

Path Analysis

Frühling Rijdsdijk

model
building

**Biometrical
Genetic
Theory**

Twin Model

**System of
Linear
Equations**

**Path
Diagrams**

**Covariance
Algebra**

**Path Tracing
Rules**

**Predicted Var/Cov
of the Model**

**Observed Var/Cov
of the Data**

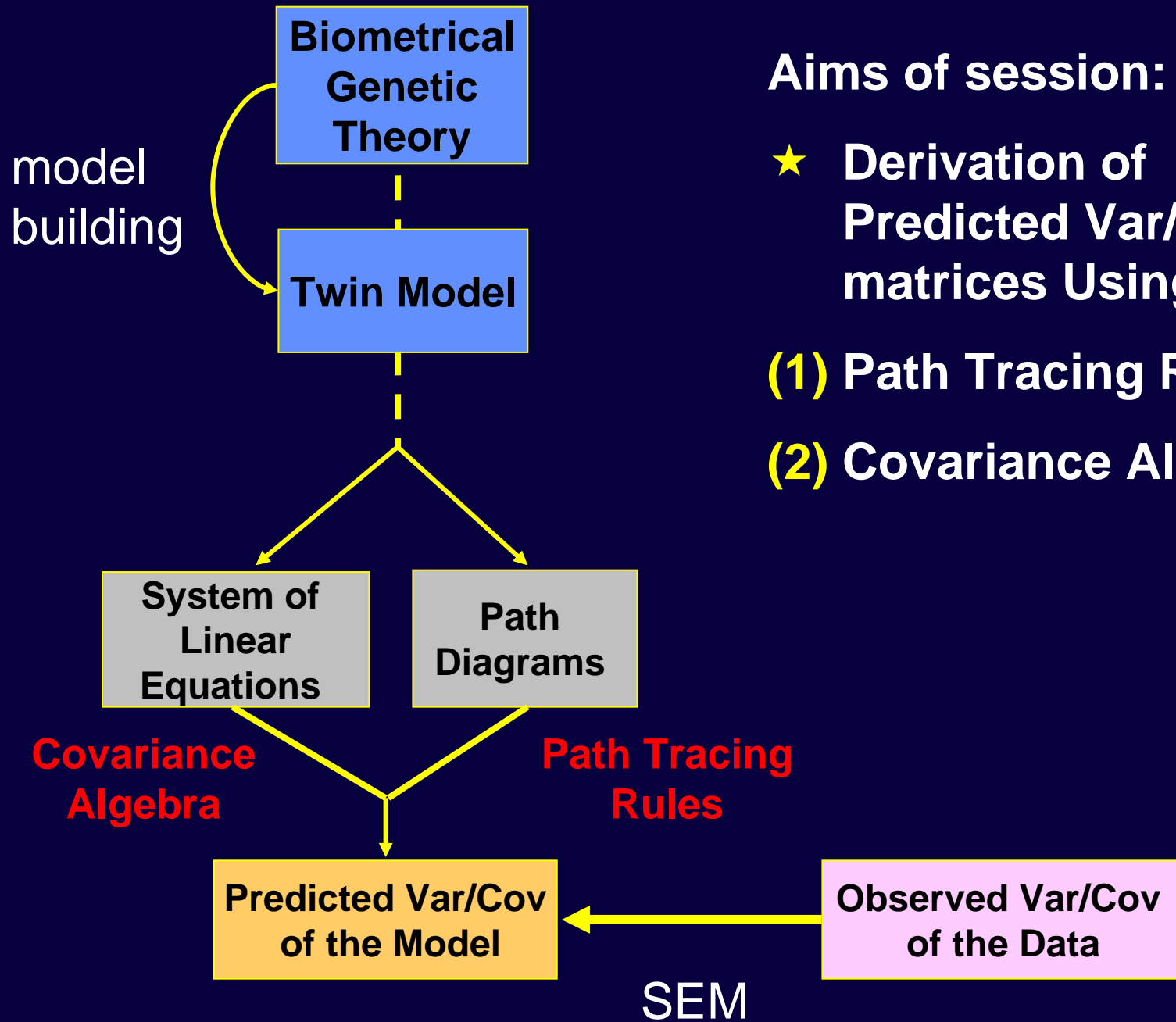
SEM

Aims of session:

★ **Derivation of
Predicted Var/Cov
matrices Using:**

(1) Path Tracing Rules

(2) Covariance Algebra

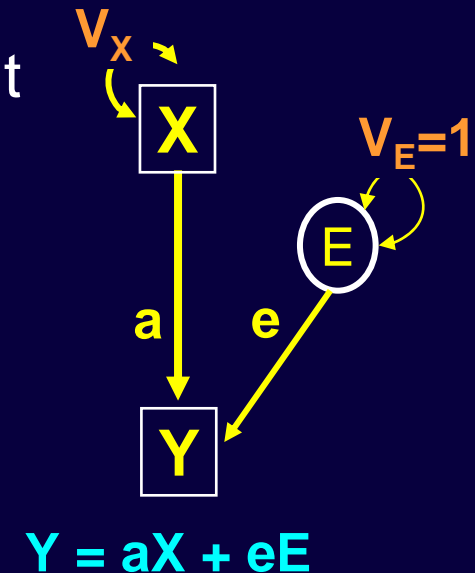


Method of Path Analysis

- Allows us to represent linear models for the relationship between variables in diagrammatic form, e.g. a genetic model; a factor model; a regression model
- Makes it easy to derive expectations for the variances and covariances of variables in terms of the parameters of the proposed linear model
- Permits easy translation into matrix formulation as used by programs such as Mx, OpenMx.

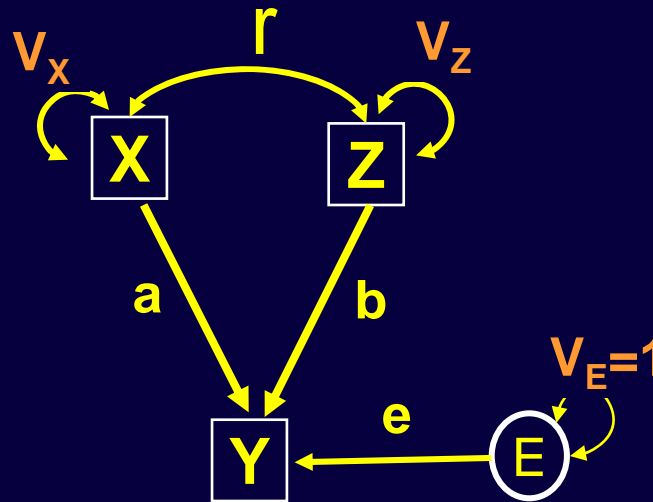
Conventions of Path Analysis I

- Squares or rectangles denote observed variables
- Circles or ellipses denote latent (unmeasured) variables
- Upper-case letters are used to denote variables
- Lower-case letters (or numeric values) are used to denote covariances or path coefficients
- Single-headed arrows or paths (\rightarrow) represent hypothesized **causal** relationships
 - where the variable at the tail is hypothesized to have a direct causal influence on the variable at the head



Conventions of Path Analysis II

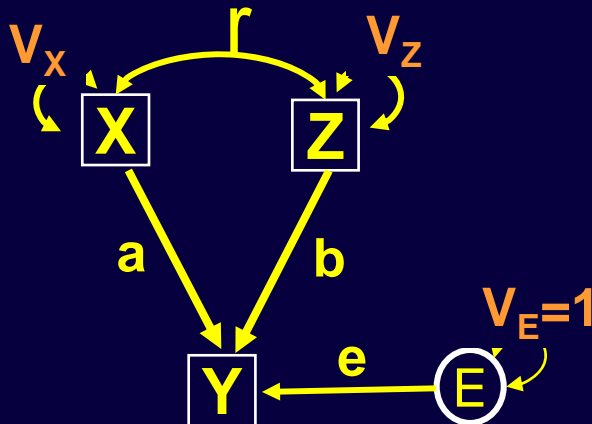
- Double-headed arrows (\leftrightarrow) are used to represent a covariance between two variables, which may arise through common causes not represented in the model.
- Double-headed arrows may also be used to represent the variance of a variable.



$$Y = aX + bZ + eE$$

Conventions of Path Analysis III

- Variables that do not receive causal input from any one variable in the diagram are referred to as **independent**, or **predictor** or **exogenous** variables.
- Variables that do, are referred to as **dependent** or **endogenous** variables.
- Only independent variables are connected by double-headed arrows.
- Single-headed arrows may be drawn from independent to dependent variables or from dependent variables to other dependent variables.



