Bioinformatics
(Graph Products and Phenotypes)

Marc Hellmuth

Phenotypes and Characters (Traits ,Merkmale)

Biology

Biology (greek: bios ‚live') Biology is a natural science concerned with the study of life and living organisms, including their structure, function, growth, evolution, distribution, identification and taxonomy.

In other words, want to understand the structure and processes in living organisms

A central point to understand living organisms is to understand their phenotype.

To describe a living organisms, we must be able to describe their phenotype.

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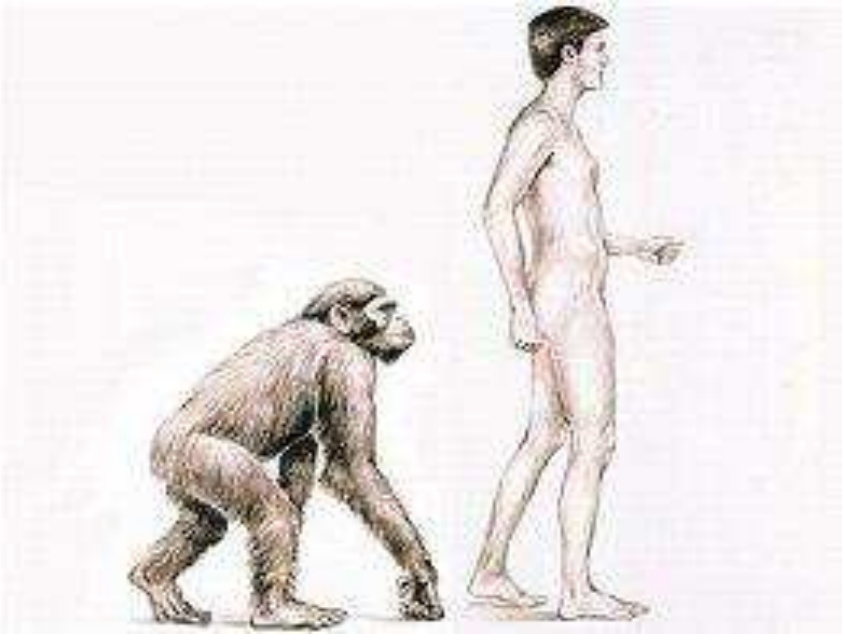
Phenotype via Example: Great Ape (Menschenaffe) vs Human



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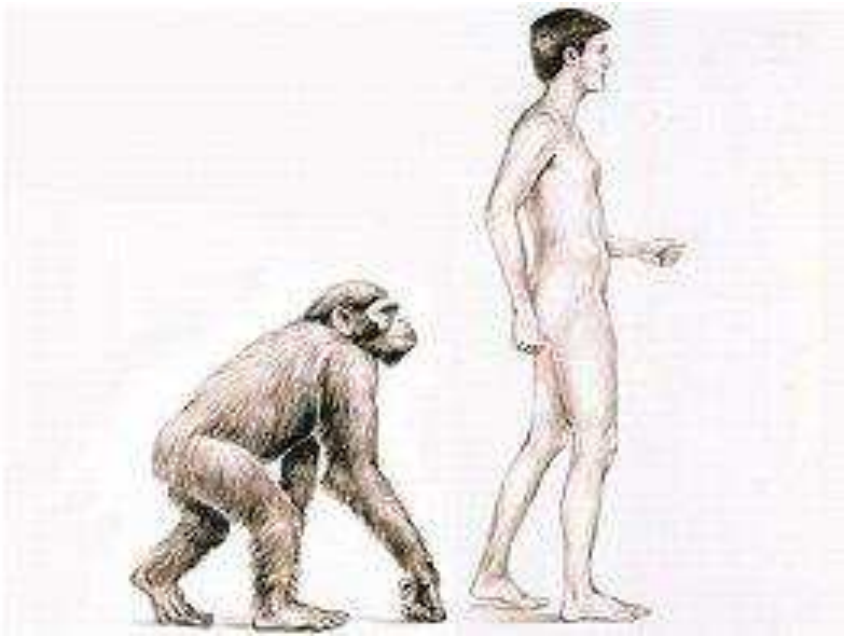

Many Similarities:
nose, eyes, 32 teeth, skeleton, bones, liver, mammal, . . .
But there are also big differences:
- hairiness
- apes don't have a nose bone (Nasenbein)
- Differences in skeleton $\rightarrow$ (Non)upright walking
- Humans have more effective spittle (= "water with enzyme") $\rightarrow$ improved predigestion (Vorverdauung) $\rightarrow$ evolutionary benefit?

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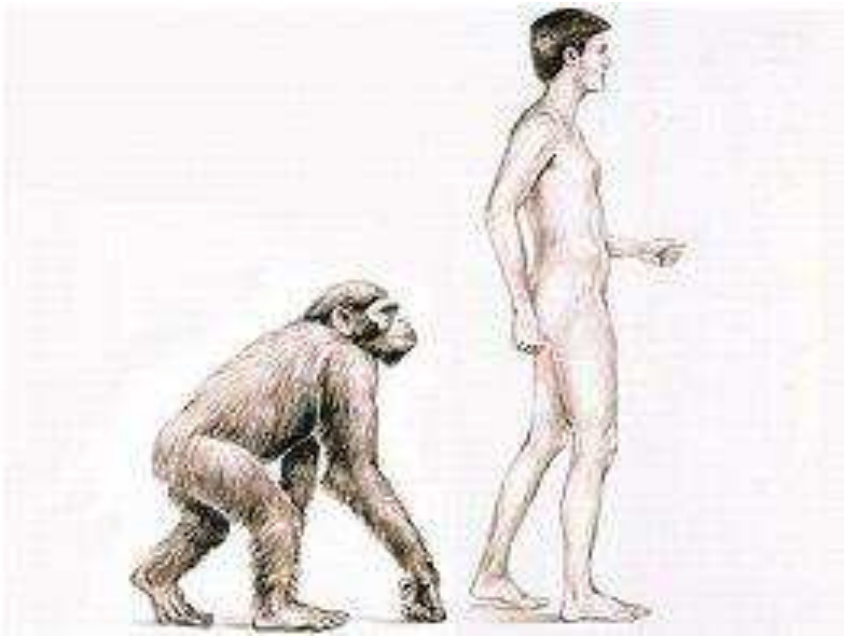


Phenotye is more that what one can see on a first glance. (skeleton, internal organs, production of enzymes, mammal, ..)

