

For a complex disease, no single site will be 100% correlated with presence/absence of the trait

- Each complex disease or trait is associated with multiple DNA sites
- Each site increases/decreases risk of having the disease/trait
- Environment also can have large effects

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Examples of traits with lots of both kinds of influences

- Diabetes
- Heart disease
- High cholesterol
- Schizophrenia
- Bipolar Disease
- Autism
- Height

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Influences on common complex human traits

- Diabetes: diet & exercise, genetic pre-disposition
- Schizophrenia: marijuana use, genetic pre-disposition
- Height: nutrition, genetic pre-disposition
- Malaria: exposure to parasite, genetic resistance

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Refresher: Twin studies yield concordance rates

- A concordance rate is the probability that the other twin will display a trait, if one does
- To determine types of influences on a trait, we:
 - Look at MZ twin rates
 - Compare rates of MZ twins and DZ twins



Figure 18.1
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How to interpret concordance rates

- If both MZ and DZ twins have similarly high rates...
- If MZ twins have higher rates than DZ twins...
- If MZ and DZ twins have similarly low rates...

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Actual concordance rates for common complex traits

Trait	Concordance (%)	
	MZ twins	DZ Twins
Heart Attack (males)	39	26
Heart Attack (females)	44	14
Bronchial Asthma	47	24
Cancer (All sites)	12	15
Schizophrenia	52	15
Rheumatoid Arthritis	32	6
Multiple sclerosis	28	5

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Actual concordance rates for common complex traits

Trait	Concordance (%)	
	MZ twins	DZ Twins
Schizophrenia	52	15
Autism	64	9
Tourette's Syndrome	53	8
Epilepsy	59	19
Rheumatoid Arthritis	32	6
Multiple sclerosis	28	5
Juvenile Delinquency	91	73
Cancer (All sites)	12	15
Heart Attack (males)	39	26
Heart Attack (females)	44	14
Bronchial Asthma	47	24

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Basic facts about schizophrenia

- Prevalence is 1 in ~100-200 world-wide
- Diagnosis is based on meeting a certain number of criteria from a list published in the DSM (Diagnostic and Statistical Manual of Mental Disorders)
- Symptoms (such as hallucinations, delusions, and thought disorders) usually start between 16-30yrs old

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Results about mutations that are correlated with schizophrenia

- Patients have more deletions/duplications than controls
- Each patient has dozens of these mutations
- Each patient's set of mutations is different (100s total)
- Some are associated with multiple disorders
- Many are spontaneous (new in that person)