## Gene interactions: Epistasis

<u>Physiological interactions</u>; biochemical pathways alter phenotypic expression.

<u>Morphological interactions</u>; the phenotype of one character prevents us from viewing the phenotype of another character.

Typical Mendelian ratios are obscured, but Mendel's postulates are still the underlying rules.

Sutton (1902) noted that there were more genes than chromosomes. Thus some genes on the same chromosome should not be transmitted independently of each other. Morgan and co-workers (1910 and earlier) proved genes were on chromosomes Progression of thought: \*All genes are independent (particles). Concept: Mendel's Law of Independent Assortment

\*Chromosomes independently assort but genes do not. Concept: Chromosome Theory of Inheritance

Gene interactions: Linkage - Physical Interactions

\*Genes which lie at specific positions on chromosomes (locus) are more independent as the distance between a pair of loci is increased. Concept: Linkage and Recombination



T. H. Morgan and students used Drosophila to demonstrate that **Mendel's genes were on chromosomes** and to show that "sex" was a genetic trait.









Are there X-linked genes in humans?

Yes!

Who expresses these genes most often?

Males!

Feel bad for males day!!















## OK, back to history.

It was pretty evident that genes were on chromosomes and Sutton made that astute observation:

## # chromosomes <<< # genes;</pre>

So why did everyone always see Mendel's Independent Assortment???

They didn't!!



Punnet and Batson 1906 Discover Linkage in Peas									
Purple vs. Red Flowers Long vs. Round Pollen									
Parent 1Parent 2 $\rightarrow$ F1Red, LongxWhite, RoundRed, LongRR LLxrr IIRr LI									
F2 $3/4 \text{ L}_{-} \longrightarrow 9/16 \text{ Red Long}$ $3/4 \text{ R}_{-} \longrightarrow 1/4 \text{ II} \longrightarrow 3/16 \text{ Red Round}$									
$1/4 \text{ rr} \longrightarrow 3/4 \text{ L} \longrightarrow 3/16 \text{ White Long}$ $1/4 \text{ rr} \longrightarrow 1/4 \text{ II} \longrightarrow 1/16 \text{ White Round}$									

Batesor	and Punnett 1	906 discover lin	kage in swee	et peas
<ul> <li>Red vs wh</li> </ul>	nite flower, Long	vs. round pollen		
<ul> <li>Parentals</li> </ul>	RRLL	х	rrll	
• F1		RrLI		
• <u>F2</u>	Observed	Expe	cted	
<ul> <li>R_L_</li> </ul>	583	9/16 * 803 =	451.7	
• R_II	26	3/16 * 803 =	150.6	
<ul> <li>rr L_</li> </ul>	24	3/16 * 803 =	150.6	
• rr ll	170	1/16 * 803 =	50.2	
•	803		803	
• X2 = <u>(5</u>	<u> 83 – 451.7)</u> ² + <u>(2</u>	<u>6 – 150.6)</u> ² + <u>(24 -</u>	<u>-150.6)</u> <sup>2</sup> + (170	<u>– 50.2)</u> <sup>2</sup> =533.6
•	451.7 <sup>·</sup>	150.6 1	50.6	50.2
• d.f. =3	P<<0.001			
<ul> <li>Reject the</li> </ul>	Null Hypothesis	Whati	s the null	
		hypot	nesis??	

p										
df	0.995	0.975	0.9	0.5	0.1	0.05	0.025	0.01	0.005	df
1	.000	.000	0.016	0.455	2.706	3.841	5.024	6.635	7.879	
2	0.010	0.051	0.211	1.386	4.605	5.991	7.378	9.210	10.597	
(3)	0.072	0.216	0.584	2.366	6.251	7.815	9.348	11.345	12.838	
4	0.207	0.484	1.064	3.357	7.779	9.488	11.143	13.277	14.860	
5	0.412	0.831	1.610	4.351	9.236	11.070	12.832	15.086	16.750	:
6	0.676	1.237	2.204	5.348	10.645	12.592	14.449	16.812	18.548	.
7	0.989	1.690	2.833	6.346	12.017	14.067	16.013	18.475	20.278	
8	1.344	2.180	3.490	7.344	13.362	15.507	17.535	20.090	21.955	:
9	1.735	2.700	4.168	8.343	14.684	16.919	19.023	21.666	23.589	
10	2.156	3.247	4.865	9.342	15.987	18.307	20,483	23.209	25.188	1
11	2.603	3.816	5.578	10.341	17.275	19.675	21.920	24.725	26.757	1
12	3.074	4.404	6.304	11.340	18.549	21.026	23.337	26.217	28.300	1
13	3.565	5.009	7.042	12.340	19.812	22.362	24.736	27.688	29.819	1
14	4.075	5.629	7.790	13.339	21.064	23.685	26.119	29.141	31.319	14
15	4.601	6.262	8.547	14.339	22.307	24.996	27.488	30.578	32.801	1:



Bateson and Punnett 1906 discover linkage in sweet peas Red vs white flower, Long vs. round pollen RRLL Х rrll Parentals F1 RrLI Observed Expected F2 Independence 9:3:3:1 Complete Linkage RL 583 9/16 \* 803 = 452 3/4 \* 803 = 602 RII 26 3/16 \* 803 = 151 = 0 rr L\_ 24 0 3/16 \* 803 = 151 = rr II 170 1/16 \* 803 = 501/4 \* 803 = 201803 You can see that what Bateson and Punnett found was somewhere in between independent assortment and complete linkage!! How do you quantify this?