Phenotyope = set of all characters (with its characteristics) of an organism.

What is a character ??

Phenotypes and Characters (Traits, Merkmale)

Phenotyope = set of all characters (with its characteristics) of an organism.

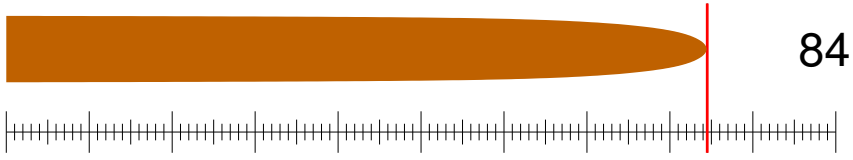
What is a character?

- Characters can vary
- Characters are independent and can be freely combined
- Genes and their variants determine the characteristics of a character

Discretization of Characters

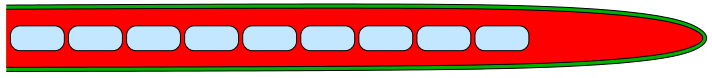
We consider the following “simplification”:

kontinuierlich (x cm)



84,5cm

diskret (0 n Wirbel)

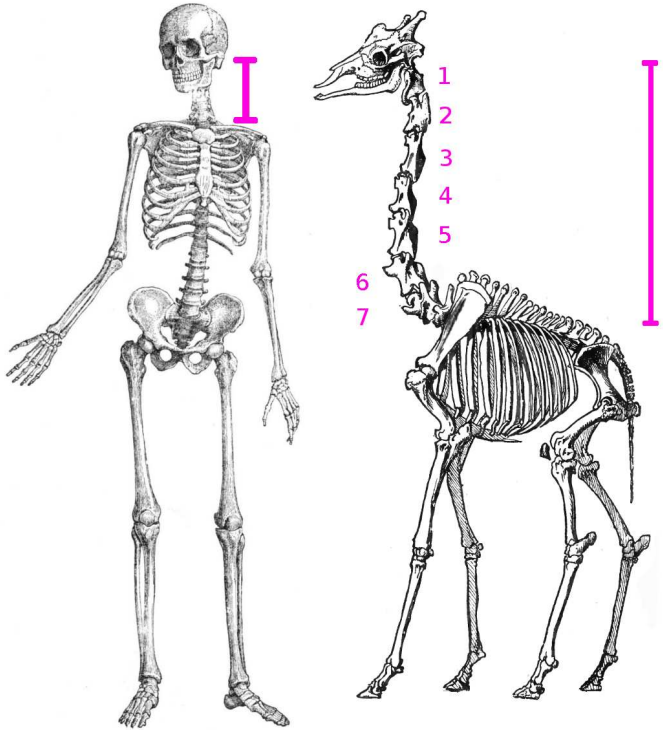


9 Wirbel

diskret (kurz oder lang)

