GENETICS

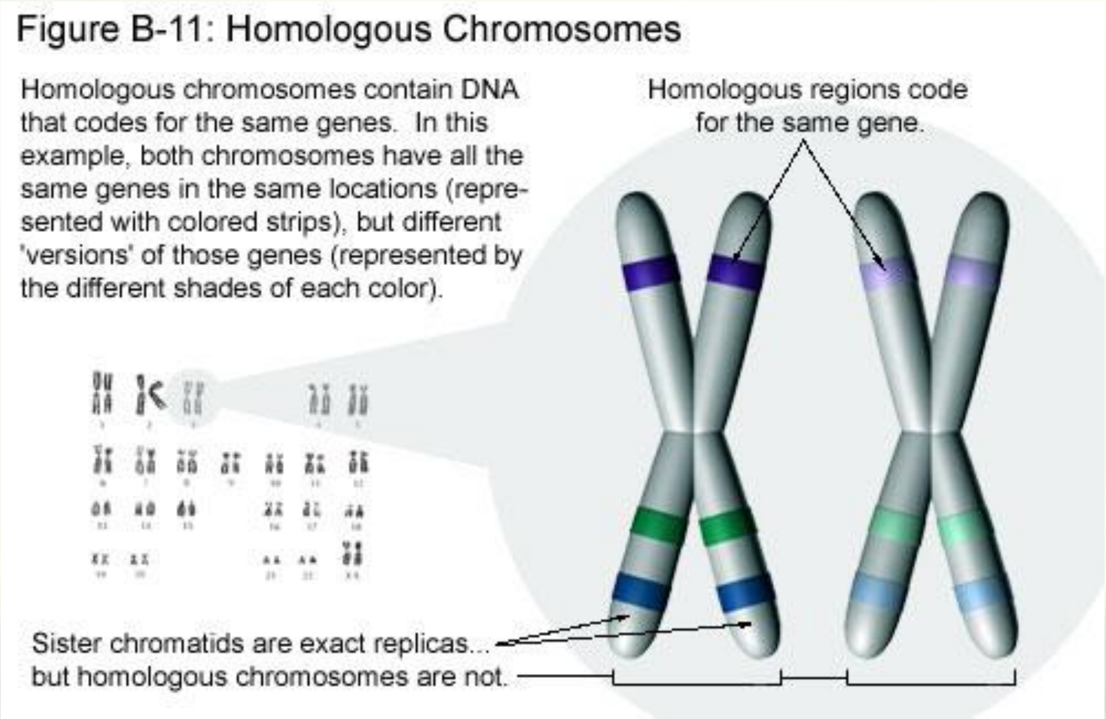
- Introduction to Genetics and heredity
- Gregor Mendel – a brief bio
- Genetic terminology (glossary)
- Monohybrid crosses
- Patterns of inheritance
- Dihybrid crosses
- Test cross
- Beyond Mendelian Genetics – incomplete dominance

Introduction to Genetics

- **GENETICS** – branch of biology that deals with heredity and variation of organisms.

- **Chromosomes** carry the hereditary information (genes)
  - Arrangement of nucleotides in DNA
  - DNA → RNA → Proteins

- Chromosomes (and genes) occur in pairs **Homologous Chromosomes**
- New combinations of genes occur in sexual reproduction
  - Fertilization from two parents



Gregor Johann Mendel

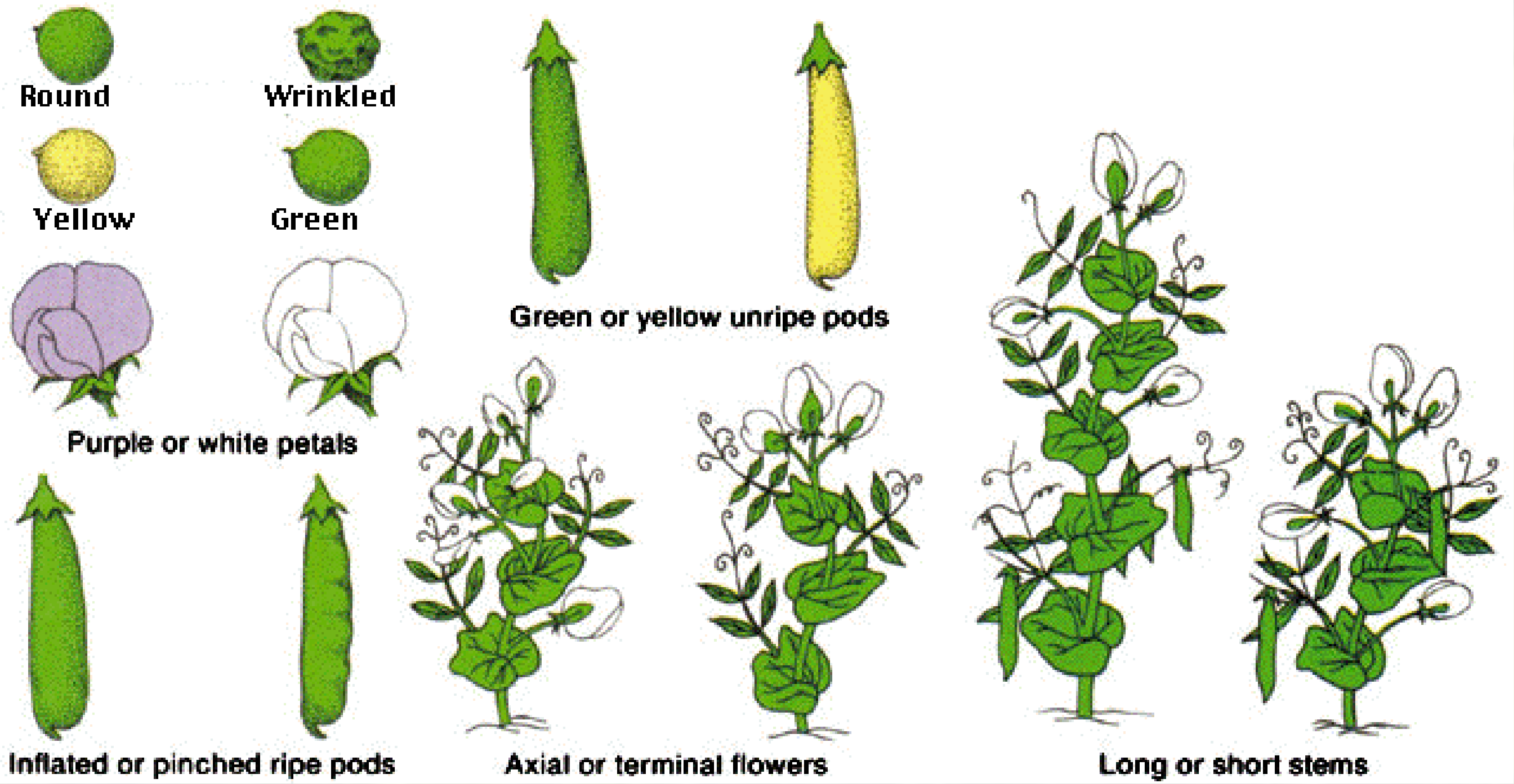
- Austrian Monk, born in what is now Czech Republic in 1822
- Son of peasant farmer, studied Theology and was ordained priest Order St. Augustine.

