Fundamentals of… (ABG 300) 2 Units

Martha N. Bemji
Department of Animal Breeding & Genetics
University of Agriculture
Abeokuta

Course requirements:

CAT: 40%

Exam: 60%

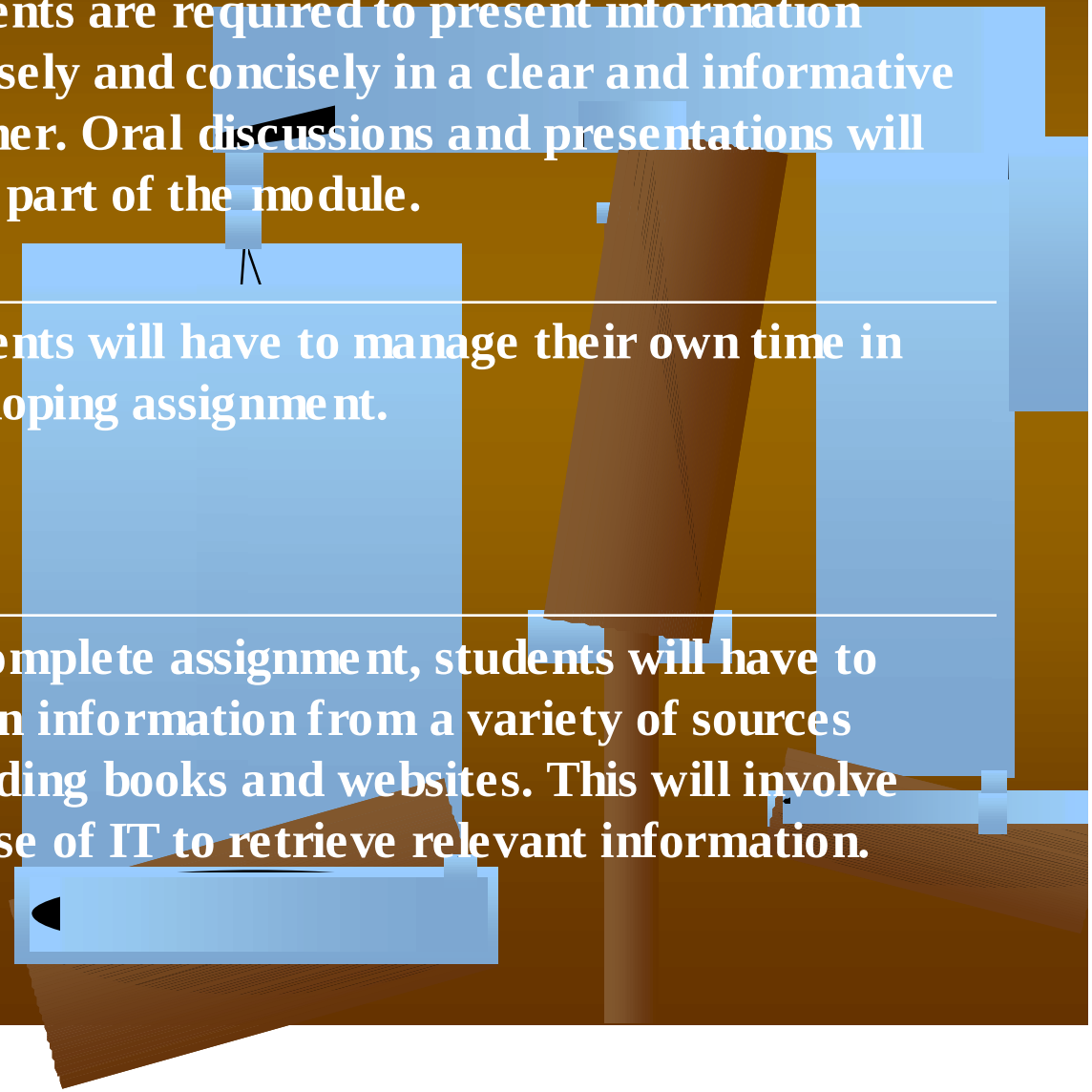
Class attendance compulsory

*Contact Lecturer ahead of time if any cogent reason will keep you away from lectures.

*Present medical reports if absence from lectures/CAT was due to ill health.

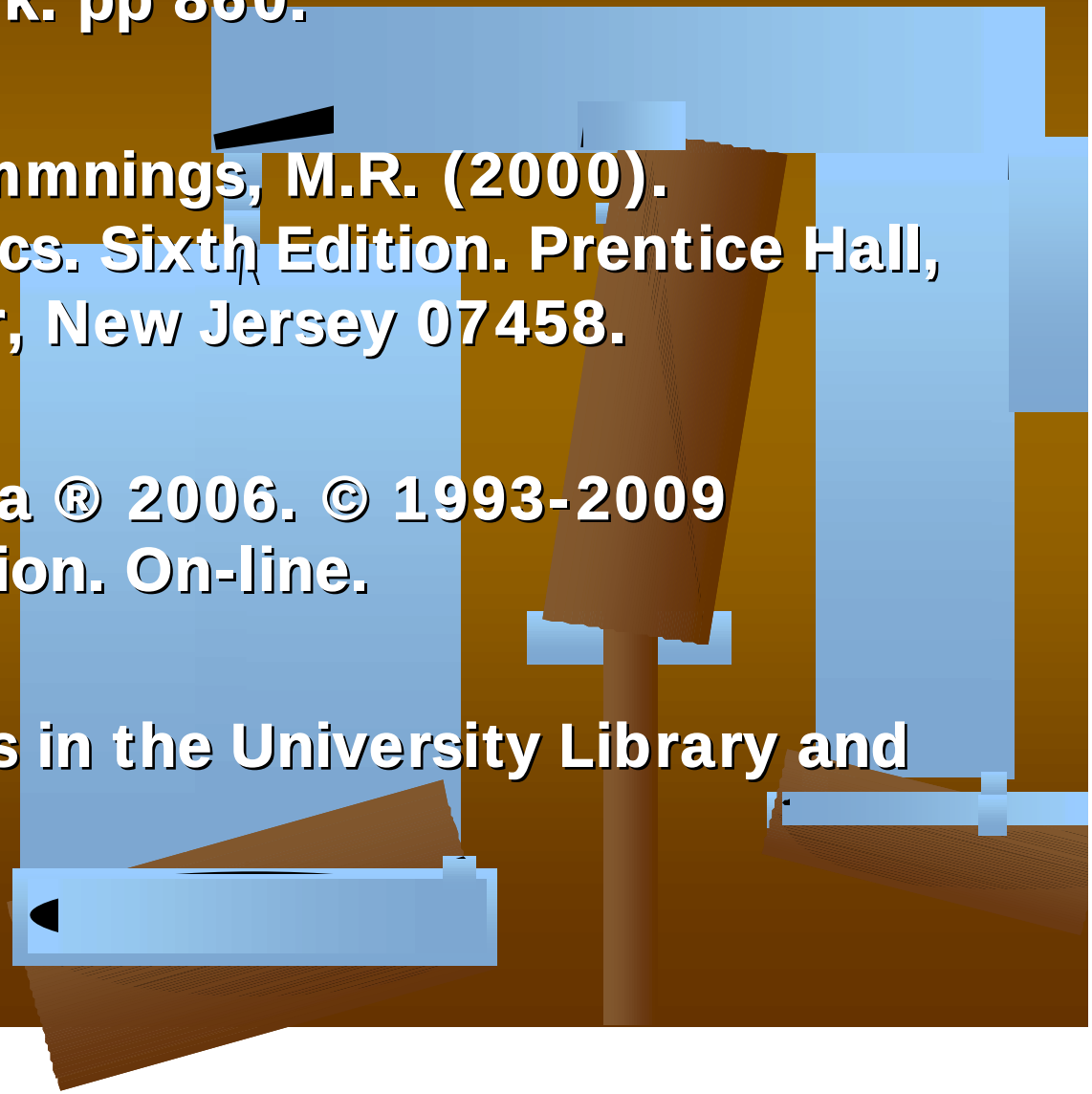
Module Skills

Research skills	**Students' ability to work independently and solve problems will be developed by assignments.**
Communication	**Students are required to present information precisely and concisely in a clear and informative manner. Oral discussions and presentations will form part of the module.**
Improving Learning and Performance	**Students will have to manage their own time in developing assignment.**
Information Technology	**To complete assignment, students will have to obtain information from a variety of sources including books and websites. This will involve the use of IT to retrieve relevant information.**



