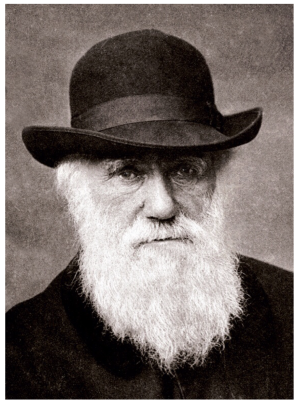
Mendelism

Evolution and the mechanism of heredity

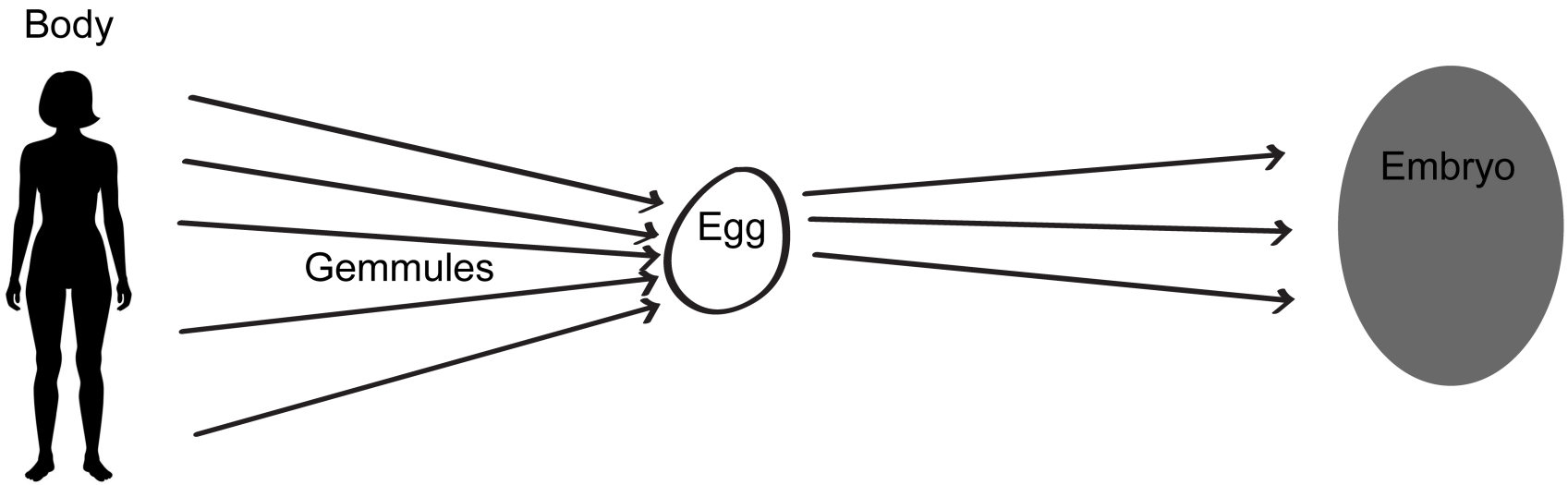


Charles Darwin and the nature of variation:
Where does the feedstock for selection come from?

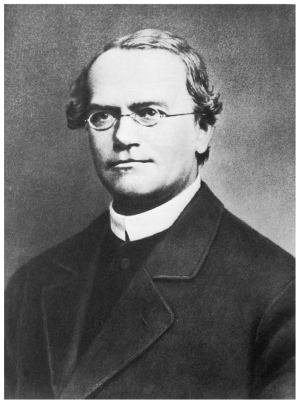
Two types of variation:

- Continuous variation
- Discontinuous variation

Pangenesis (adapted from Hippocrates)



“Experiments on Plant Hybridization” 1866



	Flower Colour	Plant Height	Seed Color	Seed Shape	Pod Colour	Pod Shape	Flower Position
Dominant Trait	Purple	Tall	Yellow	Round	Green	Inflated (full)	Axial
Recessive Trait	White	Short	Green	Wrinkled	Yellow	Constricted (flat)	Terminal

(Bioninja.com)

Summary of Mendel’s findings:

- Uniformity of F1 (dominance)
- 3:1 ratio of dominant: recessive
  - 1/4 purebred dominant, 1/4 purebred recessive, 2/4 hybrid
  - hybrid x hybrid crosses give 3:1, recessive x hybrid give 1:1