Gregor Mendel

- Went to the university of Vienna, where he studied botany and learned the Scientific Method
- Worked with pure lines of peas for eight years
- Prior to Mendel, heredity was regarded as a "blending" process and the offspring were essentially a "dilution"of the different parental characteristics.

Mendel's peas

- Mendel looked at seven traits or characteristics of pea plants:



- In 1866 he published *Experiments in Plant Hybridization*, (*Versuche über Pflanzen-Hybriden*) in which he established his three Principles of Inheritance

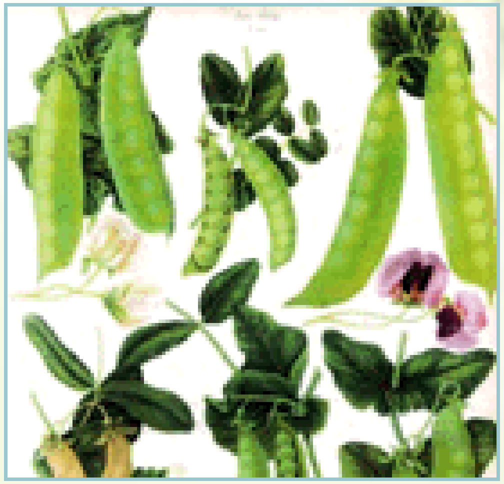
- He tried to repeat his work in another plant, but didn't work because the plant reproduced asexually! If…
- Work was largely ignored for 34 years, until 1900, when 3 independent botanists rediscovered Mendel's work.

- Mendel was the first biologist to use Mathematics – to explain his results quantitatively.
- Mendel predicted

  The concept of genes

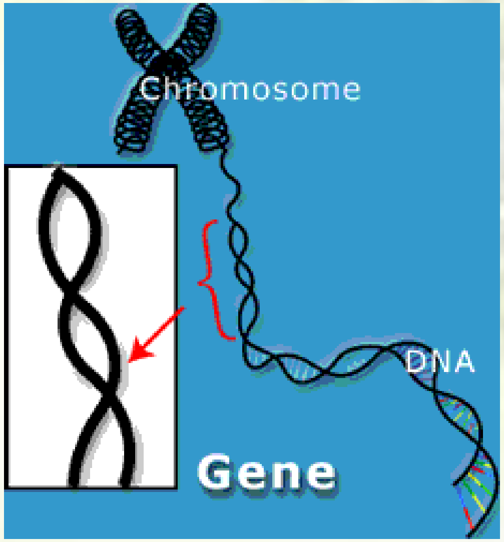
  That genes occur in pairs

  That one gene of each pair is present in the gametes



Genetics terms you need to know:

- **Gene** – a unit of heredity; a section of DNA sequence encoding a single protein
- **Genome** – the entire set of genes in an organism



- **Alleles** – two genes that occupy the same position on homologous chromosomes and that cover the same trait (like ‘flavors’ of a trait).
- **Locus** – a fixed location on a strand of DNA where a gene or one of its alleles is located.

- **Homozygous** – having identical genes (one from each parent) for a particular characteristic.
- **Heterozygous** – having two different genes for a particular characteristic.

- **Dominant** – the allele of a gene that masks or suppresses the expression of an alternate allele; the trait appears in the heterozygous condition.
- **Recessive** – an allele that is masked by a dominant allele; does not appear in the heterozygous condition, only in homozygous.

