# Assignment A1: HWE

Choose 3 out of the 5 exercises.

[A1-1] The Agouti Gene

[A1-2] Scarlet Tiger-Moth

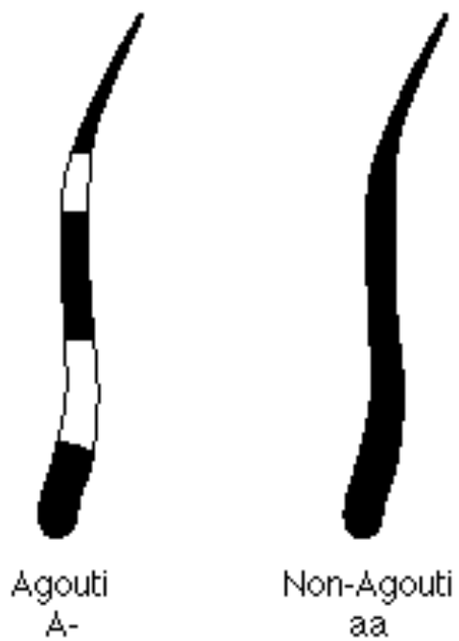
[A1-3] 1 Locus, 3 Alleles

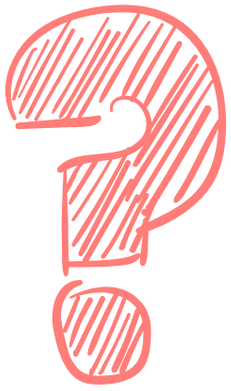
[A1-4] Brown Hare

[A1-5] Bitter Taste

# [A1-1] The Agouti Gene

The **agouti gene** is responsible for determining whether a mammal coat color is banded agouti coat color or a solid non-agouti coat color. The dominant, wild-type A causes the agouti shift phenomenon which causes hairs to be black pigmented at the tips and red pigmented at the roots (revealing the underlying tabby pattern), while the recessive non-agouti or "hypermelanistic" allele prevents this shift in the pigmentation pathway. In its homozygous form (aa) this results in black pigment production throughout the growth cycle of the hair.





We have **60 agouti** and and **51 non-agouti** individuals in a local rabbit population. How many % of the individuals are **homozygote for the agouti gene**?

# [A1-2] - Scarlet Tiger-Moth

Scarlet Tiger Moth (*Callimorpha dominula)*

# PopGen ▷ Assignments HWE

*Callimorpha dominula*

| | AA (white-spotted) | Aa (intermediate) | aa (little spotting) | sum |
|---|---|---|---|---|
| $N_{observed}$ | 1469 | 138 | 5 | 1612 |
| $N_{expected}$ | ? | ? | ? | ? |
| $\frac{(obs-exp)^2}{exp}$ | ? | ? | ? | ? |

(a) Allele Frequency ?

(b) HW expectation ?

(c) Pearson's chi-square test ?

# [A1-3] 1 Locus, 3 Alleles

| Example: 1 locus 3 alleles | | | | | | | |
|---|---|---|---|---|---|---|---|
| Genotypes | AA | AB | AC | BB | BC | CC | Sum |
| Counts | 17 | 86 | 5 | 61 | 9 | 0 | 178 |
| Frequencies | 0.096 | 0.483 | 0.028 | 0.343 | 0.051 | 0 | 1.000 |

Calculate allele frequencies

***f*(A) = ?%**

***f*(B) = ?%**

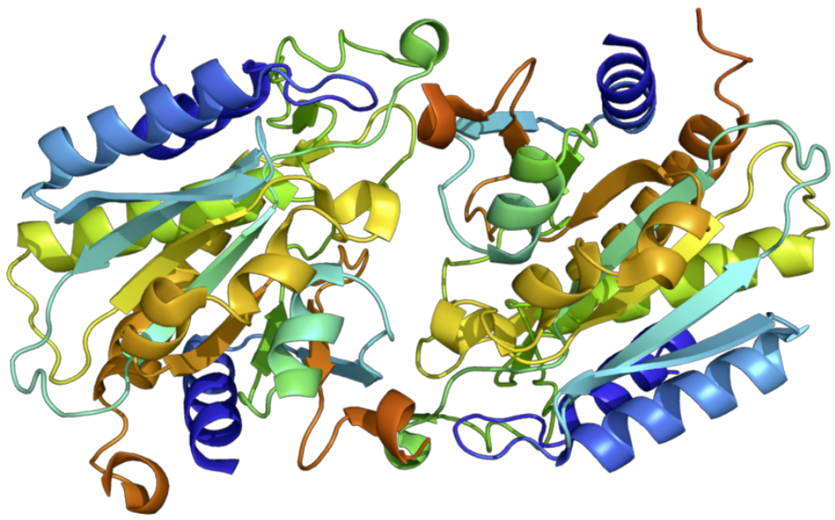
***f*(C) = ?%**

# [A1-4] Brown Hare


**Genetic diversity in the Polish brown hare *Lepus europaeus* Pallas, 1778: implications for conservation and management**

