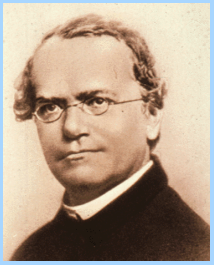
Mendelian Genetics

Augustinian Monk at Brno Monastery in Austria (now Czech Republic)

Not a great teacher but well trained in math, statistics, probability, physics, and interested in plants and heredity.



Gregor Mendel
"Father of Genetics"

While assigned to teach, he was also assigned to tend the gardens and grow vegetables for the monks to eat.

Mountains with short, cool growing season meant pea (***Pisum sativum***) was an ideal crop plant.

Contributions in 1860s (US Civil War Era)

- Discovered Genes as Particles of Inheritance
- Discovered Patterns of Inheritance
- Discovered Genes Come from **Both** Parents
  - Egg + Sperm = Zygote
  - Nature vs Nature
  - Sperm means Seed (Homunculu

http://forensicservices.utah.gov/biology/pictures/sperm.jpg

- Discovered One Form of Gene (Allele) Dominant to Another
- Discovered Recessive Allele Expressed in Absence of Dominant Allele

Mendel worked with peas (***Pisum sativum***)

- Good choice for environment of monastery
- Network provided unusual varieties for testing
- Obligate self-pollination reproductive system
  - Permits side-by-side genetic barriers
  - Cross-pollinations require intentional process
- Crosses meticulously documented
- Crosses numerically/statistically analyzed
- Scientists of 1860s could not understand math
- Work lost in journals for 50 years!
- Rediscovered in 1900s independently by 3 scientists
- Recognized as landmark work!

One Example of Mendel's Work

P Tall (DD, Homozygous Dominant) x Dwarf (dd, Homozygous Recessive) — Phenotype / Genotype

F1 All Tall, Dd, Heterozygous

Clearly Tall is Inherited… What happened to Dwarf?
1. Tall is dominant to Dwarf
2. Use D/d rather than T/t for symbolic logic

F2 F1 x F1 = F2

Punnett Square: possible gametes D, d

possible gametes	D	d
D	Tall DD	Tall Dd
d	Tall Dd	Dwarf dd

$^3/_4$ Tall
$^1/_4$ Dwarf

Dwarf is not missing…just masked as "recessive" in a diploid state… there IS a female contribution.

Mendel as a Scientist

Test Cross:
Unknown Tall x Dwarf (dd)

If Unknown is DD:

possible gametes	d	d
D	Tall Dd	Tall Dd
D	Tall Dd	Tall Dd

Test Progeny All Tall

If Unknown is Dd:

possible gametes	d	d
D	Tall Dd	Tall Dd
d	Dwarf dd	Dwarf dd

Test Progeny Half Tall Half Dwarf

$^1/_3$ of F2 Tall are DD
$^2/_3$ of F2 Tall are Dd

F2 F1 x F1 = F2

Punnett Square: possible gametes D, d

possible gametes	D	d
D	Tall DD	Tall Dd
d	Tall Dd	Dwarf dd

Another Example of Mendel's Work

P Green (gg, Homozygous Recessive) x Yellow (GG, Homozygous Dominant) — Phenotype / Genotype

F1 All Yellow, Gg, Heterozygous

Clearly Yellow is Inherited… What happened to Green?
1. Yellow is dominant to Green
2. Use G/g rather than Y/y for symbolic logic

NEVER use G/Y or g/y

F2 F1 x F1 = F2

Punnett Square: possible gametes G, g

possible gametes	G	g
G	Yellow GG	Yellow Gg
g	Yellow Gg	Green gg

$^3/_4$ Yellow
$^1/_4$ Green

Green is not missing…just masked as "recessive" in diploid state

Mendel as a Scientist

Test Cross:
Unknown Yellow x Green
gg

If Unknown is GG:

possible gametes	g	g
G	Yellow Gg	Yellow Gg
G	Yellow Gg	Yellow Gg

Test Progeny All Yellow

If Unknown is Gg:

possible gametes	g	g
G	Yellow Gg	Yellow Gg
g	Green gg	Green gg

Test Progeny Half Yellow Half Green

F2 F1 x F1 = F2

Punnett Square: possible gametes G g

possible gametes	G	g
G	Yellow GG	Yellow Gg
g	Yellow Gg	Green gg

$^1/_3$ of F2 Yellow are GG
$^2/_3$ of F2 Yellow are Gg

Mendel as a Scientist

Test Cross:
Unknown Yellow x Green
gg

If Unknown is GG:

possible gametes	g	g
G	Yellow Gg	Yellow Gg
G	Yellow Gg	Yellow Gg

If Unknown is Gg:

possible gametes	g	g
G	Yellow Gg	Yellow Gg
g	Green gg	Green gg

Actual Results	Decision
3 Yellow 2 Green	Gg
2 Yellow 3 Green	Gg
1 Yellow 4 Green	Gg
0 Yellow 5 Green	Gg
4 Yellow 1 Green	Gg
5 Yellow 0 Green	GG

Small families do not follow expected ratios perfectly!
Rare, but it can happen!
It only takes 1 green to be sure the unknown is Gg!
<5% chance unknown is Gg
$^1/_2 \cdot {}^1/_2 \cdot {}^1/_2 \cdot {}^1/_2 \cdot {}^1/_2 = {}^1/_{32}$
You could be wrong (rarely)!

Genetics After Mendel

After 1900 several scientists tried to replicate Mendel's crosses using other species including snapdragon.

P Red P^RP^R x Yellow P^YP^Y

F1 All Orange P^RP^Y

When these alleles go walking, they both do some talking (codominance)!
OK, so we cannot use R/r nor Y/y so we pick a third letter…P for the petal color gene.
Notice: we do NOT mix R/Y or r/y!

F1 x F1 = F2

F2 Punnett Square: possible gametes P^R P^Y

possible gametes	P^R	P^Y
P^R	Red P^RP^R	Orange P^RP^Y
P^Y	Orange P^RP^Y	Yellow P^YP^Y

This F2 will NOT have a 3:1 ratio of phenotypes.
Instead it shows a 1:2:1 ratio!
The exception here proves the rule.

In addition to this, there are multiple alleles possible:
P^R = red P^Y = yellow p = no pigment

The combination of alleles in a diploid determine the flower color:
P^RP^R = red P^Rp = pink
P^RP^Y = orange P^Yp = cream
P^YP^Y = yellow pp = white

Human hair color follows a similar pattern:
Alleles: H^{Bn} = brown H^{Bd} = blonde h^R = red h^{bk} = black

The combinations of these alleles determine the base hair color:
$H^{Bn}H^{Bn}$ = dark brown $H^{Bd}H^{Bd}$ = blonde h^Rh^R = red
$H^{Bn}H^{Bd}$ = sandy brown $H^{Bd}h^R$ = strawberry blonde h^Rh^{bk} = red
$H^{Bn}h^R$ = auburn
$H^{Bn}h^{bk}$ = dark brown $H^{Bd}h^{bk}$ = blonde

Dominant does NOT mean frequent!

$h^{bk}h^{bk}$ = black
Recessive can be common!

Another Example of Recessive Being Common: ***Pisum sativum***

Garden Peas: green seed, wrinkled seed, dwarf stature, white flower

gg ww dd aa

In other words: a quadruple double-recessive is the most common garden pea on Earth!

Quantitative Inheritance: multiple genes control trait

Highest Crop Yield: AABBCCDDEE
Intermediate Crop Yield: AabbCCDdEe
Lowest Crop Yield: aabbccddee

Darkest Skin Color: AABBCCDDEE
Intermediate Skin Color: AaBbCcDdEe
Lightest Skin Color: aabbccddee

AaBbCcDdEe x AaBbCcDdEe can produce a huge range of colors! Yet TV talk show guests argue this point for Maury, etc.

Phenotype = Genotype + Environment

Crop Yield = Genotype
+ Minerals
+ Water
+ Light
- Pests
etc.

Optimizing these factors determines agricultural productivity…last part of our course!

Human Skin Color = Genotype
+ Sun (UV) Exposure
- Aging Factors

The sun exposure effect is most obvious in people of intermediate skin base color but everyone can have "tan lines."