- **<u>Genotype</u>** – the genetic makeup of an organisms
- **<u>Phenotype</u>** – the physical appearance of an organism (Genotype + environment)

- **Monohybrid cross**: a genetic cross involving a single pair of genes (one trait); parents differ by a single trait.
- **P** = Parental generation
- $\mathbf{F_1}$ = First filial generation; offspring from a genetic cross.
- $\mathbf{F_2}$ = Second filial generation of a genetic cross

7 Characteristics in Peas

Trait	Stem length	Pod shape	Seed shape	Seed color	Flower position	Flower color	Pod color
Characteristics	Tall	Inflated	Smooth	Yellow	Lateral	Purple	Green
	Dwarf	Constricted	Wrinkled	Green	Terminal	White	Yellow

Monohybrid cross

- Parents differ by a single trait.
- Crossing two pea plants that differ in stem size, one tall one short

T = allele for Tall

t = allele for dwarf

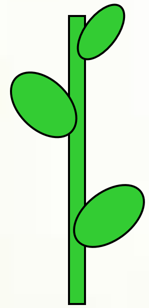
TT = homozygous tall plant

t t = homozygous dwarf plant

T T × t t

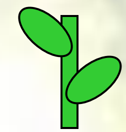
Monohybrid cross for stem length:

P = parentals
true breeding,
homozygous plants:



T T × *t t*

(tall) (dwarf)

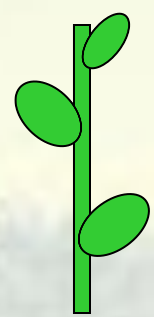


↓

F_1 generation
is heterozygous:

T *t*

(all tall plants)



Punnett square

summarize results (genotypes & phenotypes of offspring)

T T × ***t t***

	T	**T**
t	**T *t***	**T *t***
t	**T *t***	**T *t***

Genotypes:
100% T t

Phenotypes:
100% Tall plants

Monohybrid cross: F_2 generation

- If you let the F1 generation self-fertilize, the next monohybrid cross would be:

T *t* × **T *t***
(tall) (tall)

	T	***t***
T	**T T**	**T *t***
t	**T *t***	***t t***

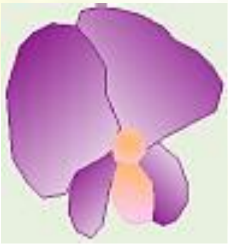
<u>Genotypes:</u>
1 TT= Tall
2 T*t* = Tall
1 *tt* = dwarf
Genotypic ratio= 1:2:1

<u>Phenotype:</u>
3 Tall
1 dwarf
Phenotypic ratio= 3:1

Another example: Flower color

For example, flower color:

P = purple (dominant)



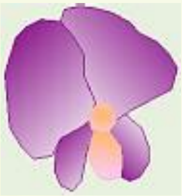
p = white (recessive)

If you cross a homozygous Purple (PP) with a homozygous white (*pp*):

P P × *p p*

↓

P *p*



ALL PURPLE (P*p*)

Cross the F1 generation:

P *p* × P *p*

	P	***p***
P	**P P**	**P *p***
p	**P *p***	***p p***

Genotypes:

1 PP

2 Pp

1 pp

Phenotypes:

3 Purple

1 White

Mendel's Principles

- **1. Principle of Dominance**:

  One allele masked another, one allele was dominant over the other in the F_1 generation.

- **2. Principle of Segregation**:

  Also known as Law of purity. It states that when gametes are formed, the pairs of hereditary factors (genes) become separated, so that each gamete (egg/sperm) receives only one kind of gene.

Law of segregation or purity of gametes or monohybrid cross

phenotypes of parents: tall   dwarf

genotypes of parents: Tt × Tt

gametes: (T) (t) × (T) (t)

punnett square

	(T)	(t)
(T)	TT	Tt
(t)	Tt	tt

F_1 genotypes: 1 TT, 2 Tt, 1 tt

F_1 phenotypes: tall tall dwarf

ratio: 3 tall : 1 dwarf

Principle of Independent Assortment

- Based on these results, Mendel postulated the

- **3. Principle of Independent Assortment**:

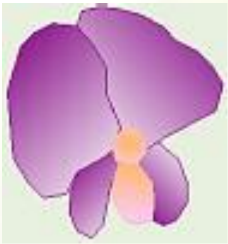
  According to the law of independent assortment, during the inheritance of two different traits, the alleles of both the traits assort and are inherited independently of one another during gamete formation. This gives both the trait equal chances of being inherited.

Dihybrid crosses

- Matings that involve parents that differ in **two** genes (two independent traits)

For example, flower color:

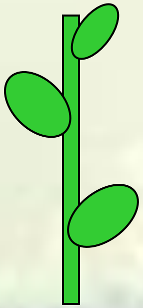
P = purple (dominant)



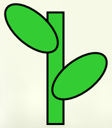
p = white (recessive)

and stem length:

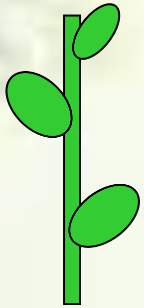
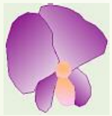
T = tall



t = short

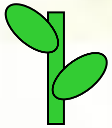


Dihybrid cross: flower color and stem length

TT PP × *tt pp*

(tall, purple) (short, white)



Possible Gametes for parents

T P and *t p*

	tp	*tp*	*tp*	*tp*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross: flower color and stem length (shortcut)

TT PP × *tt pp*

(tall, purple) (short, white)

Possible Gametes for parents

T P *t p*

	T P
t p	T*t* P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross F_2

If F_1 generation is allowed to self pollinate, Mendel observed 4 phenotypes:

T*t* P*p* × T*t* P*p*

(tall, purple) (tall, purple)

Possible gametes:

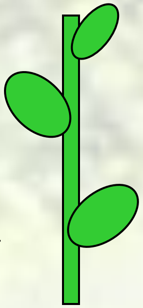
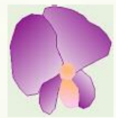
TP T*p* *t*P *tp*

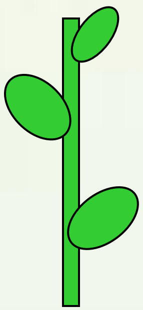
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

