# Biosocial interactions in modernization

# 2. Evolutionary determinants of biosocial processes

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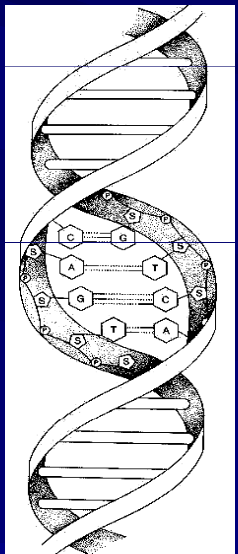
- 2.1. Evolutionary mechanisms of biosocial processes
- 2.2. Genetic-environmental interactions in biosocial processes

## 2.1. Evolutionary mechanisms of biosocial processes

- A few reminders of some basic genetic concepts :
  - At the molecular level: DNA
    *(Watson and Crick, 1953)*
  - At the cytological level: chromosome *(Strasburger, 1875)*
  - At the individual/family level: Laws of Mendel *(1865)*
  - At the population level: Hardy-Weinberg law *(1908)*

# DNA (*desoxyribonucleic acid*):



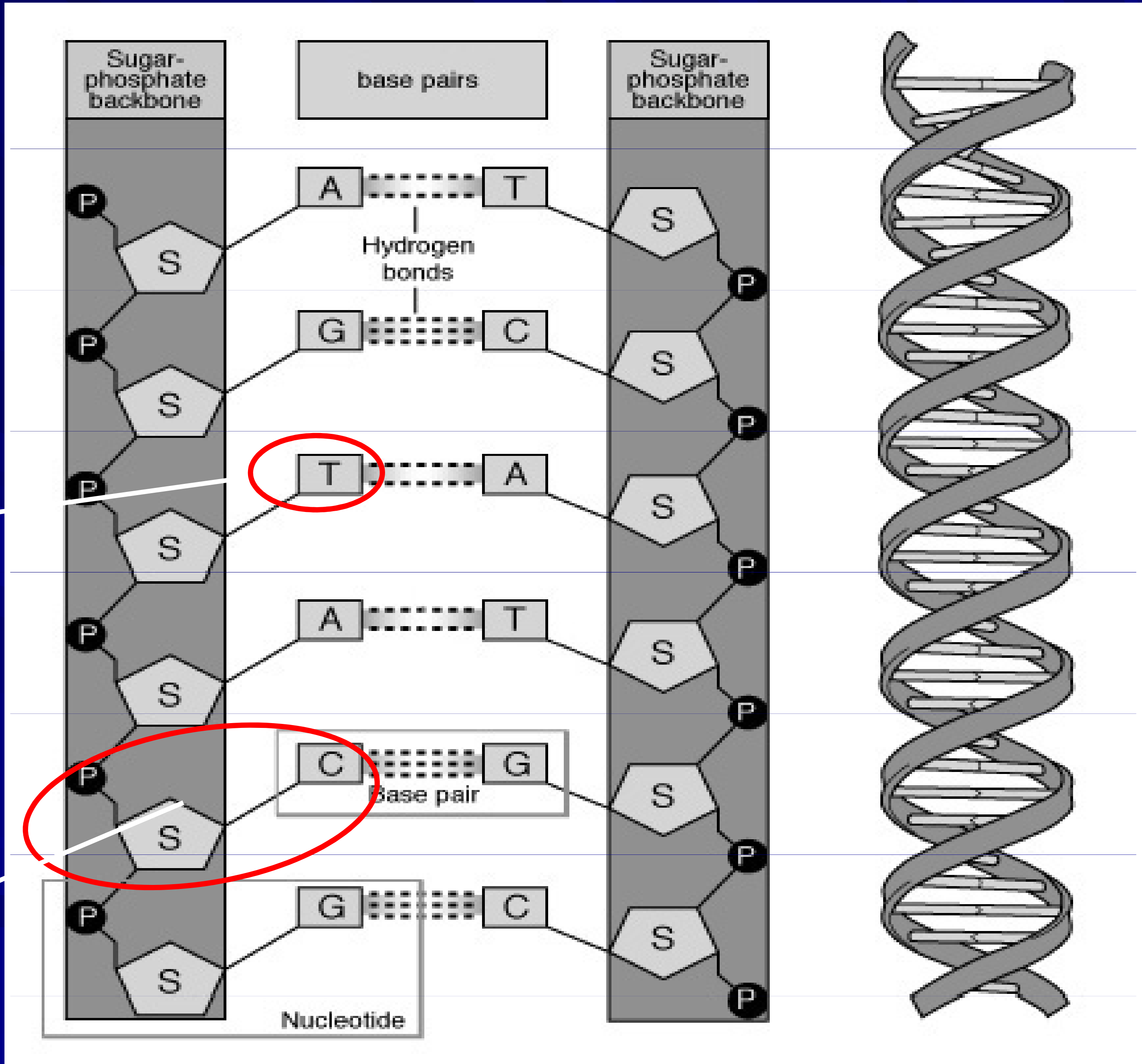

The duplication process of the genes

The enormous genetic variability which exists in nature

The duplication process of the genes

Base

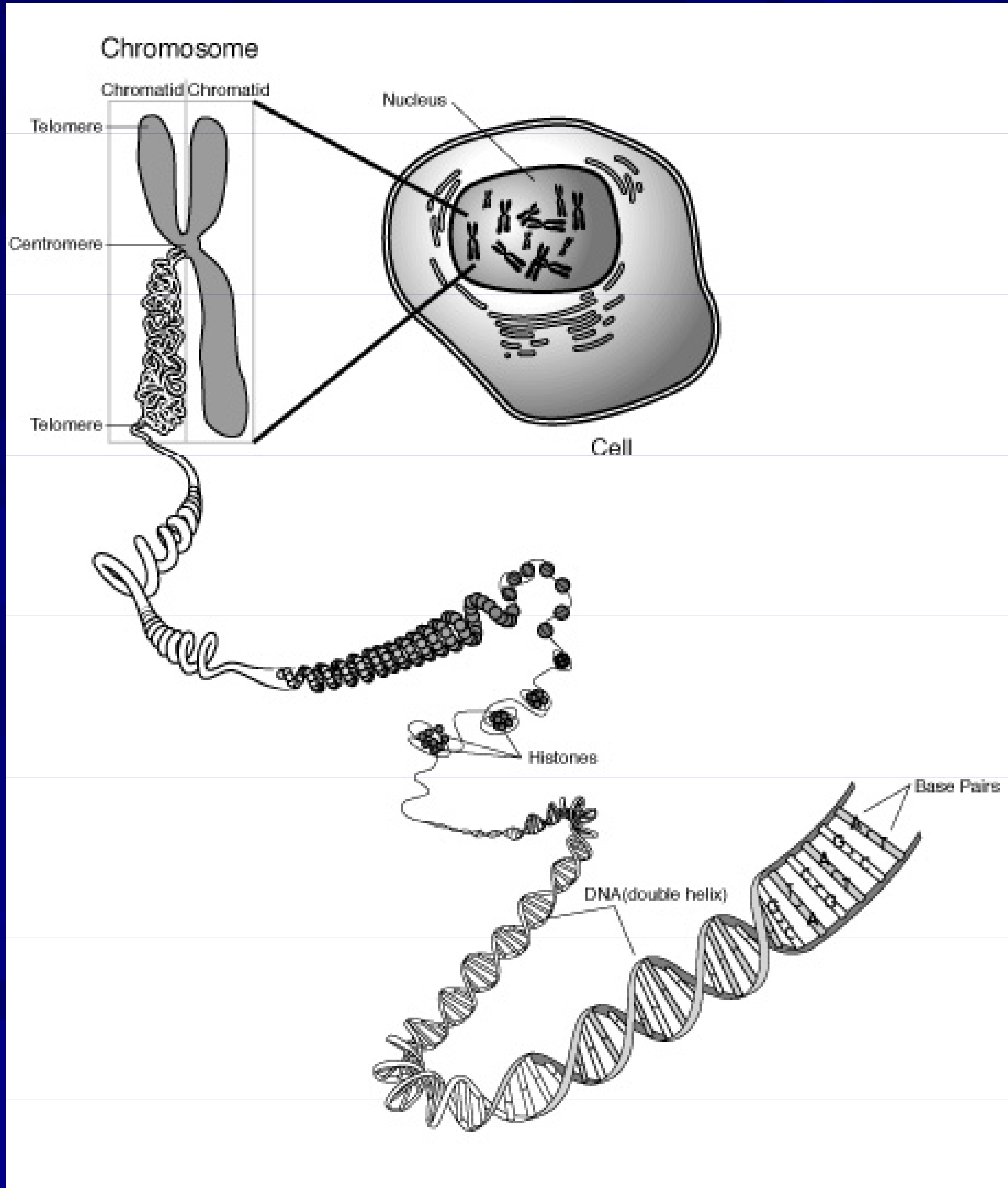
Nucleotide



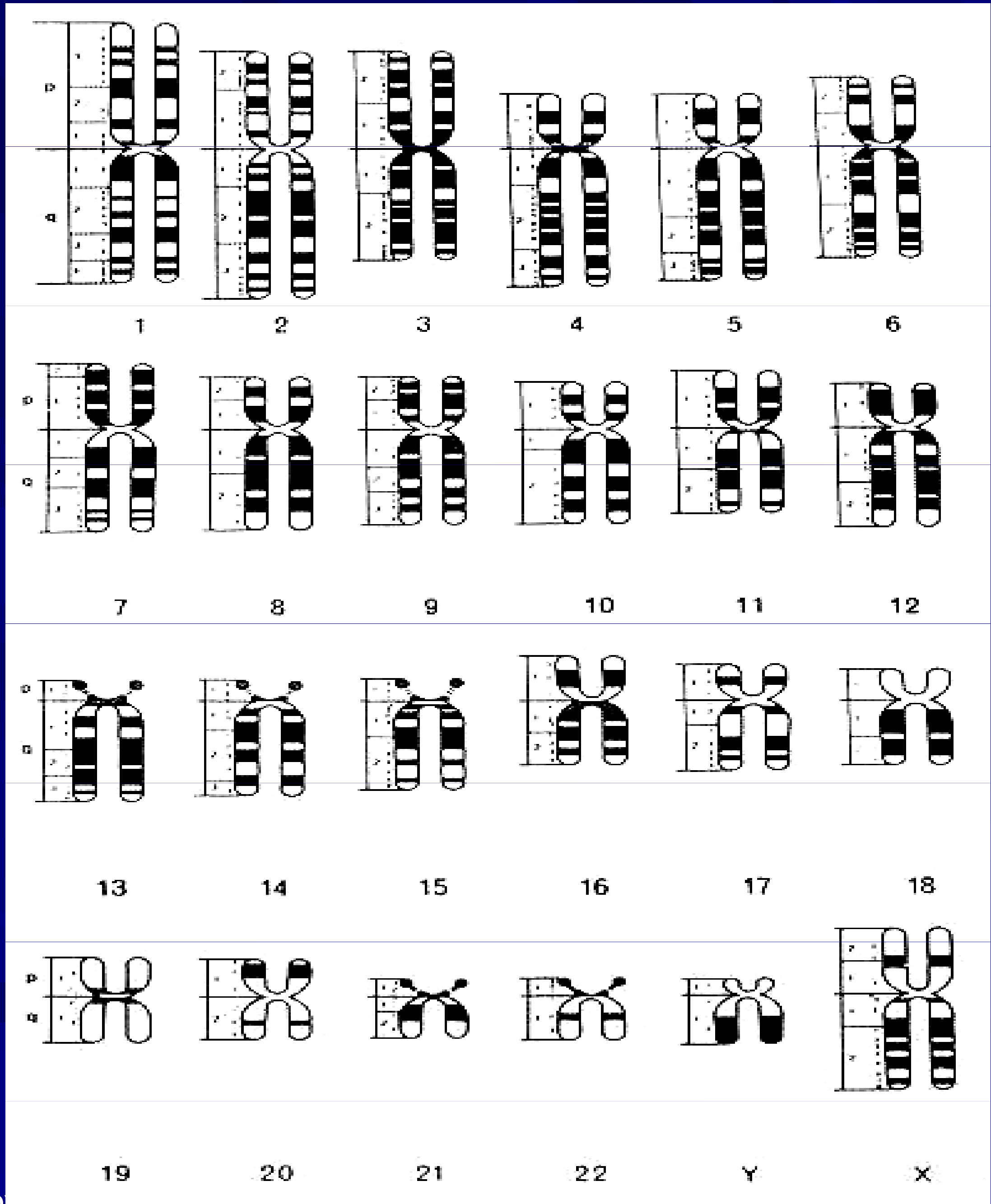
# The enormous genetic variability which exists in nature