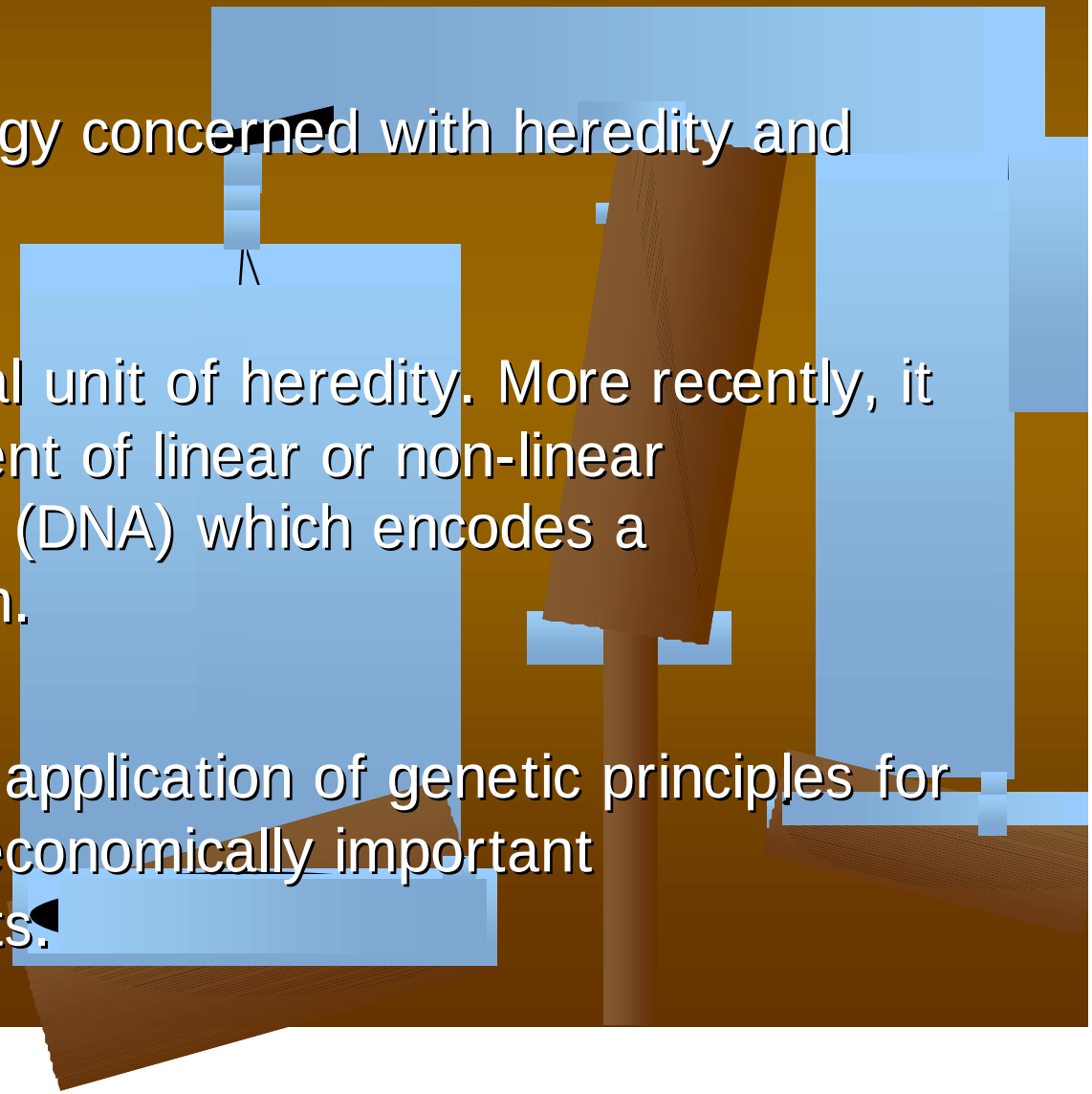
Recommended reading

- **Griffiths, A.J.F. et al. (2003). An Introduction to genetic analysis. 7th Edition. W.H. Freeman and Company, New York. pp 860.**
- **Klug, W.S. and Cummnings, M.R. (2000). Concepts of Genetics. Sixth Edition. Prentice Hall, Upper Saddle River, New Jersey 07458.**
- **Microsoft ® Encarta ® 2006. © 1993-2009 Microsoft Corporation. On-line.**
- **...and lots of others in the University Library and Internet.**



Introduction

- **Genetics** - Science of heredity concerned with behaviour of **genes** passed from parents to offspring in the reproductive process.
- It is a branch of Biology concerned with heredity and variation.
- **Gene** is the functional unit of heredity. More recently, it is defined as a segment of linear or non-linear deoxyribonucleic acid (DNA) which encodes a polypeptide or protein.
- **Breeding** deals with application of genetic principles for the improvement of economically important characteristics or traits.



Importance of Genetics

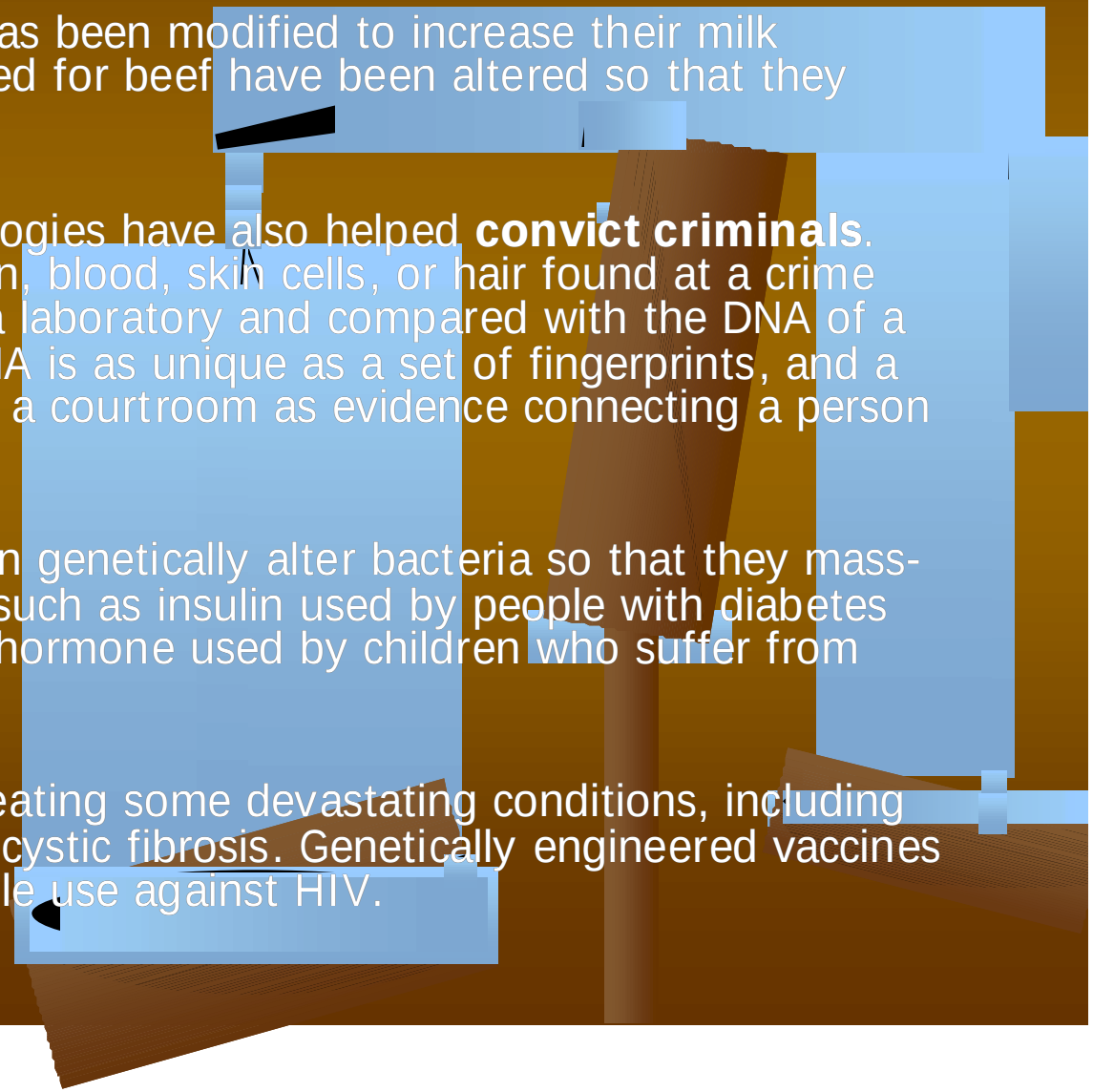
- 1. In **Agriculture**, some food crops (oranges, potatoes, wheat, and rice) have been genetically altered to withstand insect pests, resulting in a higher crop yield. Tomatoes and apples have been modified so that they resist discoloration or bruising.

- Genetic makeup of cows has been modified to increase their milk production, and cattle raised for beef have been altered so that they grow faster.