Conventions of Path Analysis IV

- Omission of a two-headed arrow between two independent variables implies the assumption that the covariance of those variables is zero
- Omission of a direct path from an independent (or dependent) variable to a dependent variable implies that there is no direct causal effect of the former on the latter variable

Path Tracing

The covariance between any two variables is the sum of all **legitimate chains** connecting the variables

The numerical value of a chain is the product of all traced path coefficients in it

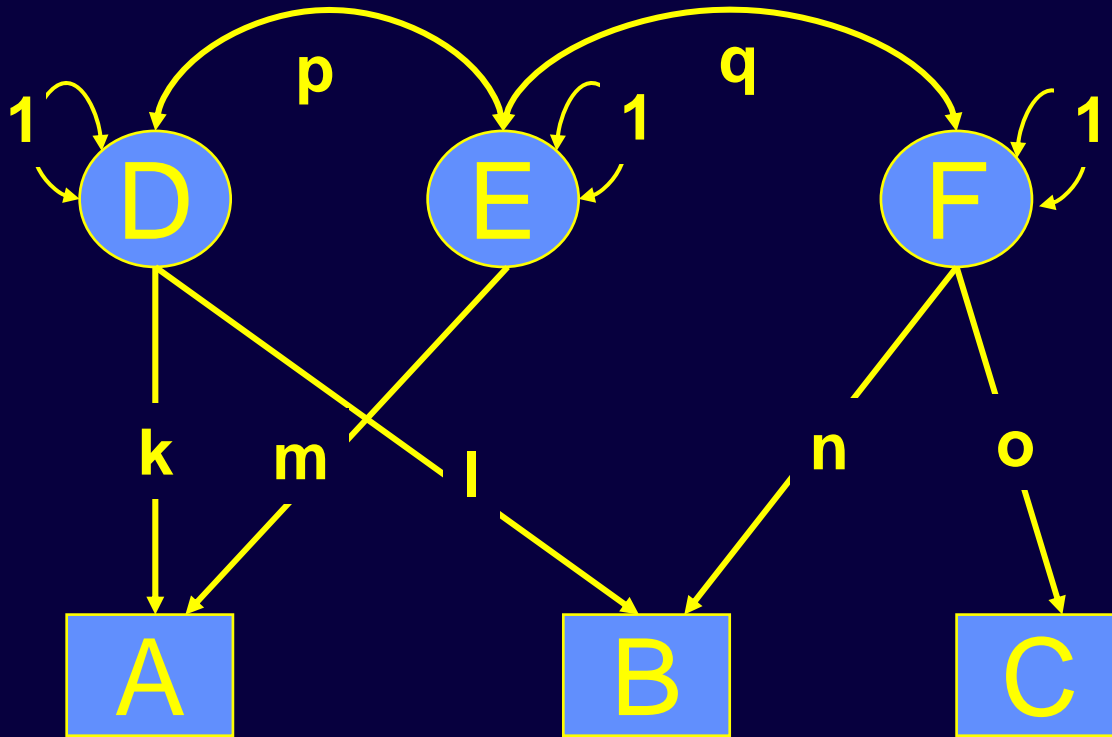
A **legitimate chain**

is a path along arrows that follow **3 rules**:

- (i) Trace backward, then forward, or simply forward from one variable to another.
NEVER forward then backward!
Include double-headed arrows from the independent variables to itself. These variances will be **1** for **latent variables**
- (ii) Loops are not allowed, i.e. we can not trace twice through the same variable
- (iii) There is a maximum of **one curved** arrow per path.
So, the double-headed arrow from the independent variable to itself is included, unless the chain includes another double-headed arrow (e.g. a correlation path)

The Variance

Since the variance of a variable is the covariance of the variable with itself, the expected variance will be the sum of all paths from the variable to itself, which follow the path tracing rules

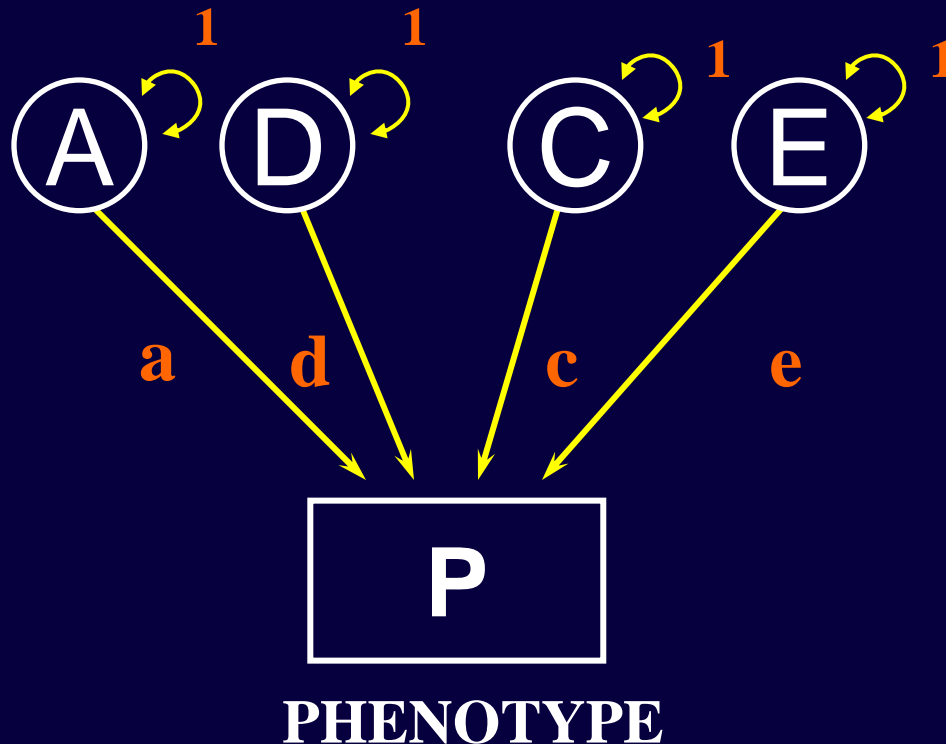


- $\text{Cov } AB = kl + mqn + mpl$
- $\text{Cov } BC = no$
- $\text{Cov } AC = mqo$
- $\text{Var } A = k^2 + m^2 + 2 kpm$
- $\text{Var } B = l^2 + n^2$
- $\text{Var } C = o^2$