AAACGTGCTTAGCTGGATAGGCTAGAGCTGATTCTCTTTCGATATAGGCCCTAGAA
ATCGATTCGCTAGAGATCGCATCGATACGTACAGTACATGATGCTACTAGCATTAGA
CTAGACTAGGATTTATTCTAGGCATGCTAGGACTAGGACCCCCCTAGTCTAGCCAT
GGTATGCAAACGTGCTTAGCTGGATAGGCTAGAGCTGATTCTCTTTCGATATAGGC
CCTAGAAATCGATTCGCTAGAGATCGCATCGATACGTACAGTACATGATGCTACTA
GCATTAGACTAGACTAGGATTTATTCTAGGCATGCTAGGACTAGGACCCCCCTAGT
CTAGCCATGGTATGCAAACGTGCTTAGCTGGATAGGCTAGAGCTGATTCTCTTTCG
ATATAGGCCCTAGAAATCGATTCGCTAGAGATCGCATCGATACGTACAGTACATGA
TGCTACTAGCATTAGACTAGACTAGGATTTATTCTAGGCATGCTAGGACTAGGACC
CCCCTAGTCTAGCCATGGTATGCAAACGTGCTTAGCTGGATAGGCTAGAGCTGATT
CTCTTTCGATATAGGCCCTAGAAATCGATTCGCTAGAGATCGCATCGATACGTACA
GTACATGATGCTACTAGCATTAGACTAGACTAGGATTTATTCTAGGCATGCTAGGAC
TAGGACCCCCCTAGTCTAGCCATGGTATGAGCTCTTAGCTACGTGCTTAGCTGGAT
AGGCTAGAGCTGATTCTCTTTCGATATAGGCCCTAGAAATCGATTCGCTAGAGATC
GCATCGATACGTACAGTACATGATGCTACTAGCATTAGACTAGACTAGGATTTATTC
TAGGCATGCTAGGACTAGGACCCCCCTAGTCTAGCCATGGTATGCAGTCGTCGATA
TAGGCTATAGGAAATCGATGTCGATGCTAGCTAGTAGCTGATCGATCGCTGATCGA
TGATGTATAGATCTCATCGATATAGGGCTCGATCGCTCGATATACTAGACTAGAGAA
TATATACACTCCCCGCTCGCTCGATAGCTAGCTCTAGGAGATTCGATAGGTA

# From DNA to the chromosome:



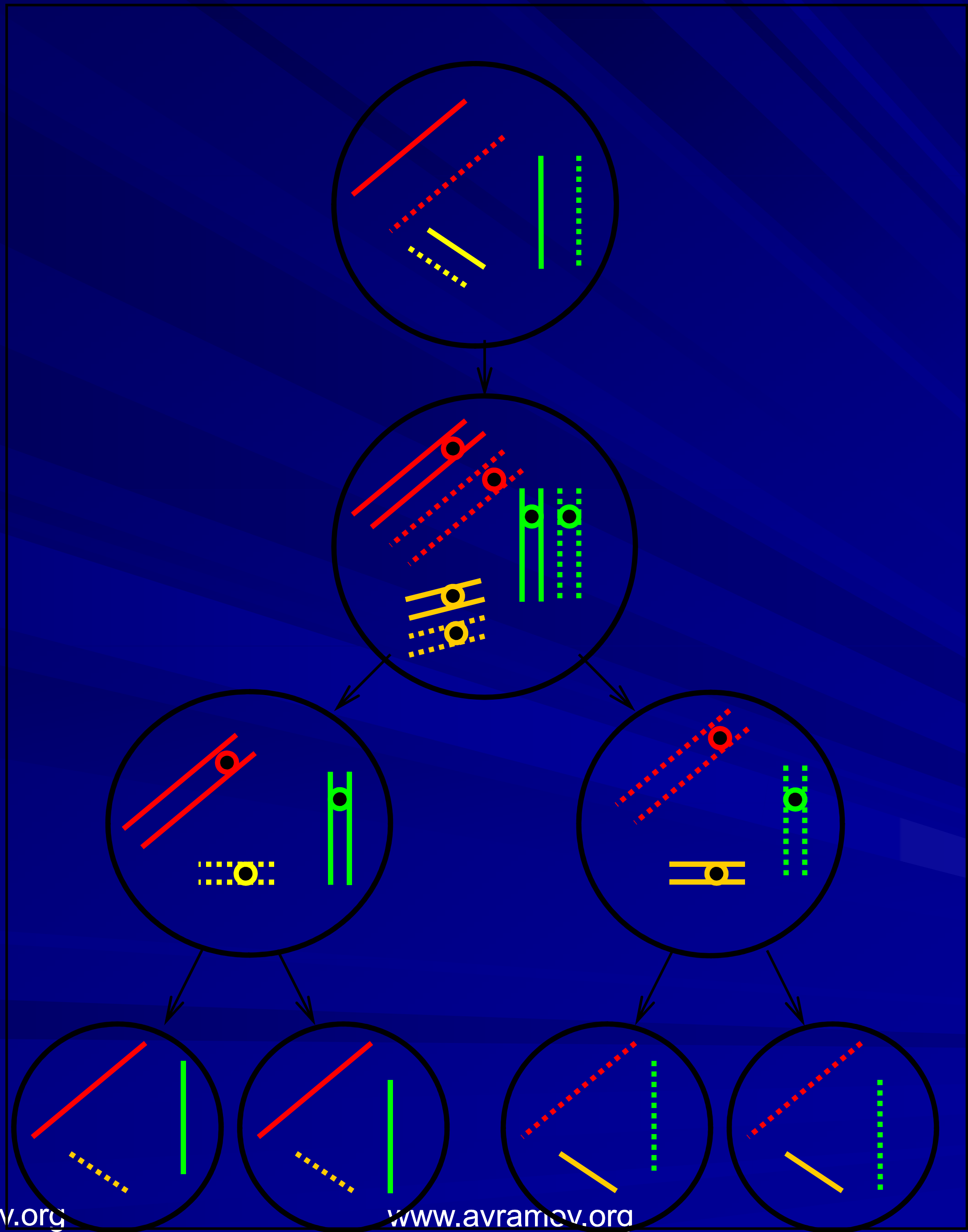
# The 46 human chromosomes:
# 22 pairs of autosomes and 1 pair of sex chromosomes



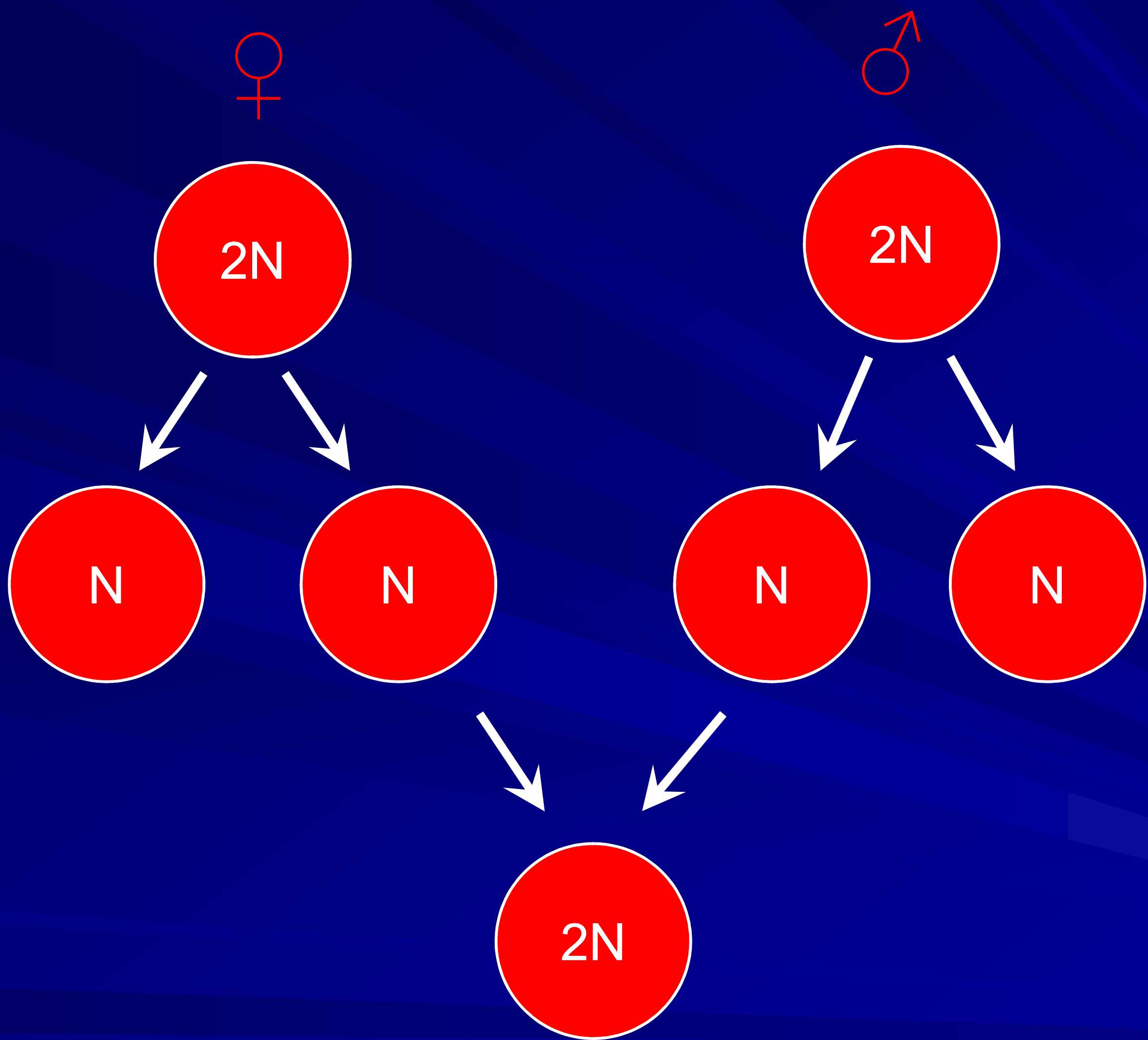
## Some additional genetic concepts

- Gene
- Monogenes and polygenes
- Allele
- Dominant and recessive alleles
- Genotype
- Homozygote genotype
- Heterozygote genotype
- Genome
- Phenotype

# Meiosis: segregation of genes



# Meiosis and fertilization: segregation and recombination of genes



# Variability at meiosis and fertilization

- n chromosome pairs = 23
- $2^{23}$ = 8.388.608 different gametes
- Crossing-over between homologeous chromosomes= >> $2^{23}$
- Recombination at fertilization: >> $2^{46}$

↓

**Biosocial impact:**

**children partly differ from parents**

# Genetic variability

- Humans = 99.6 to 99.8% of nucleotides are identical, but
- Other 0.2−0.4% nucleotides (± 10 million DNA variants) can potentially occur in different combinations

↓

**The biosocial impact:**

**The genetic unique identity of the individual**

# Mendelian laws
*(Brno, 1865):*

Uniformity
Segregation
Independence

Mendel, G.J. (1865), *Versuche über Pflanzenhybriden*. *Verhandlungen des Naturforschenden Vereines in Brunn*, IV, 3: 3-47.