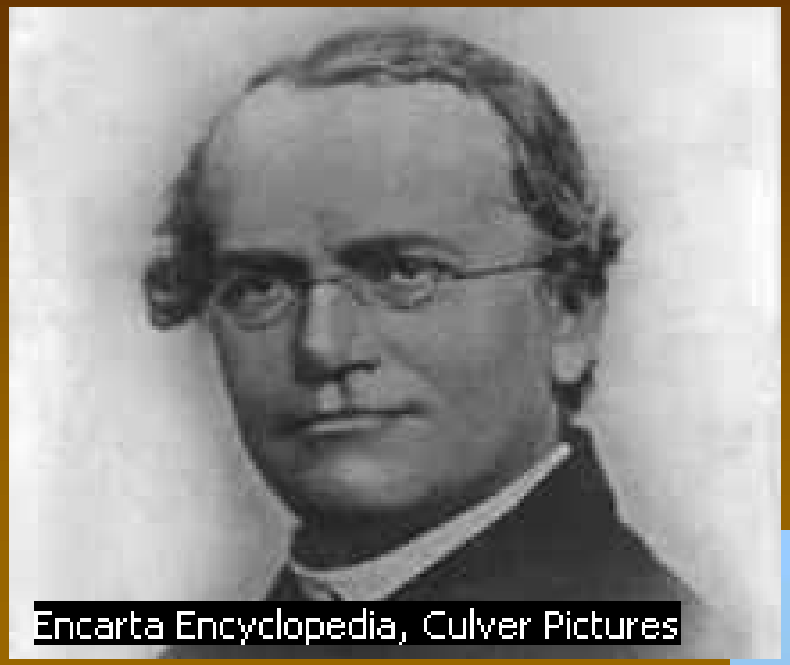
- 2. In **Law**, genetic technologies have also helped **convict criminals**. DNA recovered from semen, blood, skin cells, or hair found at a crime scene can be analyzed in a laboratory and compared with the DNA of a suspect. An individual's DNA is as unique as a set of fingerprints, and a DNA match can be used in a courtroom as evidence connecting a person to a crime.

- In **medicine**, scientists can genetically alter bacteria so that they mass-produce specific proteins, such as insulin used by people with diabetes mellitus or human growth hormone used by children who suffer from growth disorders.

- Gene therapy is used in treating some devastating conditions, including some forms of cancer and cystic fibrosis. Genetically engineered vaccines are being tested for possible use against HIV.



Simple Mendelian inheritance



Gregor J. Mendel (1865)
'Father of modern genetics'

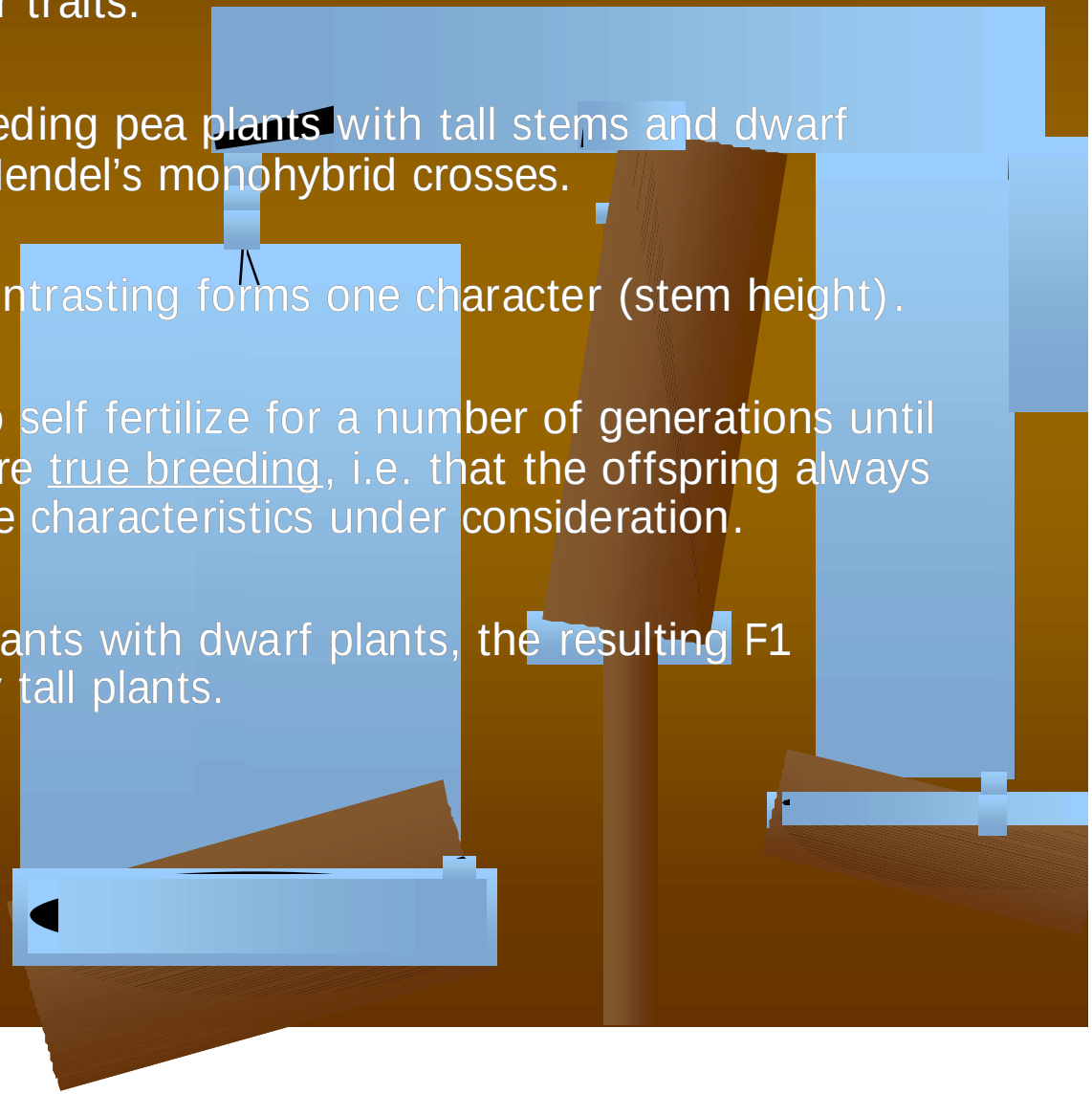
First scientist to discover the principles of heredity

Mendel's work showed that:

1. There is existence of some factors now called **genes** which are responsible for the inheritance of traits or characteristics.
2. Genes occur in pairs: Alternative phenotypes of a character are determined by different forms of a single type of gene called **alleles**.
3. Each parent contributes one factor of each trait shown in offspring.
4. The two members of each pair of alleles segregate during gamete formation so that each gamete receives one of the alleles.
5. Genes are transmitted unchanged from generation to generation.

Single gene inheritance (Monohybrid cross)

- The monohybrid cross revealed how one trait is transmitted from generation to generation.
- The simplest experiments Mendel performed involved only one pair of contrasting characteristics or traits.
- The cross between true breeding pea plants with tall stems and dwarf stems is representative of Mendel's monohybrid crosses.
- *Tall* and *dwarf* represent contrasting forms one character (stem height).
- Mendel allowed his plants to self fertilize for a number of generations until he was certain that they were true breeding, i.e. that the offspring always resembled the parent for the characteristics under consideration.
- When Mendel crossed tall plants with dwarf plants, the resulting F1 generation consisted of only tall plants.



Single gene inheritance (Monohybrid cross)

Parental phenotype: Tall Short
Parental genotype (2n): **T T** X **t t**

Gamete (n) **T** **T** **t** **t**

F1 Genotype (2n): **T t** **T t** **T t** **T t**
Phenotype: All Tall

- **When members of the F1 generation were selfed,**
  P2 phenotype: Tall Tall
  P2 genotype (2n): **T t** X **T t** (*Selfing*)

  P2 gamete (n) : **T** **t** **T** **t**

  F2 Genotype (2n): **T T** **T t** **T t** **t t**

  F2 Phenotype: (3/4 tall: ¼ dwarf)
  (787/1064): (227/1064)