Path Diagrams for the Classical Twin Model

Quantitative Genetic Theory

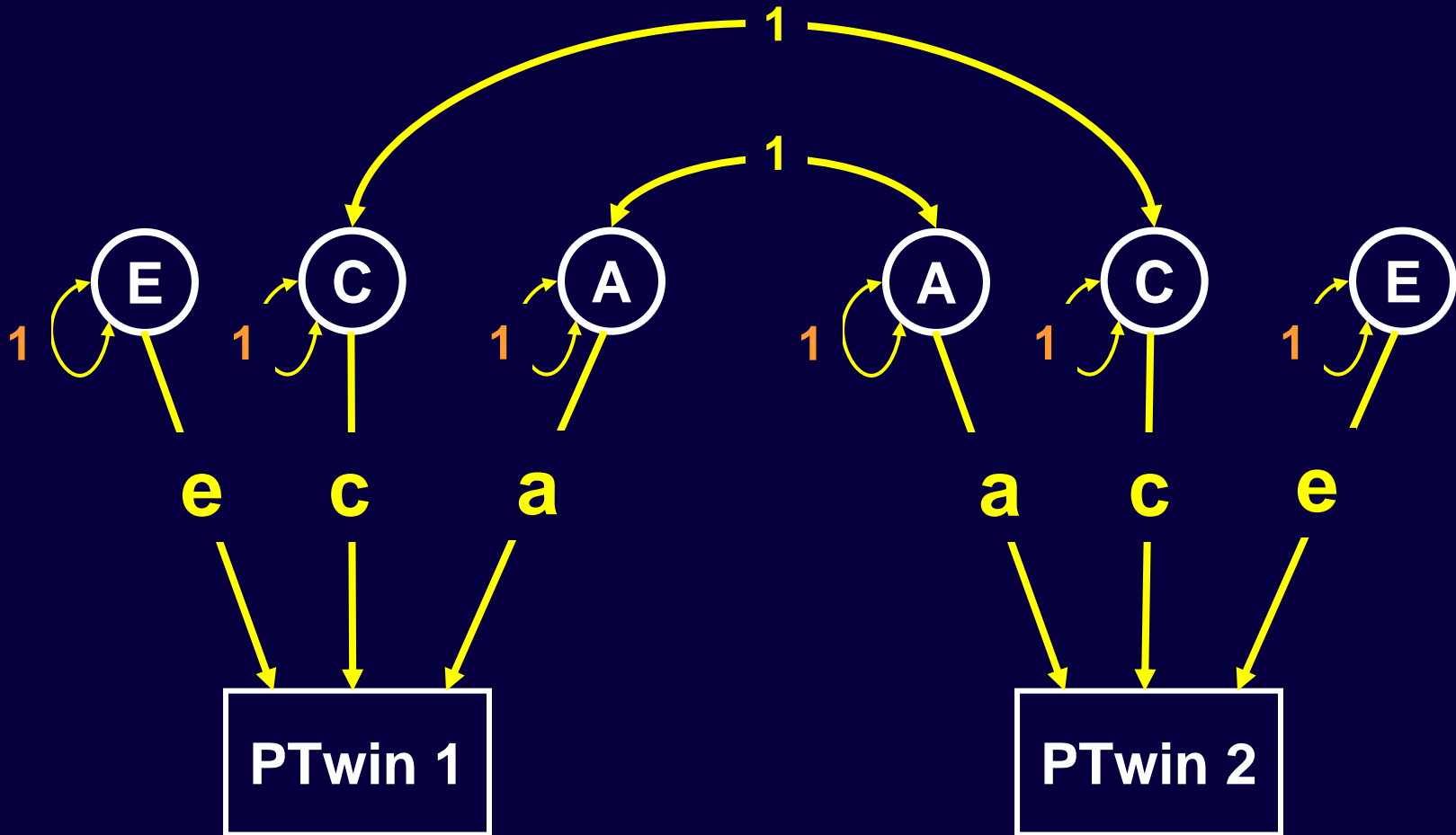
- There are two sources of Genetic influences: **Additive (A)** and **non-additive or Dominance (D)**
- There are two sources of environmental influences: **Common or shared (C)** and **non-shared or unique (E)**



In the preceding diagram...

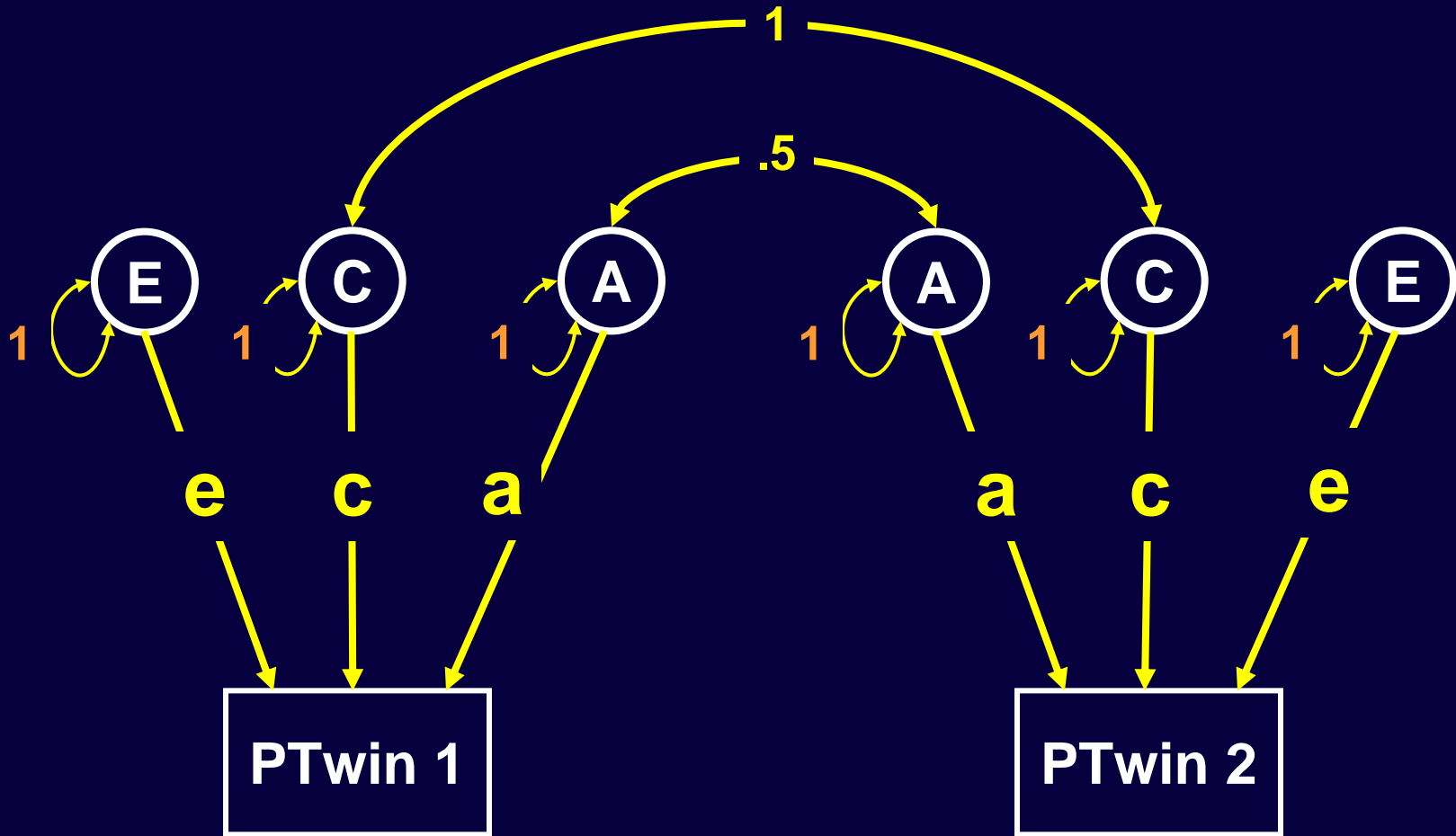
- A, D, C, E are *independent* variables
 - A = Additive genetic influences
 - D = Non-additive genetic influences (i.e., dominance)
 - C = Shared environmental influences
 - E = Non-shared environmental influences
 - A, D, C, E have variances of 1
- Phenotype is a dependent variable
 - P = phenotype; the measured variable
- a, d, c, e are parameter estimates