# Mendel's uniformity law

P generation:

| | | |
|---|---|---|
| Genotypes: | AA | aa |
| Gametes: | A | a |

$F_1$ generation:

| | | |
|---|---|---|
| Genotype: | Aa | |
| Gametes: | A | a |

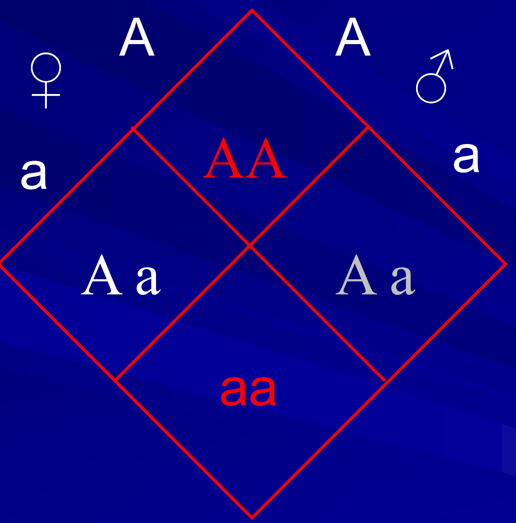
# Mendel’s segregation law

FI generatie:

Genotypen: Aa

Gameten: A a

F2 generatie:




1 AA : 2 Aa : 1 aa

# Mendel's independence law

P-generation:

Genotypes: AABB aabb

Gametes: AB ab

F1-generation:

Genotype: AaBb

Gametes: AB Ab aB ab

F2-generation:

AB AB

Ab AABB Ab

aB AABb AABb aB

ab AaBB AAbb AaBB ab

AaBb AaBb AaBb AaBb

Aabb aaBB Aabb

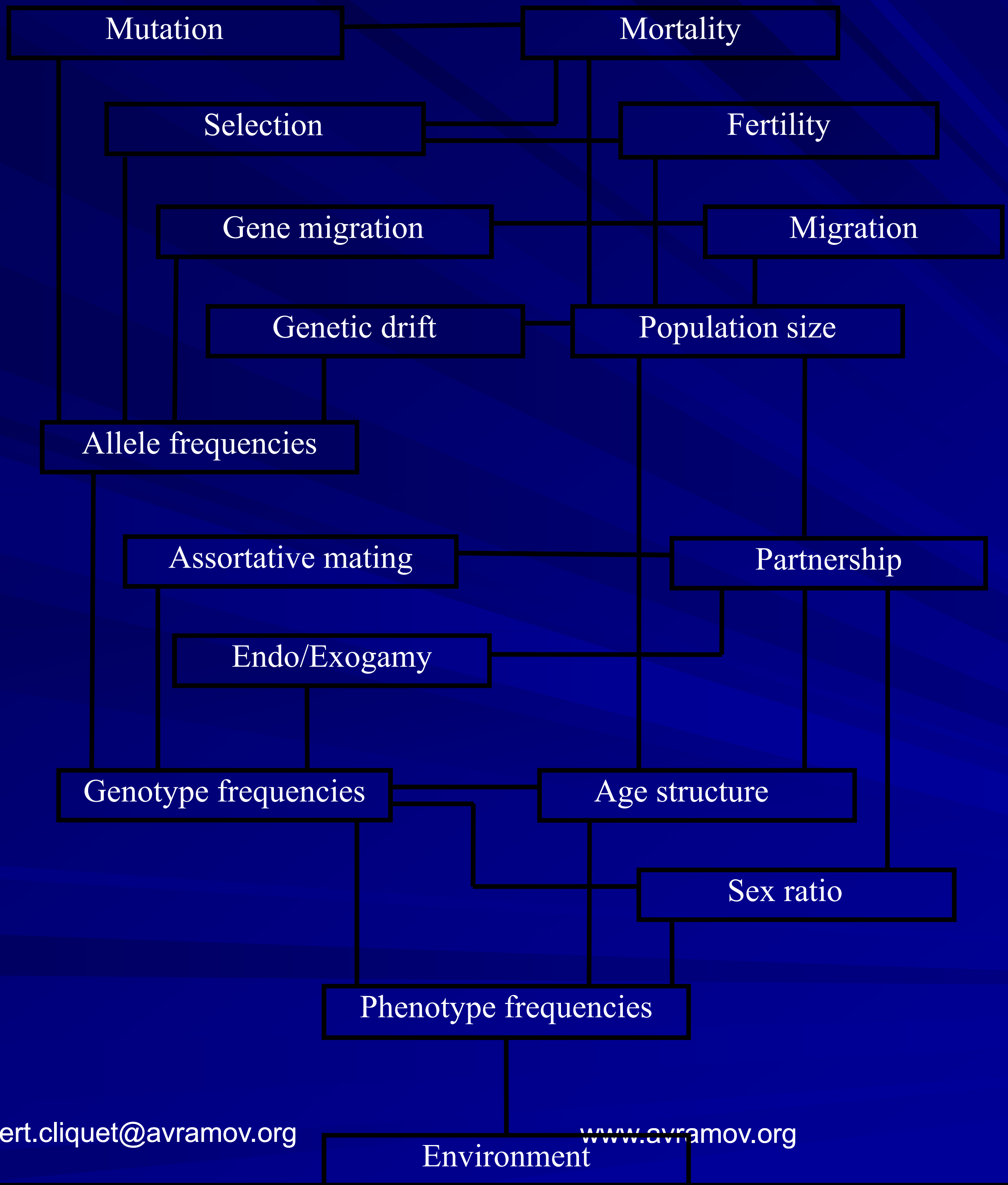
aaBb aaBb

aabb

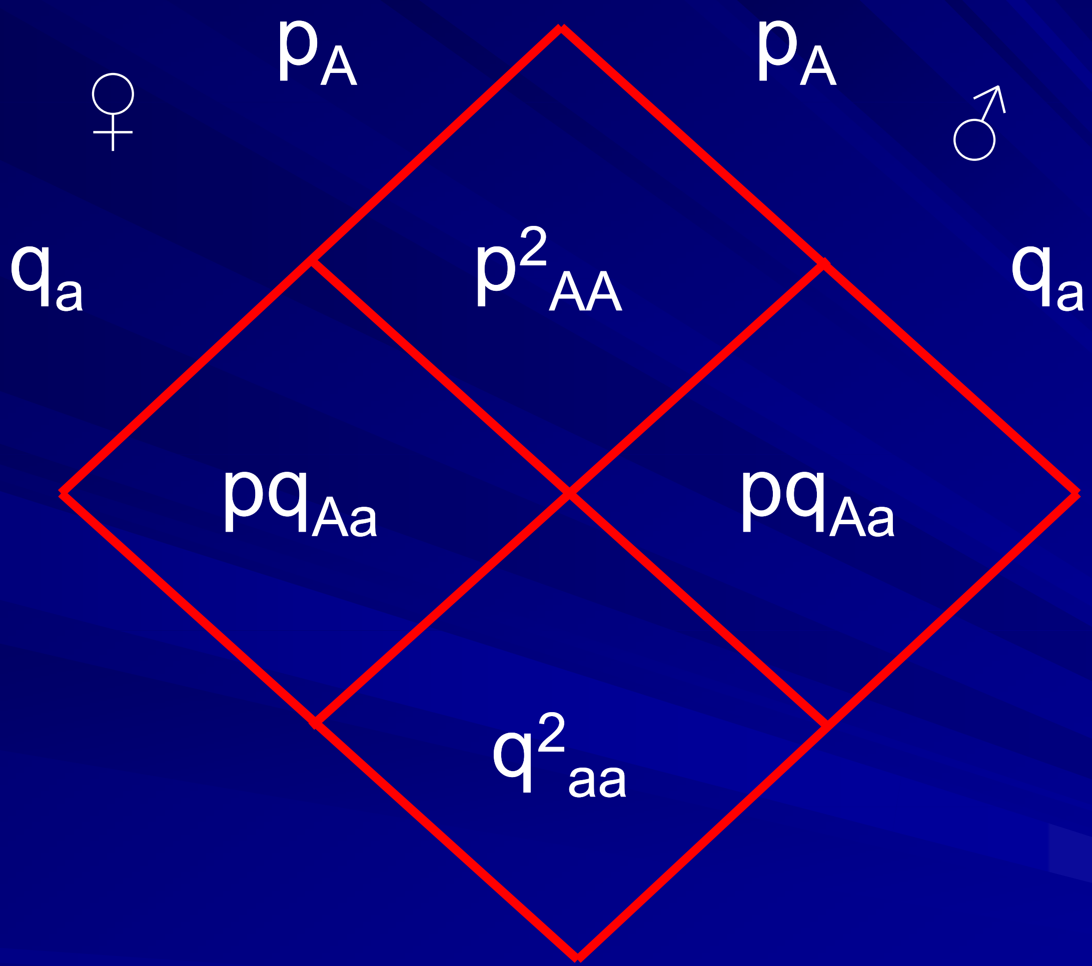
- The Hardy-Weinberg law
- Mutation
- Selection
- Genetic drift
- Genetic migration
- Partner choice

# The intertwining of demography and genetics

- Demography and genetics are closely intertwined: all of the basic demographic mechanisms and processes on the one hand and the population genetic mechanisms and processes on the other hand interact, often in multiple ways.
- The basic demographic variables - mating, fertility, migration and mortality - are the proximate instruments of population genetic changes, eventually leading to biological evolution.