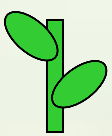
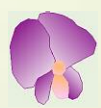
Four phenotypes observed

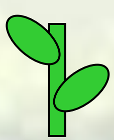
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Dihybrid cross

9 Tall  purple 

3 Tall  white 

3 Short  purple 

1 Short  white 

	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

Phenotype Ratio = 9:3:3:1

Cross: *TtYy* x *TtYy*

♂ / ♀	*TY*	*Ty*	*tY*	*ty*
TY	*TTYY* Tall, yellow	*TTYy* Tall, yellow	*TtYY* Tall, yellow	*TyYy* Tall, yellow
Ty	*TTYy* Tall, yellow	*TTyy* Tall, green	*TtYy* Tall, yellow	*Ttyy* Tall, green
tY	*TtYY* Tall, yellow	*TtYy* Tall, yellow	*ttYY* Dwarf, yellow	*ttYy* Dwarf, yellow
ty	*TtYy* Tall, yellow	*Ttyy* Tall, green	*ttYy* Dwarf, yellow	*ttyy* Dwarf, green

Genotype: 1 *TTYY* : 2 *TTYy* : 4 *TyYy* : 2 *TtYY* : 1 *TTyy* : 2 *Ttyy* : 1 *ttYY* : 2 *ttYy* : 1 *ttyy*

Phenotype: 9 tall plants with yellow seeds; 3 tall plants with green seeds; 3 dwarf plants with yellow seeds; 1 dwarf plant with green seeds

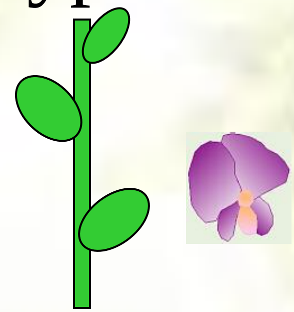
Dihybrid cross: 9 genotypes

Genotype ratios (9):

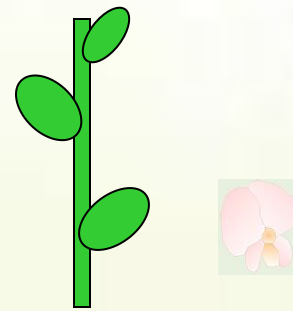
1	TTPP
2	TTP*p*
2	T*t*PP
4	T*t*P*p*
1	TT*pp*
2	T*tpp*
1	*tt*PP
2	*tt*P*p*
1	*ttpp*

Four Phenotypes:

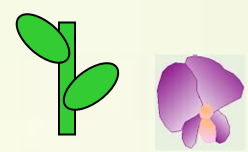
Tall, purple (9)



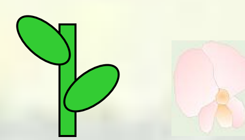
Tall, white (3)



Short, purple (3)



Short, white (1)



Test cross

When you have an individual with an unknown genotype, you do a **test cross.**

Test cross: Cross with a homozygous recessive individual.

For example, a plant with **purple** flowers can either be **PP** or **P*p***… therefore, you cross the plant with a *pp* (white flowers, homozygous recessive)

P ?   ×   *pp*

Test cross

- If you get all 100% purple flowers, then the unknown parent was PP…

	P	P
p	**P*p***	**P*p***
p	**P*p***	**P*p***

- If you get 50% white,
  50% purple flowers,
  then the unknown
  parent was P*p*…

	P	*p*
p	**P*p***	***p p***
p	**P*p***	***p p***

Beyond Mendelian Genetics: Incomplete Dominance/ Limitations of Mendelian Genetics

Mendel was lucky!

Traits he chose in the pea plant showed up very clearly…

One allele was dominant over another, so phenotypes were easy to recognize.

But sometimes phenotypes are not very obvious…

It's not always Dominant/Recessive!

Non-Mendelian Inheritance Patterns

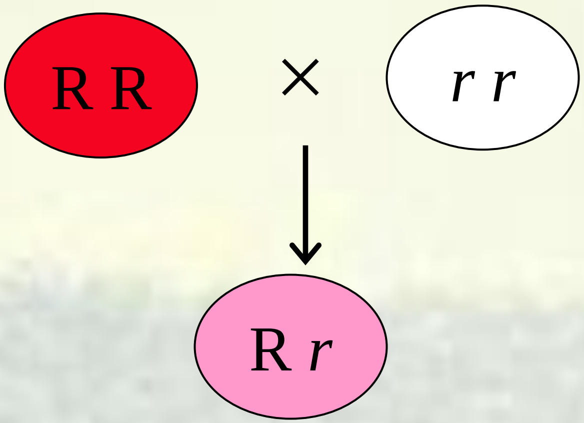
- ❖Incomplete Dominance
- ❖Codominance
- ❖Multiple Alleles
- ❖Polygenic Traits
- ❖Sex-Linked Traits

Incomplete Dominance

Incomplete dominance is **a form of Gene interaction in which both alleles of a gene at a locus are partially expressed, often resulting in an intermediate or different phenotype**. It is also known as Incomplete dominance.

It was described by **Correns**

Antirrhinum (Snapdragon) /*Mirabilis jalapa* (4 o,clock plant) flowers come in many colors.