Who Gets To Mate With Whom? …Two Extremes

- Inbreeding Depression: related parents give same recessives to children
  - Hemophilia: Queen Victoria's Mutation and Diseased Grandchildren
  - Tay-Sachs: Jewish Populations
  - Bipolar: Irish Populations
  - Dog Diseases: German Shepherd hip dysplasia

Incest Taboo…but

- Hybrid Vigor: recessives of one family are "covered" by dominant of other family

Wild Corn A x Wild Corn B

High Yield Hybrid Corn!

"Half-breed" is better

Are Human Cultures of "Great Melting Pots" Superior?

"Mutt" is best dog!

Mechanisms Preventing Inbreeding in Plants

- Evolution of Unisexuality: **Dioecious** Holly and Kiwi
  Common Gardener Error at Nursery Pickup
- Timing Separation of Bisexuality
  - Protandry: Male First, Then Female (***Alstromeria, Campanula***)
  - Protogyny: Female First (***Oenothera***)
  - Sequence in ***Cucumis***: Male, Bisexual, Female, Parthenocarpic
- Self-Incompatibility: Sweet Cherry, Almond, Filbert
  - pollen germination prevented
  - pollen tube growth halted
  - synergid action (sperm release) prevented
  - sperm deactivated

Going Outside the Box of a Species: Bread Wheat

AA (diploid wheat) x DD (diploid grass)
Triticum urartu ***Aegilops tauschii***

AD (sterile diploid) → colchicine or spontaneous → AADD (fertile tetraploid)
similar to mule!

BB (diploid grass)
Aegilops speltoides

BAD (sterile triploid) → colchicine or spontaneous → BBAADD
Fertile hexaploid
Bread Wheat
Triticum aestivum

Bread Wheat Created 7500 BC with spontaneous doubling of DNA

Levy A. A. and M. Feldman. 2002. The impact of polyploidy on grass genome evolution. Plant Physiol. 130: 1587-1593.

Yet Another Example of Mendel's Work

P Wrinkled x Round — Phenotype
ww — WW — Genotype
Homozygous Recessive — Homozygous Dominant

F1 All Round
Ww
Heterozygous

1. Round is dominant to Wrinkled
2. Use W/w rather than R/r for symbolic logic: in handwriting make it legible! W ω

NEVER use W/R or w/r

F1 x F1 = F2

Punnett Square:

possible gametes	W	w
W	Round WW	Round Ww
w	Round Ww	Wrinkled ww

F2

$^3/_4$ Round
$^1/_4$ Wrinkled

Mendel as a Scientist

Test Cross:
Unknown Round x Wrinkled
ww

If Unknown is WW:

possible gametes	w	w
W	Round Ww	Round Ww
W	Round Ww	Round Ww

Test Progeny All Round

If Unknown is Ww:

possible gametes	w	w
W	Round Ww	Round Ww
w	Wrinkled ww	Wrinkled ww

Test Progeny Half Round Half Wrinkled

F2 F1 x F1 = F2

Punnett Square:

possible gametes	W	w
W	Round WW	Round Ww
w	Round Ww	Wrinkled ww

Mendel as a Scientist

Test Cross:
Unknown Round x Wrinkled
ww

If Unknown is WW:

possible gametes	w	w
W	Round Ww	Round Ww
W	Round Ww	Round Ww

If Unknown is Ww:

possible gametes	w	w
W	Round Ww	Round Ww
w	Wrinkled ww	Wrinkled ww

Actual Results	Decision
3 Round 2 Wrinkled	Ww
2 Round 3 Wrinkled	Ww
1 Round 4 Wrinkled	Ww
	Small families do not follow expected ratios perfectly!
0 Round 5 Wrinkled	Ww
	Rare, but it can happen!
4 Round 1 Wrinkled	Ww
	It only takes 1 wrinkled to be sure the unknown is Ww!
5 Round 0 Wrinkled	WW
	<5% chance unknown is Ww

$^1/_2 \cdot {}^1/_2 \cdot {}^1/_2 \cdot {}^1/_2 \cdot {}^1/_2 = {}^1/_{32}$

You could be wrong (rarely)!