Mutation
Mortality
Selection
Fertility
Gene migration
Migration
Genetic drift
Population size
Allele frequencies
Assortative mating
Partnership
Endo/Exogamy
Genotype frequencies
Age structure
Sex ratio
Phenotype frequencies
Environment

# Hardy-Weinberg law




$$p^2{}_{AA} + 2pq_{Aa} + q^2{}_{aa} = 1$$

# Hardy-Weinberg law

- describes the relations between the individual gene and genotype frequencies from one generation to another;
- allows understanding the complex genetic interrelations and interdependencies between the individual and population levels of genetic organization;
- shows that the allele and genotype frequencies in a reproductive community remain constant from generation to generation, maintaining a genetic equilibrium expressed by the binomial formula

$$p^2_{AA} + 2pq_{Aa} + q^2_{aa} = 1$$

# Hardy-Weinberg law: allele and genotype frequencies remain constant

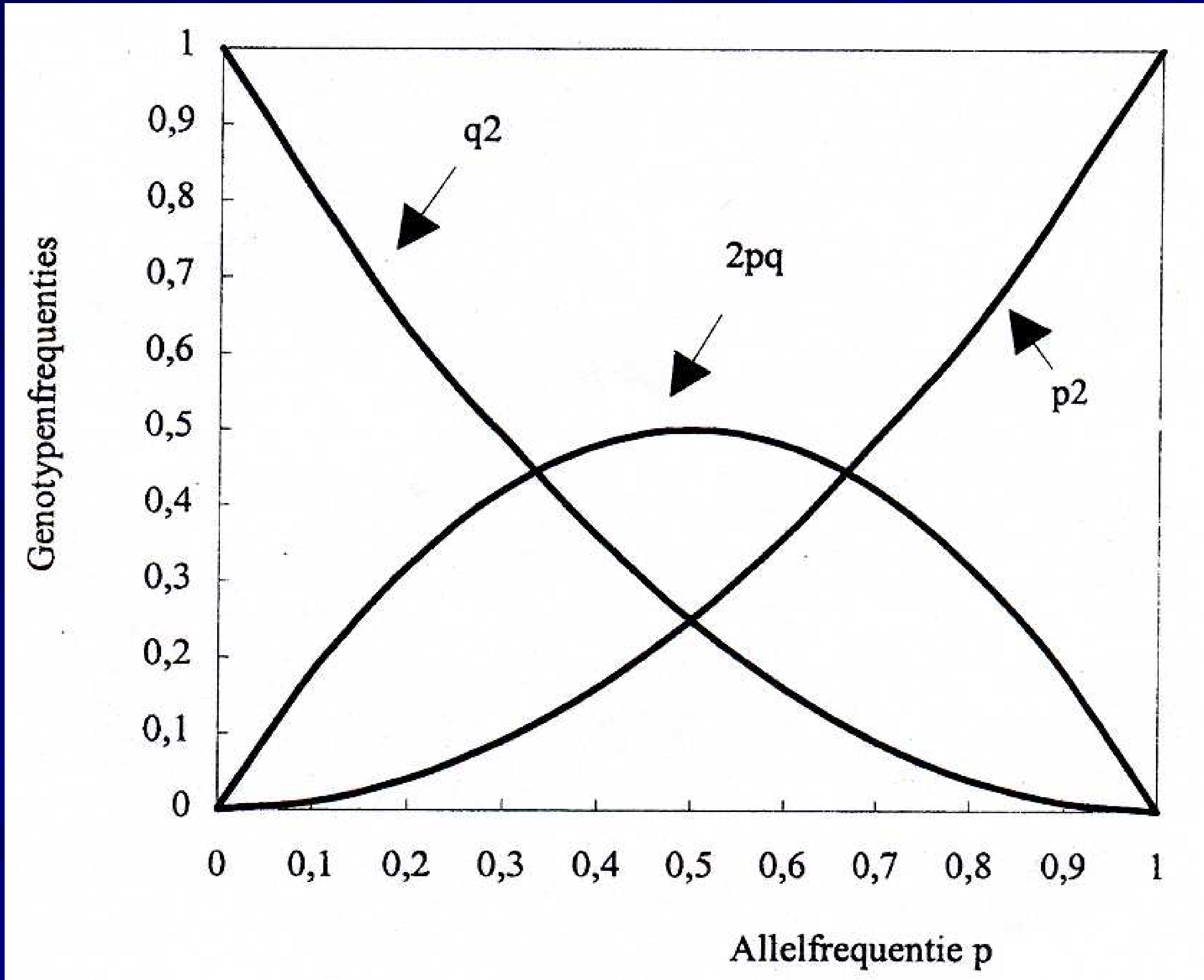
Parentes generation:

$$p^2_{AA} + 2pq_{Aa} + q^2_{aa} = 1$$

Filius generation:

$$A = p^2 + pq$$
$$A = p^2 + p(1 - p)$$
$$A = p^2 + p - p^2 = p$$
$$a = q^2 + pq$$
$$a = (l - p)^2 + p(l - p)$$
$$a = l - 2p + p^2 + p - p^2$$
$$a = l - p = q$$

# Hardy-Weinberg law



The Hardy-Weinberg equilibrium can be changed by all of the known evolutionary mechanisms:

- Mutation
- Selection
- Genetic drift
- Genetic migration
- Partner choice

# Mutation

- Mutation is a change in the chemical structure - the DNA - of a gene or a group of genes;
- Mutations are at the basis of the genetic variability;
- Mutations can have different effects:
  - Neutral;
  - Deleterious;
  - Favourable.
- In the human, less favourable ( = inadaptive) mutants can thrive because they have sufficient survival value in the culturally or economically protected environment or are even fostered by such environments

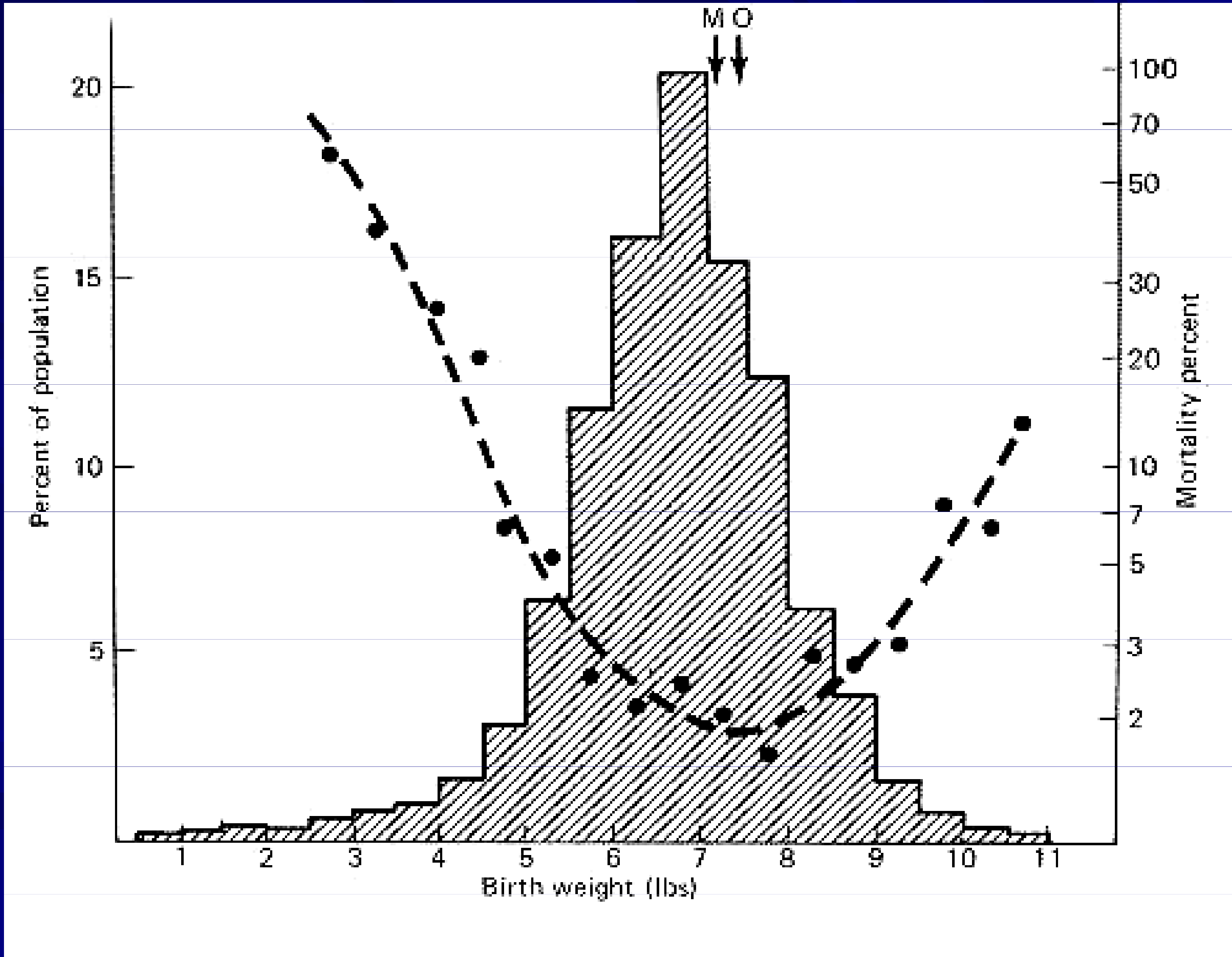
# Selection

- = differential reproduction of carriers of different alleles;

- Two major forms of selection:
  - Directional selection: continuous elimination or promotion of particular alleles;
  - Balanced selection: selective advantage of the heterozygote genotypes

- Many causes or levels of selection: e.g.
  - Natural selection
  - Social selection
  - Gene selection
  - Kin selection
  - Group selection
  - Sexual selection

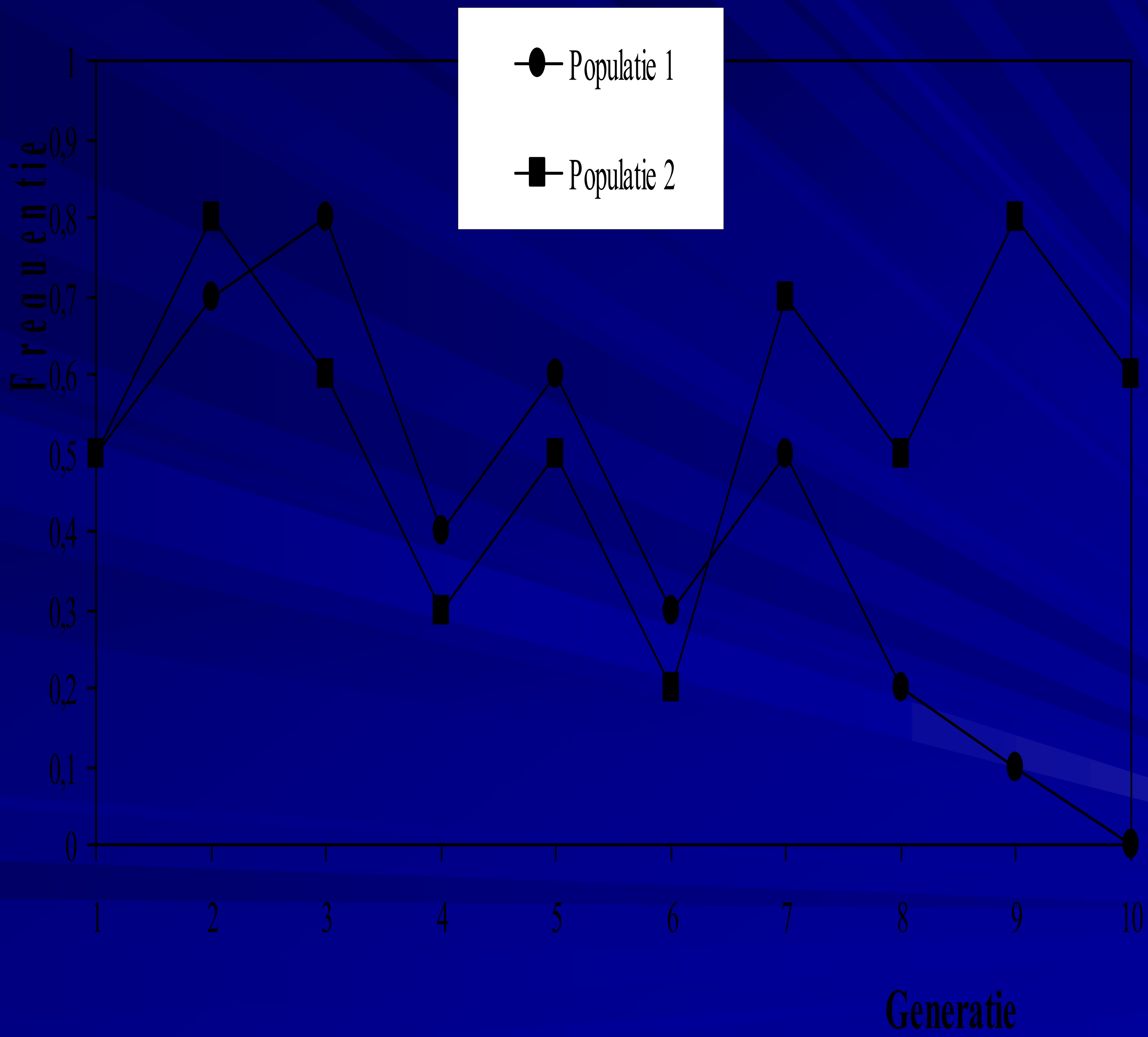
# Directional and balanced selection



# Genetic drift ('Sewell Wright' effect)

Changes the allele frequencies as a result of the accumulation of random fluctuations in the intergenerational transmission of the alleles in small populations

# Genetic drift: a computer simulation



# Genetic migration

- A genetically different population section leaves or joins a population.

