



1

Galton

1875 Francis

“ 973”

“ _____ ”

2014 11

“ ”

2

.

twin study

nature versus nurture

monozygotic MZ

dizygotic DZ

50%

MZ DZ

1

candidate gene association study CGAS

nucleotide polymorphism SNP SNP

90%

500

single DNA 1000



300
SNP
haplotype
variable number of tandem repeats VNTR

10%

— gene set association analysis

SNP
2 3 5
24 rs1487275 rs8076005 rs2192371 rs6658108 rs732050
19%

genome-wide association study GWAS

SNP

CGAS

CGAS

GWAS

GWAS

GWAS

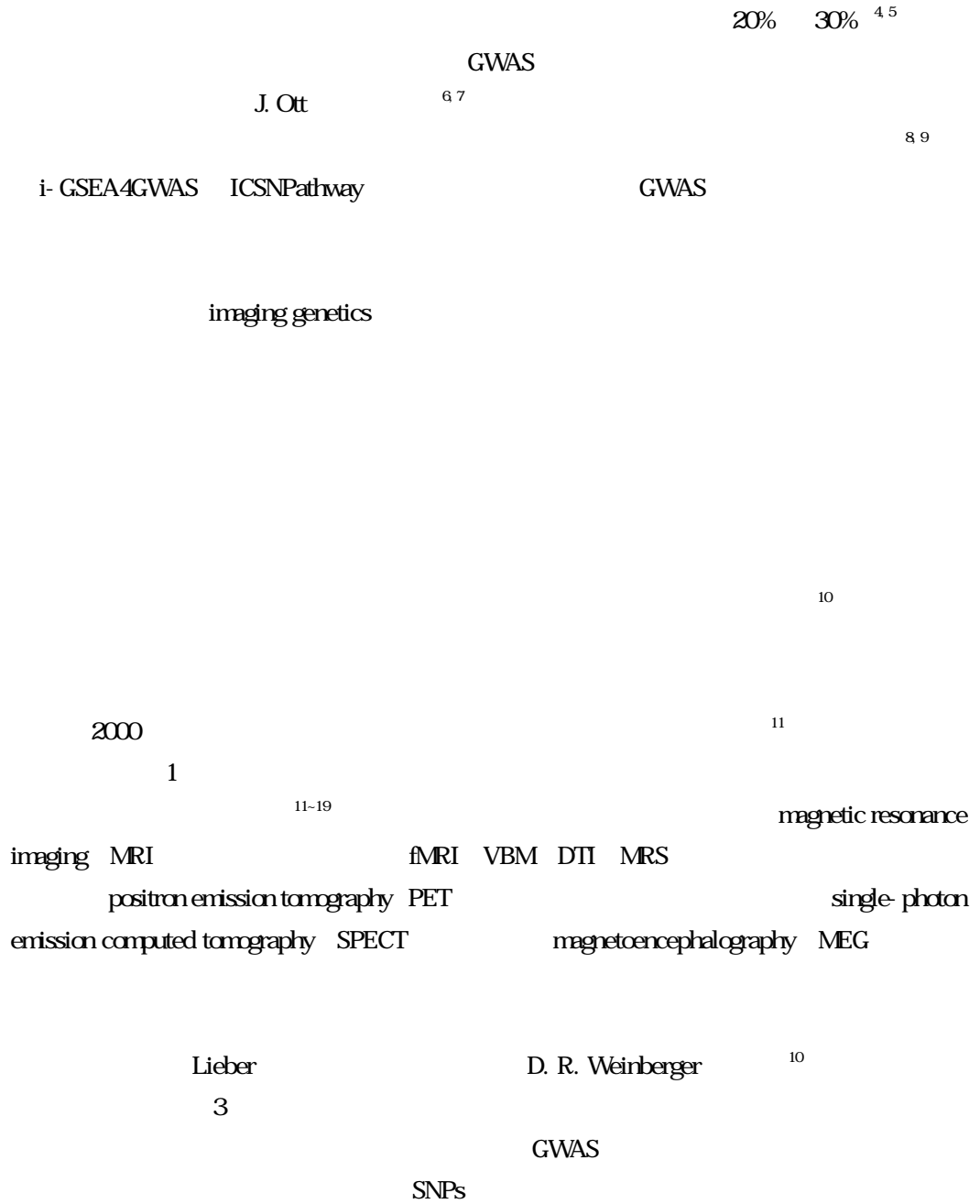
GWAS

SNP

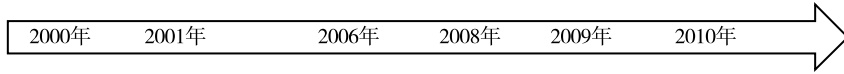
copy number variation CNV

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



1

10

case- control study

family study

animal study

knockout

3

.

2001

Chinese National Twin Registry CNTR

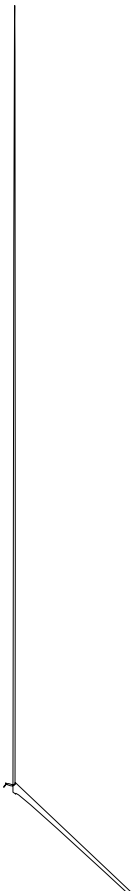
Twin Registry in Southwestern China

20

TRiSC

The Beijing Twin Study BeTwiSt

21





generalist genes

22

FRA

FIE

CFE

23

24

25

Val66Met

26 27

SLEs

Val

Met/Met

28

30%

50%

29

2

SNP rs42352

30

RAPM

rs7435958

31

32-35

35

rs4334545 C

rs4334545

CC

CT/TT

表 2 国内认知能力的遗传基础研究的主要进展

30	169 SNP	RAPM	SNP rs42352 rs42352 TT RAPM
31	49 SNP - -	WAIS- R	rs7435958 GG rs7435958GG
32	Val158Met rs4680 COMT		Met Val 2- back
33	5 SNP		2- back rs4334545 C rs6090453 G rs4334545 CT/TT CC
34	Val158Met 140G/ T - 48A/G TaqIA TaqIB Ser9Gly - C521T - G809A VNIR		TaqIA TaqIB Ser9Gly TaqIA
35	Val166Met rs6265 5		Val BDNF Met
42	7 DDC 5		SNP rs3887825 rs7786398 rs10499695 rs6969081 rs7786398 rs10499695 rs6969081 SNP G rs10499695 rs6969081
			st 798

Val

BDNF

37	- 1021C/T 19bp Ins/Del 30bp VNTR rs1799836		- 19bp Ins/Del VNTR
40	VNTR A 5-		4r/4r 3r/3r 3r/4r " "
39			AAGG/AAGG SSRT Stop- signal reaction time CCAC/AAGG CCAC/CCAC AAGG/AAGG
41	30 SNP 5- 2A		rs1923888 rs1745837 rs9567739 rs3742279 rs655888 rs655854 and rs2296972
38	13		5- T267C T267C TT CT TT

36-42

3

3

5-

5-

18%

19%

43

5- HTTLPR

44-45

表 3 国内情绪及社会行为的遗传基础研究的主要进展

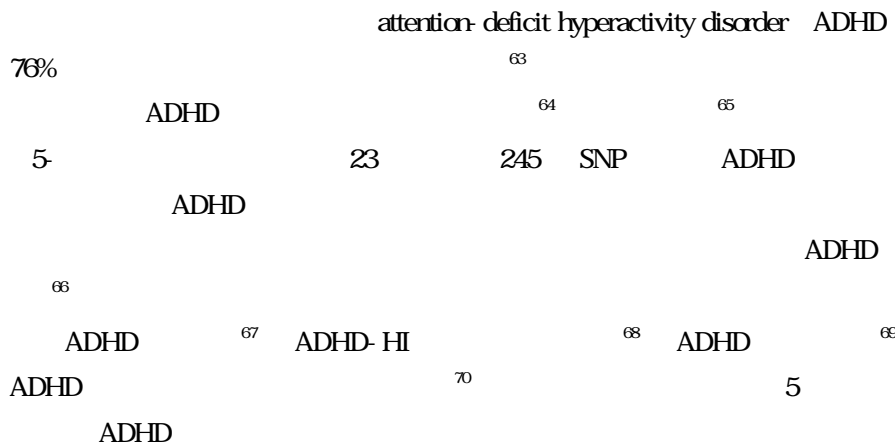
3	127 SNP 2 VNIR 5		8 5 12 SNP HTR6 rs6658108 HTR5A rs732050 TPH2 rs1487275 s8076005 rs2192371 19%
45	5 5-HTTLPR 5		5-HTTLPR M s/s
44	5 5-HTTLPR 5		5-HTTLPR M s/s HA 5-HTTLPR HA
57	5 5-HTTLPR 5	5	M s/s 5
46	rs53576		rs53576 A rs53576 rs53576



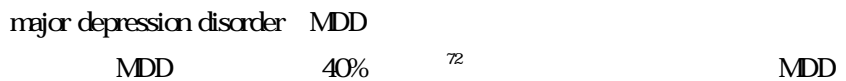
			A/A	G/G
47	rs53576		A/A	G/G
48	rs53576		A/A	G
			G/G	A/A
43	2	96 SNP		18%
58	5- 5- HTILPR	5-	5- HTILPR LAT 1	IGT s IGT 40
59	Val158Met rs4680			

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

3.4.1 注意缺陷多动障碍



3.4.2 重度抑郁症



BDNF 73, 74

Val66Met

Val Val/Val Val/Met Met/Met

28, 75 76

rs6782799 MDD

MDD 77, 78

5- SSRI

20 29 5- HTTLPR rs6295

MDD 79 5- HTTLPR

S 80 5- HTTLPR

81, 82 44, 45

83 fMRI 5- HTTLPR

2 TPH2

5- TPH2 rs4570625

MDD 84

MAOA DRD DBH COMT MDD

3.4.3 焦虑症

anxiety disorders

32% 48% 85 86

2 86 87

5- HTTLPR L S

88 S/S L

S/S

-

3.4.4 双相情感障碍

bipolar disorder BPD manic-depressive

insanity

57% 14% 89

BDNF BPD

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 - - -

147

5

5 10

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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