


# 认知、情绪与社会行为的遗传基础 及其神经相关物的研究进展



1

Galton

1875 Francis

“ 973”

“  
2014 11  
”

2

twin study

nature versus nurture

monozygotic MZ  
dizygotic DZ  
50%

MZ DZ

0

0

1

candidate gene association study CGAS

→

|

→

nucleotide polymorphism SNP SNP

single  
DNA

90%

500

1000

300  
SNP  
haplotype  
variable number of tandem repeats VNTR

10%

— gene set association analysis

SNP  
2 3 5  
24  
*TPH2* rs1487275 *SLC6A4* rs8076005 *HTR2C* rs2192371  
*HTR6* rs6658108 *HTR5A* rs732050

19%

genome-wide association study GWAS

SNP

CGAS

CGAS

GWAS

GWAS

GWAS

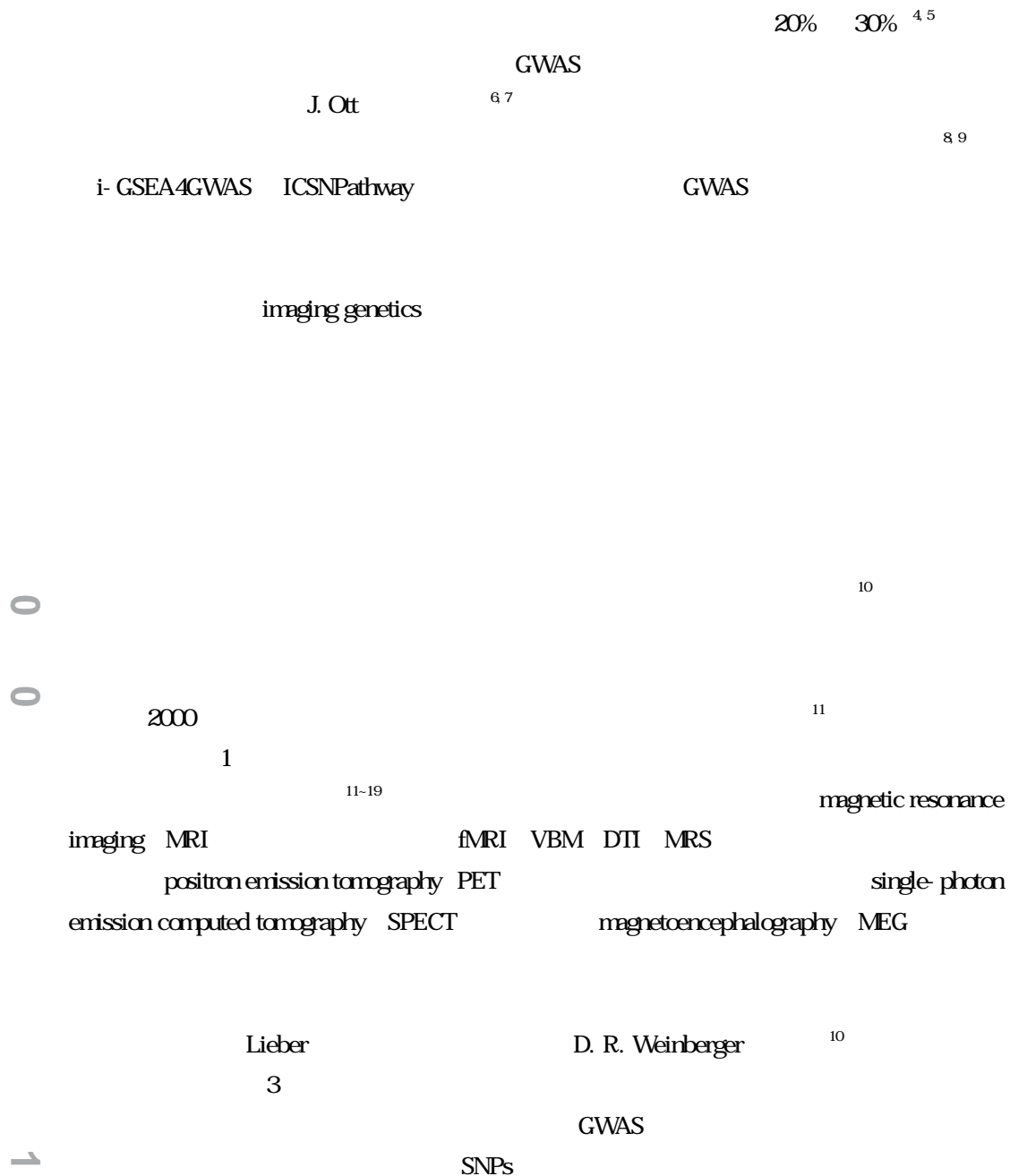
GWAS

SNP

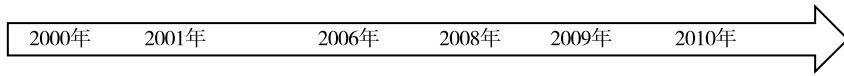
copy number variation CNV

GWAS

GWAS



fMRI和APOE (Bookheimer et al., 2001) /  
COMT (Egan et al., 2001) /5-HTTLPR  
(Hariri et al., 2002) ;  
第一次基于MRI的神经影像遗传学



case-control study

family study

animal study

knockout

3

2001

Chinese National Twin Registry CNTR

Twin Registry in Southwestern China

TRiSC

20

The Beijing Twin Study BeTwiSt

21



generalist genes

22

FRA

FIE

CFE

23

24

25

*BDNF* Val66Met

26 27

SLEs

*BDNF*

Val

Met/Met

28

30%

50%

29

2

*SEMA5A*

SNP rs42352