- Genetic migration can occur on a small scale, at the level of individual (mate) exchange, or on a large scale, as a massive population invasion.

- It can occur as a single, non-recurring population move, or as a continuous gene flow between two or more populations

# Partner choice

- Random mating = Hardy-Weinberg equilibrium
- Selective mating = changes allele frequencies
- Assortative mating = changes genotype frequencies
  - Positive assortative mating
    - Inbreeding
  - Negative assortative mating
    - Outbreeding ('racial' admixture)

Assortative mating and inbreeding change the Hardy-Weinberg equilibrium to Wright's equilibrium

[p2+ Fpq] AA + [2pq(1-F)] Aa + [q2+ Fpq] aa = 1

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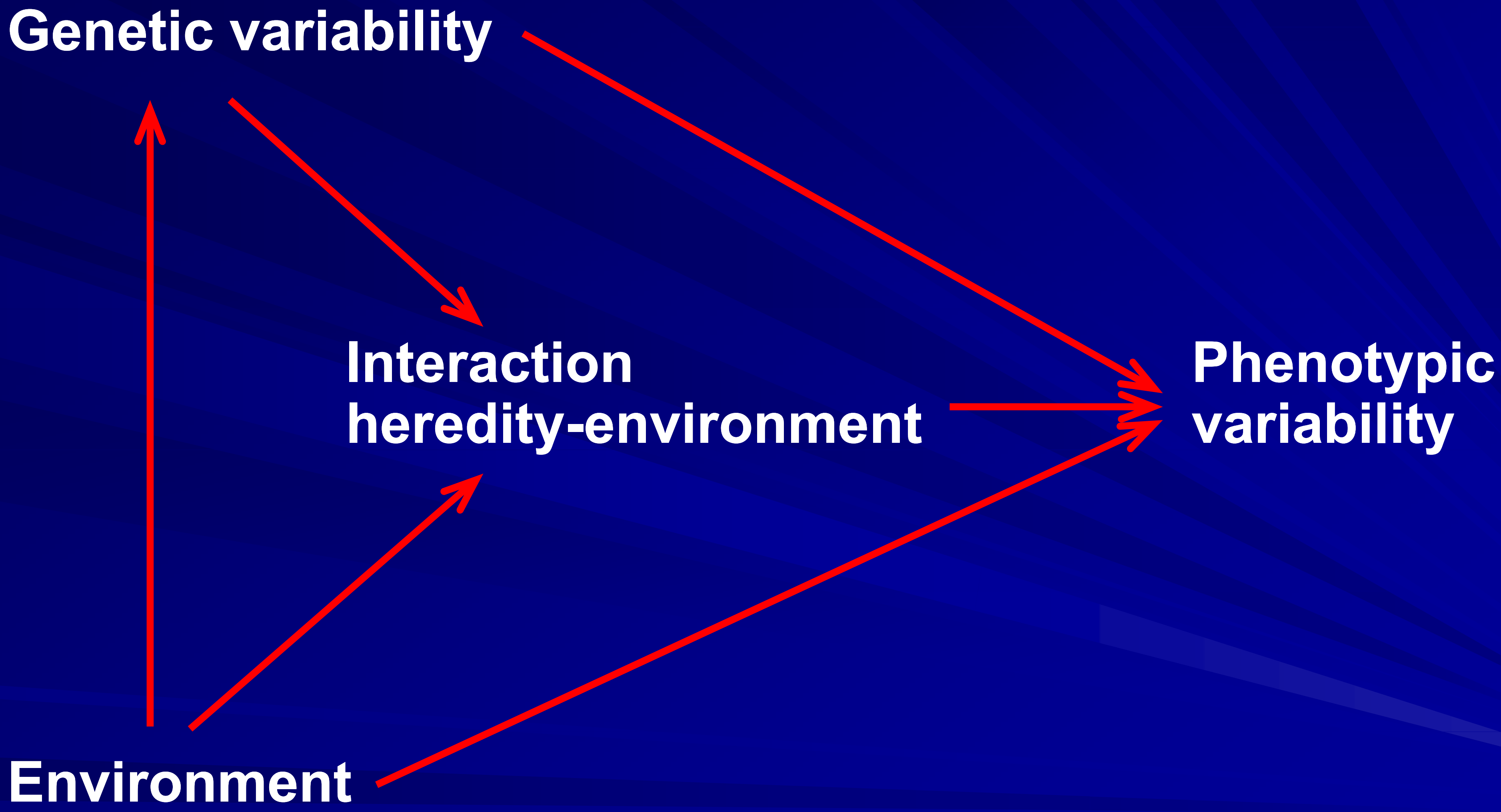
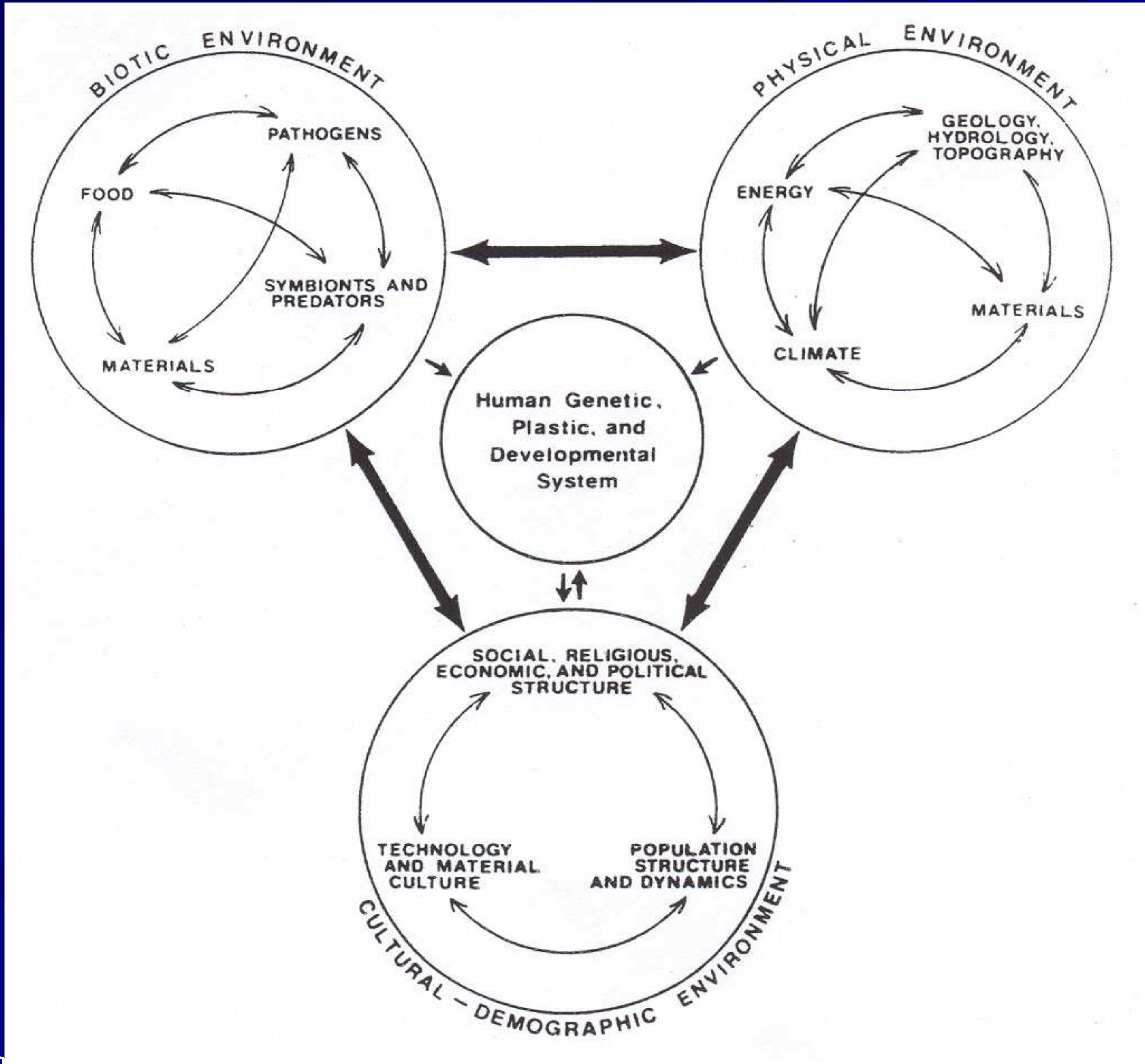


# The nature-nurture controversy

- Hereditarianism
  - Obsolete
  - Persists among extreme right wing and conservative thinkers
  - Absent in present-day biological sciences
- Environmentalism
  - Obsolete
  - Persists partially in social and behavioural sciences (= '*Standard Social Science Model*') and among many Marxist and left wing thinkers
- Integrated Causal Model (ICM)
  - Genetic and environmental (physical, non-genetic biological and socio-cultural/socio-economic) factors influence the human phenotypes
  - Generalised in human biological sciences, progresses slowly in social sciences, largely absent in politics.

# Interrelations between the human organism and its physical, biotic, and social environment



## Measuring the relative impact of genetics and environment

- Phenotype exclusively determined by genetic factors (e.g. blood groups);
- Phenotypic expression influenced both by genetic and environmental factors (e.g. most characteristics showing quantitative variability);
- Behavioural patterns exclusively determined by environmental circumstances (e.g. fashion?)

# QUANTITATIVE VARIABILITY

= Often socially important performance characteristics, e.g.

- Body characteristics;
- Cognitive abilities;
- Personality characteristics;
- Sexual and reproductive features;
- Maturation characteristics;
- Health characteristics