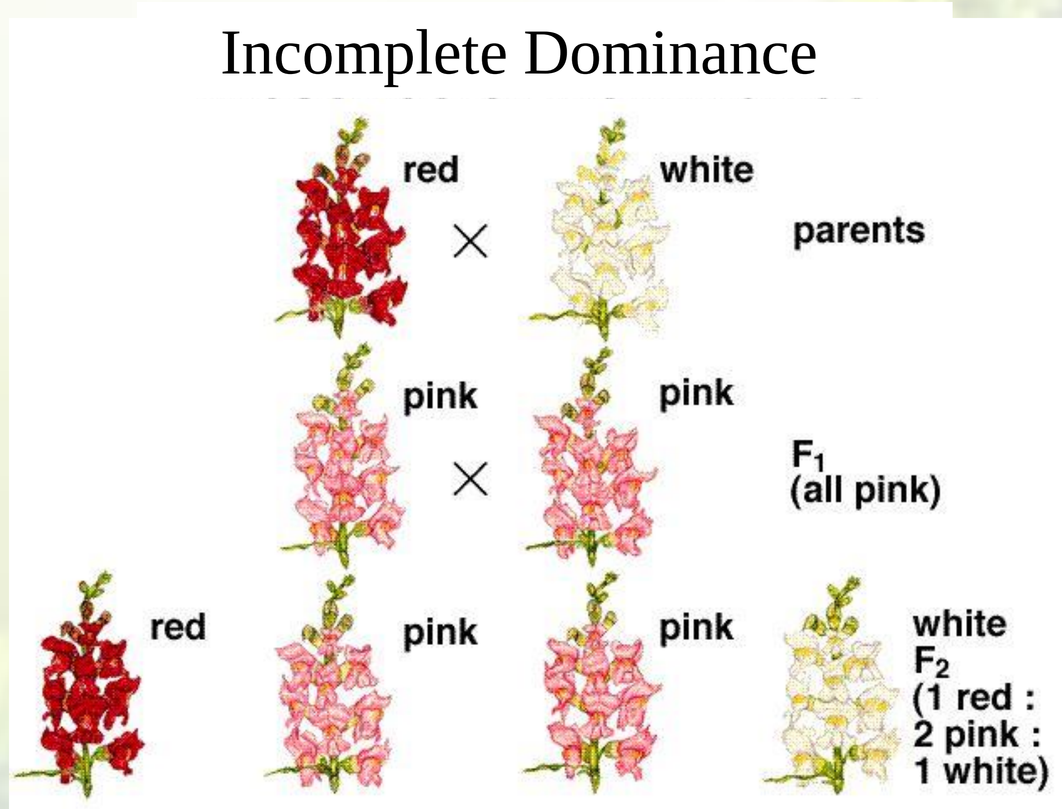


- ✓ Genes show incomplete dominance when the heterozygous phenotype is intermediate.
- ✓ If you cross a red snapdragon (RR) with a white snapdragon (rr)
- ✓ PINK flowers (Rr)

Incomplete dominance

When F1 generation (all pink flowers) is self pollinated, the F2 generation is 1:2:1 red, pink, white

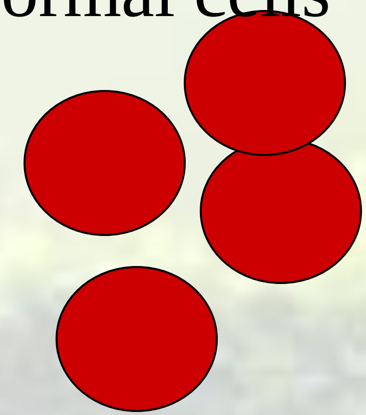
	R	***r***
R	**R R**	**R *r***
r	**R *r***	***r r***



Codominance

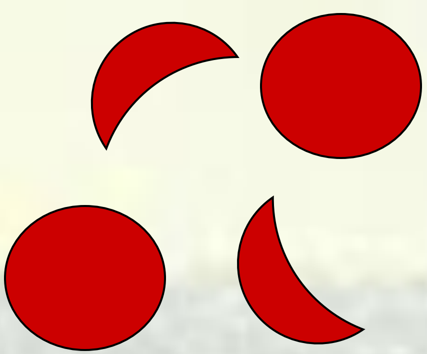
- in the heterozygous condition, **both** alleles are expressed **equally** with NO blending! Represented by using two DIFFERENT capital letters.
- Example - Sickle Cell Anemia: All Normal Cells (NN) + All sickled cells (SS) = half normal/half sickle carrier (NS)

NN = normal cells



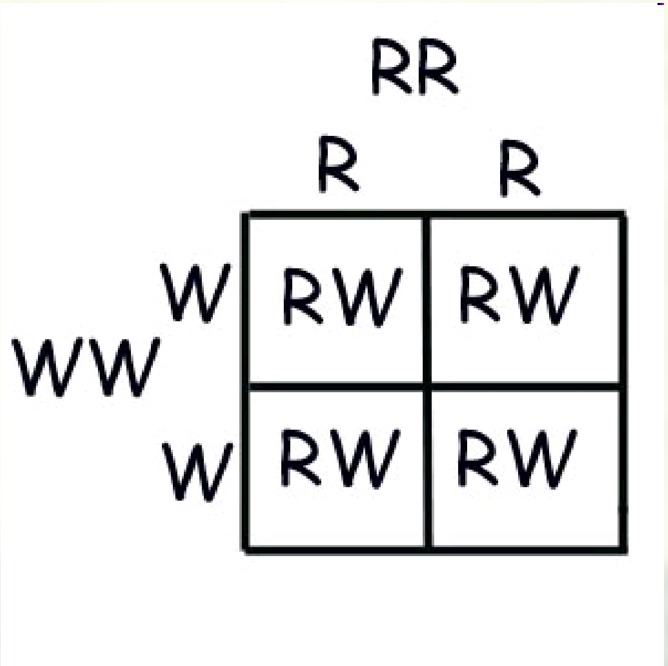
SS = sickle cells

NS = some of each



Codominance Example: Rhodedendron

- R = allele for red flowers
- W = allele for white flowers
- Cross a homozygous red flower with a homozygous white flower.