kurz

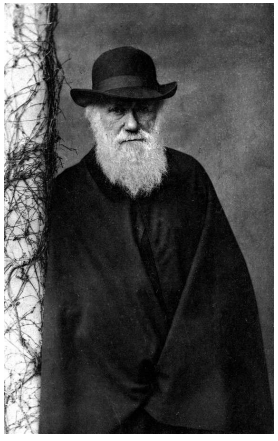
lang

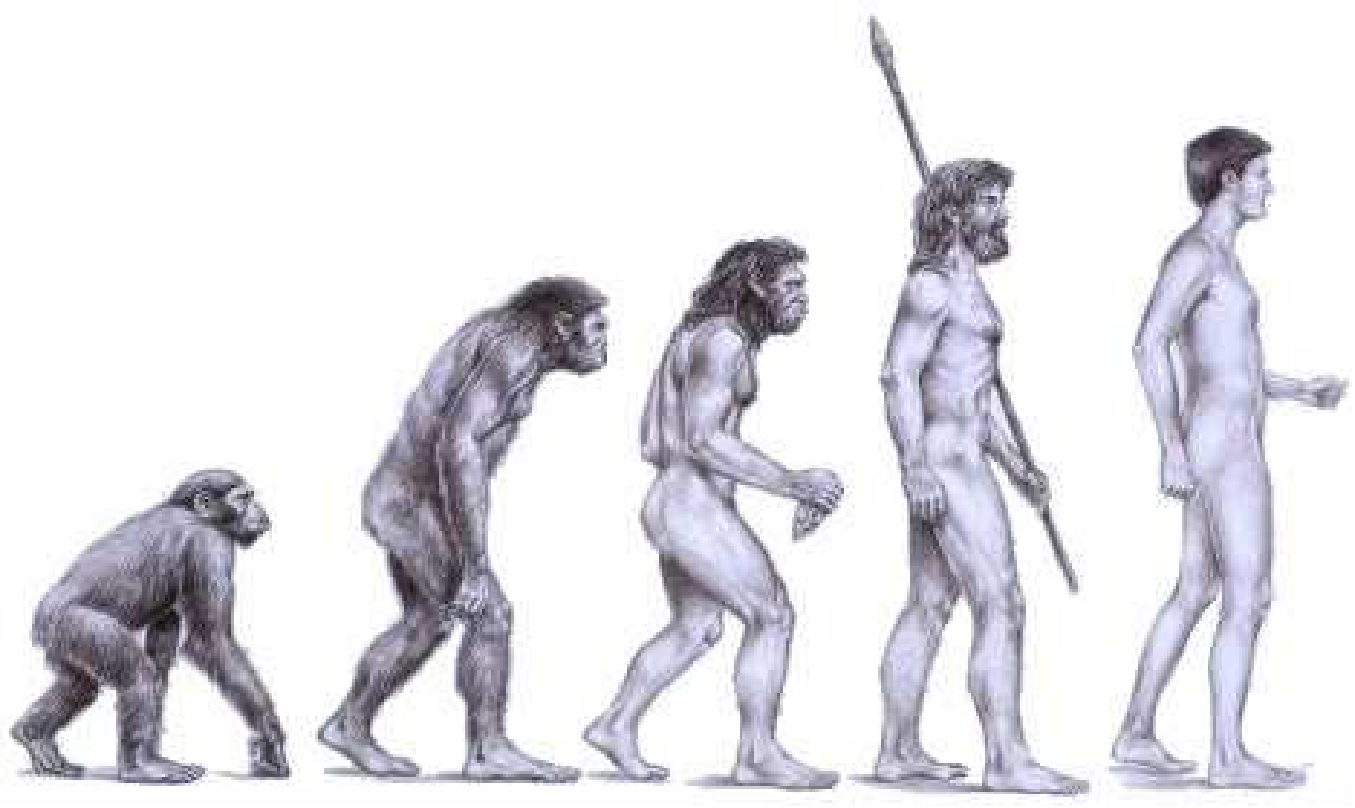


Judgening from the number of cervical vertebras the length of the neck of a giraffe and a human is equal.

Variability in Evolution

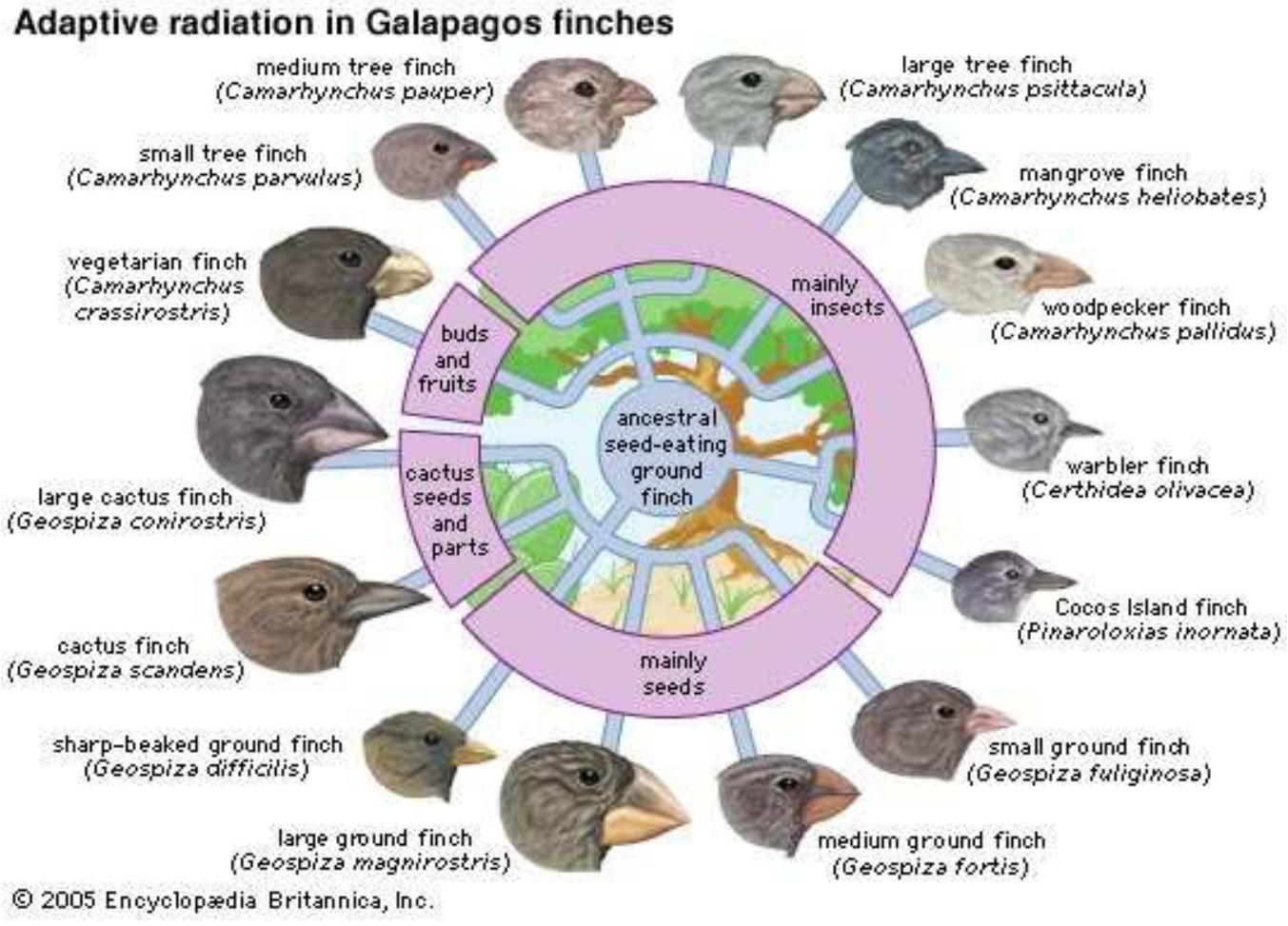
"The Origin of Species" by Charles Darwin (1859)