# The polygenic inheritance system

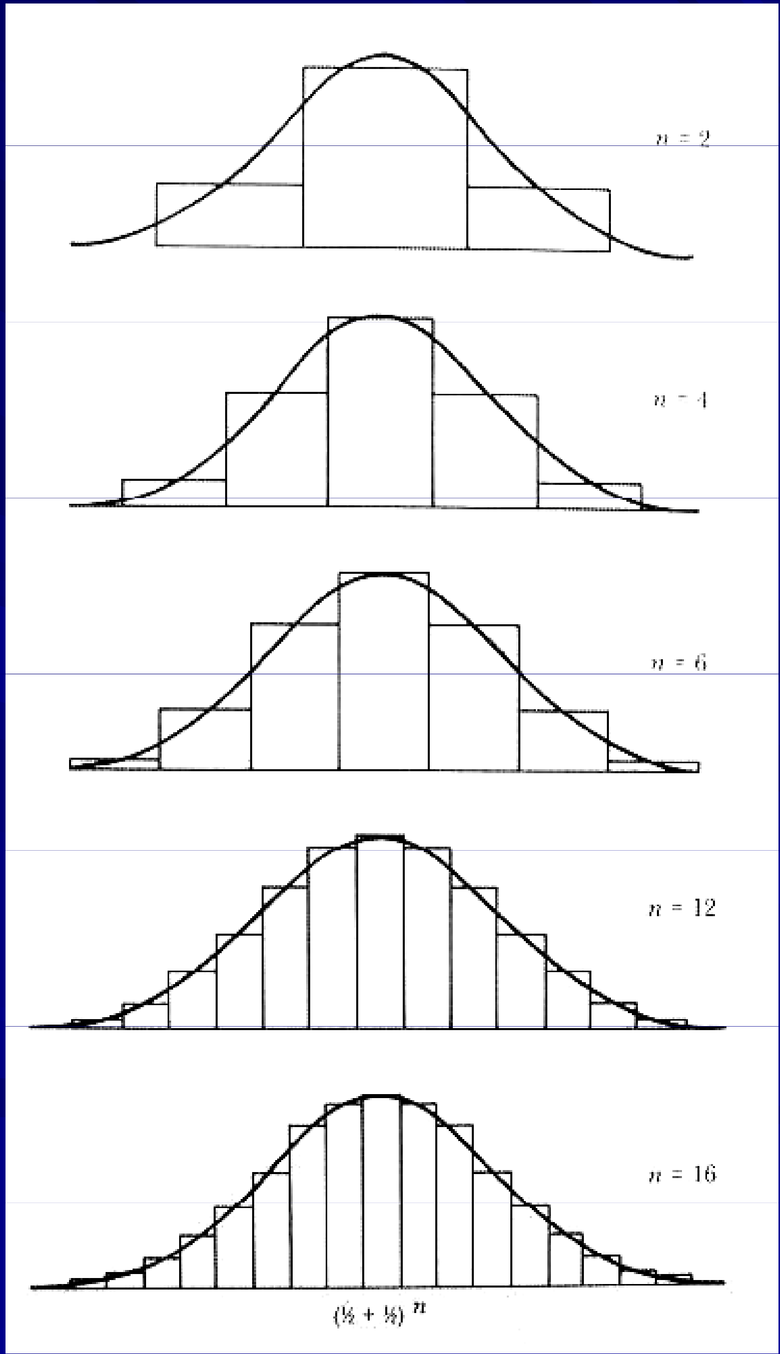
- Multiple factor hypothesis (Yule, 1906)
- Polygenes (Mather, 1941)
- Quantitative trait loci (QTLs) (Gelderman, 1975)

# The polygenic inheritance system

- Quantitative traits are affected by genes at several loci;
- The effect of alternative alleles at each of the segregating loci are relatively small and interchangeable;
- The phenotypic expression of most polygenic traits is subject to considerable influence of environmental factors during the ontogenetic development;
- Most populations include a large genetic variability for polygenic traits;
- Polygenetic characteristics abide by the same Mendelian laws as monogenes.

# The frequency distribution of a polygenic trait according to the number of allele pairs

(Fuller & Thompson, 1978)



# The binomial distribution for a polygenic trait with two allele pairs

$$[(p + q)^2]^2 =$$

$$(p + q)^4 = p^4 + 4p^3q + 6p^2q^2 + 4pq^3 + q^4 = 1$$

# Inheritance of a polygenic trait with two allele pairs

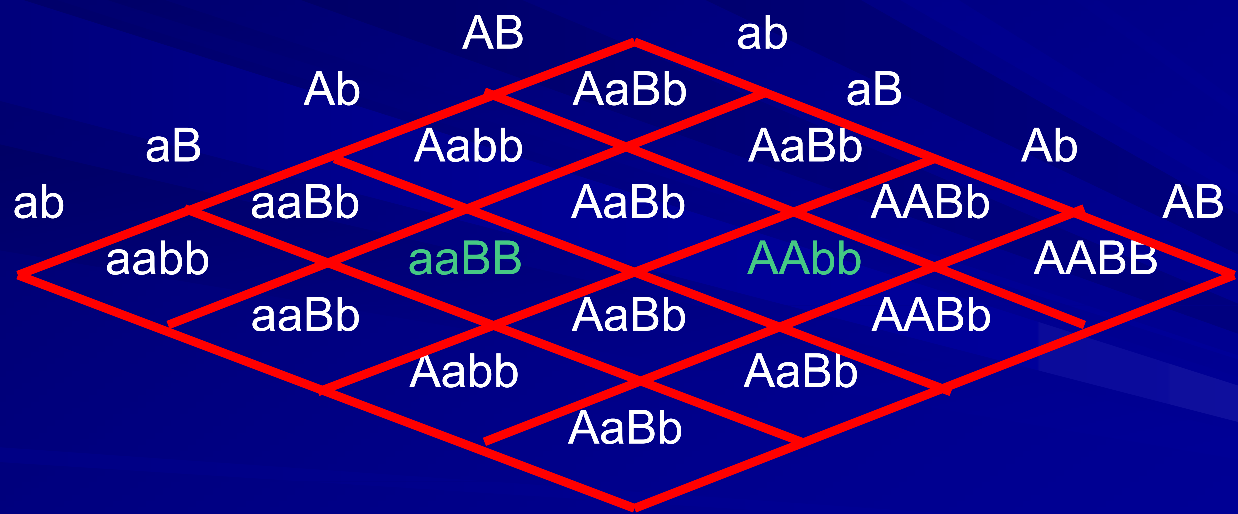
P-generation:

| | | |
|---|---|---|
| Genotypes: | AABB | aabb |
| Gametes: | AB | ab |

F1-generation:

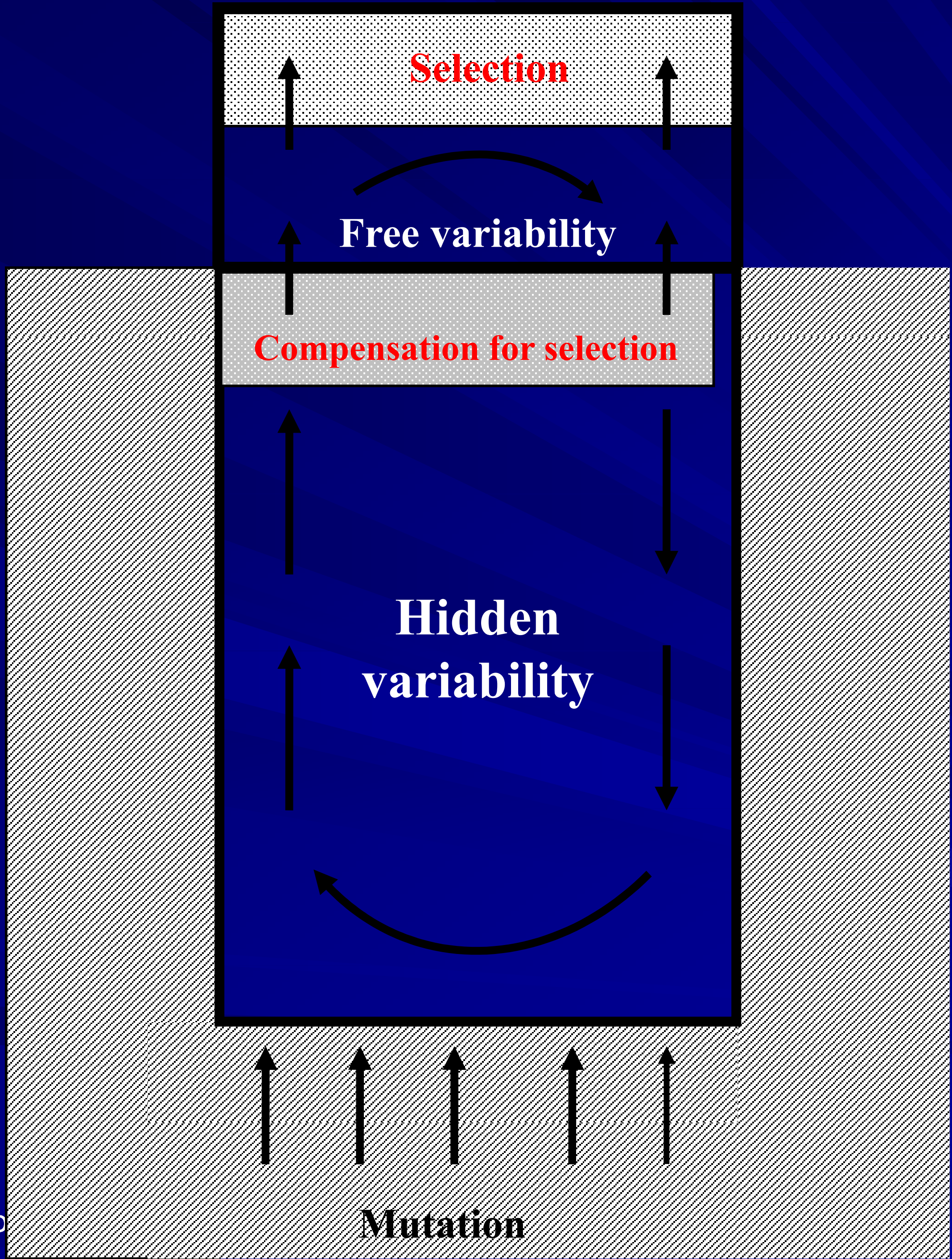
| | |
|---|---|
| Genotype: | AaBb |
| Gametes: | AB Ab aB ab |

F2-generation:




- **Phenotypes: 1 4 6 4 1**

# *The ‘iceberg’ effect of polygenes with 2 gene pairs*



# Number of allele pairs and the nature of variability

- Simple polygenetic model: more than one allele pair is responsible for the quantitative phenotypic variability of the characteristic, - e.g. body height, I.Q., blood pressure;

- Monogenetically inherited characteristics show, probably under influence of environmental factors, a quantitative phenotypic variability, - e.g. phenylketonuria (PKU);

- Polygenetic characteristics show phenotypically an alternative variability. It are the so-called *threshold characters*, known in the form of a number of diseases such as anencephaly, spina bifida, hare-lip, diabetes and schizophrenia.

# Relative importance of genetic and environmental factors

- At the individual level it is currently still impossible to answer the question about the relative importance of genetic and environmental factors in the realization of the phenotypes that fall within the range of the normal variation of a polygenetic frequency distribution (= depends on future developments in molecular genetics);

- At the population level, quantitative genetics has developed variance-analytic techniques that allow to estimate the relative importance of the effects of genetic factors, environmental factors, and their covariance and interaction in the production of within-population biological variability. Essentially it concerns the share of genetic endowment and environment in the realization of the differences between individuals in a population.

# Fractioning genetic and environmental effects on polygenic variation

$$\sigma_P^2 = \sigma_G^2 + \sigma_E^2$$

(P = phenotype, G = genetic factors, E = environmental factors, and $\sigma^2$ = variance)

A somewhat more realistic formula sounds as follows:

$$\sigma_P^2 = \sigma_A^2 + \sigma_D^2 + \sigma_{AM}^2 + \sigma_i^2 + \sigma_E^2 + \sigma_I^2 + 2\operatorname{cov}_{GE} + \sigma_e^2$$

A = additive genetic variance
D = variance caused by dominance deviation
AM = variance caused by assortative mating
i = epistasis variance
E = environmentally determined variance (to be subdivided in many subfractions)
I = gene-environment interaction-variance
2 covGE = covariance between genetic and environmental factors
e = variance due to measurement errors

# Heritability and modificability

- Heritability = the fraction of the phenotypic differences between individuals which can be attributed to genetic differences (Lush, 1940);

$$h_b^2 = \frac{\sigma_G^2}{\sigma_P^2} = \frac{\sigma_G^2}{\sigma_G^2 + \sigma_E^2}$$

- Modificability = the fraction attributed to environmental factors (Cattell, 1971).