Law of Segregation

- Two different traits will segregate independently

Law of Independent Assortment

Mendel-Fisher Controversy

Chi-squared test (1900)

$$\sum \frac{(\text{Observed} - \text{Expected})^2}{\text{Expected}} = \chi^2$$

Degrees of freedom (df) = categories -1

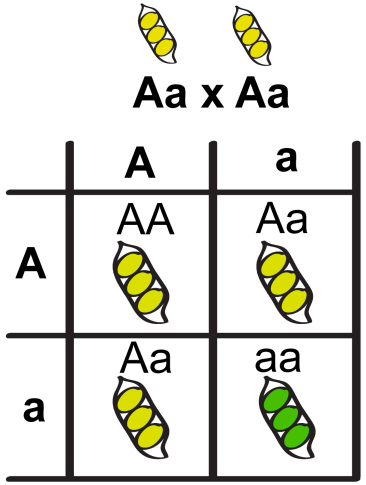


TABLE 1. F_2 RESULTS, PEA CROSSES

Source	Yellow	Green	Total	Dev. from 3 in 4	Prob. Error	Dev. ÷ P.E.
Mendel, 1866	6,022	2,001	8,023	+ .0024	± .0130	.18
Correns, 1900	1,394	453	1,847	+ .0189	± .0272	.70
Tschermak, 1900	3,580	1,190	4,770	+ .0021	± .0169	.12
Hurst, 1904	1,310	445	1,775	– .0142	± .0279	.51
Bateson, 1905	11,902	3,903	15,806	+ .0123	± .0093	1.32
Lock, 1905	1,438	514	1,952	– .0533	± .0264	2.04
Darbishire, 1909	109,060	36,186	145,246	+ .0035	± .0030	1.16
Winge, 1924	19,195	6,553	25,748	– .0180	± .0125	1.44
Total	153,902	51,245	205,147	+ .0008	± .0038	.21

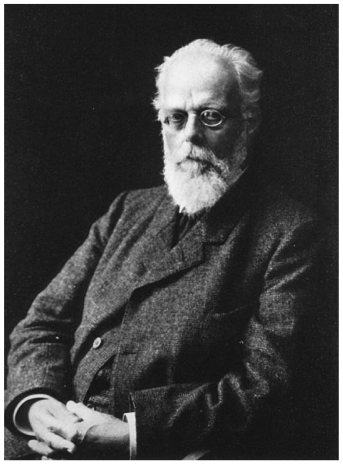
SOURCE: Johannsen, 1926.

Mendel-Fisher Controversy

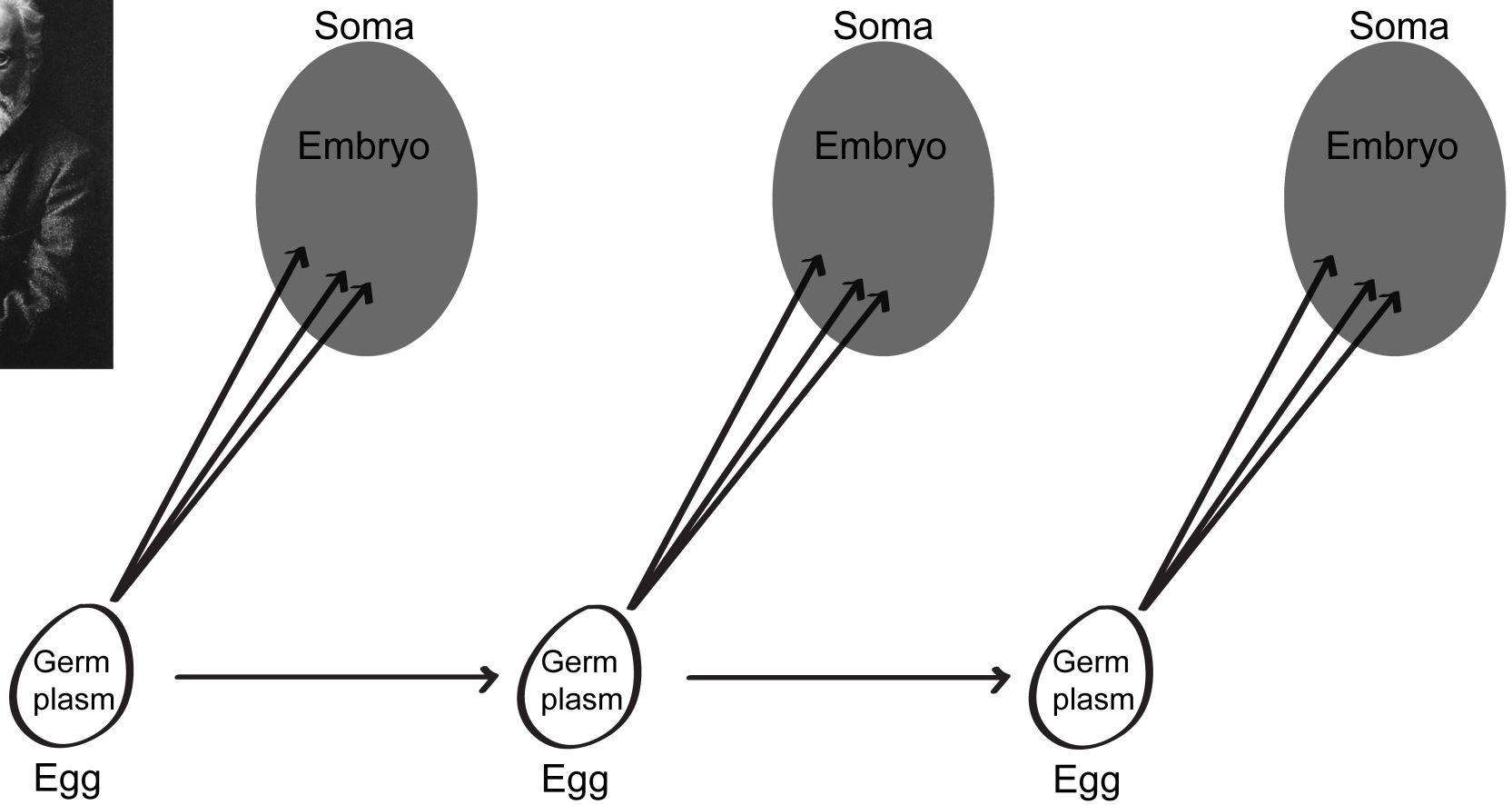
	Trait	"A"	"a"	n	Obs. freq. $n_{\text{"A"}}$	Obs. freq. $n_{\text{"a"}}$	Theor. ratio "A" : "a"
	Seed shape	round	wrinkled	7324	5474	1850	3 : 1
	Seed color	yellow	green	8023	6022	2001	3 : 1
	Flower color	purple	white	929	705	224	3 : 1
F_2	Pod shape	inflated	constricted	1181	882	299	3 : 1
	Pod color	yellow	green	580	428	152	3 : 1
	Flower position	axial	terminal	858	651	207	3 : 1
	Stem length	long	short	1064	787	277	3 : 1
	Trait	A	a	n	n_{Aa}	n_{AA}	$Aa : AA$
	Seed shape	round	wrinkled	565	372	193	2 : 1
	Seed color	yellow	green	519	353	166	2 : 1
	Flower color	purple	white	100	64	36	2 : 1
(F_3)	Pod shape	inflated	constricted	100	71	29	2 : 1
	Pod color	yellow	green	100	60	40	2 : 1
	Flower position	axial	terminal	100	67	33	2 : 1
	Stem length	long	short	100	72	28	2 : 1
	Pod color (rep.)	yellow	green	100	65	35	2 : 1

Germ plasm theory of heredity

August Weissman



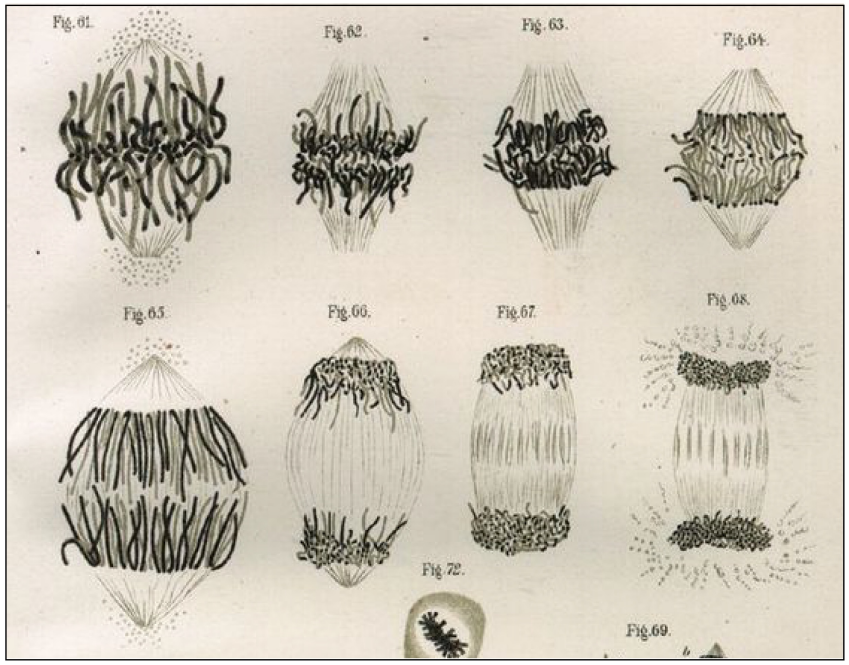
Germ plasm theory (1883-1889)



Chromosome theory

Technological Advances:

- Microtome
- Dyes
- Organisms for study:
  - Ascaris
  - Salamander

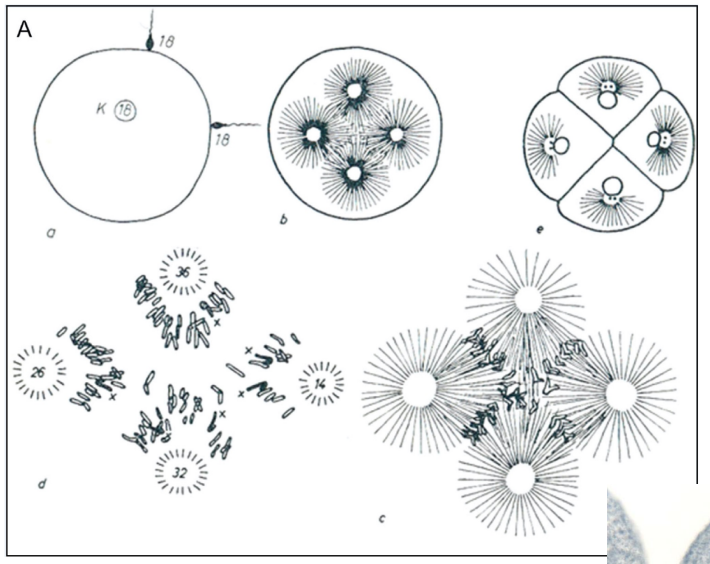


Chromosomes and
mitotic phases (Flemming 1882)

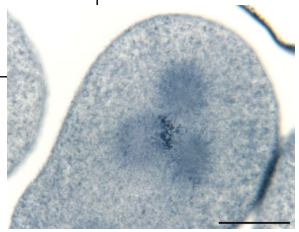
Theoretical Advances:

1883 Roux argues structure of chromosomes and their divisions argue them as bearers of heredity

- Chromosomes occur in pairs (1901-2)
  - Fertilized egg receives equal chromosomes from each parent
  - Meiotic divisions halve chromosome number
- Boveri polyspermy experiments (1902):



From Baltzer 1962

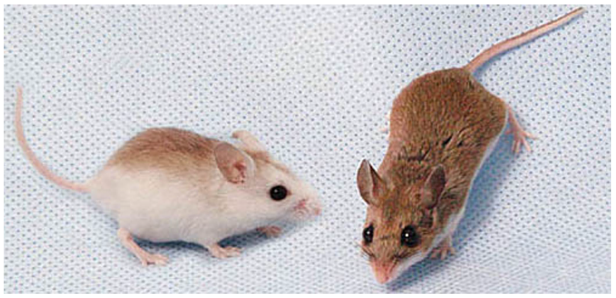


Rediscovery of Mendel ~1900

- Correns: Maize
- De Vries : several plants “This memoir, very beautiful for its time, has been misunderstood and then forgotten.”
- Tshermak: Peas

Two genes, parallel redundant pathways

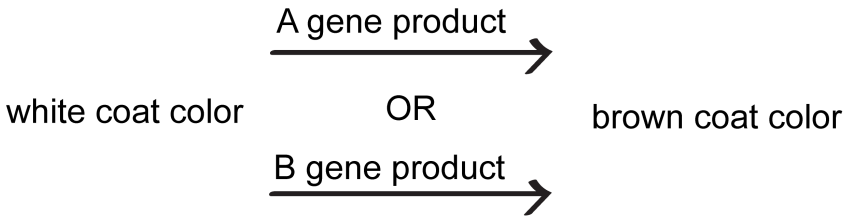
Mus musculus



AaBb x AaBb

	AB	**Ab**	**aB**	**ab**
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Two genes, redundant parallel pathways = 15 : 1 phenotypic ratio



Two genes, one pathway

Sweet pea (*Lathyrus odoratus*)

AaBb x AaBb

	AB	**Ab**	**aB**	**ab**
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Two genes, one pathway = 9 : 7 phenotypic ratio

white flower color → (A gene product) → (B gene product) → pink flower color

Bateson and Punnett

Mendelian inheritance in humans

Blood type

♂

♀	I^A	I^B	I^O
I^A	I^AI^A A	I^AI^B AB	I^AI^O A
I^B	I^AI^B AB	I^BI^B B	I^BI^O B
I^O	I^AI^O A	I^BI^O B	I^OI^O O

Huntington's disease

♂

♀	h	h
H	Hh disease	Hh disease
h	hh	hh

Genetics terminology needed

- Gene
- Genotype/Phenotype
- Allelomorph (allele)
- Homozygote/Heterozygote

Other non-Mendelian ratios

Sweet pea (*Lathyrus odoratus*)

P AABB x aabb

F1 AaBb x AaBb

F2

	expected	observed
	1199	1528
	400	106
	400	117
	113	381
totals	2132	2132

Bateson, Saunders, and Punnett 1905