Model for MZ Pairs Reared Together



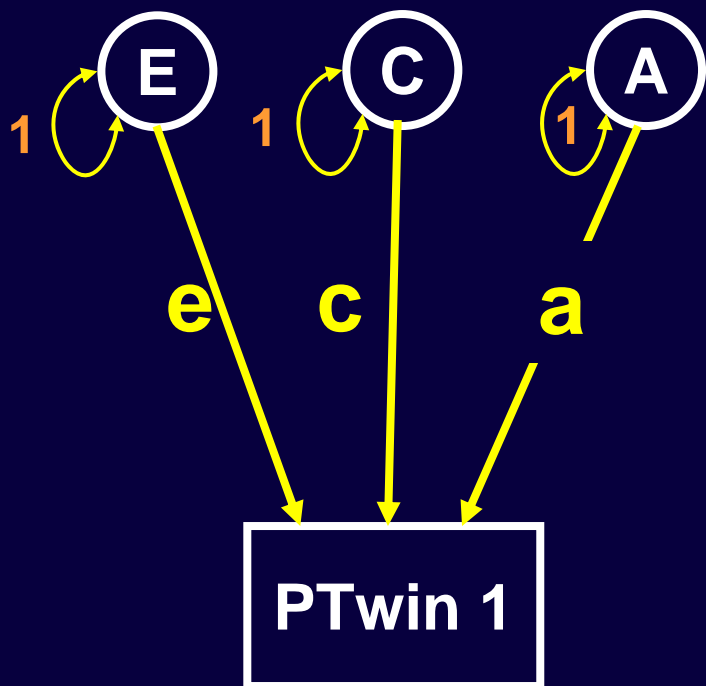
Note: a, c and e are the same cross twins

Model for DZ Pairs Reared Together

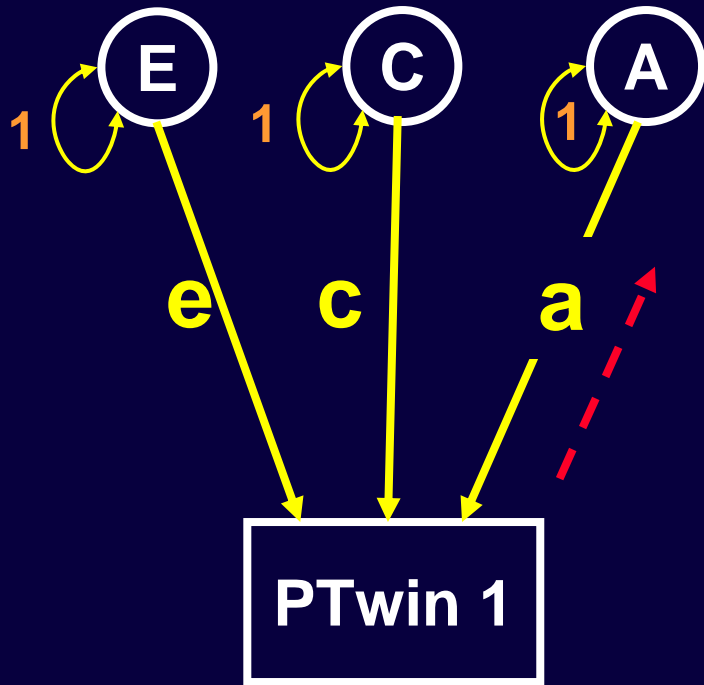


Note: a, c and e are also the same cross groups

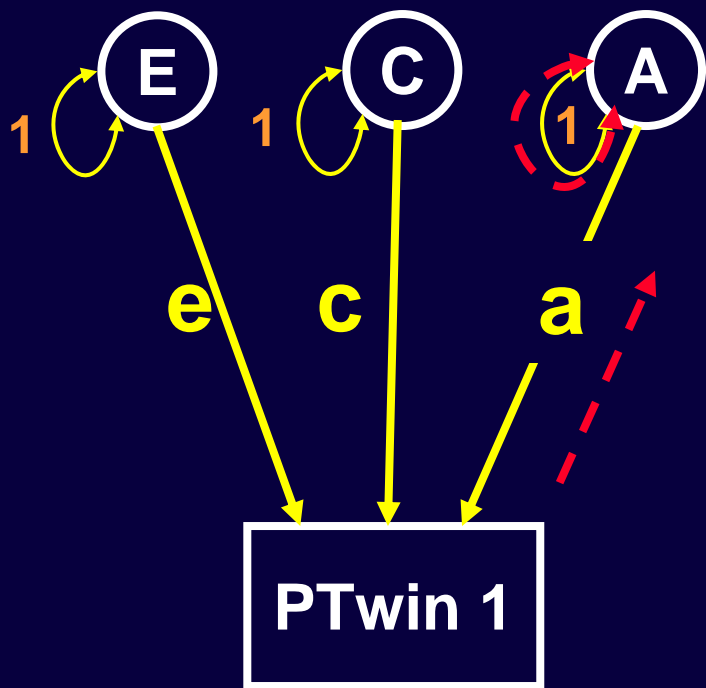
Variance of Twin 1 AND Twin 2 (for MZ and DZ pairs)



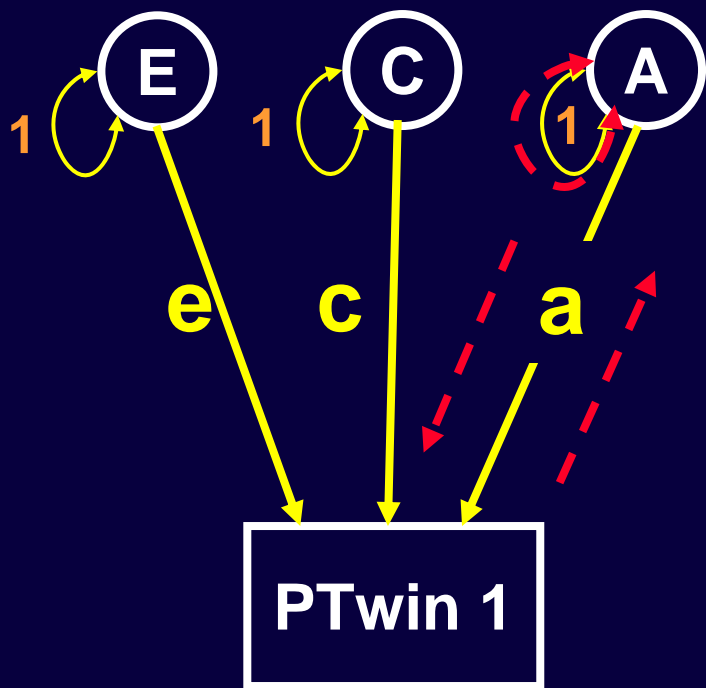
Variance of Twin 1 AND Twin 2 (for MZ and DZ pairs)



Variance of Twin 1 AND Twin 2 (for MZ and DZ pairs)

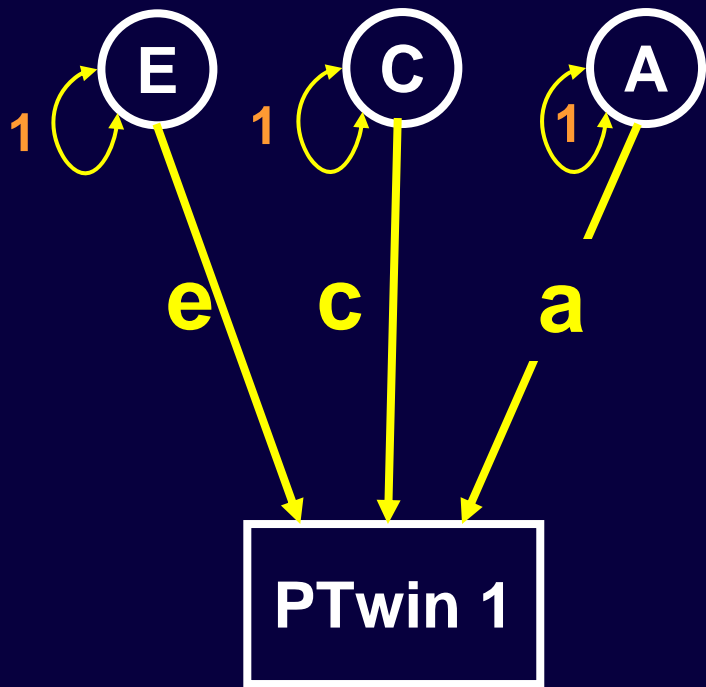


Variance of Twin 1 AND Twin 2 (for MZ and DZ pairs)



