Psych 3102
Introduction to Behavior genetics

Lecture 11

Methodology continued

Human studies

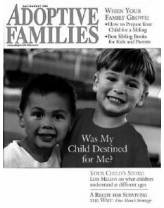
Problems with human studies

- no direct breeding studies possible – methods are therefore not as powerful or as direct as animal studies
- genetically defined populations not available
- environment cannot be controlled

Family studies

provide data on similarities between relatives but genetic and environmental influences are confounded

- separated by addition of twin and adoption data



Family studies
with twins and adoptions

Components of variance and covariance

RELATIONSHIP	(resemblances) Covariance source	(differences) Variance source
Biological *parent/offspring* *sib/sib* *MZ twin pairs*	shared genes + shared environment	segregating genes + non-shared env. non-shared env only
Adoptive *p/adopted child* *sib/adopted sib*	shared environment only	segregating genes + non-shared env.
Adopted away *p/adopted away child* *sib/adopted away sib*	shared genes only	segregating genes + non-shared env.

- we can get some estimates of genetic and environmental variance components

Model-fitting

- using variance and covariance data from family, twin, adoption studies
- constructing an explanation in the form of a model that describes the observed data

1. models are constructed by hypothesizing that certain variables (eg additive genetic influence) are present at certain levels of influence
2. expected variance and covariance values are computed and compared to observed data
3. model with the fewest parameters that best fit the data is chosen

path analysis visual way of analyzing the model and discovering which variable parameters best explain the data

paths =

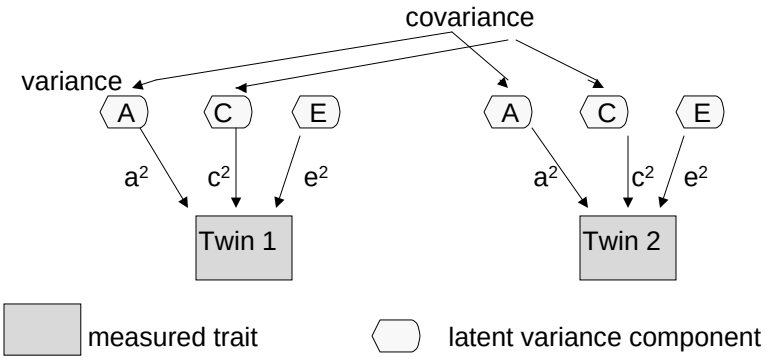
variables =

ACE model

most commonly used model in behavior genetics
A = additive genetic effects
C = common (shared) environment effects
E = non-shared (individual-specific) environmental effects

$$V_{(P)} = V_{G_A} + V_{E_C} + V_{E_E}$$

ACE path diagram for twin data



SUMMARY Sources of variation in human family studies

Genetic influences G
- A = additive genetic influences
  - a^2 = variance due to additive effects of genes
- D = dominance effects
  - d^2 = variance due to dominance effects of genes
- I = epistasis
  - i^2 = variance due to epistasis

Environmental influences
- E = non-shared environmental influences
  - e^2 = variance due to individual experiences of family members
- C = shared (common) environmental influences
  - c^2 = variance due to environmental differences between families

Problems with adoption studies

1. many fewer adoptions than in the past

current US adoption rate: 30,000 per year

1960: 1% of all babies born (around 200,000)

2. are adoptive parents and adopted children representative of general population?

Colorado Adoption Project (CAP)

3. may be unknown prenatal influences

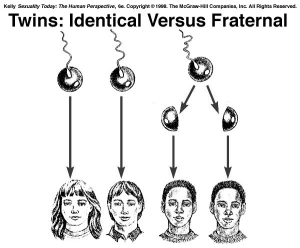
data on both biological parents needed as well as adoptive parents

4. selective placement

data on bio and adoptive parents needed to assess this

may inflate both genetic and environmental correlations

Twin Studies



monozygotic (MZ) twins identical

dizygotic (DZ) twins fraternal

MZ similarities >DZ similarities for any phenotype influenced by genes, assuming equal environments