Codominance Example: Roan cattle

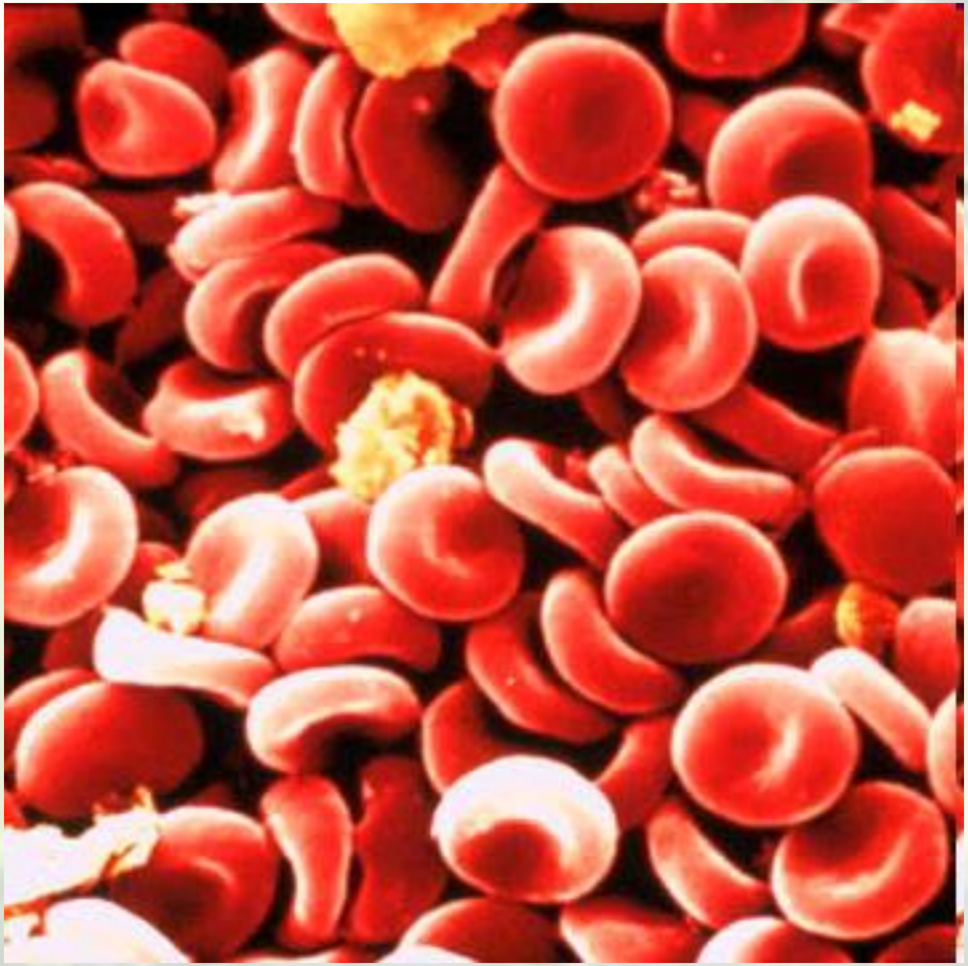
- cattle can be

red
(RR – all red hairs)
white
(WW – all white hairs)
roan
(RW – red and white hairs **together**)

Multiple Alleles

- Sometimes there are **more than two alleles** present in the gene pool for a gene. Ex – blood type consists of two dominant and one recessive allele in humans

Allele A (I_A) and B (I_B) are dominant over Allele O (i).