$$a * 1 * a = a^2$$
$$+$$

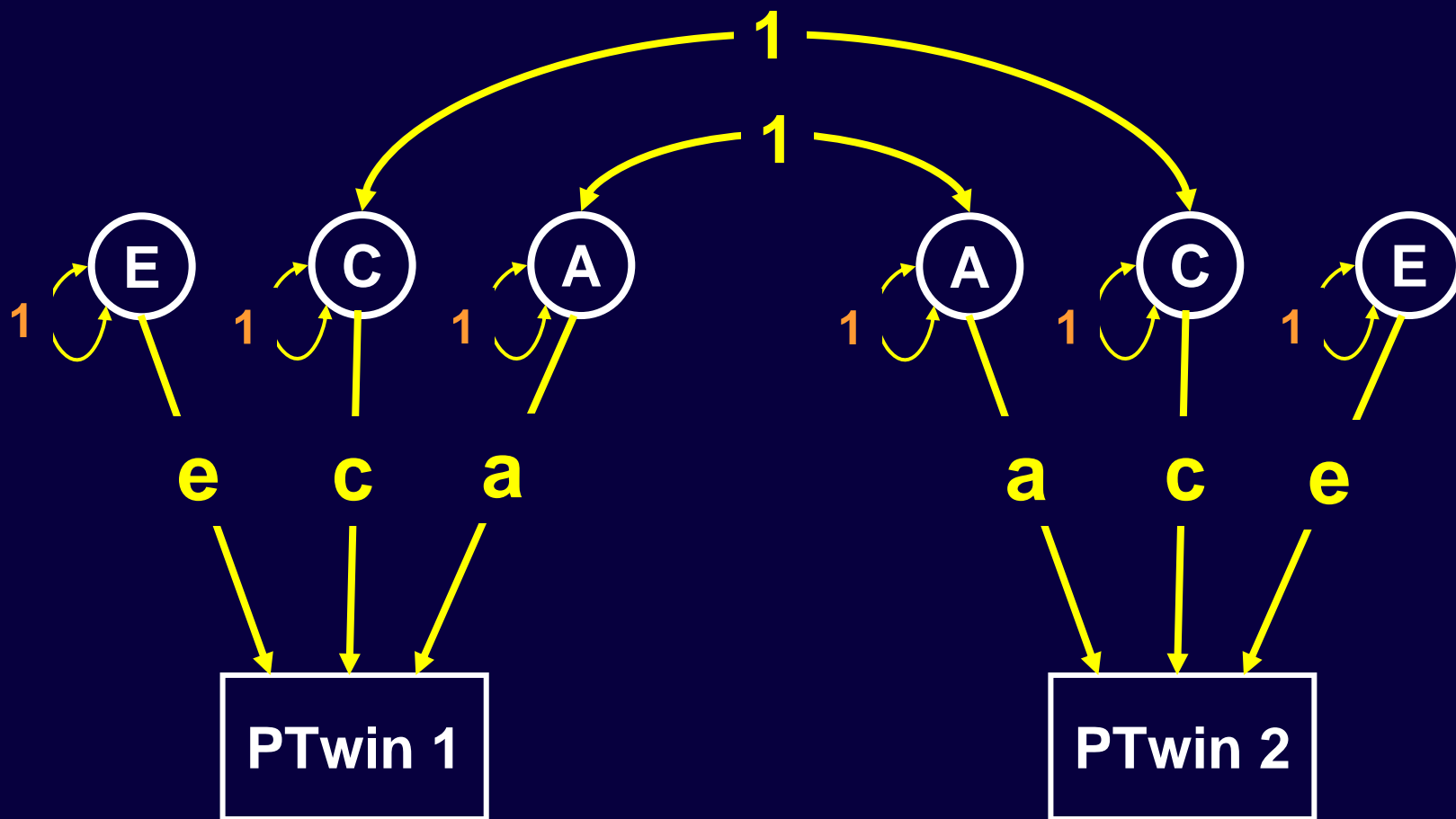
Variance of Twin 1 AND Twin 2 (for MZ and DZ pairs)



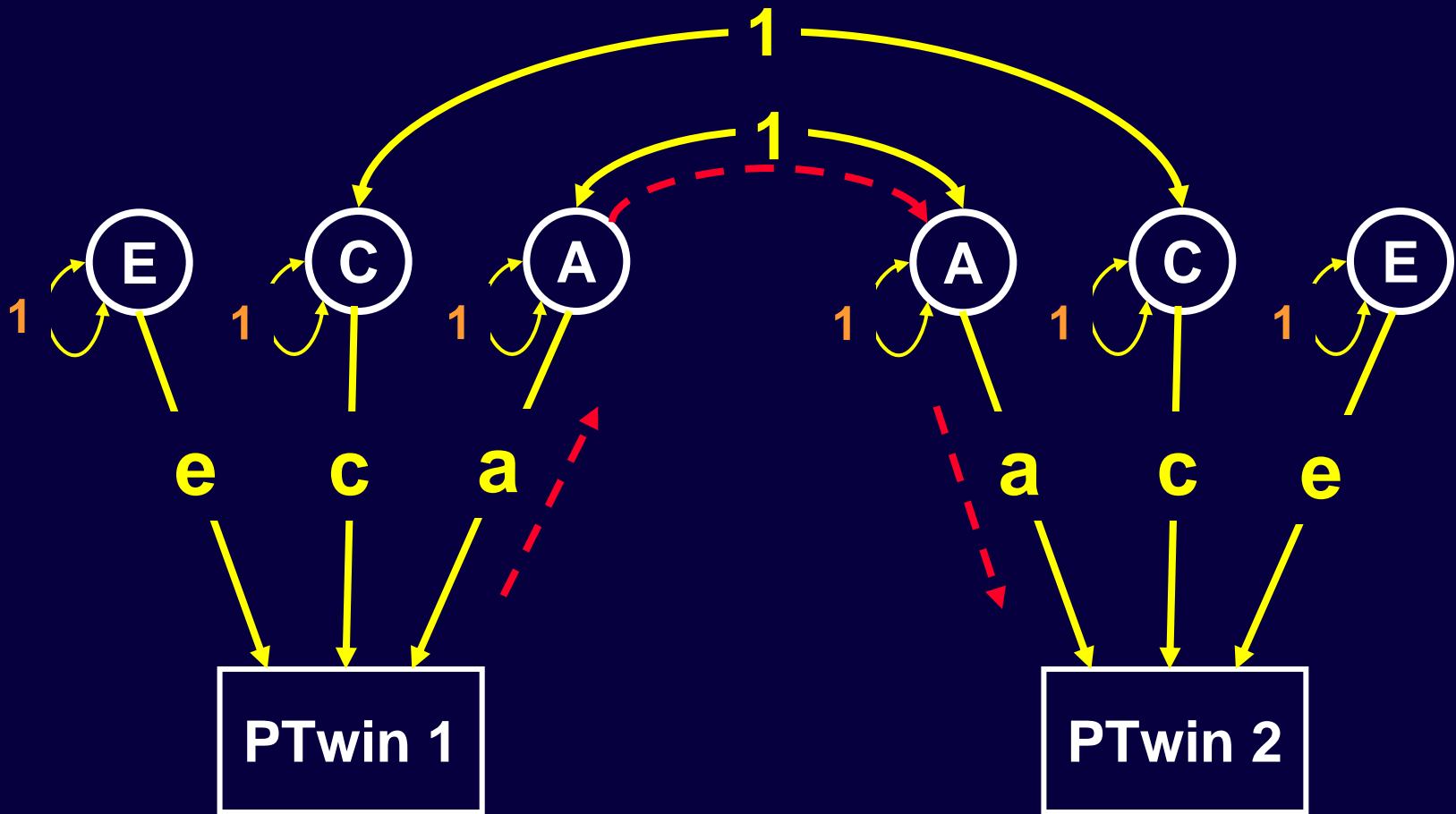
$$\begin{aligned} a * 1 * a &= a^2 \\ + \\ c * 1 * c &= c^2 \\ + \\ e * 1 * e &= e^2 \end{aligned}$$