Günther B. HARTL, Janusz MARKOWSKI, Aleksander ŚWIĄTECKI, Tomasz JANISZEWSKI and Rudolf WILLING

Hartl et al. (1992) Acta Theriologica 37:15–25.



Aminoacylase-1 is an enzyme that in humans is encoded by the **ACY1** gene*

| | Genotype** | Observed |
|---|---|---|
| 1 | 100/100 | 4 |
| 2 | 100/81 | 6 |
| 3 | 81/81 | 14 |
| 4 | 100/66 | 4 |
| 5 | 81/66 | 7 |
| 6 | 66/66 | 3 |
| | Total | 38 |

* The enzyme aminoacylase 1 (ACY1) is involved in the breakdown of proteins. ACY1 performs the final step in the breakdown of these proteins by removing the acetyl group from certain protein building blocks (amino acids). The amino acids can then be recycled and used to build other proteins. — ghr.nlm.nih.gov/gene/ACY1

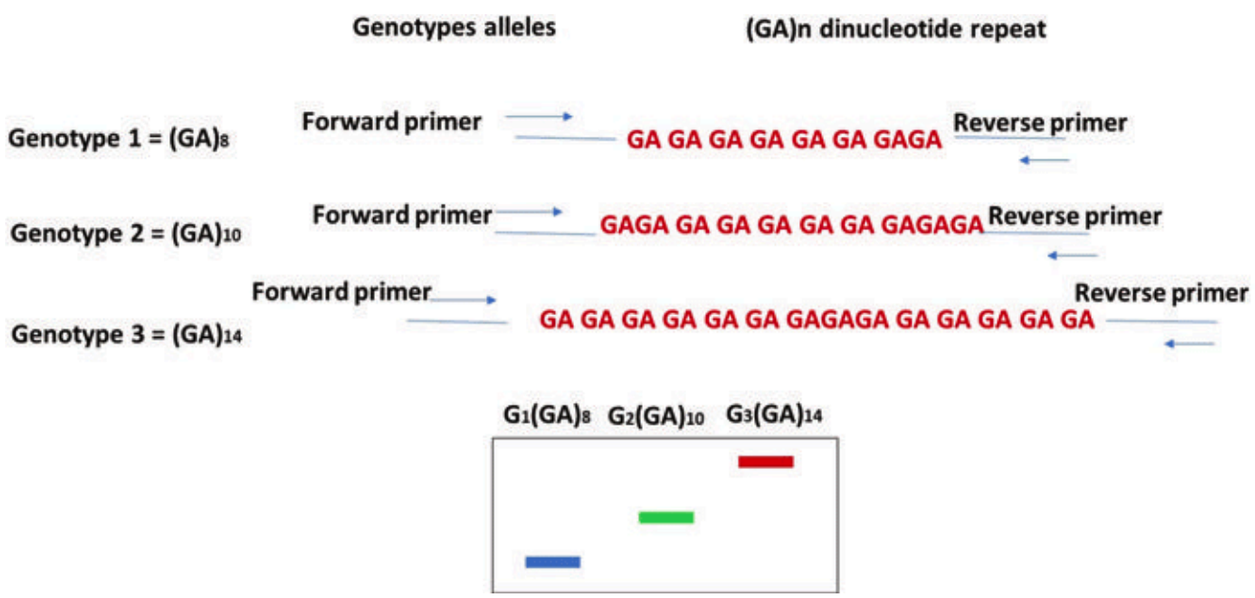
** Fragment size based on microsatellite markers.

Is the sampled population in Hardy-Weinberg proportions at locus ACYI?

**Microsatellites**, also known as simple sequence repeats (SSRs), are genetic markers characterised by variations in fragment size based on the number of repeated DNA sequences within a given genomic region. These repetitive sequences consist of short DNA motifs, typically 1 to 6 base pairs in length, which are repeated in tandem.

The variability in the number of these repeats between individuals results in fragments of different sizes when analysed by techniques such as polymerase chain reaction (PCR) and gel electrophoresis. By measuring the size differences in these fragments, researchers can use microsatellites to study genetic diversity, relatedness and inheritance patterns in populations or individuals.