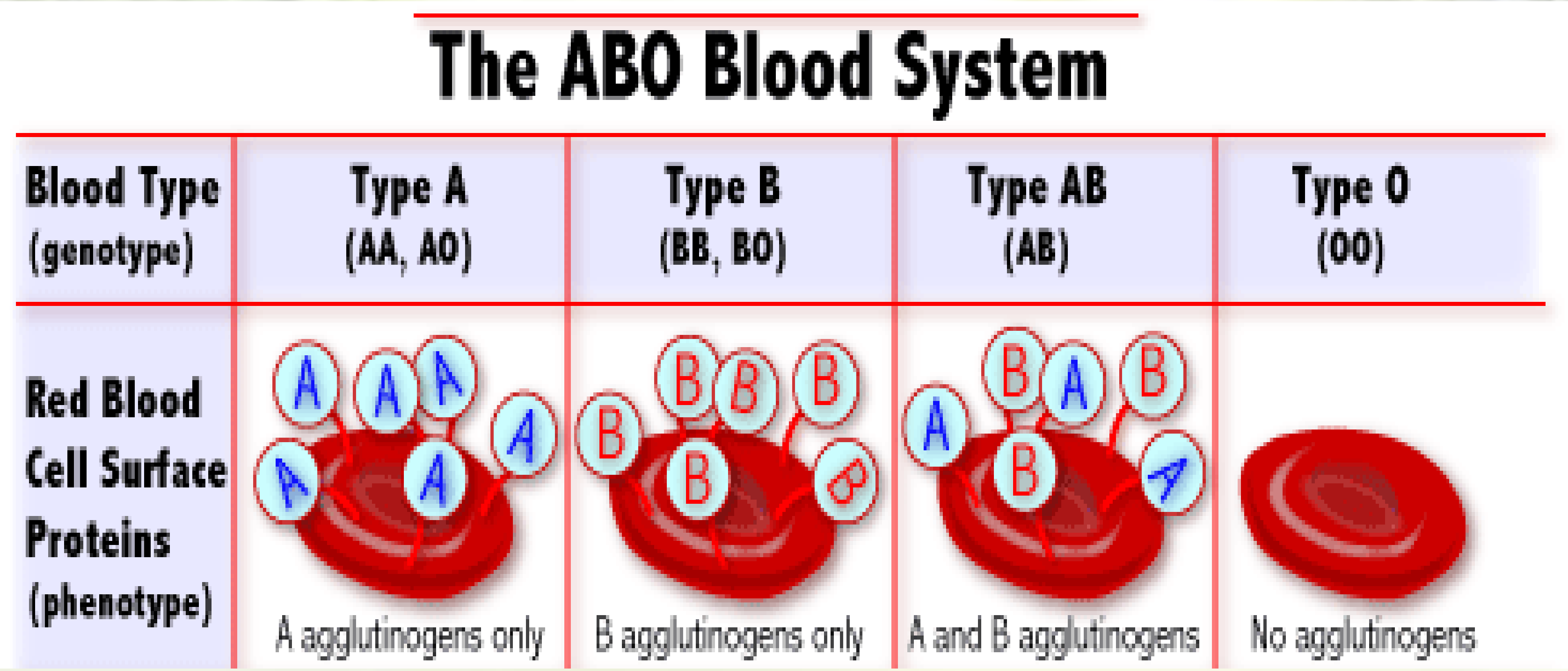


(NOTE: You still only get TWO alleles!!! One from mom and one from dad)

Multiple Alleles: Blood Types (A, B, AB, O)

- Rules for Blood Types: *(geno = pheno)*
  - A and B are co-dominant *(Both show)*
    - $I^{A}I^{A}$ = type A
    - $I^{B}I^{B}$ = type B
    - $I^{A}I^{B}$ = type AB
  - A and B are both dominant over O *(Regular dom/rec)*
    - $I^{A}i$ = type A
    - $I^{B}i$ = type B
    - ii = type O

Multiple Alleles: Blood Types (A, B, AB, O)



Phenotype	Possible Genotype(s)	Allele (antigen) on RBC surface	Can Donate Blood To	Can Receive Blood From
A	I^Ai I^AI^A	A	A, AB	A, O
B	I^Bi I^BI^B	B	B, AB	B, O
AB	I^AI^B	AB	AB	A, B, AB, O
O	ii	O	A, B, AB, O	O

Polygenic inheritance

- When one phenotypic character is controlled by more than one gene, it is called **polygenic inheritance**
- **Kolerenter** is known as father of polygenic inheritance

- It is also called **Quantitative inheritance**

- The quantity of inheritance depends on dominant alleles

- Dominant alleles have **cumulative effect** each expressing part of trait

- Gene involved in quantitative inheritance is known as **polygenes**

- Polygenic inheritance don't follow the **mendelian ratio**

- Eg; **skin color of man, wheat kernel colour**

Ratio :

- when 2 polygene are considered-

  1:4:6:4:1

- when 3 polygene are considered-

  1:6:15:20:15:6:1

Skin colour of man

- It was first studied by **Devenport** (1913) in case of Negro-Europeian intermarriage.

- Skin colour is due to pigment melanin. **More pigment, darker is the colour.**

- There are **three genes (polygene)** controlling the production of melanin.
- But, for convenience we consider only **two pair of genes.**
- For **Negro** :AABB (maximum melanin)
- For **Albino** :aabb (minimum melanin)

- **Parents-** **Negro** (high melanin) AABB — **Albino** (no melanin) aabb
- **Gametes-** **AB** × **ab**