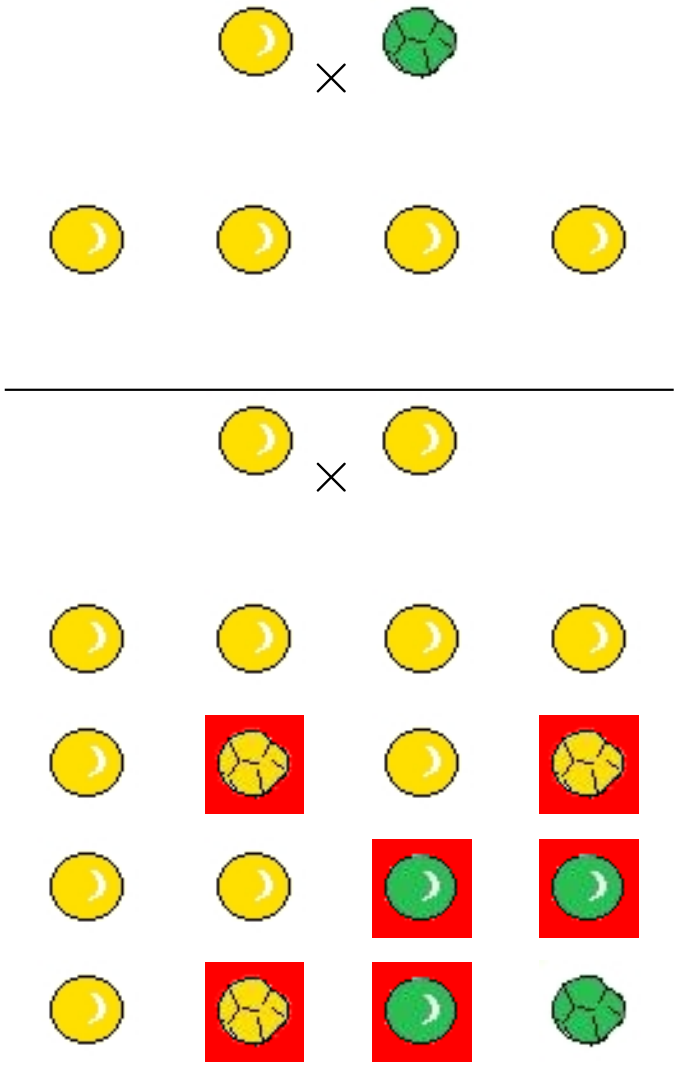
About the variability of phenotypic characters.

Variability in Evolution



The shape of the beak is a character. Multiple characteristics exist.

Independence of Characters

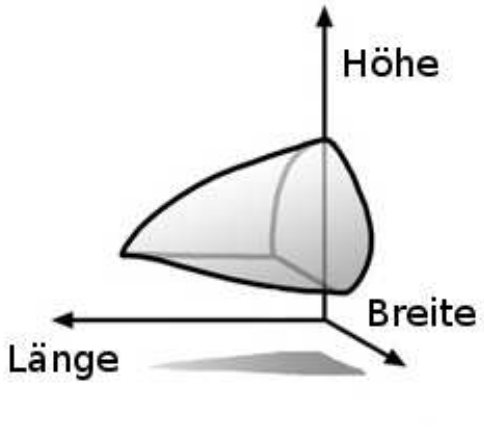


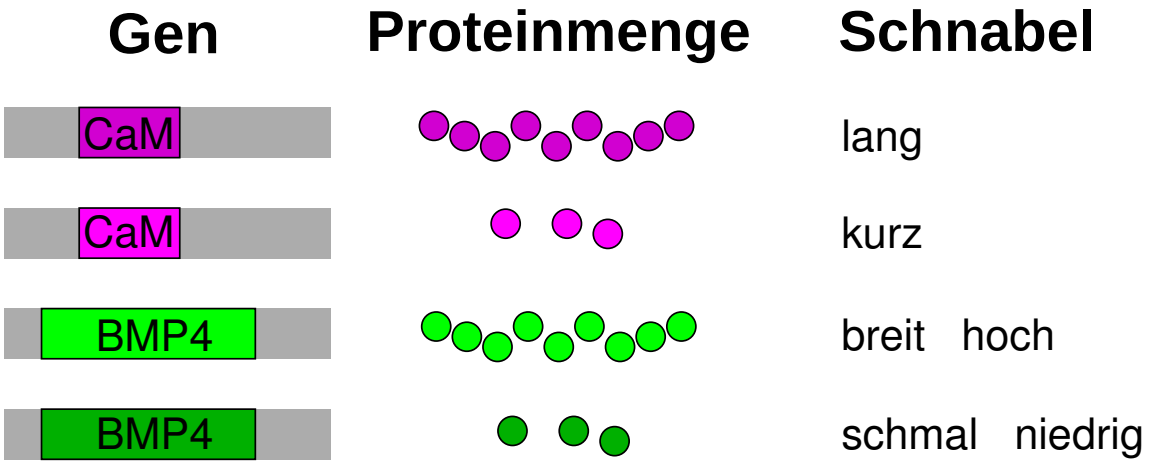
Gregor Mendel (1822-1884) discovered that the characters

- **“Surface”**
  with the characteristics “smooth” and “wrinkled”
- and **“Seed color”**
  with the characteristics “yellow” and “green”

are independent.

