



# Categorical Data

HGEN619 2006



## Univariate Genetic Analysis

- Saturated Models
  - Free thresholds
- Univariate Models
  - Variances partitioned in a, c/d and e
  - Free thresholds (or not)



## Raw Dataset usmdd.ord

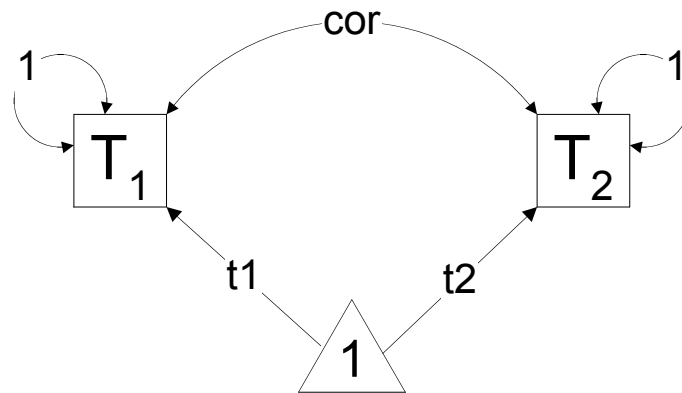
100	200
100	200
100	200
100	201
100	201
101	201
101	210
110	210
110	210
110	210
111	210
111	211
111	211

## Frequency Data: usmdd?.ctf

MZ  
0 0 329  
0 1 83  
1 0 95  
1 1 83

DZ  
0 0 201  
0 1 94  
1 0 82  
1 1 63

# Saturated Model



! Estimate thresholds & correlations – Saturated  
! US MDD data – females **contingency tables**

- #NGroups 2
- #define nvar2 2
- Title 1: MZ data
- Data NInput=2
- **C**Table 2 2
- 329 83 95 83
- Begin Matrices;
- T Full nvar2 1 Free
- X **S**td nvar2 nvar2 Free
- End Matrices;
- Start 1 T 1 1 - T nvar2 1
- **T**hreshold T;
- **C**orrelation X;
- Option RSiduals
- End
- Title 2: DZ data
- Data NInput=2
- CTable 2 2
- 201 94 82 63
- Begin Matrices;
- T Full nvar2 1 Free
- X Std nvar2 nvar2 Free
- End Matrices;
- Start 1 T 1 1 - T nvar2 1
- Threshold T;
- Correlation X;
- Option RSiduals
- Option Multiple Issat
- End

usmddsatct.mx

! Estimate thresholds & correlations – Saturated  
! US MDD data – females **raw ordinal data**

- #NGroups 2
  - #define nvar2 2
  - Title 1: MZ data
  - Data NInput=3
  - **Ordinal File=usmdd.ord**
  - Labels zyg mdd1 mdd2
  - Select if zyg=1
  - Select mdd1 mdd2 ;
  - Begin Matrices;
  - T Full 1 nvar2 Free
  - X Stnd nvar2 nvar2 Free
  - End Matrices;
  - Start 1 T 1 1 - T 1 nvar2
  - Threshold T;
  - Correlation X;
  - Options RSiduals
  - End
- Title 2: DZ data
  - Data NInput=3
  - Ordinal File=usmddmz.ord
  - Labels zyg mdd1 mdd2
  - Select if zyg=2
  - Select mdd1 mdd2 ;
  - Begin Matrices;
  - T Full 1 nvar2 Free
  - X Stnd nvar2 nvar2 Free
  - End Matrices;
  - Start 1 T 1 1 - T 1 nvar2
  - Threshold T;
  - Correlation X;
  - Options RSiduals
  - End

usmddsatord.mx

! Estimate thresholds & correlations – Saturated  
! US MDD data – females **frequency data**

- #NGroups 2
  - #define nvar2 2
  - Title 1: MZ data
  - Data NInput=3
  - **Ordinal File=usmddmz.ctf**
  - Labels mdd1 mdd2 frq
  - **Definition frq ;**
  - Begin Matrices;
  - T Full 1 nvar2 Free
  - X Stnd nvar2 nvar2 Free
  - F Full 1 1
  - End Matrices;
  - Start 1 T 1 1 - T 1 nvar2
  - **Specify F frq**
  - Threshold T;
  - Correlation X;
  - **Frequency F;**
  - End
- Title 2: DZ data
  - Data NInput=3
  - Ordinal File=usmdddz.ctf
  - Labels mdd1 mdd2 frq
  - Definition frq ;
  - Begin Matrices;
  - T Full 1 nvar2 Free
  - X Stnd nvar2 nvar2 Free
  - F Full 1 1
  - End Matrices;
  - Start 1 T 1 1 - T 1 nvar2
  - Specify F frq
  - Threshold T;
  - Correlation X;
  - Frequency F;
  - End

usmddsatfrq.mx

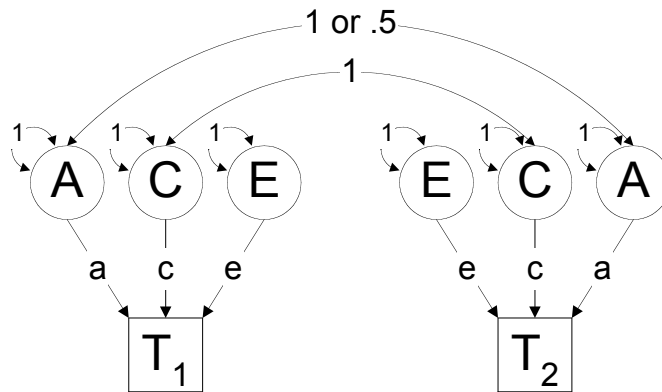
## Dat File: usmdd.dat

- Data NInput=3
- Rectangular File=usmdd.ord
- Labels zyg mdd1 mdd2

## Submodels: Equality of Thresholds

	MZ (group 1)					DZ (group 2)					par
	t1	t2	v1	cor	v2	t3	t4	v3	cor	v4	
Full	1	2	0	3	0	4	5	0	6	0	6
II	1	1	0	3	0	4	4	0	6	0	4
III	1	1	0	3	0	1	1	0	6	0	3