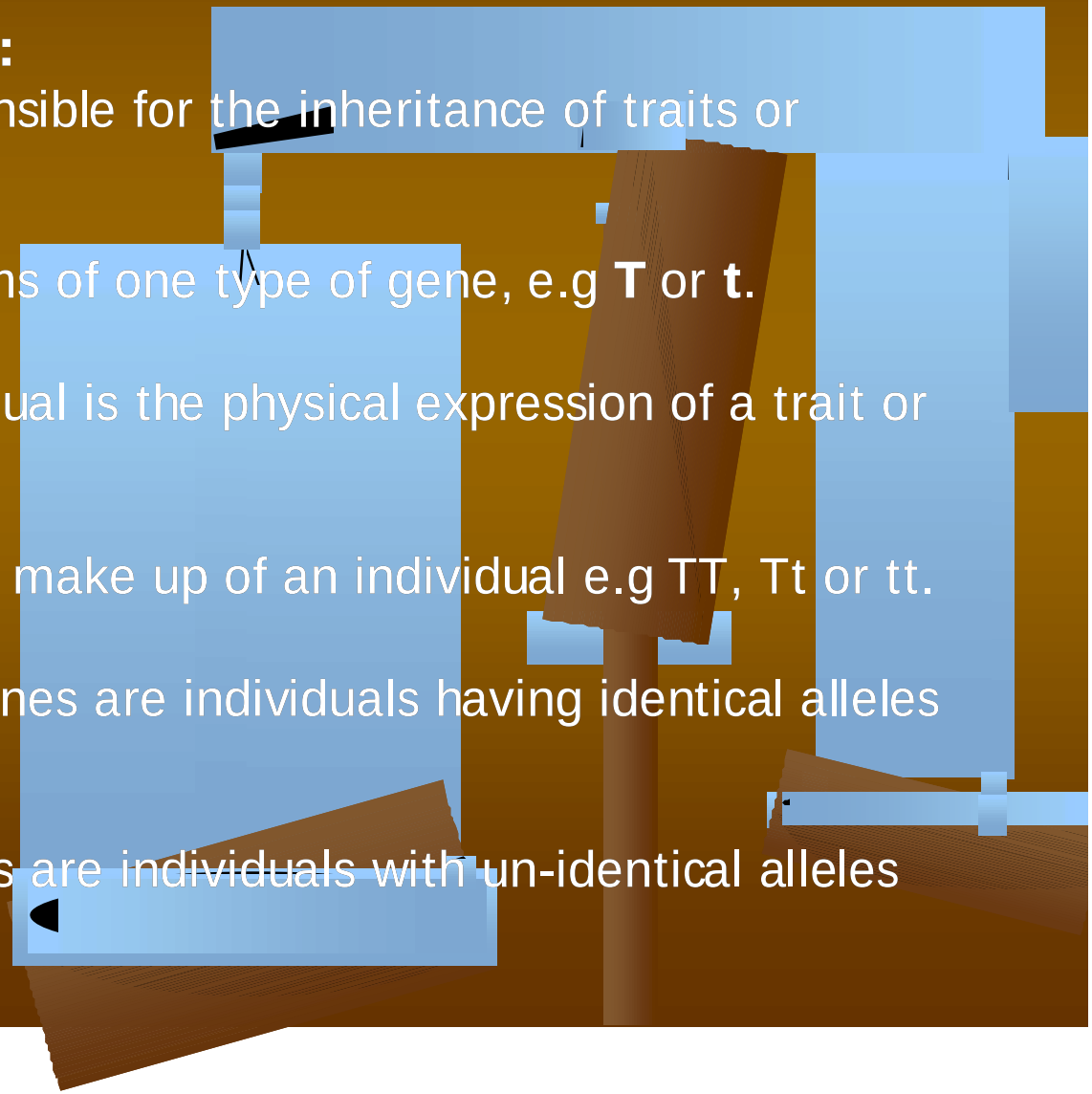
- To explain these results, Mendel proposed the existence of what he called particulate **unit factors** for each trait which served as the basic unit of heredity and are passed unchanged from generation to generation.

Mendel's first law of inheritance

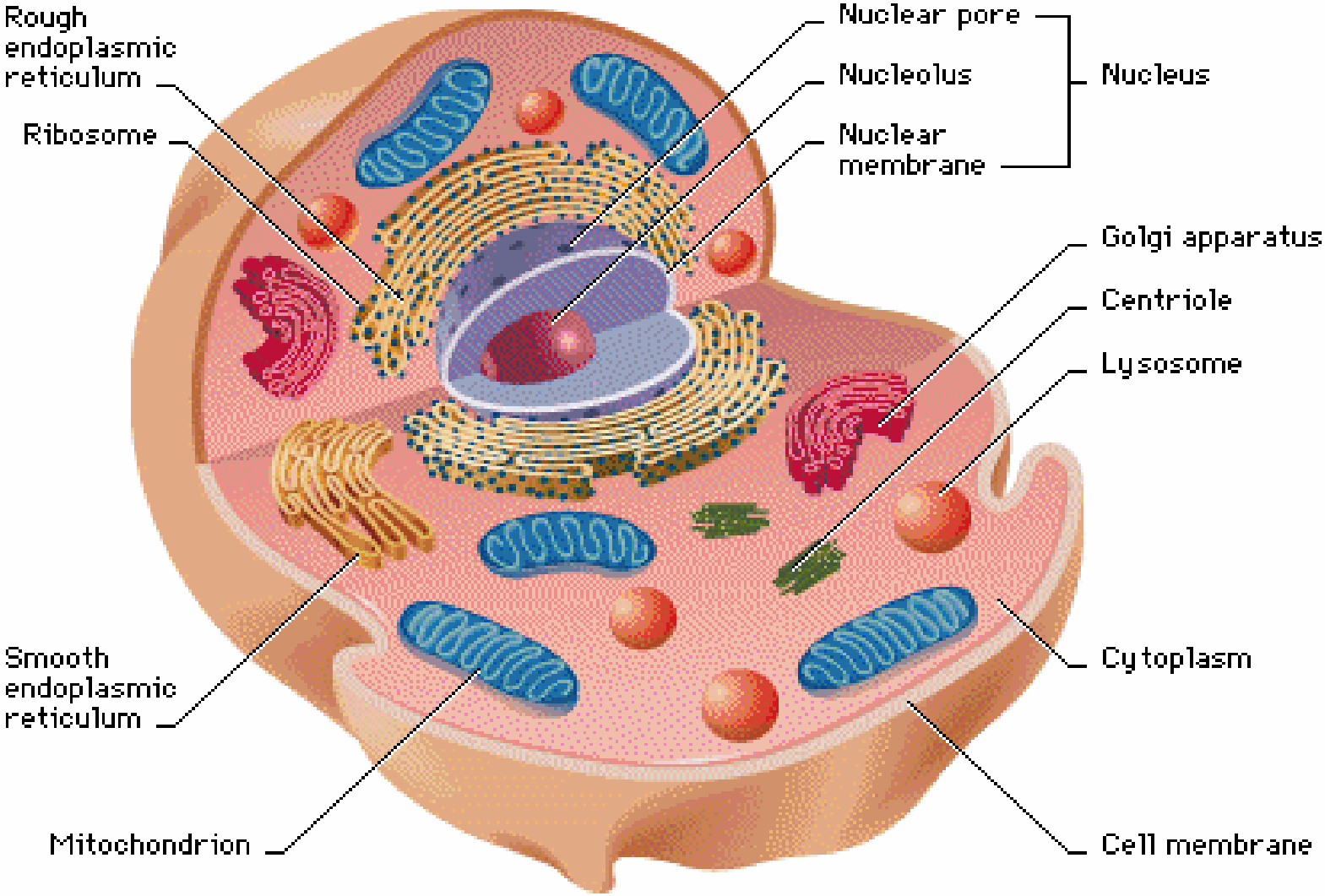
- **It states that two members of a gene pair segregate from each other into the gametes, so that half of the gametes carry one member of the pair and the other half of the gametes carry the other member of the pair.**

Modern genetic terms:

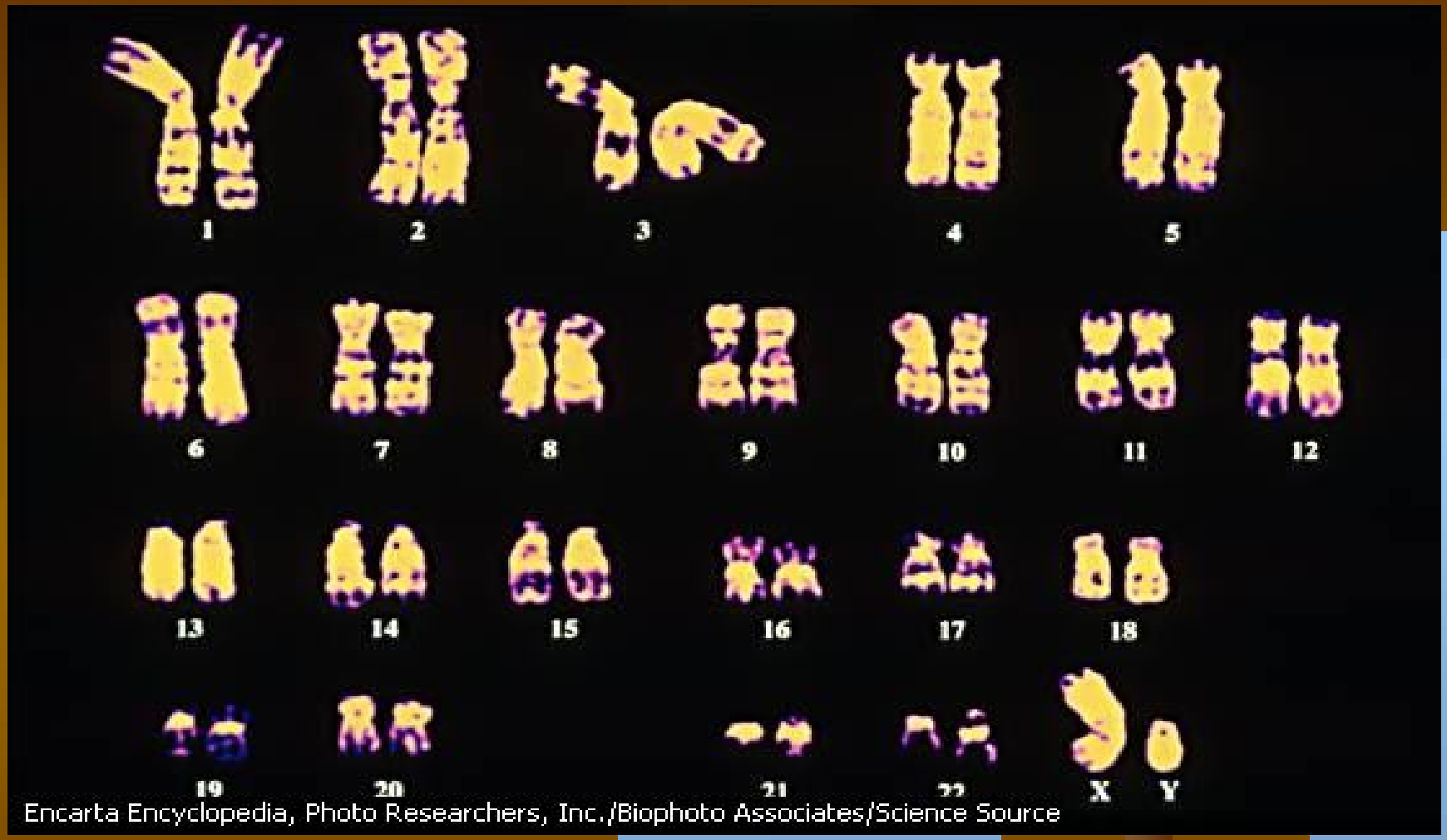
- **Genes** are factors responsible for the inheritance of traits or characteristics.
- **Alleles** are different forms of one type of gene, e.g **T** or **t**.
- **Phenotype** of an individual is the physical expression of a trait or outward appearance.
- **Genotype** is the genetic make up of an individual e.g TT, Tt or tt.
- **Homozygotes** or pure lines are individuals having identical alleles (TT or tt).
- **Heterzygotes** or hybrids are individuals with un-identical alleles (Tt).