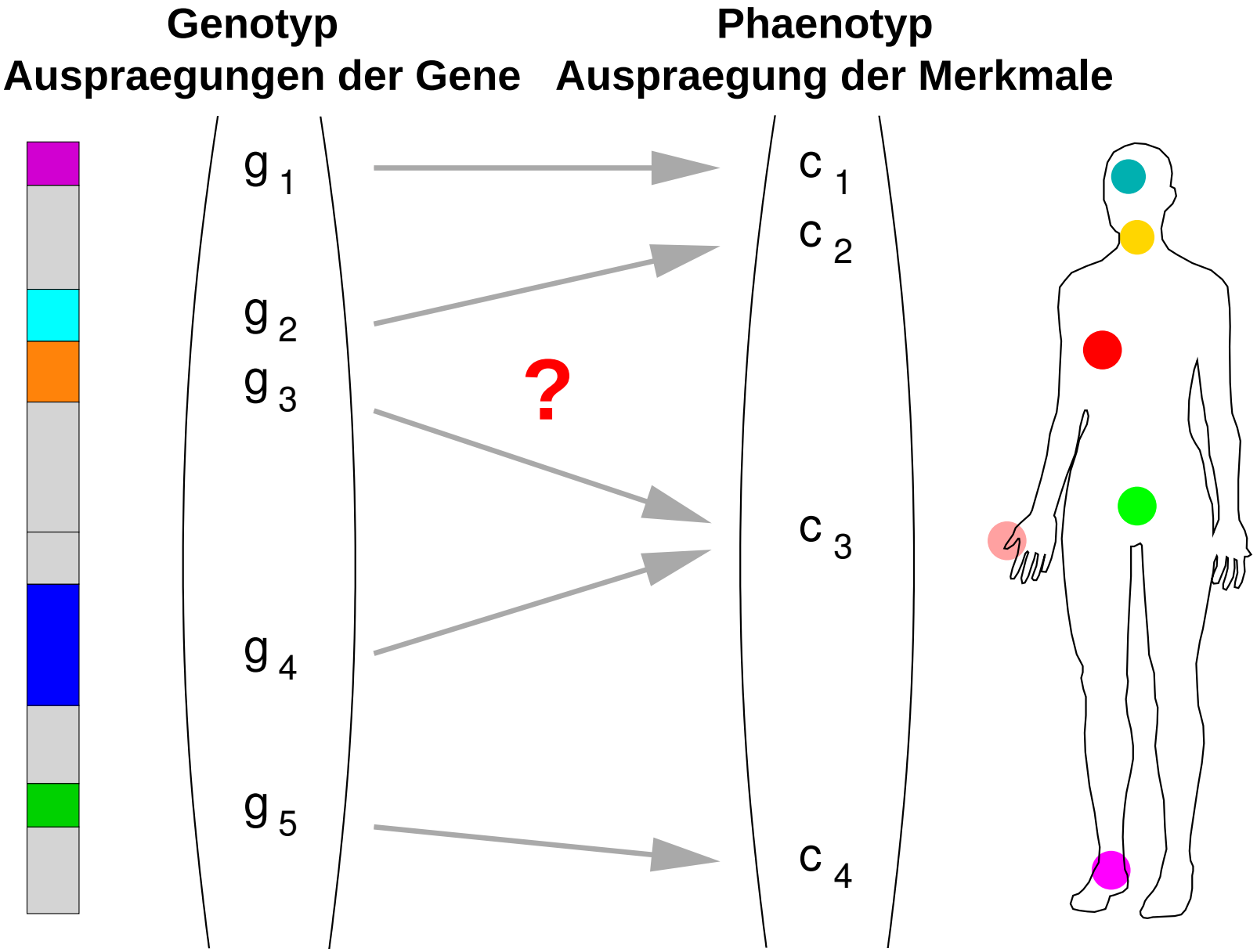
How are the Characteristics of a Character Determined?





Genes and their variants determine the characteristics of a character.

Genotyp ⇔ Phenotype



What is a character (Merkmal)?

Darwin's finches

Seeds Insects

Characters can vary

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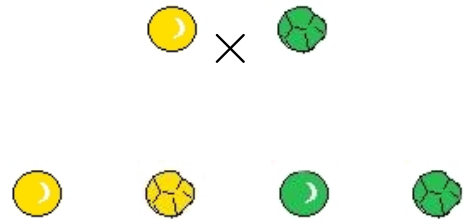
Mendel's peas



Characters are independent and can be freely combined

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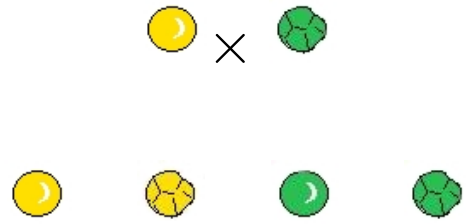

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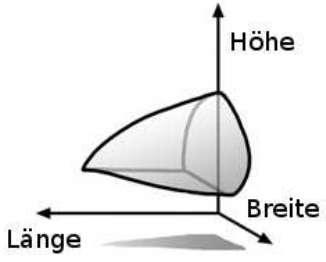
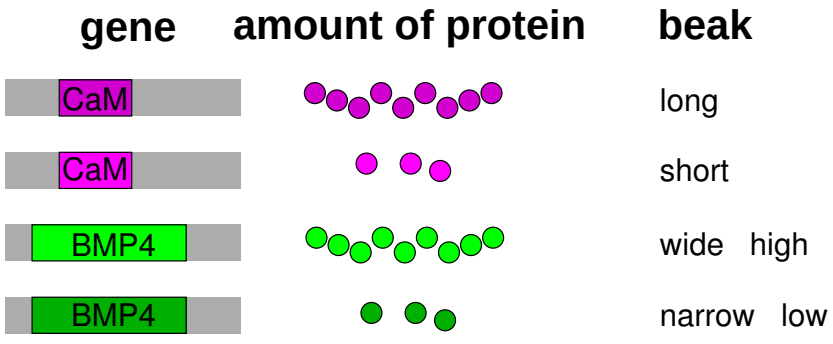




Genes and their variants determine the characteristics of a character

Determine a Character: Genotype-Phenotype-Mapping

Accessibility of genotypes based on "variational operators" $u \in \mathcal{U}$:

$$x \curvearrowright_{\mathcal{U}} y$$

U-neighborhood of genotypes $y \in \mathbb{X}$:

$$N_{\mathcal{U}}(y) := \{x \in \mathbb{X} \mid x \curvearrowright_{\mathcal{U}} y\}$$

