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# Introduction to Human Genetics

## Module 1 Mendelian genetics

# Mendel's crosses

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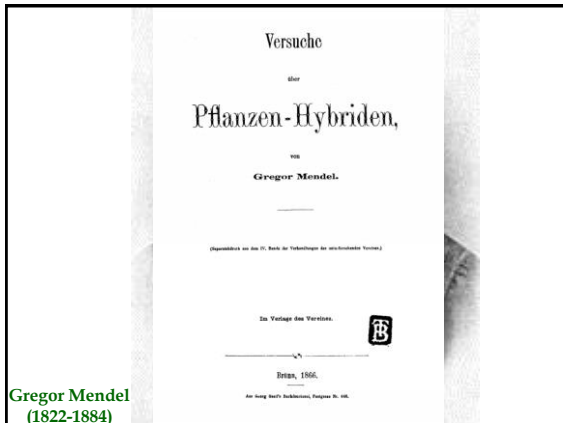
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## Mendel's scientific approach to the study of heredity

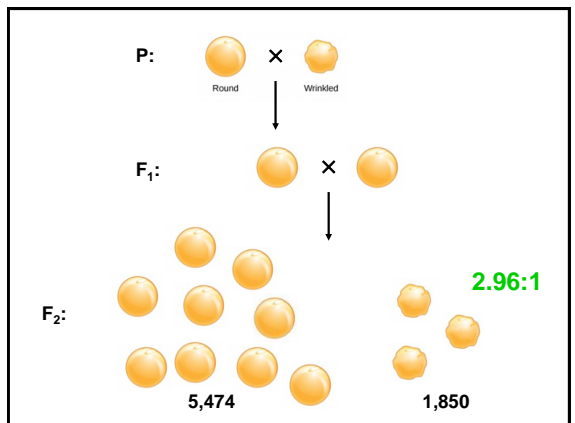
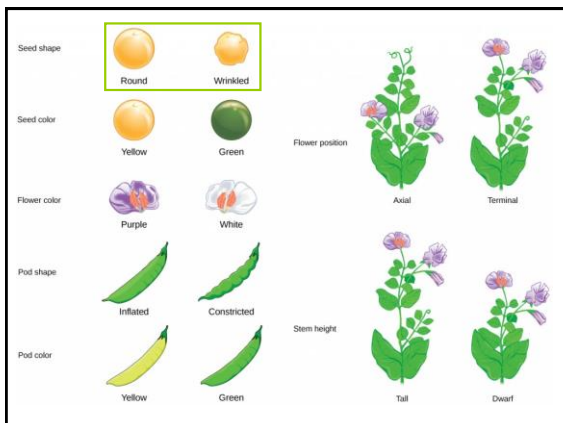
- well characterized materials
- accurate quantitative records
- data analysis => model



Gregor Mendel  
(1822-1884)



Gregor Mendel  
(1822-1884)



The Results of Mendel's Garden Pea Hybridizations				
Characteristic	Contrasting P <sub>0</sub> Traits	F <sub>1</sub> Offspring Traits	F <sub>2</sub> Offspring Traits	F <sub>2</sub> Trait Ratios
Flower color	Violet vs. white	100 percent violet	705 violet 224 white	3.15:1
Flower position	Axial vs. terminal	100 percent axial	651 axial 207 terminal	3.14:1
Plant height	Tall vs. dwarf	100 percent tall	787 tall 277 dwarf	2.84:1
Seed texture	Round vs. wrinkled	100 percent round	5,474 round 1,850 wrinkled	2.96:1
Seed color	Yellow vs. green	100 percent yellow	6,022 yellow 2,001 green	3.01:1
Pea pod texture	Inflated vs. constricted	100 percent inflated	882 inflated 299 constricted	2.95:1
Pea pod color	Green vs. yellow	100 percent green	428 green 152 yellow	2.82:1

## Mendel's conclusions

- UNIT FACTORS IN PAIRS
- DOMINANCE / RECESSIVENESS
- SEGREGATION

## Definitions

### Gene

The fundamental unit of heredity and the basic structural and functional unit of genetics.

### Dominant trait

The trait expressed in the F<sub>1</sub> (or heterozygous) condition.

### Phenotype

The observable properties of an organism.

### Genotype

The specific genetic constitution of an organism.

### Recessive trait

The trait unexpressed in the F<sub>1</sub> but expresses in some members of the F<sub>2</sub> generation

### Allele

One of the possible alternative forms of a gene, usually distinguished from other alleles by its phenotypic effect.

P:

RR

rr

### Homozygous

Having identical alleles for one or more genes.



F<sub>1</sub>:

Rr

Rr

### Heterozygous

Carrying two different alleles for one or more genes.

### Segregation

The separation of members of a gene pair from each other during gamete formation.

## Punnett square

P:

RR

rr

F<sub>1</sub>:

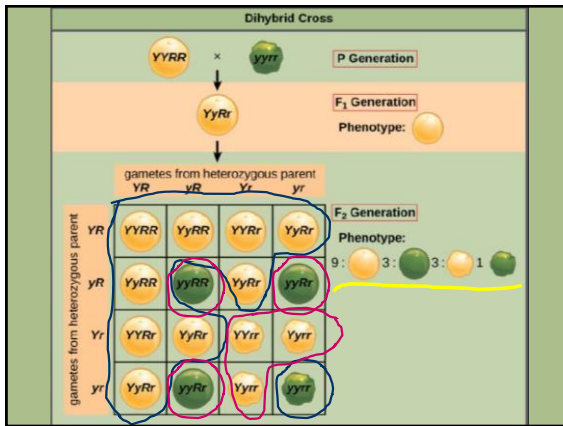
Rr

Rr

F<sub>2</sub>:

	R	r
R	RR	Rr
r	Rr	rr

Studying the inheritance of two characters simultaneously



### Variations on a Theme by Mendel

- incomplete dominance
- codominance
- multiple alleles
- epistasis