Animal Cell



Chromosome



Encarta Encyclopedia, Photo Researchers, Inc./Biophoto Associates/Science Source

This karyotype of a human male shows the 23 pairs of chromosomes that are typically present in human cells. The chromosome pairs labeled 1 through 22 are called autosomes, and have a similar appearance in males and females. The 23rd pair, shown on the bottom right, represents the sex chromosomes. Females have two identical-looking sex chromosomes that are both labeled X, whereas males have a single X chromosome and a smaller chromosome labeled Y..

Table 1. A karyotype of eight common domestic animals

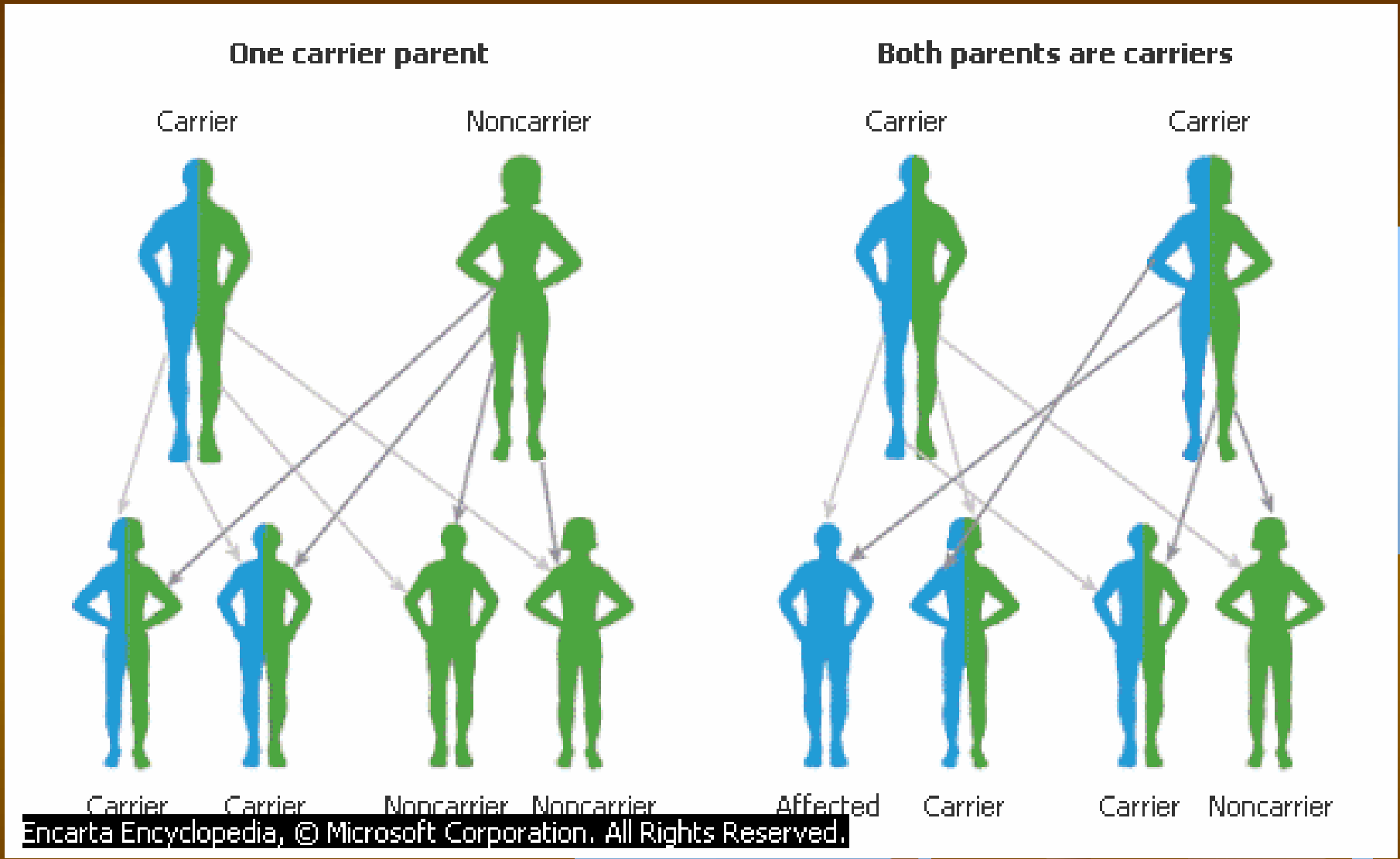
Common name	Specific name	Haploid No. (n)	Diploid No. (2n)	No of Metacentrics	No of Telocentrics	X	Y
Dog	Canis familiaris	39	78	0	38	M	A
Cat	*Felis catus*	19	38	16	2	M	M
Pig	*Sus scrofa*	19	38	12	6	M	M
Goat	*Capra hircus*	30	60	0	29	A	M
Sheep	*Ovis aries*	26	54	3	23	A	M
Cattle	*Bos Taurus*	30	60	0	29	M	M
Horse	*Uquus caballus*	32	64	13	18	M	A
Donkey	*Equus asinus*	31	62	24	6	M	A

Table 1. A karyotype of eight common domestic animals

Albinism

Albinism, the lack of normal pigmentation, occurs in all races. A rare condition, albinism occurs when a person inherits a recessive allele, or group of genes, for pigmentation from each parent. In this case, production of the enzyme tyrosinase is defective. Tyrosinase is necessary for the formation of melanin, the normal human skin pigment. Without melanin, the skin lacks protection from the sun and is subject to premature aging and skin cancer. The eyes, too, colorless except for the red blood vessels of the retina that show through, cannot tolerate light. Albinos tend to squint even in normal indoor lighting and frequently have vision problems

Abinism



Non-carrier: **AA**

Carrier: **Aa**

Albino: **aa**

Punnett Square

A convenient method of predicting the relative ratios of the progeny in any cross is by constructing a **Punnett Square** named after R.C. Punnett, who first devised the approached.

F1 cross : **T t** X **T t**

Pheotype: *Tall* *Tall*

Gamete formation by F1 parents: T t T t

T t T t

Male/Female	**T**	**t**
T	**TT** *Tall*	**Tt** *Tall*
t	**Tt** *Tall*	**tt** *Dwarf*

	Genotype	**Phenotype**
	1 TT	¾ Tall
	2 Tt	
	1 tt	¼ Dwarf
Ratio:	1:2:1	3:1

Test Cross (One character)

- The organism of a dominant phenotype but unknown genotype is crossed to a homozygous recessive individual (tester). Consider a test cross illustrated with a single character in the following cases:

- 1. *If the tall parent is homozygous,*

  Parental phenotype: Homozygous tall Homozygous dwarf

  Parental genotype (2n): **T T** **X** **t t**

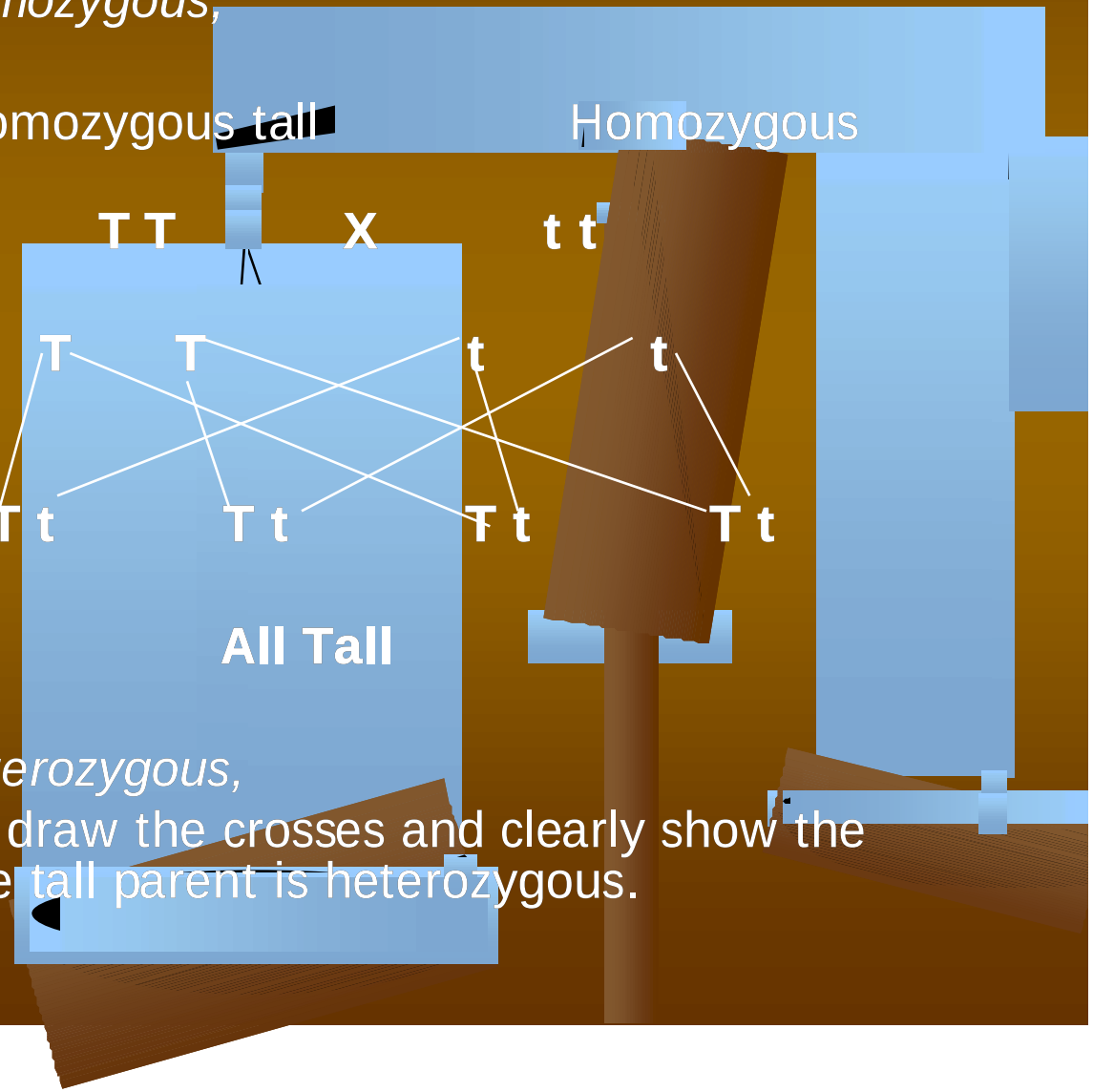
  Gametes (n): **T** **T** **t** **t**

  F1 Genotype (2n): **T t** **T t** **T t** **T t**

  Resulting phenotype: **All Tall**

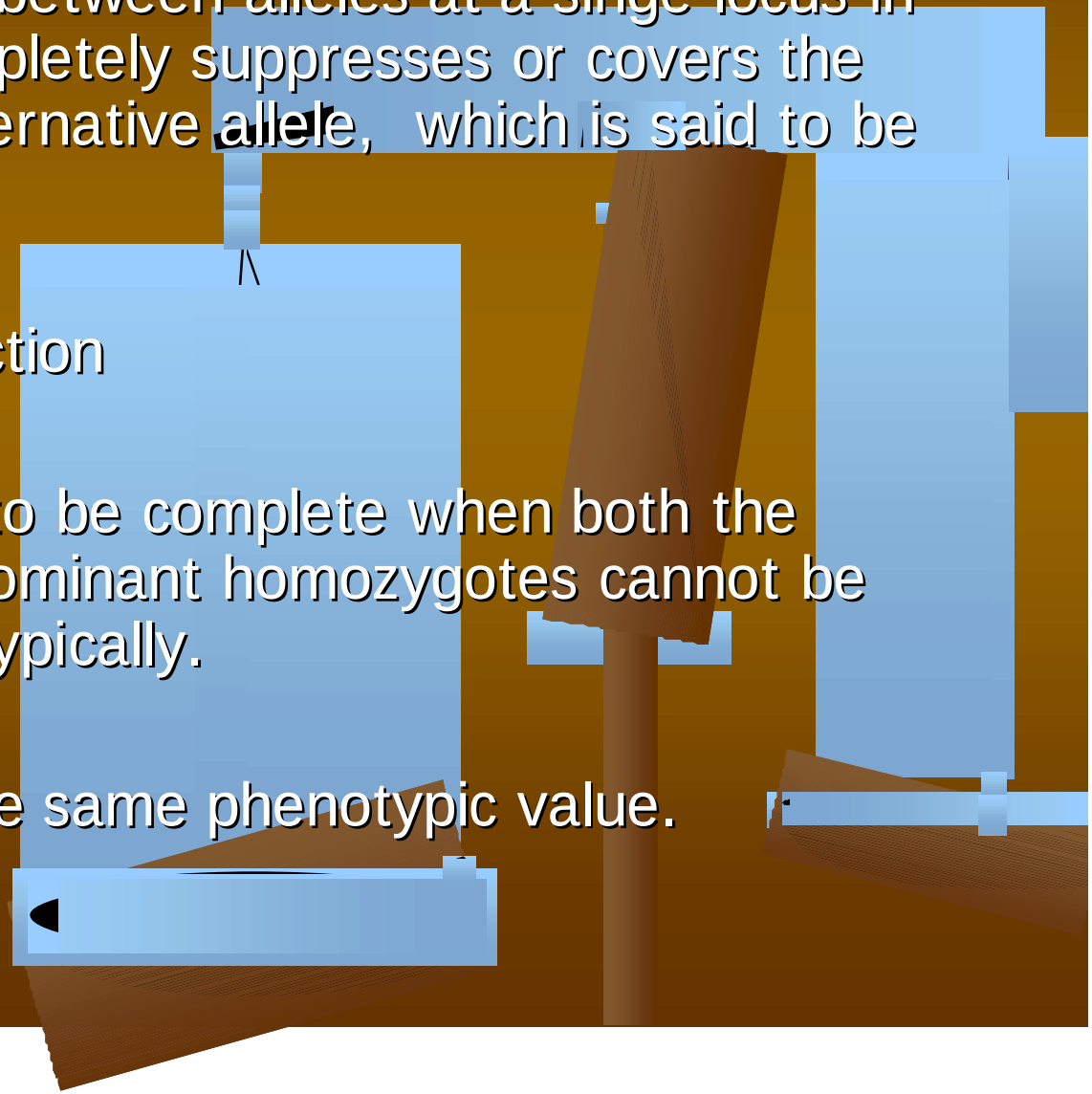
  2. *If the tall parent is heterozygous,*

- ***Assignment:** Similarly draw the crosses and clearly show the resulting phenotype if the tall parent is heterozygous.



Dominance - recessive

- Allelic interaction
- Result of interaction between alleles at a singe locus in which one allele completely suppresses or covers the expression of the alternative allele, which is said to be **recessive**.
- Non-additive gene action
- **Dominance** is said to be complete when both the heterozygotes and dominant homozygotes cannot be distinguished phenotypically.
- That is, they have the same phenotypic value.