$$\text{Total Variance} = a^2 + c^2 + e^2$$

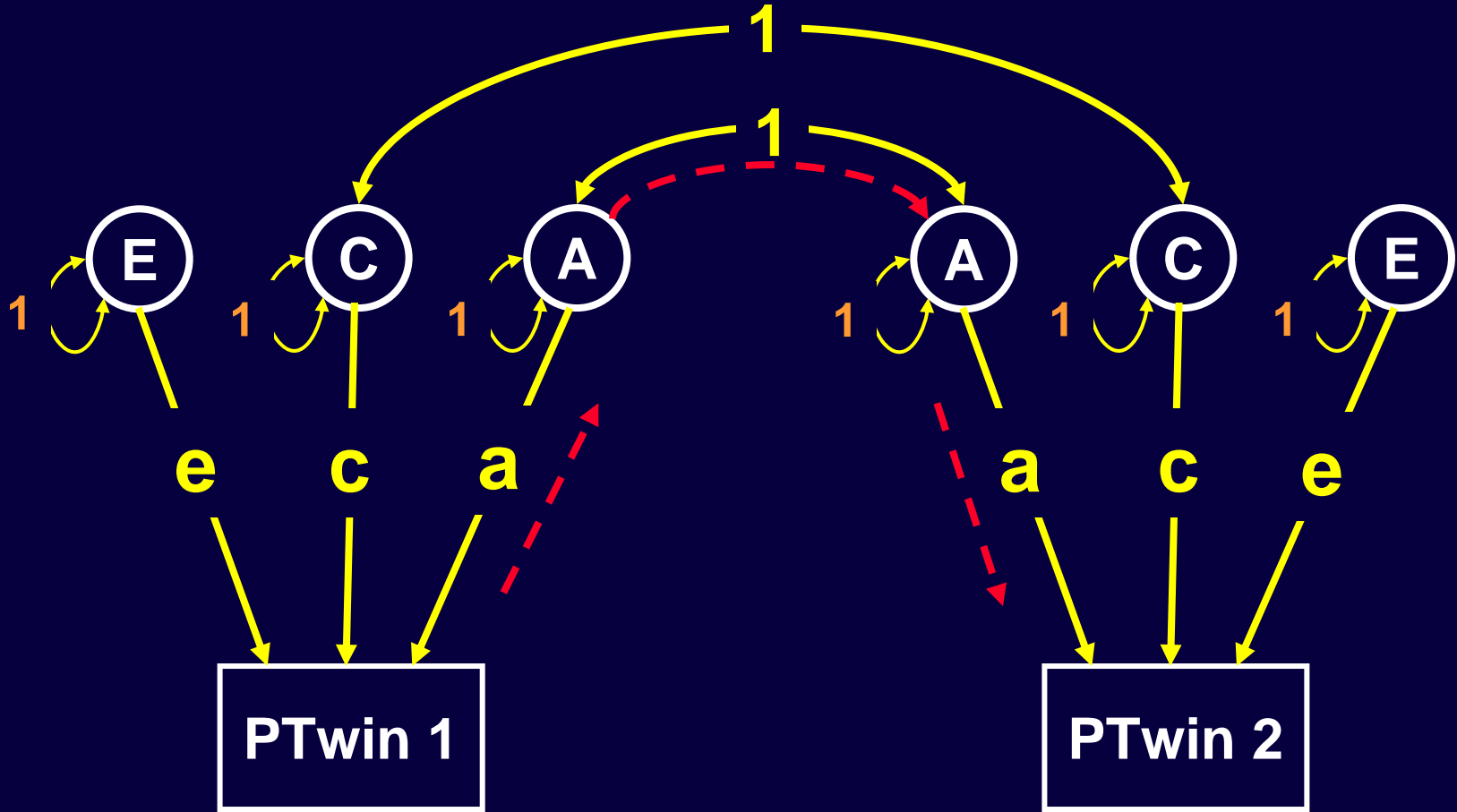
Covariance Twin 1-2: MZ pairs



Covariance Twin 1-2: MZ pairs

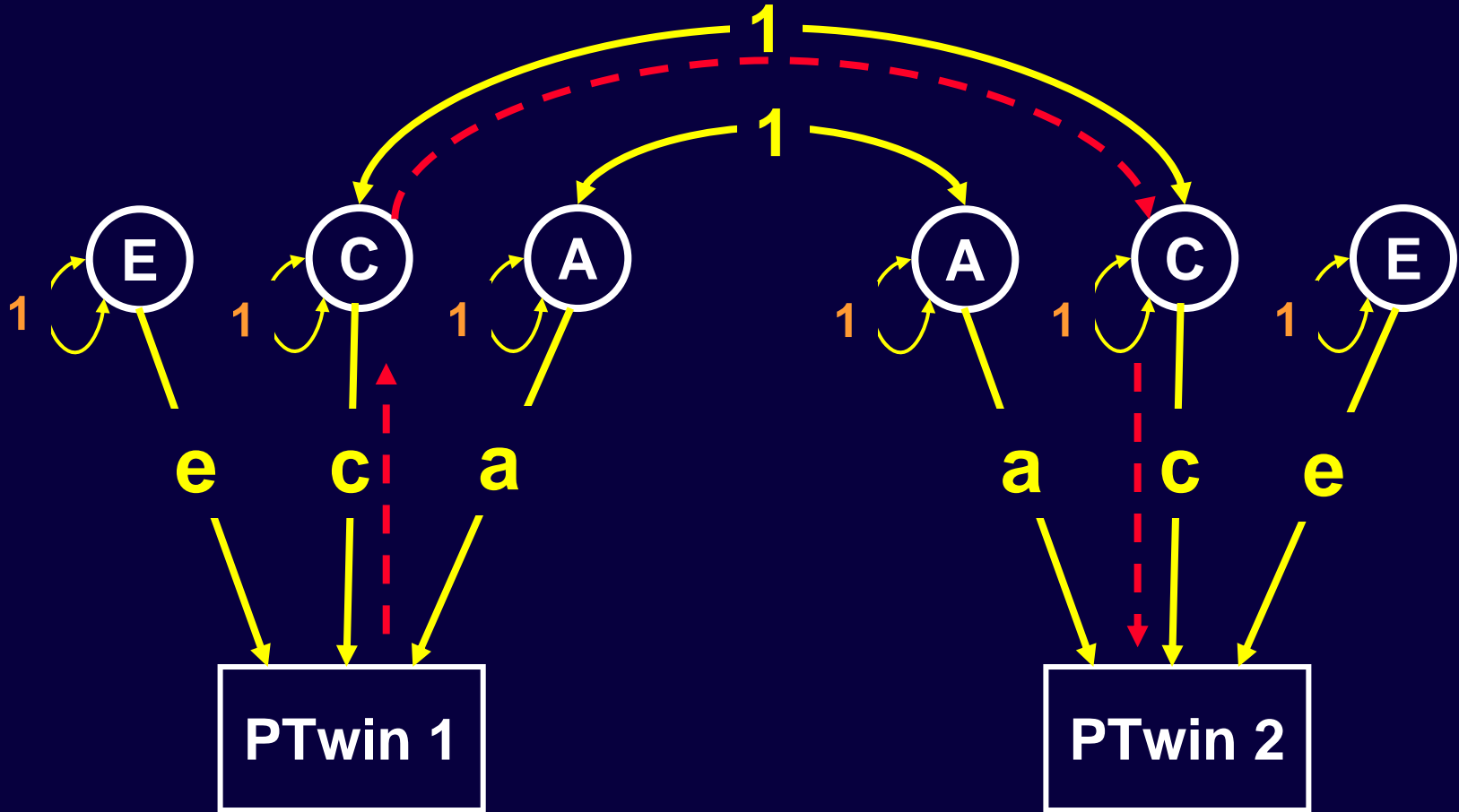


Covariance Twin 1-2: MZ pairs



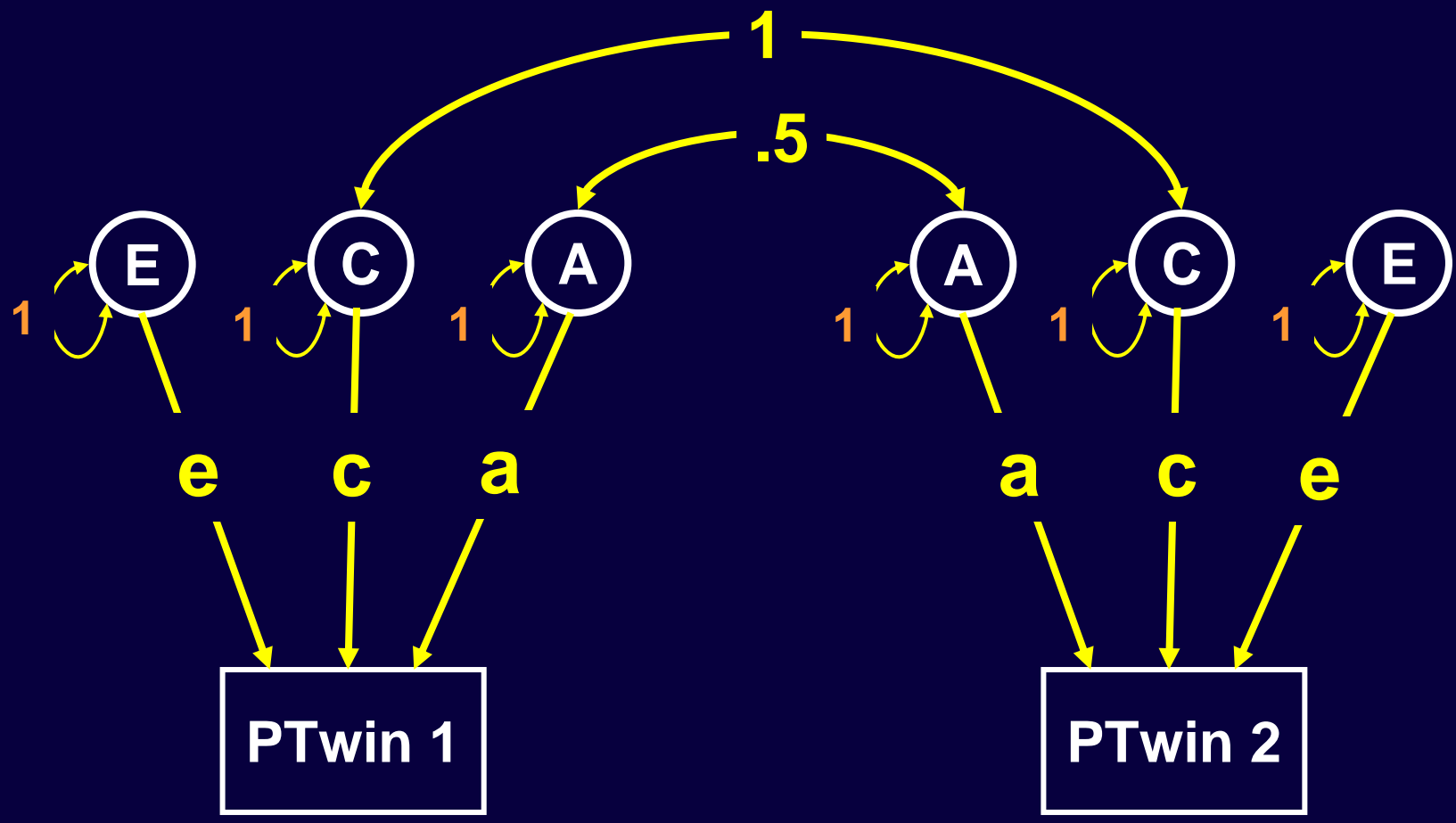
Total Covariance = $a^2 +$

Covariance Twin 1-2: MZ pairs



$$\text{Total Covariance} = a^2 + c^2$$

Covariance Twin 1-2: DZ pairs