Microsatellites are highly polymorphic, making them valuable tools in several fields, including genetics, forensics and evolutionary biology. They provide insight into the genetic differences between individuals and populations, aiding applications such as paternity testing, population genetics studies and linkage mapping for disease-associated genetic markers.



# [A1-5] Bitter Taste

## Genetic, Functional, and Phenotypic Diversity in TAS2R38-Mediated Bitter Taste Perception

**Abstract**

Mutational polymorphism in the TAS2R38 bitter taste receptor is a key determinant of threshold taste detection of isolated compounds, such as phenylthiocarbamide (PTC) and propylthiouracil (PROP), as well as complex orosensation-mediated traits such as diet choice and smoking habits. These relationships are accounted for, in part, by 2 common alleles differing in functionality, TAS2R38-PAV and TAS2R38-AVI. However, TAS2R38 harbors extensive additional polymorphism whose functional significance remains unknown. To examine this variation, we ascertained genetic diversity in 56 Caucasian subjects via whole-gene sequencing, analyzed allele-specific responses to 5 TAS2R38 agonists (PTC, PROP, goitrin, methimazole, and sinigrin) using *in vitro* assays, and assessed genotypic associations with threshold detection phenotypes. Sequencing identified 3 single-nucleotide substitutions encoding 3 amino acid changes (C145G/P49A, C785T/A262V, and A886G/I296V), which combined to form 6 haplotypes in our sample. *In vitro* assays revealed a continuous range of response across alleles, and associations with threshold were significant for all single nucleotide polymorphisms ($P < 0.002$) and PAV/AVI haplotypes ($P < 0.001$). Haplotypes other than PAV and AVI did not exhibit phenotypic associations in our sample, possibly as a result of their low frequencies. However, prior studies have indicated that these alleles are common in some global regions, suggesting that alleles rare in our sample may be phenotypically relevant in other populations.

Behrens et al. (2013) Genetic, Functional, and Phenotypic Diversity in TAS2R38-Mediated Bitter Taste Perception.

TAS2R38 SNP, haplotype (A), and diplotype (B) frequencies.

A

| Haplotype | Nucleotide 145 | 785 | 886 | Amino Acid 49 | 262 | 296 | Occ. | Freq. |
|---|---|---|---|---|---|---|---|---|
| AAI | G | C | A | A | A | I | 1 | 0.01 |
| AAV | G | C | G | A | A | V | 5 | 0.05 |
| AVI | G | T | A | A | V | I | 48 | 0.43 |
| PAI | C | C | A | P | A | I | 1 | 0.01 |
| PAV | C | C | G | P | A | V | 55 | 0.49 |
| PVI | C | C | A | P | V | I | 2 | 0.01 |
| | | | | | | | 112 | 1.00 |

B

| Diplotype | Occ. | Freq. |
|---|---|---|
| AAI/PAV | 1 | 0.02 |
| AAV/AVI | 2 | 0.04 |
| AVI/AVI | 9 | 0.16 |
| PAI/PVI | 1 | 0.02 |
| PAV/AAV | 3 | 0.05 |
| PAV/AVI | 27 | 0.48 |
| PAV/PAV | 12 | 0.21 |
| PAV/PVI | 1 | 0.02 |
| | 56 | 1.00 |

(**A**) Six haplotypes defined by 3 SNPs were observed. The composition of each haplotype with respect to nucleotide positions 145, 785, and 886 is shown, along with composition with respect to amino acid positions 49, 262, and 296. The number of occurrences of each haplotype indicates the total number of observations in the sample for a total of 112 observations. (**B**) The number of occurrences and frequency of each observed diplotype (i.e., haplotype pairing in an individual).

a. Calculate allele and genotype frequency for the 3 different loci.
b. Are the 3 loci in Hardy-Weinberg proportions?
c. Create a **Ternary Plot** with the data (see website).

## Suggestion

```
Diplotype    Genotype at locus 145
AAI / PAV ->  GC (Heterozygote)
AAV / AVI ->  GG (Homozygote)
AVI / AVI ->  GG (Homozygote)
PAI / PVI ->  CC (Homozygote)
PAV / AAV ->  CG (Heterozygote)
PAV / AVI ->  CG (Heterozygote)
PAV / PAV ->  CC (Homozygote)
PAV / PVI ->  CC (Homozygote)
```



```
HWChisq(X, cc = 0, verbose = TRUE)
```