AaBb

- **F1--** Mulleto

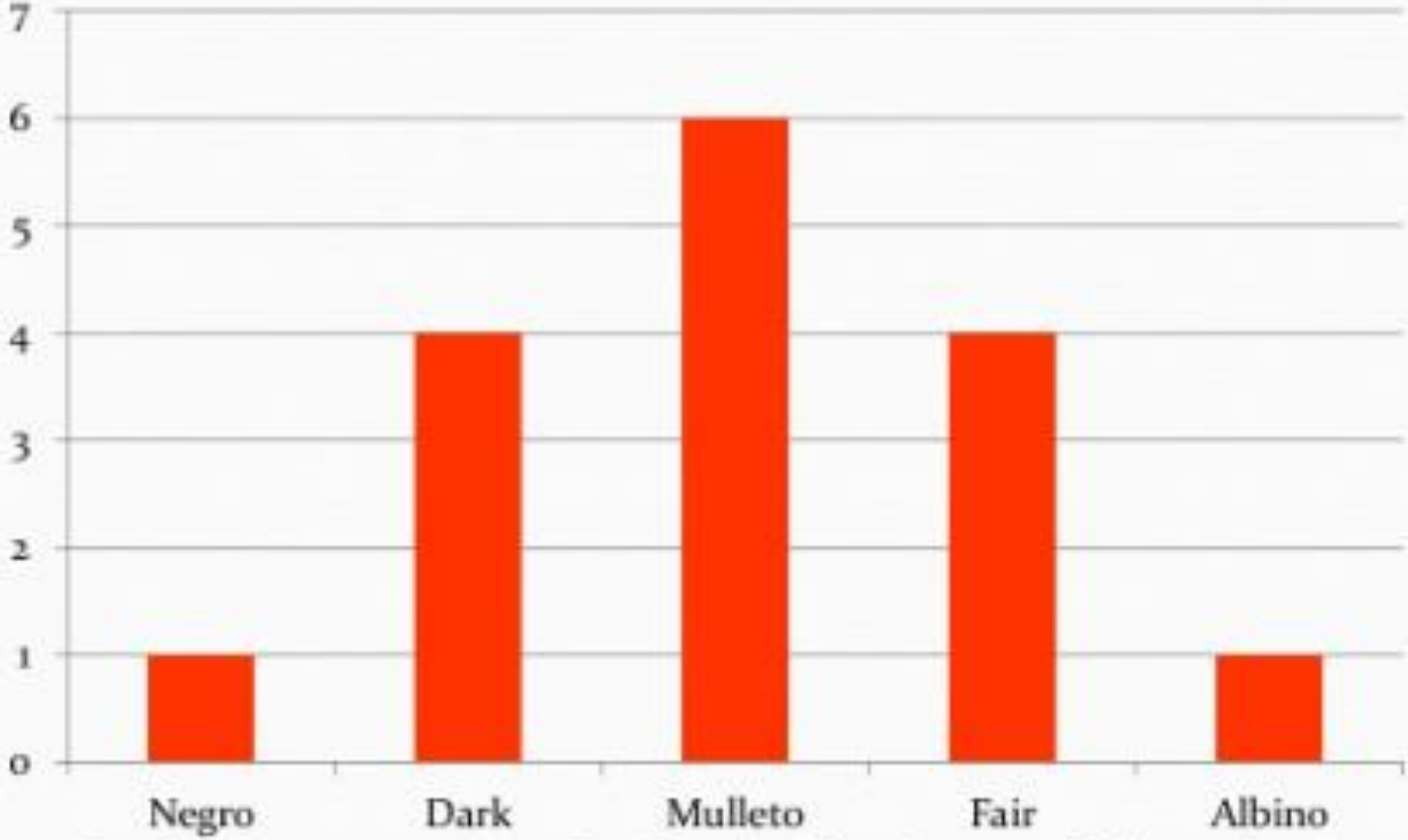
F2-

Gametes- AB Ab aB ab

♀ \ ♂	AB	Ab	aB	ab
AB	AA BB (Negro)	AA Bb (Dark)	Aa BB (Dark)	Aa Bb (Mulleto)
Ab	AA Bb (Dark)	Aa bb (Mulleto)	Aa Bb (Mulleto)	Aa bb (Fair)
aB	Aa BB (Dark)	Aa Bb (Mulleto)	aa BB (Mulleto)	aa Bb (Fair)
ab	Aa Bb (Mulleto)	Aa bb (Fair)	aa Bb (Fair)	aa bb (Albino)

F2 Ratio- 1:4:6:4:1

Number of Dominant allele	Phenotype	Ratio
No of dominant alleles	Albino	1/16
One dominant alleles	Fair	4/16
Two dominant alleles	Mulleto	6/16
Three dominant alleles	Dark	4/16
Four dominant alleles	Negro	1/16



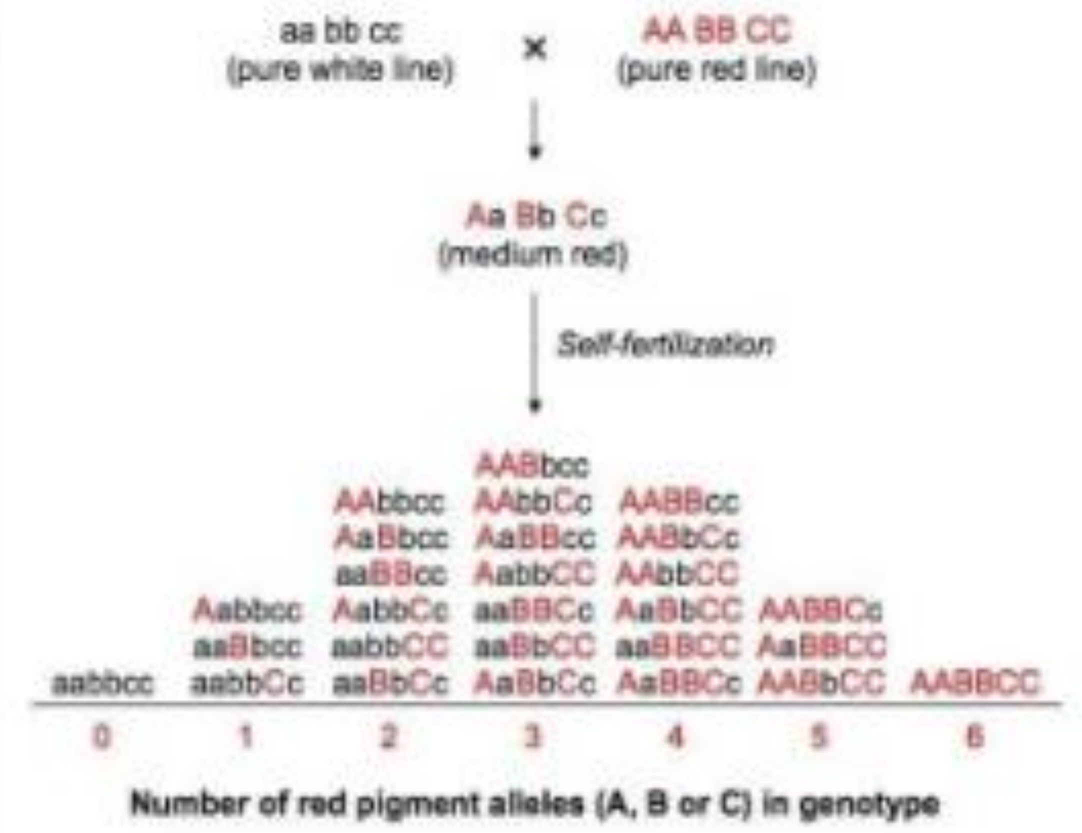
Graphical representation of polygenic inheritance of skin colour in human

Wheat kernel colour

with three genes

F1 -

F2-



Thank you