# Misunderstandings about heritability

- Heritability does not identify the degree to which genetic and environmental factors determine the phenotype of an individual

- $h^2 = 1$ does not mean that environmental factors are not involved in the development of the phenotypes, but only that all phenotypical differences within a population are of genetic origin

- Heritability estimates for a particular trait are not a constant

- The heritability of a trait can also change without a change of the genetic composition of the population

- The heritability coefficient is a within-population measure

# The fractioning of IQ: an important but sensitive matter

- Fractioning of IQ: Human cognitive (< Latin: *cognitio* = getting to know) abilities belong to the most studied and best known characteristics in psychology and behavioural genetics;

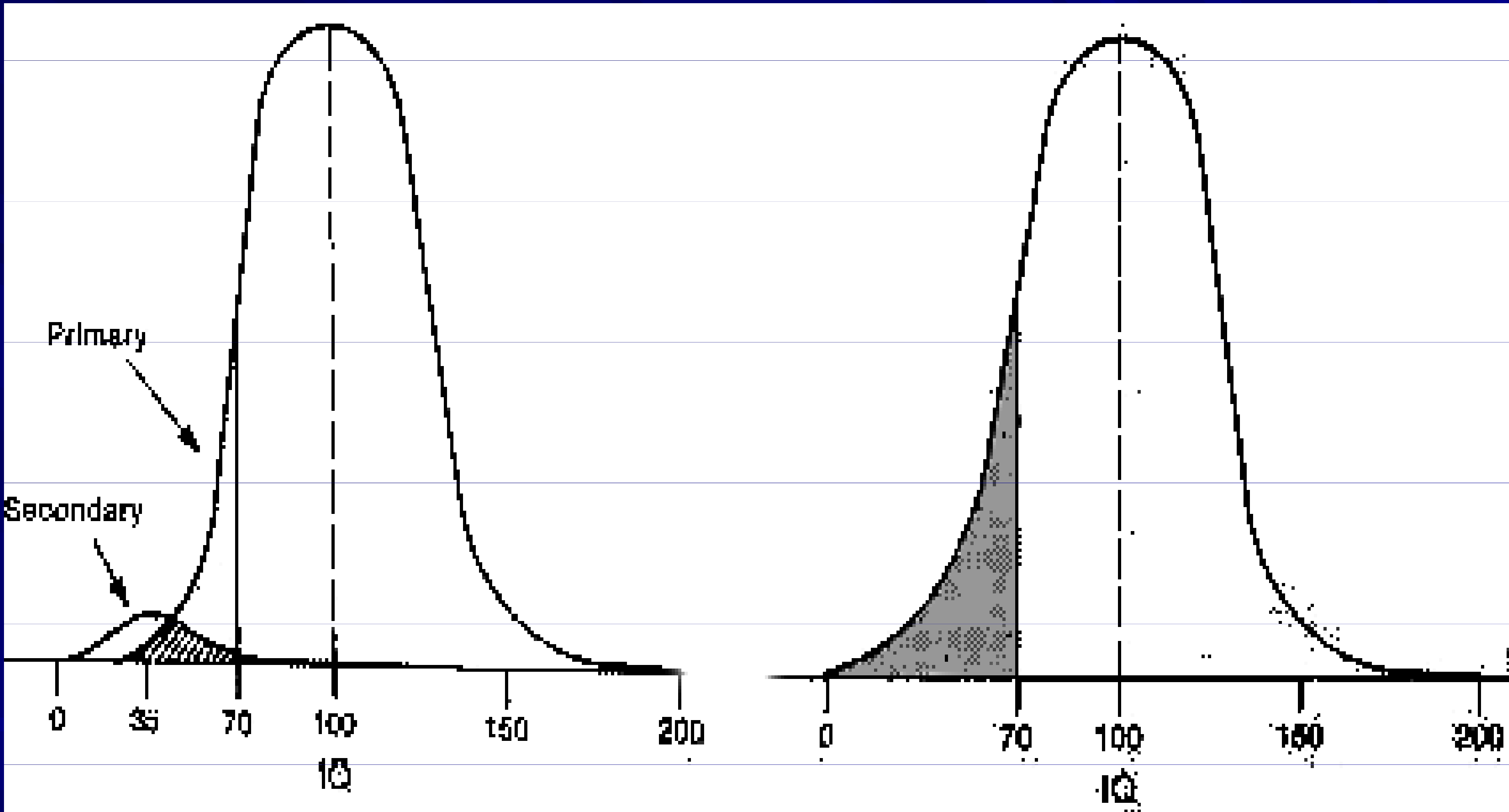
- Importance: cognitive abilities belong to the most specific human features characteristic for the hominization process resulting in the relative freedom from and control of the natural environment the human has been able to acquire;

- Sensitivity: relates partly to the ideological, hence prejudiced conceptualization people harbour about IQ, partly to societal problems related to between-group (SES and 'race') differences in measured intelligence.

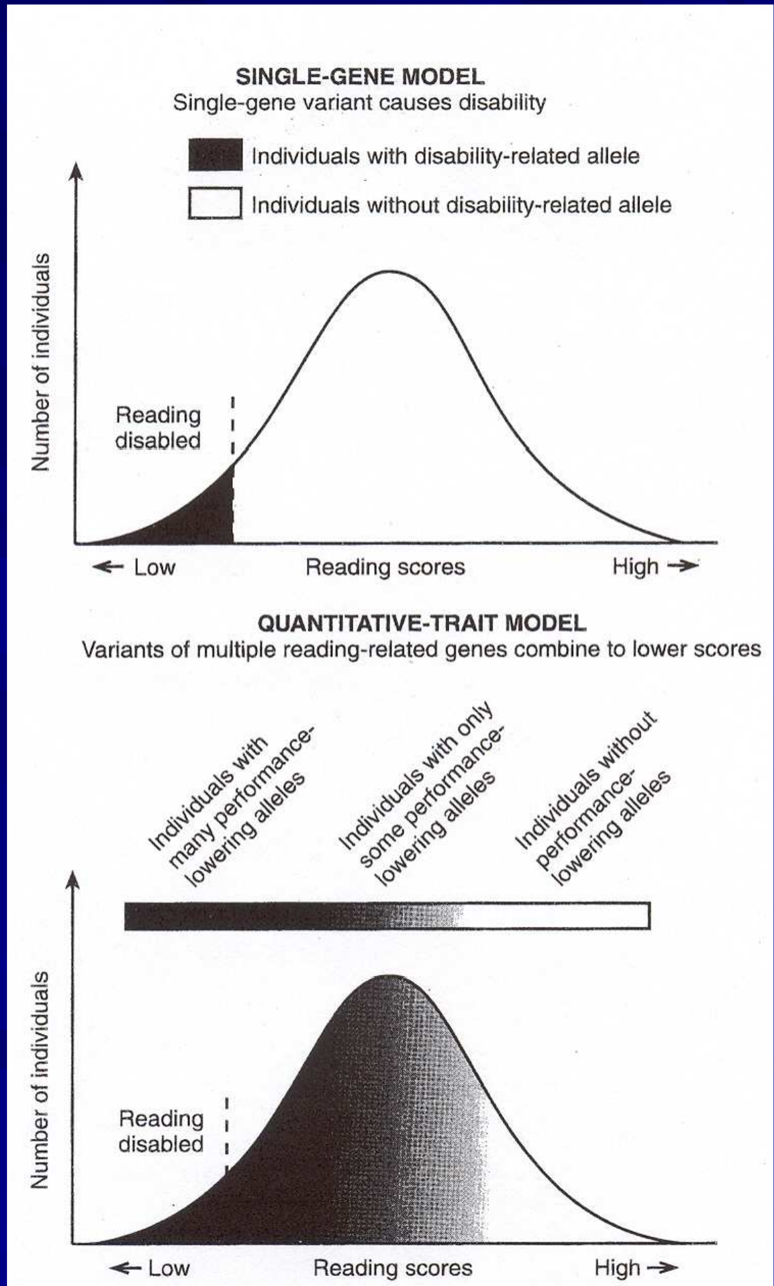
# *Defining and measuring dimensions of cognitive abilities*

- Cognitive abilities are often grouped under the comprehensive and multifactorial concept ‘intelligence’;
- General intelligence (the ‘g’ factor): positive correlation between all human cognitive skills (Spearman, 1904; Carroll, 1993; Jensen, 1998);
- Specific mental abilities: reasoning, verbal ability, spatial ability, perceptual speed, and memory;
- IQ (intelligence quotient) = a comparison of a subject's measured score with the average score of the population of the same age that is taken as norm;
- Intelligence tests = ‘fluid’ (culture-fair tests) and ‘cristallized’ intelligence (Cattell, 1963; 1971);
- Intelligence A, B and C (Eysenck, 1984): A = biological basis; B = behavioural component ; C = measured component.

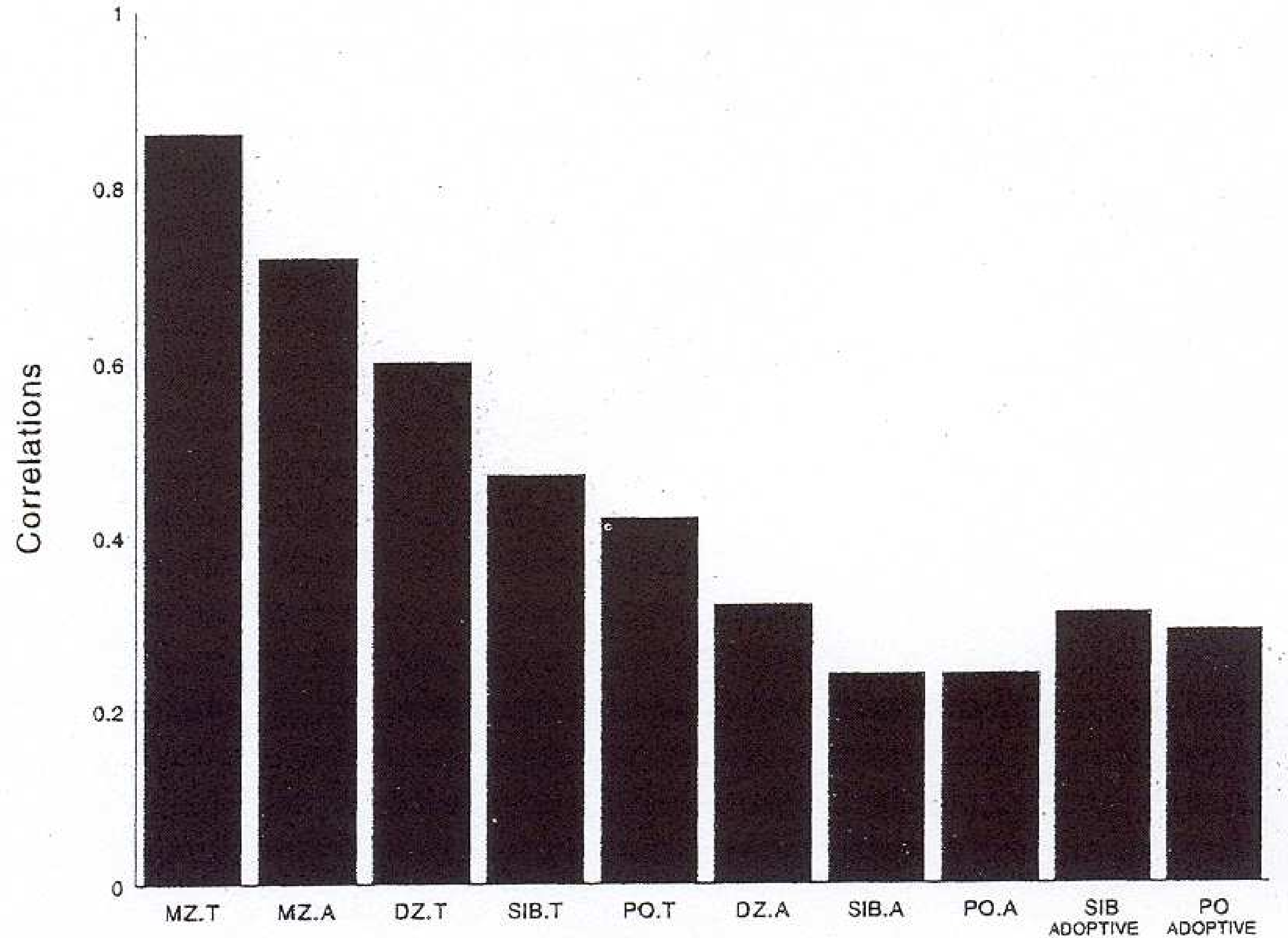
# Intelligence: monogenes, polygenes and environment



# Single gene model and polygenetic model as explanation for reading disabilities (Plomin, 1999)



# *Kinship correlations for IQ (Bouchard & McGue, 1981)*



# Heritability estimates for intelligence

- Averaging the heritability estimates derived from the different kinship correlations gives a broad heritability of ± 0.70;

- More advanced biometrical analyses in which all the data on people of various degrees of kinship reared together or apart are dealt with simultaneously, taking also into account effects of dominance, assortative mating, gene-environment interaction and covariance, lead to identical results, although with a somewhat broader range between about 0.50 and 0.80;

- A recent analysis, taking also into account the maternal effect, estimates the narrow-sense heritability at 0.34 and the broad-sense heritability at 0.48.