# ACE Model



! Estimate variance components - ACED model  
! US MDD data - females

- #NGroups 4
- #define nvar 1
- #define nvar2 2
- Title 1: Model Parameters
- Calculation
- Begin Matrices;
- X Lower nvar nvar Free !a
- Y Lower nvar nvar !c
- Z Lower nvar nvar Free !e
- W Lower nvar nvar Free !d
- H Full 1 1 !0.5
- Q Full 1 1 !0.25
- End Matrices;
- Matrix H .5
- Matrix Q .25
- Label Row X add\_gen
- Label Row Y com\_env
- Label Row Z spec\_env
- Label Row W dom\_gen
- Begin Algebra;
- A= X\*X'; !a^2
- C= Y\*Y'; !c^2
- E= Z\*Z'; !e^2
- D= W\*W'; !d^2
- End Algebra;
- End

usmddaces.mx

## ! Estimate variance components - ACED model ! US MDD data - females II

- |                                                                                                                                                                                                                                                                                                                                                                                                             |                                                                                                                                                                                                                                                                                                                                                                                                                 |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <ul style="list-style-type: none"> <li>■ Title 2: MZ data</li> <li>■ #include usmdd.dat</li> <li>■ Select if zyg =1</li> <li>■ Select mdd1 mdd2 ;</li> <li>■ Begin Matrices =Group 1;</li> <li>■ T Full 1 nvar2 Free</li> <li>■ End Matrices;</li> <li>■ Thresholds T;</li> <li>■ Covariance</li> <li>■ A+C+E+D   A+C+D _</li> <li>■ A+C+D   A+C+E+D;</li> <li>■ Option RSiduals;</li> <li>■ End</li> </ul> | <ul style="list-style-type: none"> <li>■ Title 3: DZ data</li> <li>■ #include usmdd.dat</li> <li>■ Select if zyg =2</li> <li>■ Select mdd1 mdd2 ;</li> <li>■ Begin Matrices =Group 1;</li> <li>■ T Full 1 nvar2 Free</li> <li>■ End Matrices;</li> <li>■ Thresholds T;</li> <li>■ Covariance</li> <li>■ A+C+E+D  H@A+C+Q@D _</li> <li>■ H@A+C+Q@D A+C+E+D;</li> <li>■ Option RSiduals</li> <li>■ End</li> </ul> |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

usmddaces.mx

## ! Estimate variance components - ACED model ! US MDD data - females III

- |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |                                                                                                                                                                                                                                                                                          |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <ul style="list-style-type: none"> <li>■ Title 4: Constrain var=1</li> <li>■ Constraint</li> <li>■ Begin Matrices =Group 1;</li> <li>■ I Iden 1 1</li> <li>■ End Matrices;</li> <li>■ Start .5 all</li> <li>■ St 1 T 2 1 1-T 2 1 nvar2</li> <li>■ St 1 T 3 1 1-T 3 1 nvar2</li> <li>■ Begin Algebra;</li> <li>■ P=A C E D;</li> <li>■ End Algebra;</li> <li>■ Constraint A+C+E+D=I;</li> <li>■ !ADE model</li> <li>■ Option NDecimals=4</li> <li>■ Option Sat=2508.004,2054</li> <li>■ Option Multiple</li> <li>■ End</li> </ul> | <ul style="list-style-type: none"> <li>■ !AE model</li> <li>■ Drop W 1 1 1</li> <li>■ End</li> <li>■ !ACE model</li> <li>■ Free Y 1 1 1</li> <li>■ End</li> <li>■ !CE model</li> <li>■ Drop X 1 1 1</li> <li>■ End</li> <li>■ !E model</li> <li>■ Drop Y 1 1 1</li> <li>■ End</li> </ul> |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

usmddaces.mx

## Submodels

\*1 constraint: A+C+D+E=1

Matrix / Model	X (a)	Y (c)	Z (e)	W (d)	Cor NP	Mean NP	NP	DF
Sat					2	4	6+1*	
ADE	Free		Free	Free		4	7	0
AE	Free		Free	Drop		4	6	1
ACE	Free	Free	Free			4	7	0
CE	Drop	Free	Free			4	6	1
E		Drop	Free			4	5	2

## Goodness-of-Fit

	-2LL	df	$\Pi^2$	df	p	AIC
Sat						
ADE				0		
AE				1		
ACE				0		
CE				1		
E				2		

## Thresholds: A quick review

- When using raw ordinal data, it is necessary to give  $Mx$  starting values for thresholds
- Thresholds indicate the point at which the variable transitions from one level to the next
- For a variable with  $n$  categories, there will always be  $n - 1$  thresholds

## Thresholds for Multiple Level Variables

- CUMULATIVE thresholds:
  - Threshold 1, 61% have a score below 1 → z-score of 0.28
  - Threshold 2, 77% have a score below 2 → z-score of 0.74
  - Threshold 3, 86% have a score below 3 → z-score of 1.08
- INCREMENTAL thresholds:
  - Threshold 1 = 0.28
  - Threshold 2 =  $0.74 - 0.28 = 0.46$
  - Threshold 3 =  $1.08 - 0.74 = 0.34$