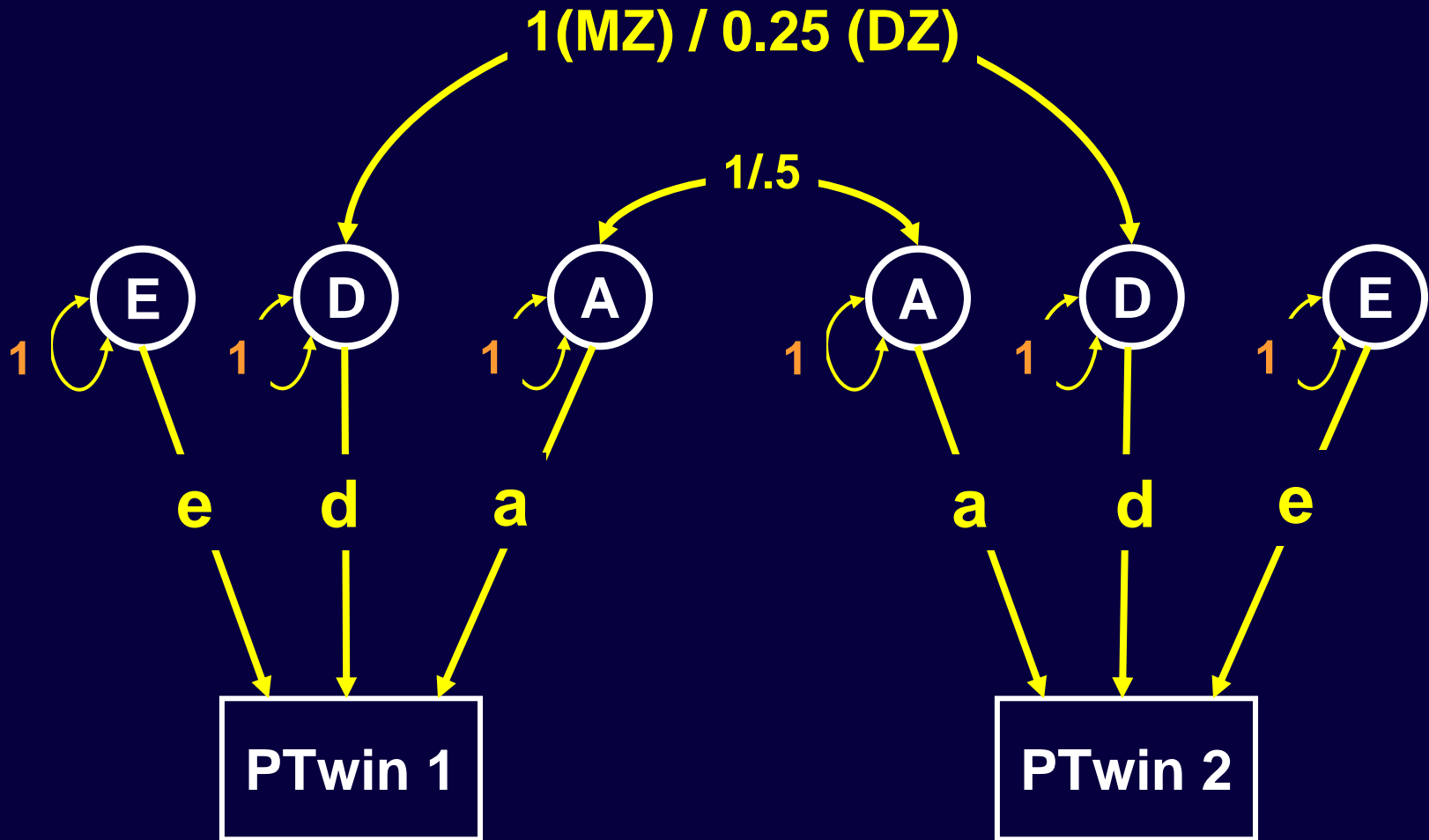
Total Covariance = $.5a^2 + c^2$

Predicted Var-Cov Matrices

$$\text{Cov } MZ \begin{matrix} \text{Tw1} \\ \text{Tw2} \end{matrix} = \begin{matrix} & \text{Tw1} & \text{Tw2} \\ \begin{matrix} \text{Tw1} \\ \text{Tw2} \end{matrix} & \begin{bmatrix} a^2 + c^2 + e^2 & a^2 + c^2 \\ a^2 + c^2 & a^2 + c^2 + e^2 \end{bmatrix} \end{matrix}$$