Twin births = 1 in 85 live births (1 in 5 conceptions)

All twins
- 1/3 MZ
- 2/3 DZ
  - 1/3 same sex
  - 1/3 opposite sex

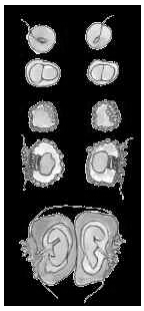
comparison of these is best

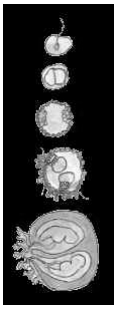
MZ twinning is independent of maternal age & fertility treatments - both of which increase DZ twinning

Types of MZ twins

Whilst all DZ twins have separate chorionic and amniotic sacs,

MZ twins may have one of 3 types of arrangements *in utero :*





1.Dichorionic MZs (DC-MZ) 32% of all MZ

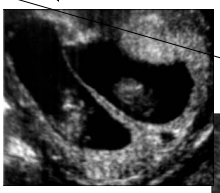
separate placentas, amnions, chorions

zygote splits before Day 4 after fertilization (before implantation)

2 .Monochorionic/diamniotic (MC-DA MZ) 66%

separate amnions but share the same chorionic sac and placenta

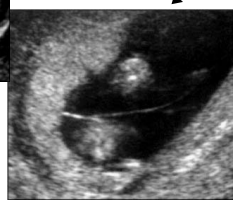
zygote splits between Day 4 and Day 7 (after implantation)



3. Monochorionic/monoamniotic (MC-MA 2-3% MZ)

share amnion, chorionic sac and placenta

zygote splits after Day 8



Effects of uterine environment:

- death rate
- birth weight
- sex ratio
- congenital deformities

- behavioral traits

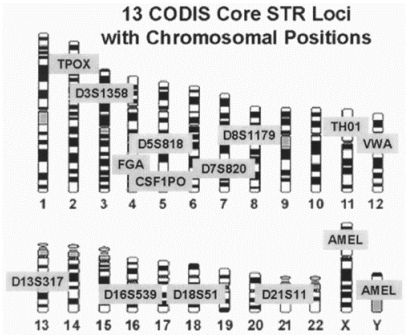
Jacobs et al (2001) Behavior Genetics Wechsler tests

Determination of zygosity

- visual appearance

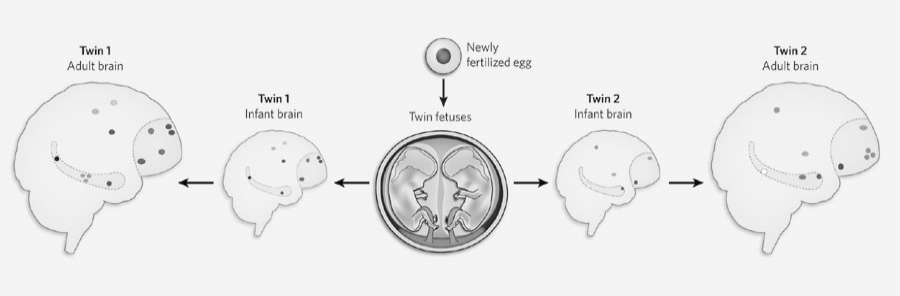
- use of DNA markers

CODIS panel
(COmbined DNA Index System)



Human brain variation by retrotransposon

from Coufal et al (2009), Nature 460)



Twins that are genetically identical at conception may later show brain cell genetic differences at birth because of new Line1 (retrotransposon) insertions that take place during the development of the nervous system in the fetus.

Ongoing retrotransposition in neural progenitor cells will further diversify the genetic makeup of their brains in adulthood. Depending on the target genes and neurons affected, the twins may differ in brain function or dysfunction

Each unique insertion is represented by a different color. Darker shaded areas highlight brain regions more likely to be affected after birth.

Assumptions underlying twin methodology

1. Equal environments assumption

Tests for this:

2. Twins are representative of the general population