

POLYGENIC INHERITANCE NONALLELIC GENE INTERACTIONS Stomatology

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Brief repetition:

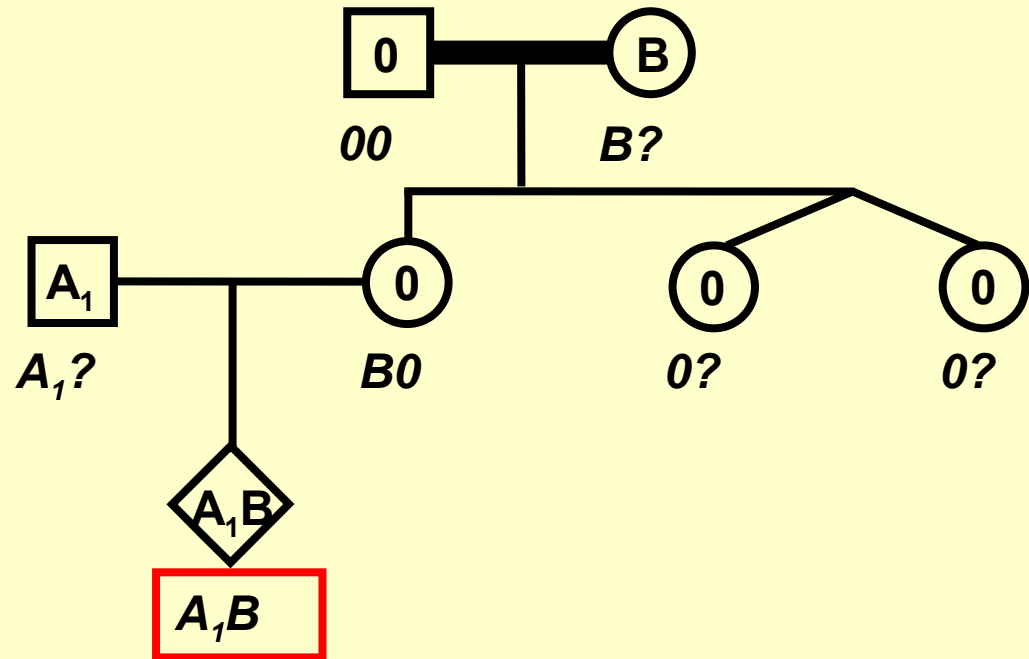
- **Monogenic inheritance**
 - participation of alleles of the same gene (locus) in hybrid phenotype formation
 - differences in allele dominance level
- **Allelic and non-allelic genes**
- **Dihybridism**
 - 16-poles table of genotype combinations
 - genotypic and phenotypic ratios

Introduction I:

- **genetic determination** of particular traits - **participation** of alleles of two, three or more genes
 - **modifying genes**
 - **gene interaction**
 - **complementary, epistatic or supplementary genes -**

Bombay phenotype in the ABO system

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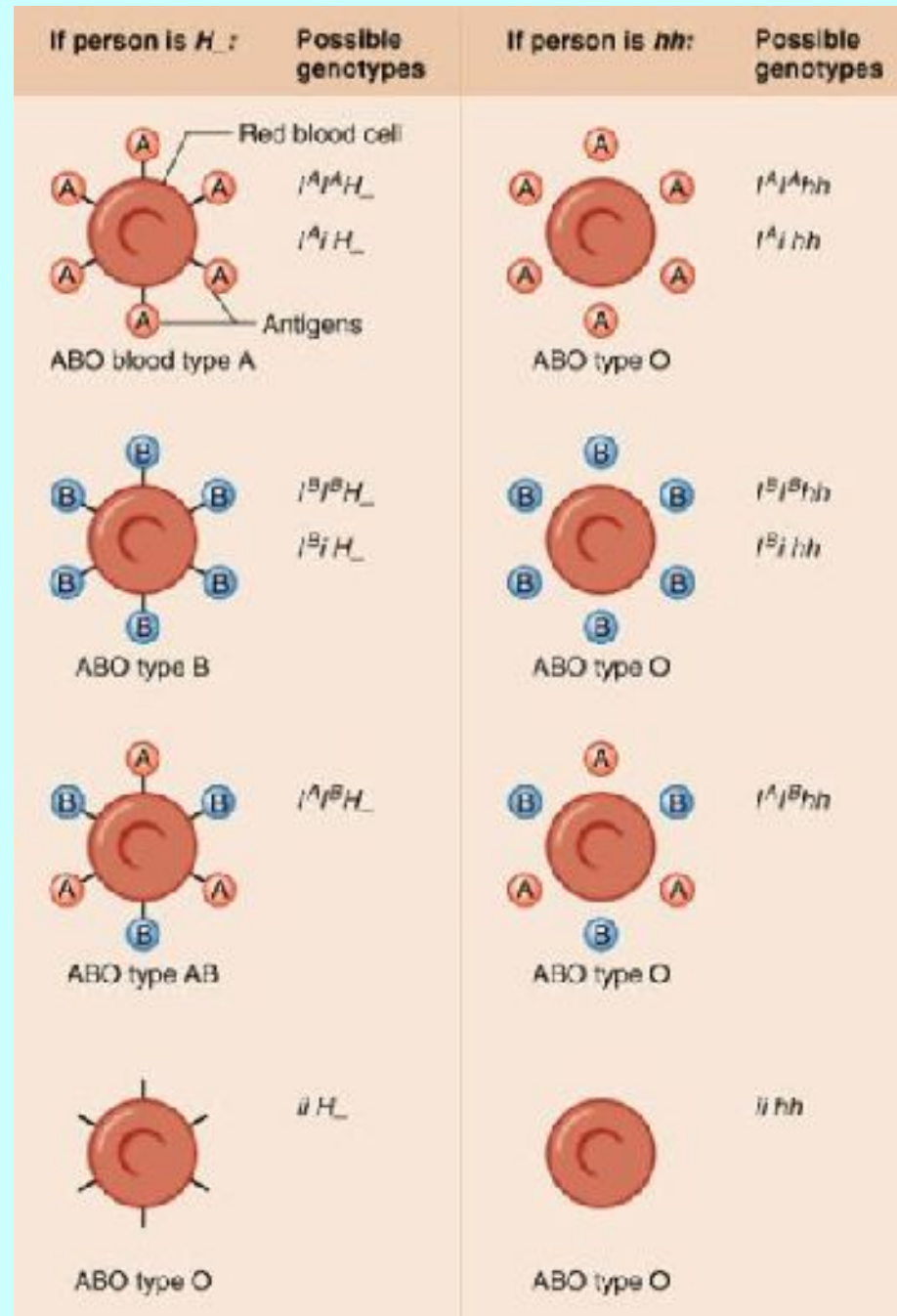


**Supplementarity, recessive epistasis,
exemple of metabolic pathway.**

H and ABO loci as recessive epistasis

Homozygosity for alleles of one gene affects the expression of a second gene.

- H gene is epistatic to the ABO gene.
- H substance is attached to the cell surface.
- *hh* genotype = no H protein.
- All ABO genotypes appear as type O.



Introduction II:

Recapitulation of key-words

- multifactorial traits
- polygenic inheritance
- major and minor genes
- modifying genes
- active and non-active alleles
- additive effect of alleles
- variability caused by genotype and environment expression

Segregation ratios

Pascal's triangle

n	$(1 + 1)^n$										Total	
1					1	1					2	
2				1	2	1					4	
3			1	3	3	1					8	
4			1	4	6	4	1				16	
5			1	5	10	10	5	1			32	
6			1	6	15	20	15	6	1		64	
7			1	7	21	35	35	21	7	1	128	
8			1	8	28	56	70	56	28	8	1	256

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7			1	7	21	35	35	21	7	1	128	
8			1	8	28	56	70	56	28	8	1	256

Number and frequencies of the genotypes for 3, 4 a 5 genes

1 : 10 : 45 : 120 : 210 : 252 : 210 : 120 : 45 : 10 : 1

Recurrence risk estimates for diseases with multifactorial etiology

- 1. Although the disorder is obviously familial, there is no distinctive pattern of inheritance within family**
- 2. The recurrence risk to first-degree relatives is approximately the square root of the population risk (incidence) – Edwards' formula**
- 3. The risk is sharply lower for second-degree than for first-degree relatives, but it declines less rapidly for more remote relatives.**
- 4. The risk is higher when more than one family member is affected – for first-degree relatives, the value calculated from Edwards' formula is multiplied by 2, 3 etc.**
- 5. The more severe the malformation, the greater the risk**
- 6. If a multifactorial trait is more frequent in one sex than in the other, the risk is higher for relatives of patients of the less susceptible sex.**
- 7. An increased risk when the parents are consanguineous (multiple factors with additive effects may be involved)**
- 8. Strongly affected by the environmental factors**

Neural tube defect recurrence risk

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Congenital malformation with multifactorial etiology and polygenic inheritance, population frequency ca 0,0009

