POPULATION GENETICS

CHW population

(limiting conditions)

- large = more than 1000 individuals
- panmictic
- no mutations
- no selection
- no migration
- monogenic traits - at least two different alleles
- non-overlapping of generations

Castle-Hardy-Weinberg law

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

Applied on panmictic population under the assumption of limiting conditions

Castle-Hardy-Weinberg law

Basic relation for a system with two alleles in a given gene

$$p_{(A)} + q_{(a)} = 1$$

$$p_{(A)} = 1 - q_{(a)}$$

approximation

$2\,pq_{(Aa)} \doteq 2q$, **if** $p_{(A)}$ **approaches** 1

Task 1/p.139 – frequencies of alleles in MN blood group system

Phenotype	Number of persons
M	406
MN	744
N	332

Task 1/p.139 – frequencies of alleles in MN blood group system

Solution:

- direct calculation of the frequency of one of alleles according to formula [3] on p. 138

$$p = \frac{2 \times \text{number of homozygotes } (AA) + \text{number of heterozygotes } (Aa)}{2 \times \text{number of all individuals in the sample}}$$

- Calculation of the frequency of the second allele as addition to 1.

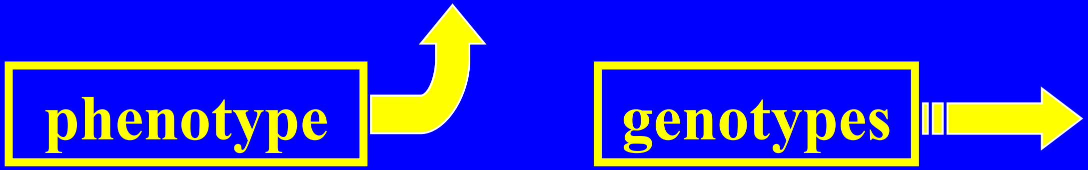
Task 1/p.139 – frequencies of alleles in MN system

phenotype	Number of		
	persons	alleles M	alleles N
M	406	812	0
MN	744	744	744
N	332	0	664
Total	1 482	1 556	1 408

$$p = \frac{2 \times 406 + 744}{2 \times 1482} = \frac{1556}{2964} = 0.525 \qquad q = 1 - p = 0.475$$

Task 2a/p. 139 – Estimation of gene frequencies in the dominant system

30 % of persons unable to recognize the bitter taste of PTC in a given population (non-tasters)



phenotype

genotypes

***TT*, *Tt* – tasters**

***tt* – non-tasters**

$q^2_{(tt)} = 0.3 \quad \Rightarrow \quad q_{(t)} = \sqrt{0.3} = 0.548$

$p_{(T)} = 1 - q = 0.452$

$p^2_{(TT)} = 0.205$

$2pq_{(Tt)} = 0.495$

Estimation of gene frequencies in two-allele polymorphism (*T*, *t*)

in the population sample of students

phenotyping of PTC tasting in students in the classroom:

one drop of saturated solution of phenylthiocarbamide (PTC) on the tip of the tongue

---

$$\text{portion of non-tasters } [q^2_{(tt),\text{ class}}] = \frac{\text{non-tasters in the class}}{\text{total of students in the class}}$$

$$\text{frequency of recessive allele } [q_{(t),\text{class}}] = \sqrt{q^2_{(tt),\text{ class}}}$$

$p_{(T),\text{class}} = 1 - q$ $\quad p^2_{(TT),\text{class}}$ $\quad 2pq_{(Tt),\text{class}}$

Task 4/p. 140 – estimates of frequencies of deleterious (recessive) alleles

Disease	Abbrev.	population frequency
phenylketonuria	PKU	1/8100
cystic fibrosis (mucoviscidosis)	CF	1/2500

Task 4/p. 140 – estimates of frequencies of deleterious (recessive) alleles

Solution: the estimate calculated according to formula [4] on p. 139 (top),

$$q = \sqrt{\frac{\text{number of recessive homozygotes}}{\text{number of all individuals in the sample}}} =$$

$$= \sqrt{\text{frequency in population}}$$

Task 4/p. 140 – estimates of frequencies of deleterious (recessive) alleles

disease	Frequency in population	estimate		
		q	p = 1 - q	2pq ≐ 2q
PKU	1/8100	1/90	89/90 ≐ 1	2 x 1 x 1/90 = 1/45
CF	1/2500	1/50	49/50 ≐ 1	2 x 1 x 1/50 = 1/25

Small population

- less than 1000 individuals

Gene drift

Simple model of gene drift I

Drift – changes in gene frequencies in small populations elicited by random processes

- **modelling of random use of alleles (gametes)**
- **random mechanism – dice throw**
- **small population $N = 3$, and original $p = q = ½$**
    - **allele A picked out … side 1, 2 or 3 cast**
    - **allele a picked out … side 4, 5 or 6 cast**

- **6 throws each time – results of series mark in the table**
- **According to the result (ratio, new frequencies) p_A / q_a change the rules for next series of throws (generation)**
- **continue until one allele fixed**

Simple model of gene drift II

– Task 14/p. 148

Into your protocols, prepare this table, at the least to the 10th generation

Generation		1		2		3			10 (20)	
Alelle		*A*	*a*	*A*	*a*	*A*	*a*		*A*	*a*
side casted		1, 2, 3	4, 5, 6							
throw No.	1									
	2									
	3									
	4									
	5									
	6									
Total										

Simple model of gene drift III

– Task 14/p. 148

An example of results after the firts series of throws and the change of the rules for next series

Generation		1		2		3			10 (20)	
Alelle		*A*	*a*	*A*	*a*	*A*	*a*		*A*	*a*
side casted		1, 2, 3	4, 5, 6	1, 2, 3, 4	5, 6					
throw No.	1	/								
	2		/							
	3	/								
	4	/								
	5		/							
	6	/								
Total		4	2							

Simple model of gene drift IV

– Task 14/p. 148

An example of results after the firts series of throws and the change of the rules for next series

Generation		**1**		**2**		**3**			**10 (20)**	
Alelle		***A***	***a***	***A***	***a***	***A***	***a***		***A***	***a***
side casted		**1, 2, 3**	**4, 5, 6**	**1, 2, 3, 4**	**5, 6**	**1, 2, 3, 4, 5**	**6**			
throw No.	**1**	/		/						
	2		/	/						
	3	/		/						
	4	/			/					
	5		/	/						
	6	/		/						
Total		**4**	**2**	**5**	**1**					

Simple model of gene drift IV

– Task 14/p. 148

- work in pairs, one is throwing, the other is recording
- remind you the change of rules, where according to the result the gene frequencies are changing for next series of throws and consequently which sides of the dice will represent the allele A or a
- continue in experiment until the point of fixation of one allele
- now, swap your roles, this way one pair will finish and evaluate at least two experiments, each experiment and result visualize in a graph
- for whole classroom calculate average time of fixation as weighted mean of results of all partial experiments, the ratio of fixed A and a alleles should be approx. 1 : 1

Simple model of gene drift V – Task 14/p. 148

Conclusion of experiments

number of generations to fixation	1	2	3	4	5	6	7	8	9	10	11
represented in practicals											

12	13	14	15	16	17	18	19	20				

Number of fixation *A*			Total of experiments	0
Number of fixation *a*			Total of *A* + *a*	0
Average time of fixation				