30

RAPM

*GABRB1*

rs7435958

31

*COMT* *DRD2* *DRD3* *NTSR1*

32-35

35

*BDNF*

*NTSR1* rs4334545

C

CT/TT

rs4334545

CC

表 2 国内认知能力的遗传基础研究的主要进展

30	SEMA5A 169 SNP	RAPM	SEMA5A SNP rs42352 SEMA5A rs42352 TT RAPM
31	GABRB1 49 SNP - -	WAIS-R	GABRB1 rs7435958 GG GABRB1 rs7435958GG
32	COMT Val158Met rs4680 COMT		COMT Met Val 2-back COMT
33	NTSR1 5 SNP		2-back rs4334545 C rs6090453 G rs4334545 CT/TT CC
33	COMT Val158Met DRD1 140G/T - 48A/G DRD2 TaqIA TaqIB DRD3 Ser9Gly DRD4 - C521T - G809A DAT1 VNIR		DRD2 TaqIA TaqIB DRD3 Ser9Gly DRD2 TaqIA
34	BDNF Val166Met rs6265 5		BDNF Val BDNF Met
42	DDC 7 DDC 5		DCC SNP rs3887825 rs7786398 rs10499695 rs6969081 rs7786398 rs10499695 rs6969081 SNP

0

0

1

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1



37	DBH - 1021C/T 19bp Ins/Del MAOA 30bp VNTR MAOB rs1799836		DBH - 19bp Ins/Del MAOA VNTR
40	MAOA VNTR A 5-		MAOA 4r/4r 3r/3r 3r/4r " "
39	ADRA2B		AAGG/AAGG SSRT Stop-signal reaction time CCAC/AAGG CCAC/CCAC AAGG/AAGG
41	5-HTR2A 30 SNP 5 2A		rs1923888 rs1745837 rs9567739 rs3742279 rs655888 rs655854 and rs2296972
38	13		5- 5-HT6 T267C T267C TT CT TT BDNF COMT DBH DRD2 DRD3 DRD4 MAOA TPH2 5-HT2A GRM1 GRIN2B

36-42

3

5-

5-

19%

3

18%

43

5-HTTLPR

44-45

OXTR

5-HT1A

表 3 国内情绪及社会行为的遗传基础研究的主要进展

3	127 SNP 2 VNIR 5		8 5- TPH2 MAOA MAOB SLC6A4 HTR2A HTR2C HTR5A HTR6 12 SNP HTR6 rs6658108 HTR5A rs732050 TPH2 rs1487275 SLC6A4 rs8076005 HTR2C rs2192371 19%
45	5- 5-HTTLPR 5-		5-HTTLPR M s/s
44	5- 5-HTTLPR 5-		5-HTTLPR M s/s HA 5-HTTLPR HA
57	5- 5-HTTLPR 5-	5-	M s/s 5-
46	OXTR rs53576		OXTR rs53576 A OXTR rs53576 OXTR rs53576

0  
0

1  
-  
1

				A/A	G/G	
47	<i>OXTR</i>	rs53576		A/A	G/G	
48	<i>OXTR</i>	rs53576		A/A	G	
				G/G	A/A	
43		96 SNP	2			18%
58	5- <i>5-HTTLPR</i>	5-		5- <i>HTTLPR</i>	LAT	IGT
				1	s IGT	40
59	<i>COMT</i>	Val158Met rs4680				

50	5-HT1A C- 1019G rs6295 5- 1A		5-HT1A CG/GG CC
49	DBH - 1021C/T 19bp Ins/Del		DBH - 1021C/T CC DBH CT/TT 2.3% 1.4%
61	OXTR 10	IRI	OXTR rs2254298 rs2268491 rs13316193 rs4686302 IRI
62	COMT 7 SNP COMT		COMT rs2020917 and rs737865 SNP rs5993883 SNP

0  
0

### 3.4.1 注意缺陷多动障碍

attention-deficit hyperactivity disorder ADHD

76% 63

ADHD 64 65

5- 23 245 SNP ADHD

ADHD MAOA DDC ADRA2C

COMT MAOA ADHD

66 BDNF MAOA DRD4 SLC6A2 ADRA2A

ADHD 67 ADHD-HI 68 ADHD 69

ADHD 70 5 71

DRD1-5 ADHD

### 3.4.2 重度抑郁症

major depression disorder MDD

MDD 40% 72 MDD

BDNF <sup>73, 74</sup>

BDNF Val Val/Val Val/Met Met/Met

rs6782799 <sup>28, 75</sup> <sup>76</sup> BDNF Val66Met GSK3B

BDNF CRHR1 MDD BDNF NTRK2

MDD <sup>77, 78</sup>

5- SSRI

20 29 5- HTTLPR HTR1A rs6295

MDD <sup>79</sup> 5- HTTLPR

S <sup>80</sup> 5- HTTLPR

<sup>81, 82</sup> <sup>44, 45</sup>

<sup>83</sup> fMRI 5- HTTLPR

2 TPH2

5- TPH2 rs4570625

MDD TPH2 <sup>84</sup>

MAOA DRD DBH COMT MDD

3.4.3 焦虑症

anxiety disorders

32% 48% <sup>85</sup> <sup>86</sup>

2 <sup>87</sup>

5- HTTLPR L S

<sup>88</sup> S/S L

S/S

-

3.4.4 双相情感障碍

bipolar disorder BPD manic-depressive

insanity

57% 14% <sup>89</sup>

BDNF BPD

BDNF Val166Met rs6265 BPD Met

Val BPD-I BPD-II





4

22

90

138

139

W. S. Kremen

140 141

J. L. Stein

GWAS

142

80%

GWAS

143-145

GWAS

GWAS

ENIGMA CHARGE IMAGEN

146

5

1

5 10

2  
GWAS

GWAS

3

GWAS

4

0

147

0

5

→ 5 10

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部分生物学专业名词解释

genetic material	RNA DNA
heritability	
gene polymorphism	2
allele	
genotype	
haplotype	
copy number variation CNV	1 kb
variable number of tandem repeats VNTR	DNA      DNA      10 10   1000
linkage	
phenotype	
gene frequency	
susceptibility	

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