Exceptions to Mendel's rules

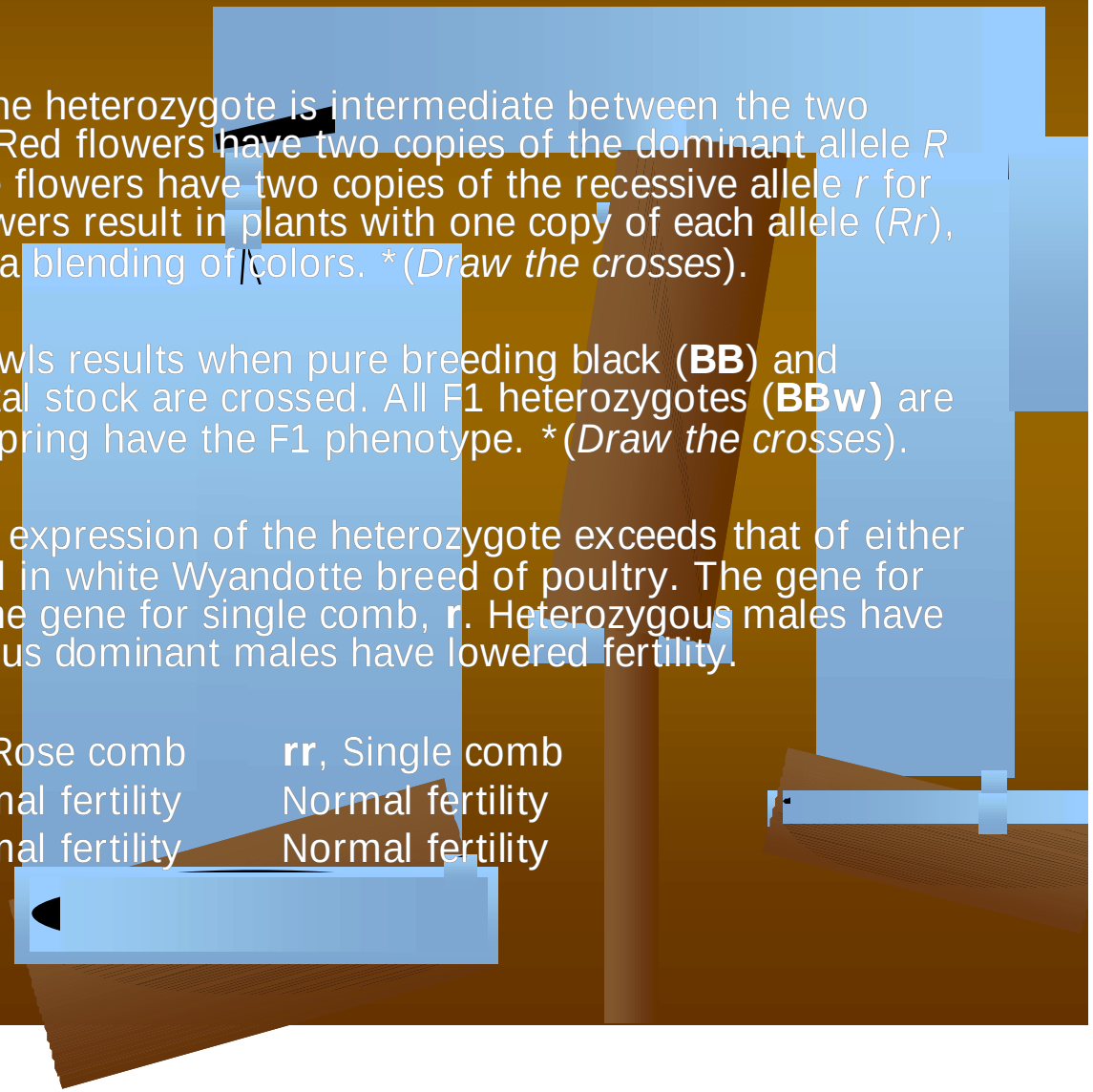
- **1. Incomplete dominance**
- Inheritance of a dominant and a recessive allele results in a blending of traits so that heterozygotes can be distinguished phenotypically from the dominant homozygotes. There are two types:

- i. Co-dominance:
- The phenotypic expression of the heterozygote is intermediate between the two homozygotes. E.g. **In plants**: Red flowers have two copies of the dominant allele *R* for red flower color (*RR*). White flowers have two copies of the recessive allele *r* for white flower color (*rr*). Pink flowers result in plants with one copy of each allele (*Rr*), with each allele contributing to a blending of colors. *(*Draw the crosses*).

- **In poultry**, blue Andalusian fowls results when pure breeding black (**BB**) and splashed white (**BwBw)** parental stock are crossed. All F1 heterozygotes (**BBw)** are 'blue', while 50% of the F2 offspring have the F1 phenotype. *(*Draw the crosses*).

- ii. Over-dominance: Phenotypic expression of the heterozygote exceeds that of either homozygotes. Example is found in white Wyandotte breed of poultry. The gene for Rose comb **R**, is dominant to the gene for single comb, **r**. Heterozygous males have normal fertility while homozygous dominant males have lowered fertility.

	RR, Rose comb	**Rr**, Rose comb	**rr**, Single comb
Male:	*Lower fertility	Normal fertility	Normal fertility
Female:	Normal fertility	Normal fertility	Normal fertility



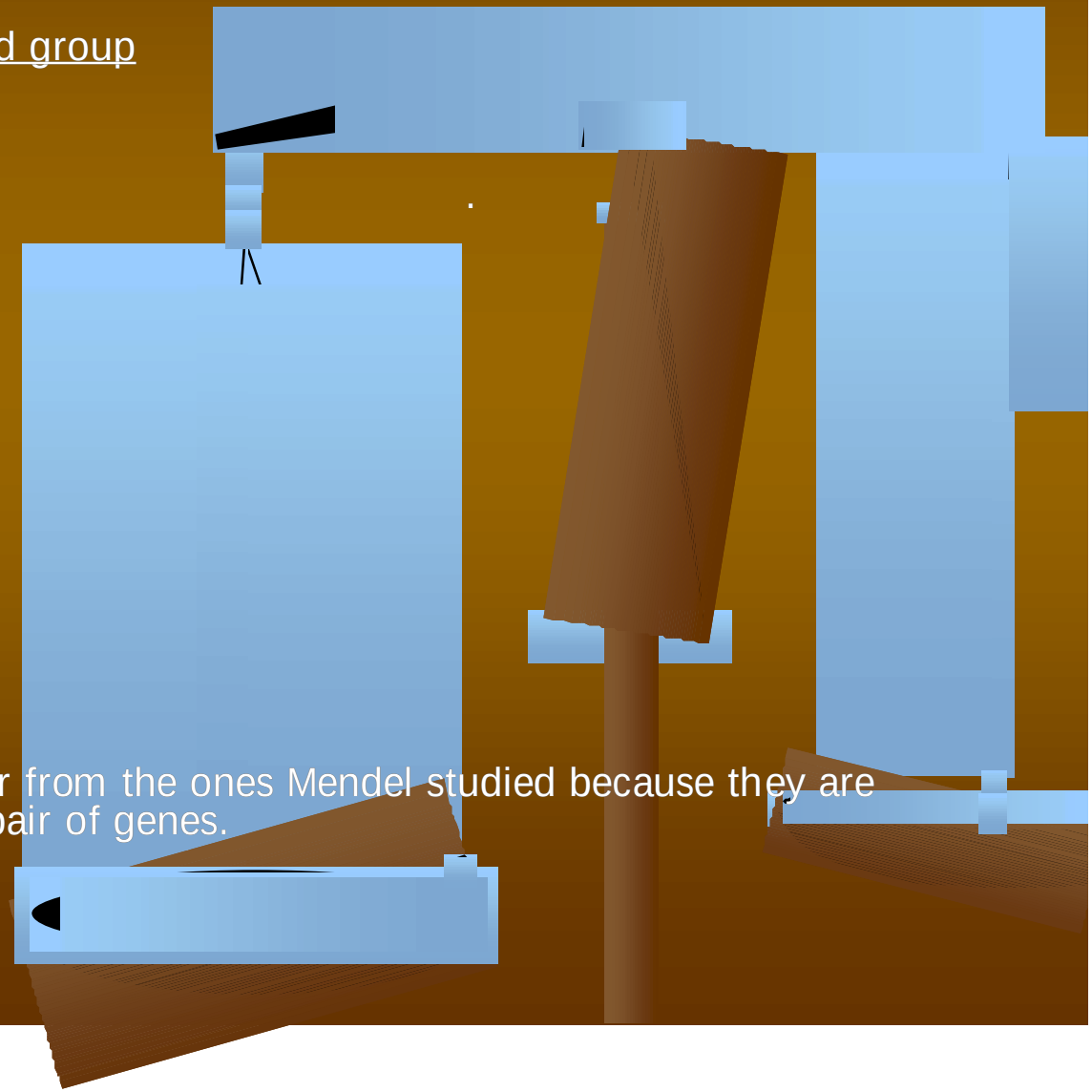
Exceptions to Mendel's rules Cont…

2. Multiple alleles

Inheritance of blood groups: Blood group is controlled by and autosomal gene locus I, standing for Isohaemaglutinogen and there are 3 alleles representing the symbols **A, B, O**. A and B are equally dominant and O is recessive to both.

Human blood group genotypes:

Genotypes	Blood group
$I^A I^A$	A
$I^A I^O$	A
$I^B I^B$	B
$I^B I^O$	B
$I^A I^B$	AB
$I^O I^O$	O



- 3. Lethal genes
- 4. Gene linkage
- 5. Sex-linked traits
- 6. **Quantitative Inheritance**

  Traits such as skin color differ from the ones Mendel studied because they are determined by more than one pair of genes.

Dihybrid inheritance

- Inheritance of 2 pairs of contrasting characteristics. e.g. Seed shape and colour in plants; Coat colour (black or red) and presence or absence of horns in cattle.

Consider seed shape (round or wrinkle) and seed colour (Yellow or green):

- Round (**R**) is dominant to wrinkle (**r**), and Yellow (**Y**) is dominant to green (**y**)

- Parental phenotypes: Round-yellow X wrinkled-green
- Parental genotypes (2n): **RRYY** X **rryy**
- Gamete (n): **RY** **ry**
- F1 Genotype (2n): **RrYy** (*Round-yellow*)
- F1 heterozygote plants were self pollinated to produce F2 generation from four kinds of gametes.