# Behavioural genetics: genes and environment

- Contrary to what is sometimes assumed, behavioural genetics provides information on the degree of environmental influence as well as it does on genetic factors.
- The heritability estimates of cognitive abilities suggest that environmental factors do still have a non-negligable effect on the within-population variation in modern societies

# The search for intelligence QTLs

- Since 1990 an IQ-QTL project was launched to systematically search for QTLs associated with normal variation in general intelligence (Plomin et al. 1994; Plomin, 2003).

- Several DNA markers have already been found to be associated with general cognitive ability (Plomin et al., 1994; 1995; Chorney et al., 1998; Plomin et al., 2001; Harlaar, et al., 2005).

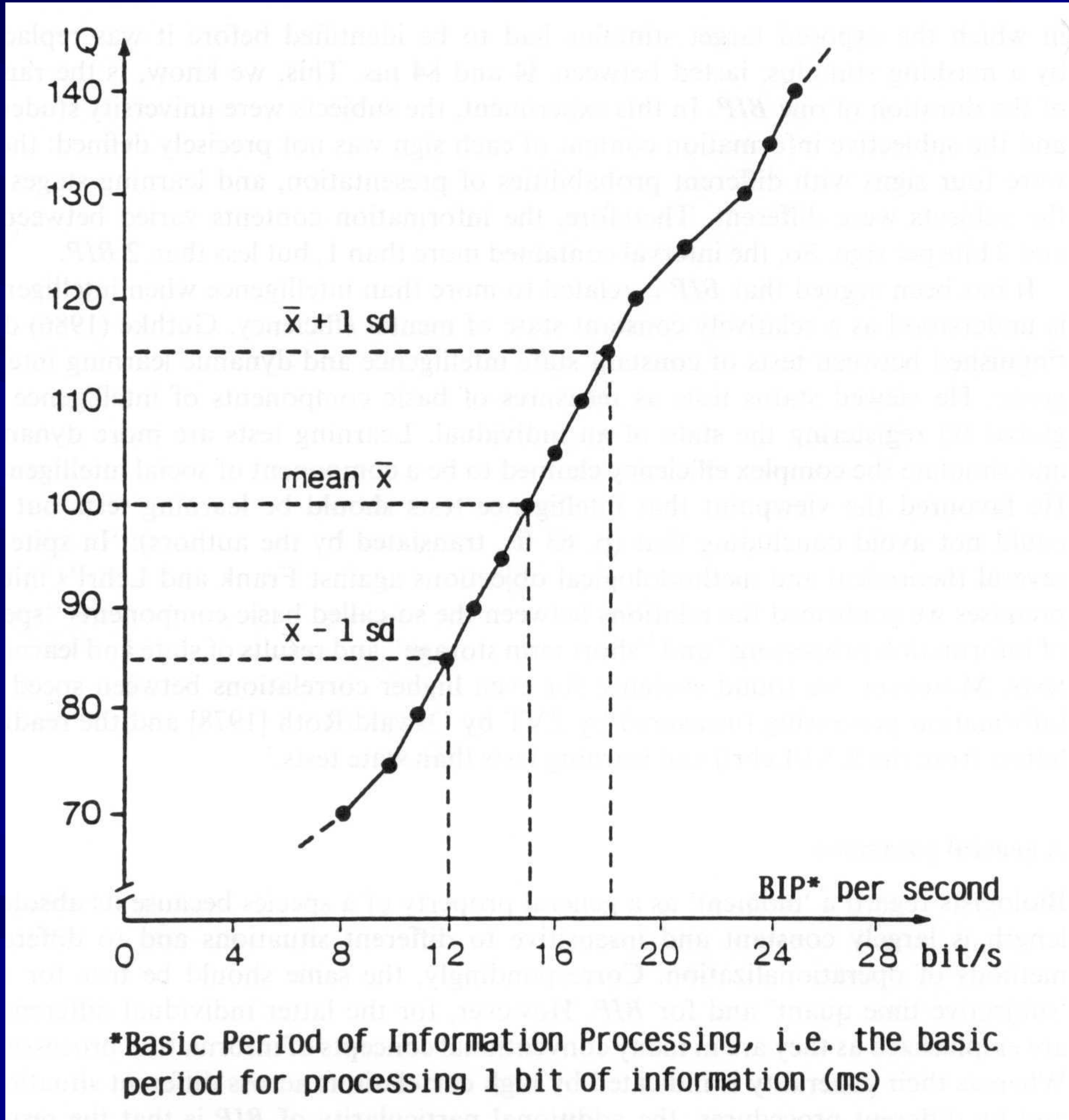
- However, progress towards identifying quantitative trait loci (QTLs) for intelligence has been slower than expected, probably because most QTL effects may be much smaller than expected and can only be detected by more powerful analyses (Plomin et al., 2006).

# Correlations between cognitive ability and neuro-physiological traits

- Brain size:  0.30 to 0.40
- Glucose metabolism : -0.7 to -0.8
- Reaction time (RT) : -0.3 to -0.4
- Inspection time : ≈ −0.30
- Speed of information processing (BIP): -0.60
- Average evoked potential (AEP): 0.3 to 0.6
- Index of neural adaptability (NA): 0.5 to 0.7
- General health or physical well-being: ≈ 0.4

# Relationship between the amount of basic period of information processing' (BIP) per second and IQ

## (Lehr and Fischer, 1990)



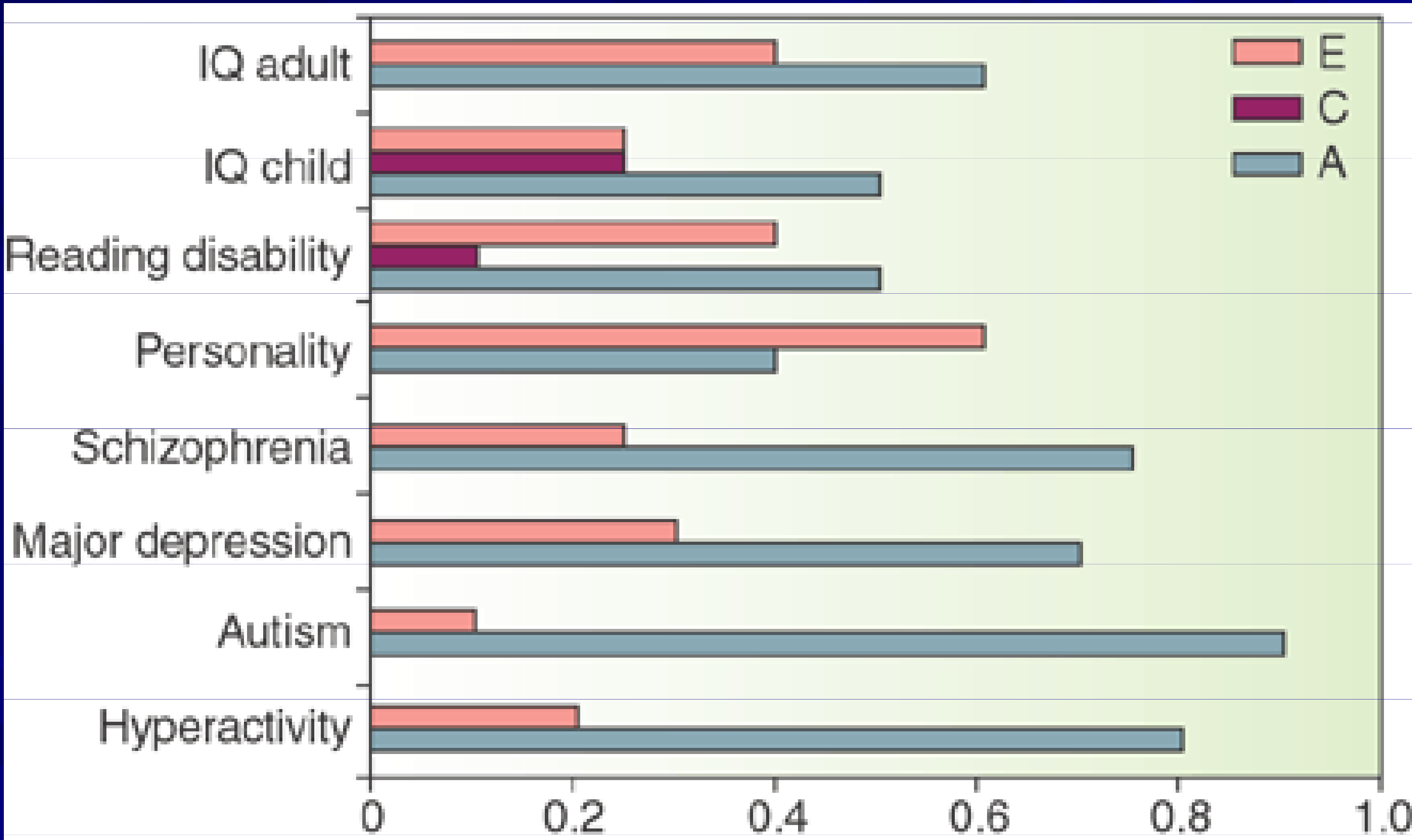
## The secular increase in measured intelligence (Flynn-Lynn effect)

- Measured intelligence levels were rising in economically prosperous countries in de period 1920-1990, but may have come to an end in the 1990s (cfr. body height);
- Possible explanations:
  - (1) cognitive stimulation provided by modern culture, especially education;
  - (2) improvements in the biological environment;
  - (3) changes in the composition of the population.

# Role of environmental factors in the development of intelligence

- Shared environmental factors such as SES, education, and other stimulating cultural factors seem to have a limited effect on innate cognitive abilities;
- Most of the non-genetic variance in IQ relates to variance within families, the so-called non-shared environmental factors, i.e. environmental factors that are specific for each child within a family.

# Heritability (A), shared- (C) and non-shared (E) environmental effects on intelligence



# The substantial role of the within-family biological micro-environment

- General health;
- Suppression of diseases;
- Improved nutrition;
- Increasing but not too high maternal age;
- Healthy life style during pregnancy;
- More favourable obstetrical care;
- Avoidance of premature birth, low birth weight;
- Breast feeding.

## Effects of compensatory intervention programmes on intelligence

- Different views of experts;
- Compensatory intervention programmes have positive effects on test learning, scholastic achievements, physical development and behavioural outcomes in general;
- Such programmes would have only limited long lasting effects on cognitive ability