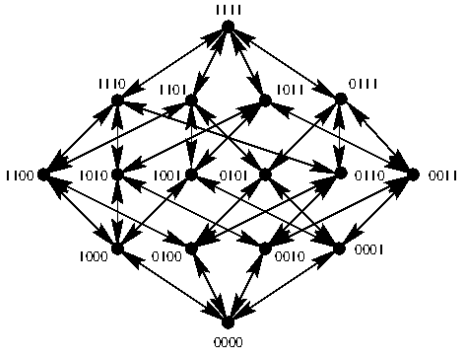
Genotype-Phenotype-Map
is mapping into phenotype space $\mathbb{P}$

$$f : \mathbb{X} \to \mathbb{P}$$

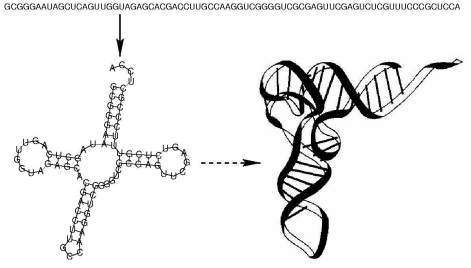
Accessibility of phenotypes $\alpha, \beta \in \mathbb{P}$:

$$\alpha \curvearrowright_p \beta \iff \frac{|f^{-1}(\alpha) \cap N_{\mathcal{U}}(f^{-1}(\beta))|}{|N_{\mathcal{U}}(f^{-1}(\beta))|} > p$$

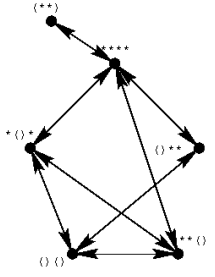
Genotype Space $\mathbb{X}$



GP-Map $f : \mathbb{X} \to \mathbb{P}$

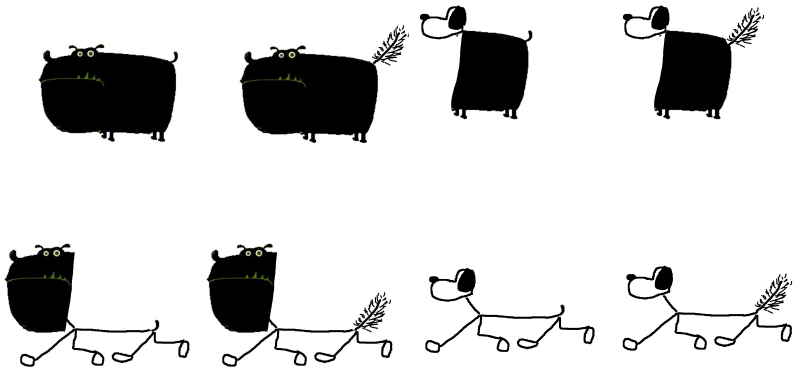


Phenotype Space $\mathbb{P}$



Determine a Character

Given: Genome of single organisms and Genotype-Phenotype-Mapping

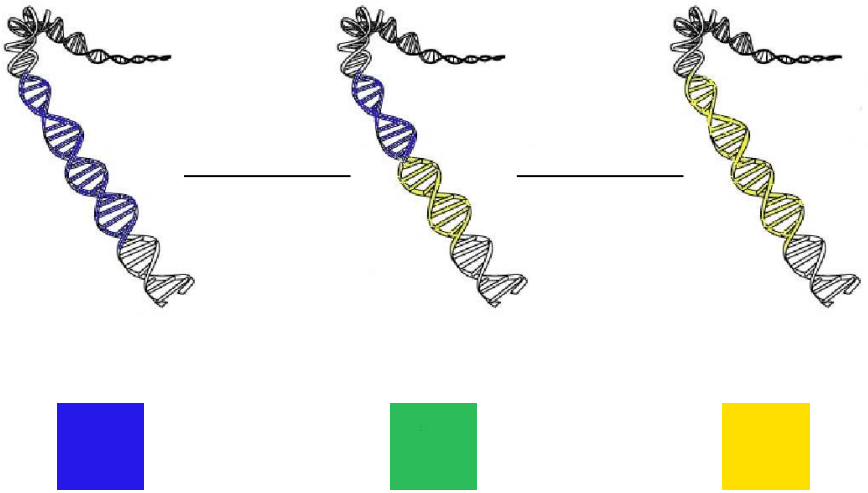
Observable Phenotype	Putative Characters
	Structure, Color, Sugar Level, ...
	Tail, Body, Head, ...

- Are characters independent?
- How can one determine "real" characters?