- F1 cross: **RrYy** X **RrYy**

- Gametes (n) **RY** **Ry** **rY** **ry**

- Note: Segregation of alleles (**R, r, Y, y**) and their independent assortment (recombination) result to RY, Ry, rY and ry which are four possible arrangements of alleles in each of the male and female gametes.

Mendel's second law (Law of independent assortment)

- The law states that gene pairs assort independently during gamete formation.

Formation of gametes

F1 cross: Male **RrYy** X Female **RrYy**

Gametes:

	R	r
Y	**RY**	**rY**
y	**Ry**	***ry***

	R	r
Y	**RY**	**rY**
y	**Ry**	***ry***

Sperms

Male/Female					
Eggs		RY	Ry	rY	ry
	RY	**RRYY** *Round-yellow*	**RRYy** *Round-yellow*	**RrYY** *Round-yellow*	**RrYy** *Round-yellow*
	Ry	**RRYy** *Round-yellow*	**RRyy** *Round-green*	**RrYy** *Round-yellow*	**Rryy** *Round-green*
	rY	**RrYY** *Round-yellow*	**RrYy** *Round-yellow*	**rrYY** *wrinkled-yellow*	**rrYy** *wrinkled-yellow*
	ry	**RrYy** *Round-yellow*	**Rryy** *Round-green*	**rrYy** *wrinkled-yellow*	**rryy** *Wrinkled-green*

Dihybrid inheritance Continue…

Male/Female	Sperms				
Eggs		RY	Ry	rY	Ry
	RY	**RRYY** *Round-yellow*	**RRYy** *Round-yellow*	**RrYY** *Round-yellow*	**RrYy** *Round-yellow*
	Ry	**RRYy** *Round-yellow*	**RRyy** *Round-green*	**RrYy** *Round-yellow*	**Rryy** *Round-green*
	rY	**RrYY** *Round-yellow*	**RrYy** *Round-yellow*	**rrYY** *wrinkled-yellow*	**rrYy** *wrinkled-yellow*
	ry	**RrYy** *Round-yellow*	**Rryy** *Round-green*	**rrYy** *wrinkled-yellow*	**rryy** *wrinkled-yellow*

F2 ratios:

Genotypic	**Phenotypic ratios**	**Actual plant counts**	**Ratios**
1/16 RRYY 2/16 RRYy 2/16 RrYY 4/16 RrYy	**16** Round-yellow (R-Y-)	315	9
1/16 RRyy 2/16 Rryy	**3/16** Round-green (**R- yy**)	108	3
1/16 rrYY 2/16 rrYy	**3/16** Wrinkled-yellow (**rr Y-**)	101	3
1/16 rryy	**1/16** wrinkled-green	32	1

Law of product probability

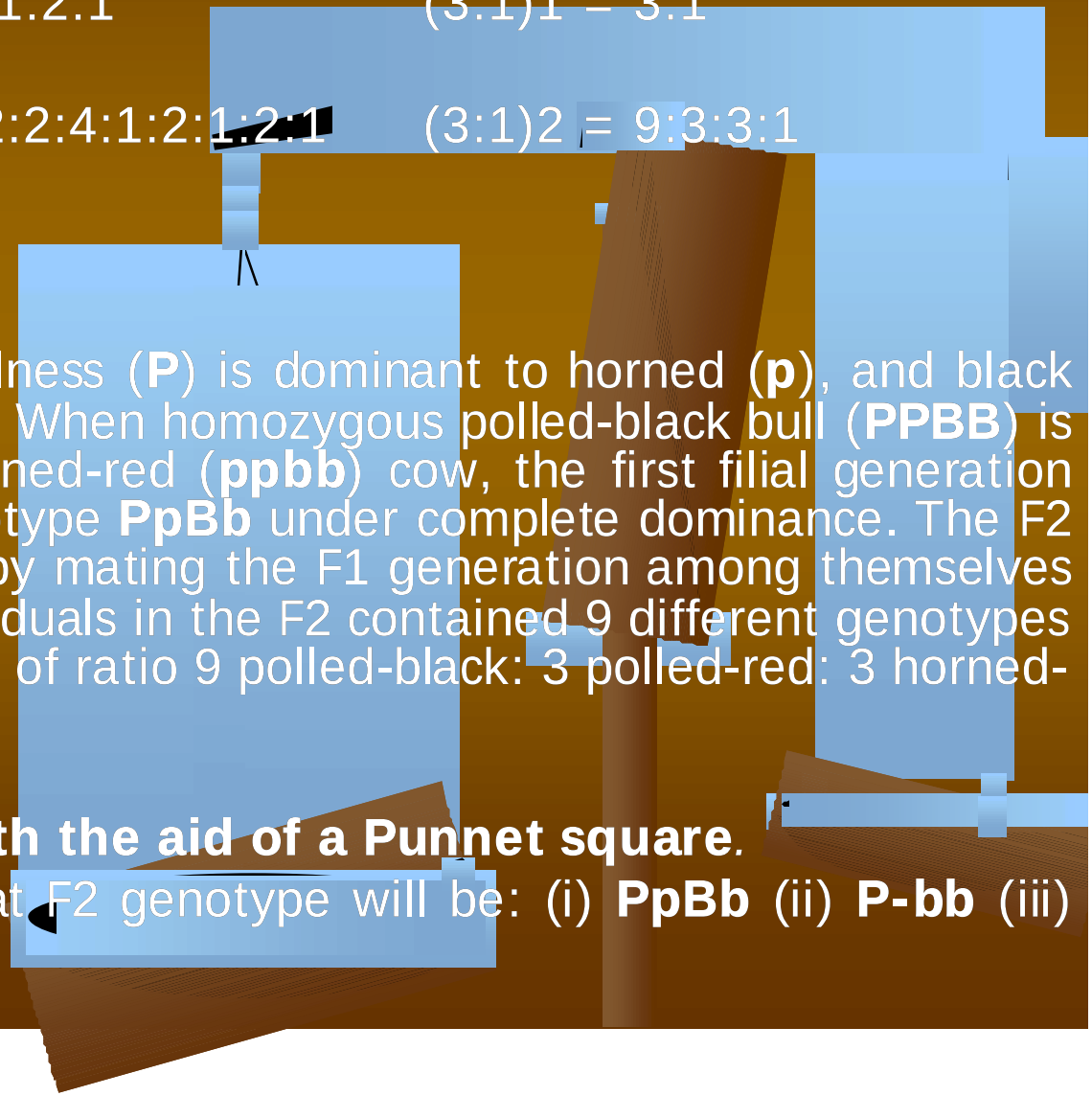
It states that “If two events are independent, the probability that both events will occur simultaneously is the product of their separate probabilities”.

The dihybrid ratio is also obtained by multiplying the expected monohybrid ratios for two gene pairs considered separately.

Ratios		3		1
	X	¼ RR	½ Rr	¼ rr
3	¼ YY	**9/16 R-Y-**		**3/16 rrY-**
	½ Yy			
1	¼ yy	**3/16 R-yy**		**1/16 rryy**

Summary

No. of genes	Gametes	F2 genotypic ratio	F2 phenotypic ratio
(n)	**(2n)**	**(3n)**	**(3:1)n**
1	21 = 2	1:2:1	(3:1)1 = 3:1
2	22 = 4	1:2:2:4:1:2:1:2:1	(3:1)2 = 9:3:3:1
3	...		



- **Example 2**: In cattle, pollness (**P**) is dominant to horned (**p**), and black (**B**) is dominant to red (**b**). When homozygous polled-black bull (**PPBB**) is mated to homozygous horned-red (**ppbb**) cow, the first filial generation was polled-black with genotype **PpBb** under complete dominance. The F2 generation was produced by mating the F1 generation among themselves (**interse** mating). 16 individuals in the F2 contained 9 different genotypes and 4 different phenotypes of ratio 9 polled-black: 3 polled-red: 3 horned-black: 1 horned-red.

  Draw these crosses with the aid of a Punnet square.

- What is the probability that F2 genotype will be: (i) **PpBb** (ii) **P-bb** (iii) **ppB- ?**

Test cross: Two characters

- It applies to individuals that express two dominant traits, but whose genotypes are unknown. E.g. The expression of a round-yellow phenotype may result from **RRYY, RRYy, RrYY or RrYy** genotypes.

- If an F2 round-yellow plant is crossed with a recessive wrinkled-green **(rryy)** plant which is the **tester,** analysis of the offspring will indicate the exact genotype of the round-yellow plant.

- 1. Test cross results of **RRYy** will be as follows:

  Parental phenotypes: Round-yellow Wrinkled-green

  Genotypes (2n): **RRYy** X **rryy**

  Gametes (n): **RY, Ry** **ry**

  Offspring genotype:

	RY	Ry
ry	**RrYy**	**Rryy**

  Phenotypic ratio: **½ Round-yellow: ½ Round-green**

  ***Assignment**

  2. Similarly draw the test cross results of RRYY, RrYY and RrYy.