genetic background (stable)**

![](_page_27_Figure_3.jpeg)

#### Environment

(Variable)

Phenotype

![](_page_28_Picture_0.jpeg)

![](_page_28_Picture_1.jpeg)

![](_page_28_Picture_2.jpeg)

![](_page_29_Picture_0.jpeg)

![](_page_29_Figure_1.jpeg)

#### **Disease Status**

![](_page_30_Figure_1.jpeg)

Genetic Protective Factors

![](_page_30_Picture_3.jpeg)

Environmental Protective Factors

# Genetic Components of Risk for Disease A

Genetic Components of Risk for Disease A	
Increased risk for Disease A	Overall Genetic Component of Risk
Decreased risk for Disease A (protection)	

![](_page_31_Picture_2.jpeg)

![](_page_32_Figure_0.jpeg)

Overall Risk

![](_page_32_Picture_2.jpeg)

![](_page_33_Figure_0.jpeg)