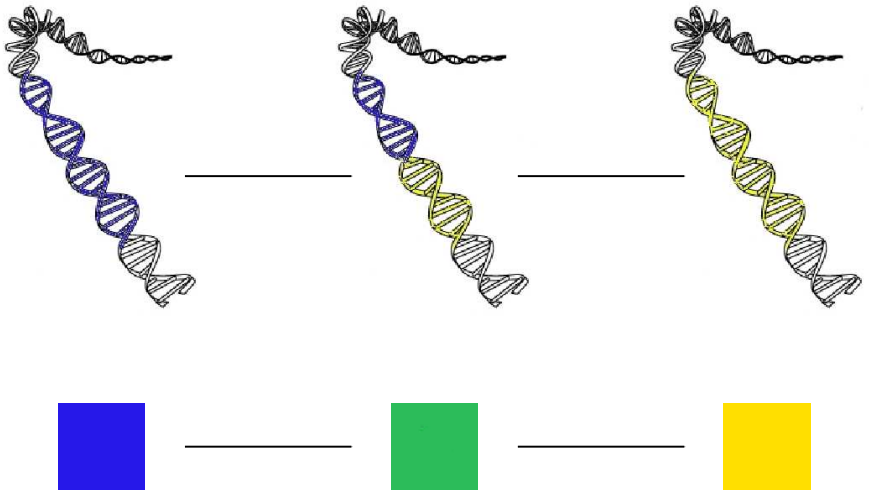
$\longrightarrow$ Graphproducts

Produktgraphs

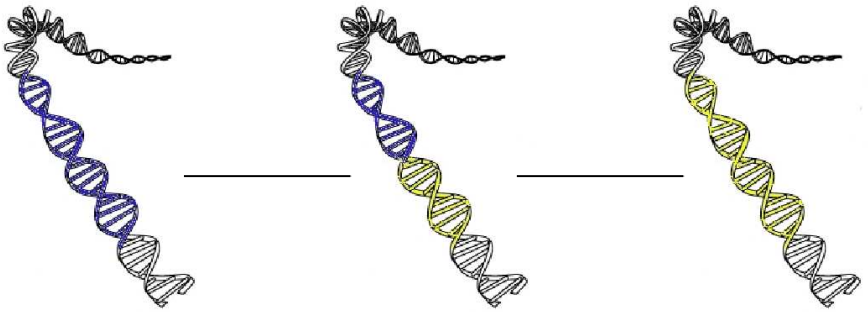
Genotyp-Raum

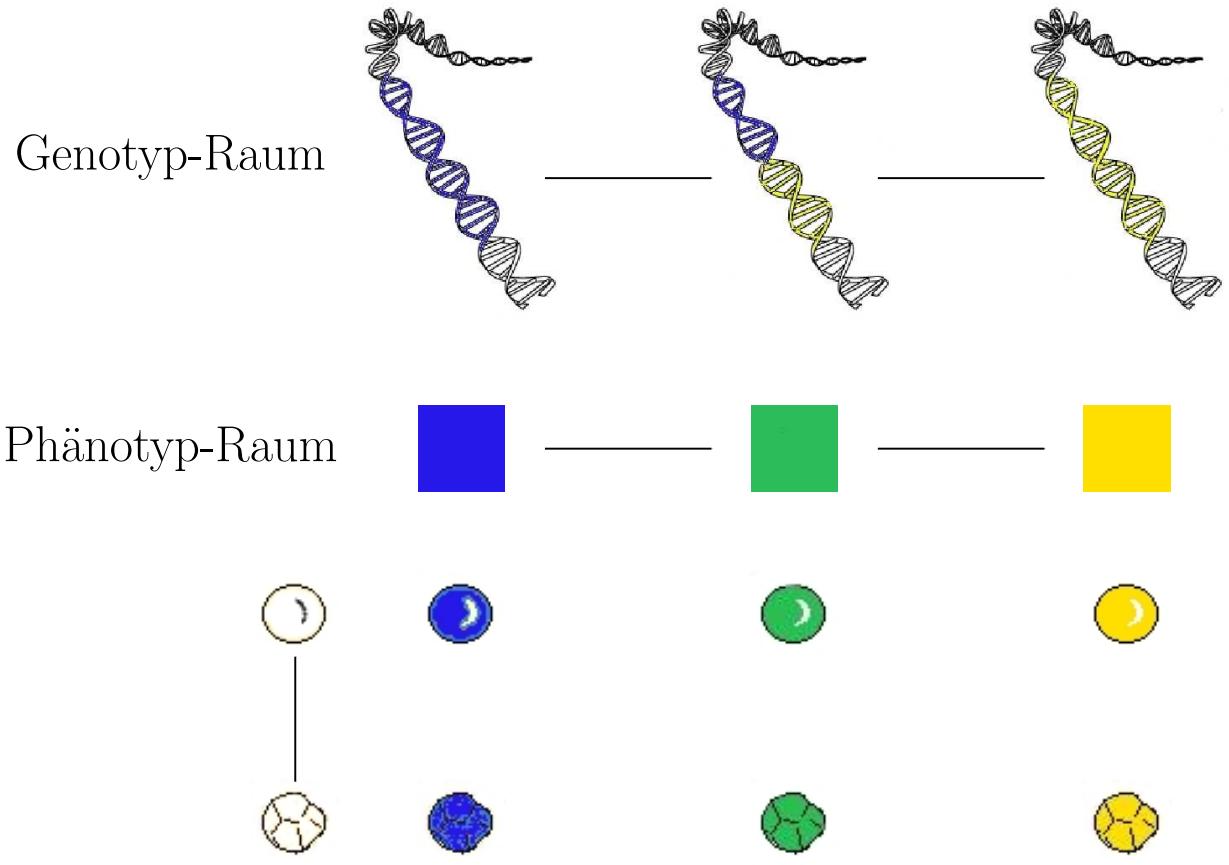


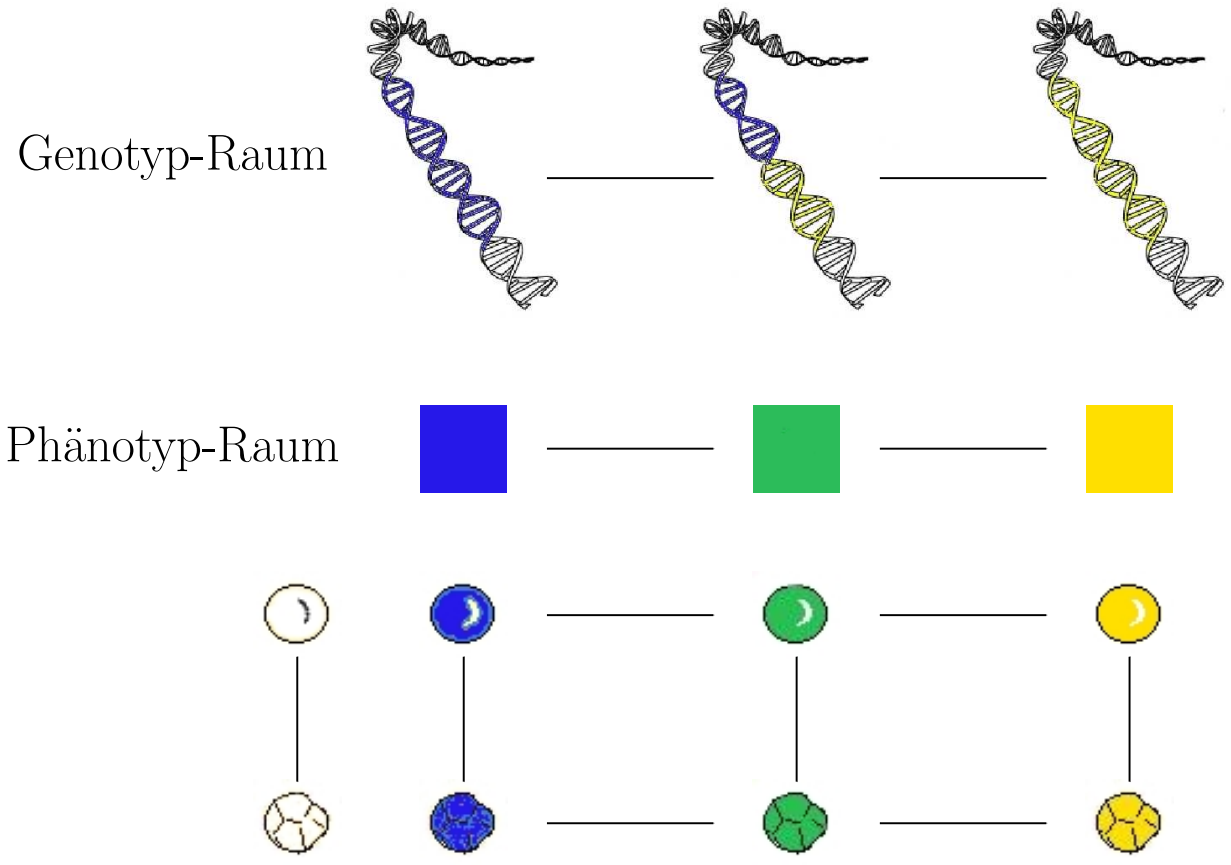
Genotyp-Raum



Genotyp-Raum



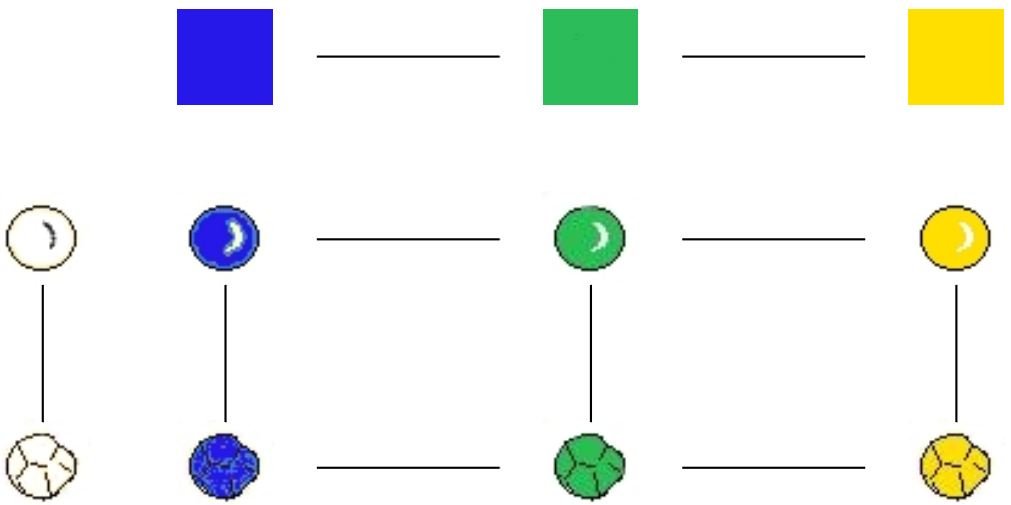

Genotyp-Raum
Phänotyp-Raum


Genotyp-Raum
Phänotyp-Raum

Characters and Graphproducts [3]


Theorem