$$\text{Cov } DZ \begin{matrix} \text{Tw1} \\ \text{Tw2} \end{matrix} = \begin{matrix} & \text{Tw1} & \text{Tw2} \\ \begin{matrix} \text{Tw1} \\ \text{Tw2} \end{matrix} & \begin{bmatrix} a^2 + c^2 + e^2 & \frac{1}{2}a^2 + c^2 \\ \frac{1}{2}a^2 + c^2 & a^2 + c^2 + e^2 \end{bmatrix} \end{matrix}$$

ADE Model



Predicted Var-Cov Matrices

$$\text{Cov } MZ \begin{array}{c} \text{Tw1} \\ \text{Tw2} \end{array} = \begin{array}{cc} & \begin{array}{c} \text{Tw1} \\ \text{Tw2} \end{array} \\ \begin{array}{c} \text{Tw1} \\ \text{Tw2} \end{array} & \begin{bmatrix} a^2 + d^2 + e^2 & a^2 + d^2 \\ a^2 + d^2 & a^2 + d^2 + e^2 \end{bmatrix} \end{array}$$

$$\text{Cov } DZ \begin{array}{c} \text{Tw1} \\ \text{Tw2} \end{array} = \begin{array}{cc} & \begin{array}{c} \text{Tw1} \\ \text{Tw2} \end{array} \\ \begin{array}{c} \text{Tw1} \\ \text{Tw2} \end{array} & \begin{bmatrix} a^2 + d^2 + e^2 & \frac{1}{2}a^2 + \frac{1}{4}d^2 \\ \frac{1}{2}a^2 + \frac{1}{4}d^2 & a^2 + d^2 + e^2 \end{bmatrix} \end{array}$$

ACE or ADE

$$\text{Cov}(mz) = a^2 + c^2 \text{ or } a^2 + d^2$$

$$\text{Cov}(dz) = \frac{1}{2} a^2 + c^2 \text{ or } \frac{1}{2} a^2 + \frac{1}{4} d^2$$

$$V_p = a^2 + c^2 + e^2 \text{ or } a^2 + d^2 + e^2$$

3 unknown parameters (a, c, e or a, d, e),
and only **3** distinct predictive statistics:

Cov MZ, Cov DZ, V_p

this model is **just identified**

Effects of C and D are confounded

The twin correlations indicate which of the two components is more likely to fit the data:

$$\text{Cor}(mz) = a^2 + c^2 \text{ or } a^2 + d^2$$

$$\text{Cor}(dz) = \frac{1}{2} a^2 + c^2 \text{ or } \frac{1}{2} a^2 + \frac{1}{4} d^2$$

If $a^2 = .40$, $c^2 = .20$

$$r_{mz} = 0.60$$

$$r_{dz} = 0.40$$

ACE

If $a^2 = .40$, $d^2 = .20$

$$r_{mz} = 0.60$$

$$r_{dz} = 0.25$$

ADE

ADCE: classical twin design + adoption data

$$\text{Cov}(mz) = a^2 + d^2 + c^2$$

$$\text{Cov}(dz) = \frac{1}{2} a^2 + \frac{1}{4} d^2 + c^2$$

$$\text{Cov}(\text{adopSibs}) = c^2$$

$$V_p = a^2 + d^2 + c^2 + e^2$$

4 unknown parameters (a, c, d, e), and 4 distinct predictive statistics:

Cov MZ, Cov DZ, Cov adopSibs, V_p

this model is **just identified**

**Path Tracing Rules are
based on
Covariance Algebra**

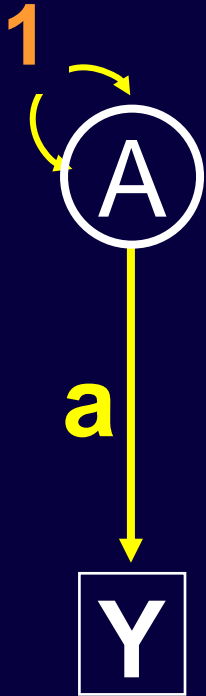
Three Fundamental Covariance Algebra Rules

$$\text{Var}(X) = \text{Cov}(X, X)$$

$$\text{Cov}(aX, bY) = ab \text{Cov}(X, Y)$$

$$\text{Cov}(X, Y+Z) = \text{Cov}(X, Y) + \text{Cov}(X, Z)$$

Example 1

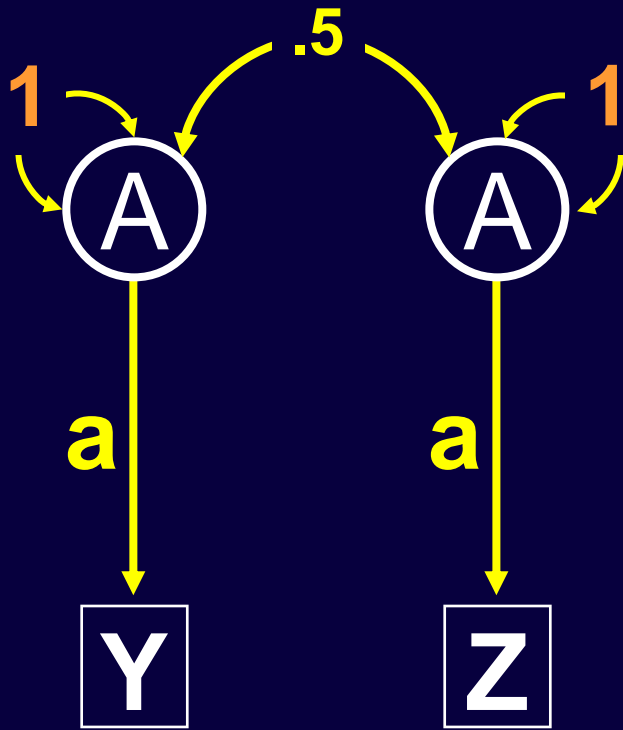


$$Y = aA$$

$$\begin{aligned} \text{Var} (Y) &= \text{Var} (aA) \\ &= \text{Cov} (aA, aA) \\ &= a^2 \text{Cov} (A, A) \\ &= a^2 \text{Var} (A) \\ &= a^2 * 1 \\ &= a^2 \end{aligned}$$

The variance of a dependent variable (Y) caused by independent variable A, is the squared regression coefficient multiplied by the variance of the independent variable

Example 2



$$Y = aA \quad Z = aA$$

$$\begin{aligned} \text{Cov}(Y, Z) &= \text{Cov}(aA, aA) \\ &= a^2 \text{Cov}(A, A) \\ &= a^2 * .5 \end{aligned}$$

Summary

- Path Tracing and Covariance Algebra have the same aim :
 - to work out the predicted **Variances** and **Covariances** of variables, given the specified model
- The Ultimate Goal is to fit Predicted Variances / Covariances to observed Variances / Covariances of the data in order to estimate model parameters :
 - regression coefficients, correlations