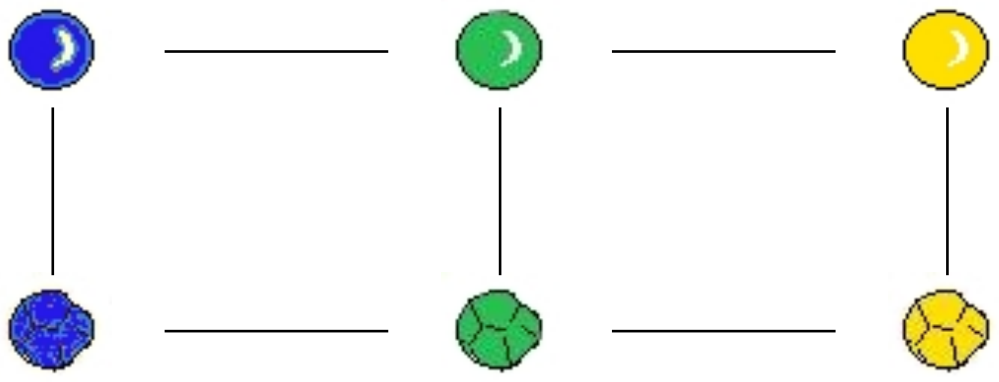
Characters **can** *vary independently* $\Longleftrightarrow$
Prime factors of phenotype space



[3] Quasi-Independence, Homology and the Unity of Type: A Topological Theory of Characters, Günter Wagner and Peter F. Stadler, J. theor. Biol., 2003

Graphproducts

Problem: We might compute the phenotype space, but we don't know the prime factors!



Another, but wrong example: