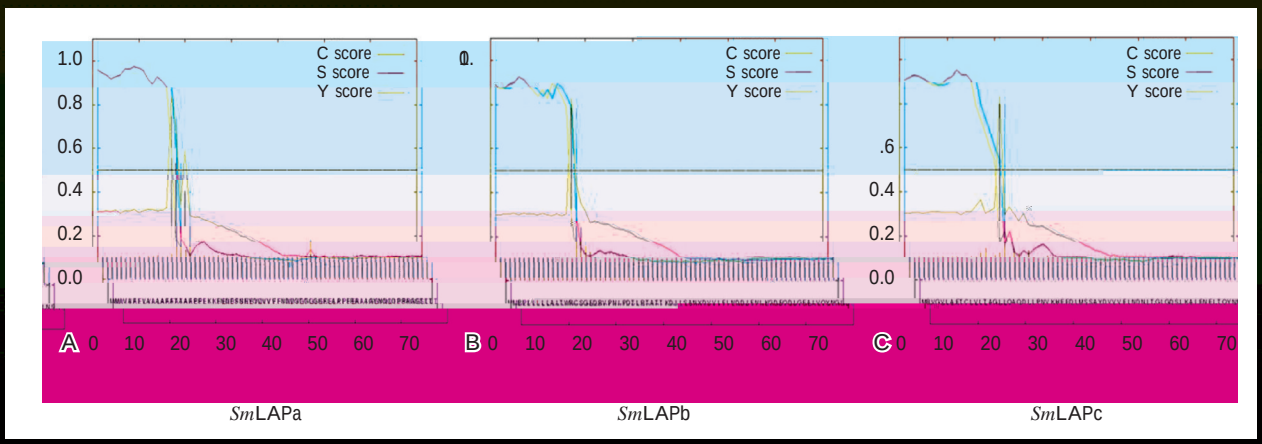
ISSN 1674-6929
CN 44-1658/R

2017 7 9 4 50

Volume 9 Number 4 July 2017



A SmLAPa B SmLAPb C SmLAPc

P244

Figure P244 Signal peptide pepdiction of *Sm*LAPs sequences

ISSN 1674-6929

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分子诊断与治疗杂志

JOURNAL OF MOLECULAR DIAGNOSTICS AND THERAPY

2017 7 9 4 50 Bimonthly Volume 9 Number 4 July 2017

179 10~11 510620
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jmdt vip.163.com
ISSN 1674-6929
CN 44-1656/R
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2017 7 18
RMB 15.00

Responsible Institution *Sun Yat sen University*
Sponsor *China Family Doctors Magazine Publisher Co. Ltd.*
Organizer *DaAn Gene Co., Ltd. of Sun Yat sen University*
Consultant *LAI Maode SHEN Ziyu*
Editor in Chief *LI Ming*
Managing Director *HAO Fen*
Editorial Office *<JOURNAL OF MOLECULAR DIAGNOSTICS AND THERAPY> Editorial Office*
Editors *LI Xiaolan YE Pingping LI Caizhen*
Editing *China Family Doctors Magazine Publisher Co. Ltd.*

Add *10~11 Fl., Xianglog Building, 179# Tian he bei Lu, Guangzhou, China 510620*
Tel *020 32290177 32290789−206*
Fax *020 32290177*
E mail *jmdt@vip.163.com*
CSSN *ISSN 1674−6929*
CN 44−1656/R
Printing *TianYi Yofus Technology Co., Ltd*
Publish Date *2017.7.18*
Price *RMB 15.00*

分子诊断与治疗杂志

2017 7 9 4

223

p.R248C TD

228

DA8600 EGFR DNA

234

241

23 STR 247

252

257

261

267

17α 272

I 278

MiRNA 3p/5p 284

289

293

JOURNAL OF
MOLECULAR DIAGNOSTICS AND THERAPY

Bimonthly Volume 9 Number 4 July 2017

CONTENTS

COMMENTS

The clinical applications of prenatal genetic diagnosis for thalassemia

QIN Danqing HE Tianwen YIN Aihua 223

ORIGINAL ARTICLES

Fast detection of thanatophoric dysplasia type p.R248C mutation hot spots and rapid prenatal diagnosis of three TD type high risk fetuses

JIANG Yu PAN Jingxin GUO Dongwei AI Yang LI Rong JIANG Weiying FANG Qun GUO Yibin 228

The lowest amount of DNA in EGFR gene sequencing by DA8600 sequencing platform

CAO Zhijia ZHANG Jiabin LI Cuiyun WU Yingsong 234

Bioinformatics analysis transcriptional level detection of leucine aminopeptidases from Spirometra mansoni

LI Yiji FU Ruijia ZHOU Xiaojun YIN Feifei LIANG Peng LV Gang LIANG Pei 241

Genetic polymorphism of 23 STR Loci in Jiangmen Han population of Guangdong

ZHANG Jie FENG Dongjiang 247

The analysis of gene spectrum of thalassemia in Chaoshan area Guangdong Province

LIN Bifen YANG Liye XING Shaoy

JOURNAL OF
MOLECULAR DIAGNOSTICS AND THERAPY

Bimonthly Volume 9 Number 4 July 2017

CONTENTS

Study on the prevalence and subtype distributions of human papillomavirus in low income women in Hainan

ZHONG Weida OU Wuying CHEN Yuanhua GUO Hong 267

The clinical comparison of three methods for 17α OH progesterone assay kit

QU Hai LV Mengmeng ZHU Yuhuang FU Guangyu 272

REVIEWS

Analysis of the mechanism of high incidence of thanatophoric dysplasia type

JIANG Yu GUO Dongwei GUO Yibin 278

MiRNA 3p/5p in tumor development

ZHANG Lingyu CHEN Changjie YANG Qingling..... 284

The application of proteomics in lung cancer related biomarkers in data mining

LIANG Weijun QIN Shini YUAN Tianzhu DAI Shengming..... 289

1 2 1 2 1 2

16.83% 6

1

14 332

2 3

40 808

α β
[4]
α α β
β 2
α Hb Bart s
β
β
DNA
[n]
11 14
16 24

1

DNA PCR
DNA α β
β

1.1 Gap polymerase chain reaction Gap PCR
Gap PCR

PCR Gap PCR

Gap PCR
α α$^{3.7}$/ α$^{4.2}$/ ^{SEA}/ ^{THAI}/
[5 6] 2 Hb F
β
$^{G}\gamma^{+}$ $^{A}\gamma\delta\beta^{0}$ HPFH [7 8]

1.2 polymerase chain reaction reverse dot blot PCR RDB
RDB
allele specific oligonucleotide ASO
PCR
DNA
[9]
20
β
17 α 3
[10]

1.3 multiplex ligation dependent probe amplification MLPA

Gap PCR

18 HRMA

α β

human R

Sanger PCR

1.5 amplification refractory mutation system ARMS

RDB

ARMS

PCR allele specific PCR AS PCR

3 ’ DNA

2

2 3 ’

PCR 3 ’

15

PCR

16 17

RDB α β

1.6 high resolution melting analysis HRMA

HRMA

PCR

PCR

5% MCC [25] MCC 2

Chiu [29] Li [30] β

RDB QF PCR RDB

MCC QF PCR STR/SNP [31] α

[23] [SEA]/ STR SNPs

[26] MCC

PCR droplet digital STR SNPs [32 33]

PCR DD PCR MCC Chan [34] β

DD PCR 1.562 5% MCC SNPs

QF PCR 6.25% MCC

MCC DD PCR

MLPA 2.2

SNP

[27] PCR digital PCR D

MLPA α1 α2 PCR DNA SNP

GAP PCR

relative mutation dosage RMD

GAP PCR α MLPA [35]

α β

[36]

2

relative

haplotype dosage RHDO

[36]

NIPT

cffDNA

DNA cell free fetal DNA cffDNA NIPT NIPT

sex detemining region Y SRY X

RH

[28] NIPT

2.1

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• •

p.R248C

TD

1 2 3 1 1 1 4 1

I thanatophoric dysplasia type TD *FGFR3*
“ p.R248C ” restriction endonuelease testing RE
amplification refractory mutation system ARMS /RE 3 TD I
preimplantation genetic diagnosis
PGD p.R248C
“ *Afe* I ” *Apa* LI ARMS /RE
A*fe* I *FGFR3*
exons 6 7 PCR 535 bp p.R248C TD 255 bp
280 bp 2 1~ 3 255 bp 280 bp 535 bp 3 E7 p.R248C
p.R248C TD p.R248C
Apa LI PCR 365 bp 1~ 3 22 bp 343 bp 2

71010025

1.
2.
3. 361102
4.
E mail: aguoabin@qq.com

thanatophoric dysplasia TD

MIM187600

autosomal dominant AD ǰ ʓ ʗ ʯ Ŏ Ŏ Ŏ

- [14]

1/40 000

2
15 —— 4
TD 96 TD
3 3
2011
70
mm 28 245 mm 132 mm 200
mm 24 23 mm 17 20
mm 16 40 mm 211 mm
>0.6 /
<0.89
4
2013
55 mm
200 mm 180 mm 40 mm
39 mm 150 mm
22 4 532±135 g
2014
62 mm 215 mm 187 mm
22 mm 19 mm 218 mm

1.2
1.2.1 DNA DNA
[12,13]
1.2.2 PCR
NM_000142 *FGFR3*
Primer Premier 5.0
1
amplification refractory mutation system ARMS [13]
c.742C>T/p.R248C “ ……GTGCTCCCCGCACCGGCCCATCC…… ”
“ *Apa* LI ” “ GTGCACCCCGCACCGGCCCATCC ” “ GTGCAC ” *Apa* LI G ↓ TGCAC
1
polyacrylamide gel electrophoresis PAGE

1 *FGFR3* exon 7 PCR
Table 1 Common primer and specific primer of exon 7 in *FGFR3* gene and PCR condition

	5′ 3′			bp
FGFR3 E 6 7	F TGCCTCCGCTCACTCACCCG R CCCAAATCCTCACGCAACCC	64	38	535
FGFR3 E7 p.R248C	F CGGCAGGGAGTTCCGCGGCGAG R GGATGGGCCGGTGCGGGGTGCA	65	38	365

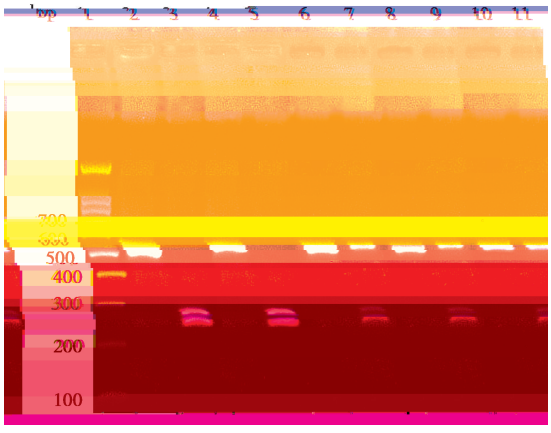
1.2.3 restriction endonuelease tesing
RE
1.2.3.1 PCR *Afe* I
p.R248C TD
1~ 3 PCR 7
μL Buffer 10× 2

1.2.4

ABI PRISM 3730

2

2.1 PCR Afe I 1 lane L L1 100 bp Marker L2 L4 L6 L8 L10 N M p.R248C 1 F1 2 F2 3 F3 535 bp L3 L5 L7 L9 L11 N M F1 F2 F3 *Afe* I p.R248C 255 bp 280 bp 2 L3 L5 p.R248C *Afe* I 255 bp 280 bp 535 bp 3 L7 L9 L11



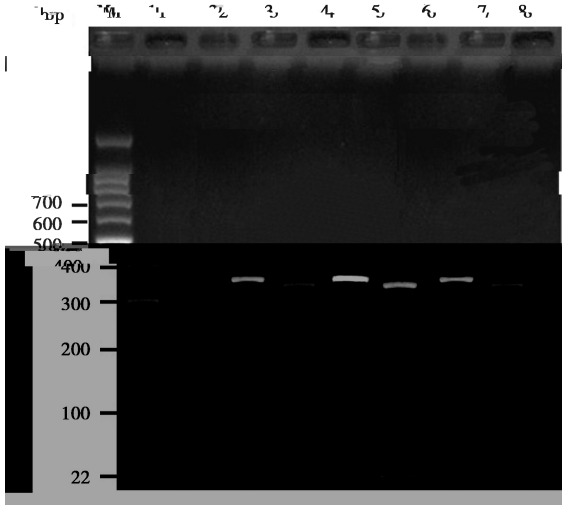
1 *Afe* I p.R248C

Figure 1 Enzyme identification of p.R248C mutation with *Afe* I

2.2 PCR *Apa* LI 2 M 100 bp Marker L1 L2 L4 L6 1 2 3 L3 L5 L7 1 2 3 L8 p.R248C 365 bp 3 22 bp 343 bp 2

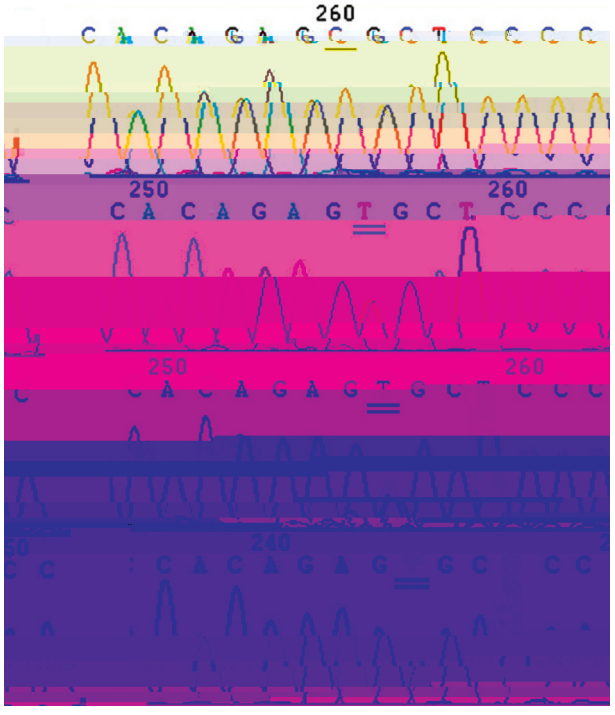
2.3

2.3.1 3 ~ 1 2 3 FGFR3 E 6 7



2 *Apa* LI p.R248C

Figure 2 Enzyme identification of p.R248C mutation with *Apa* LI



~ 1 2 3 FGFR3 E 6 7

3

Figure 3 The partial forward sequence alignment figure of the products amplified by using common primers

C/C ~ 1~ 3 C/T 1~3 *FGFR3* c.742C>T/p.R248C TD I

2.3.2

4 4 ~

C T

C/T T T

A PCR 312 317 bp

Apa LI G ↓ TGCAC “ T ” 3

“ A ”

Apa LI TD I

preimplanta

tion genetic diagnosis PGD

PGD 24 h

RE ARMS/RE

3

TD I AD

TD osteogenesis im

perfecta type OI osteogenesis imper

fecta type OI

achondrogenesis type and type ACG

ACG asphyxiating tho

racic dysplasia ATD

TD I

22 bp 343 bp 2

5~10 min

RE ARMS/RE

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13 . ARMS RE FGFR3 J . 2010 2 1 5 8.

• •

DA8600 *EGFR* DNA

1 1 1 2

DA8600 AmpliSeq *EGFR* DNA next generation sequencing NGS 10 *EGFR* 1 ng 5 ng 10 ng 20 ng DNA DA8600 10 ng 20 ng DNA *EGFR* 5 ng DNA 7 *EGFR* 1 1 *EGFR* 1 1 ng DNA 10 DNA 10 ng DNA AmpliSeq DA8600 *EGFR* DA8600 *EGFR* DNA

The lowest amount of DNA in *EGFR* gene sequencing by DA8600 sequencing platform

CAO Zhijia[1] ZHANG Jiabin[1] LI Cuiyun[1] WU Yingsong[2]

1. Guangzhou Darui Biotechnology Co. LTD Guangzhou Guangdong China

2015B020233009

2015B020233009

1. 510665

2. 510515

E mail wg@smu.edu.cn

DA8600

DNA

H^+ pH

[1 3]

/

[4 5]

EGFR 7p12

EGFR 18~24

EGFR

EGFR

50%

[6 7] *EGFR*

18~21 G719S L861Q

tyrosine kinases in

hibitors TKI [8 9] *EGFR*

EGFR TKI *EG-*

FR

[10]

[11]

EGFR

EGFR

®

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FL 1 1
FL 2 1
FL 3 1
FL 4 1
FL 5 1
FL 6 1
FL 7 1
FL 8 1
FL 9 1
FL 10 1
FL 1 5
FL 2 5
FL 3 5
FL 4 5
FL 5 5
FL 6 5
FL 7 5
FL 8 5
FL 9 5
FL 10 5
FL 1 10
FL 2 10
FL 3 10
FL 4 10
FL 5 10
FL 6 10
FL 7 10
FL 8 10
FL 9 10
FL 10 10
FL 1 20
FL 2 20
FL 3 20
FL 4 20
FL 5

2

2.1 DNA

QIAamp DNA FFPE Tissue KIT 10 DNA 100 μL Qubit®3.0 2

2.2

Ion AmpliSeq™ Library Kit 2.0 1 ng 5 ng 10 ng 20 ng 3

2.3

Qubit® 3.0 4 1 ng DNA Too Low 5 ng DNA 1 Too Low 0.084~0.783 ng/μL 10 ng DNA 0.367~1.14 ng/μL 20 ng DNA 0.707~1.31 ng/μL Too Low 4 0.402 ng/μL

2.4

Variant Caller Coverage Analysis 5 5 ng DNA 9 2 FL 5 5 COSM6224 “Frequency” 2.2% 5% FL 7 5 “ ” 0.2M “ ” 1000 10 ng DNA 10 “ ” 0.2M “ ” 1000 “ ” 5% “ ” 2500 20 ng DNA 10 “ ” 0.2M “ ” 1000 “ ” 5% “ ” 2500

2.5

4

Table 4 Quantitative and mixed the library

	ng/μL	μL
FL 1 1	Too Low	-
FL 2 1	Too Low	-
FL 3 1	Too Low	-
FL 4 1	Too Low	-
FL 5 1	Too Low	-
FL 6 1	Too Low	-
FL 7 1	Too Low	-
FL 8 1	Too Low	-
FL 9 1	Too Low	-
FL 10 1	Too Low	-
FL 1 5	0.599	4.17
FL 2 5	0.350	7.14
FL 3 5	0.084	29.76
FL 4 5	0.553	4.52
FL 5 5	0.783	3.19
FL 6 5	0.156	16.03
FL 7 5	0.208	12.02
FL 8 5	Too Low	-
FL 9 5	0.269	9.29
FL 10 5	0.151	16.56
FL 1 10	0.367	6.81
FL 2 10	1.120	2.23
FL 3 10	0.513	4.87
FL 4 10	0.768	3.26
FL 5 10	0.852	2.93
FL 6 10	1.140	2.19
FL 7 10	0.491	5.09
FL 8 10	1.020	2.45
FL 9 10	0.464	5.39
FL 10 10	0.599	4.17
FL 1 20	1.250	2.00
FL 2 20	0.707	3.54
FL 3 20	1.310	1.91
FL 4 20	0.939	2.66
FL 5 20	1.010	2.48
FL 6 20	1.110	2.25
FL 7 20	0.747	3.35
FL 8 20	0.806	3.10
FL 9 20	0.689	3.63
FL 10 20	1.100	2.27

“ - ”

DNA ng

1

5

10

20

3

2015 “ ” precision medicine initiative PMI “ ” Nature 2015 [12] “ ”

NGS

DA8600 Life Technologies

[13 15] 5 min 160M 10G

[1 2 16] DNA *EGFR* FFPE 10 ng DNA Ampliseq [17] 10 *EGFR* 1 ng 5 ng 10 ng 20 ng [18 20]

1 ng DNA 10 5 ng DNA 10 1 1 1

10 ng DNA 10 20 ng DNA 10

FFPE 10 ng DNA Ampliseq DA8600 *EGFR*

NGS PCR NGS

DA8600 *EGFR* NGS

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26 . PCR

ĩ Ÿ M

1 1 2 1 1 1 1

3 leucine aminopeptidases of *Spirometra mansoni* *Sm*LAPs
3
Real time PCR *Sm*LAPs
*Sm*LAPs *Sm*LAPa *Sm*LAPb *Sm*LAPc 38% *Sm*LAPb *Sm*LAPc 44%
*Sm*LAPs B
*Sm*LAPa 13 *Sm*LAPb 14 *Sm*LAPc 13 T *Sm*LAPa 12 *Sm*LAPb 13 *Sm*LAPc 14 3 *Sm*LAPs
*Sm*LAPb *Sm*LAPa 1.34 *Sm*LAPc *Sm*LAPa 46.53 *Sm*LAPb *Sm*LAPa 1.96 *Sm*LAPc *Sm*LAPa 56.89 *Sm*LAPc *Sm*LAPa *Sm*LAPb 3 *Sm*LAPs *Sm*LAPc

Bioinformatics analysis transcriptional level detection of leucine aminopepti dases from

LI Yiji[1] FU Ruijia[1] ZHOU Xiaojun[2] YIN Feifei[1] LIANG Peng[1] LV Gang[1] LIANG Pei[1]
1. School of Tropical Medicine and Laboratory Medicine, Hainan Medical University Haikou Hainan China 571199 2. Clinical Laboratory Hainan Province People 's Hospital, Haikou Hainan China 570311

[ABSTRACT] Objective To detect transcription levels of 3 leucine aminopeptidases from *Spirometra mansoni* in different developmental stages of the parasite, which lays a foundation for pathogenic diagnosis of further study. Methods Using bioinformatics software, the functional region, immunological characteristics, signal peptide and transmembrane domain of *Sm*LAPs were analyzed. The transcriptional levels of *Sm*LAPs at mature proglottid, gravid proglottid and sparganum stages were detected. Results In the alignment of conserved domains of *Sm*LAPs, the amino acid sequence identities of *Sm*LAPa were 38% compared with *Sm*LAPb and *Sm*LAPc. The identity between *Sm*LAPb and *Sm*LAPc was 44%. Multi functional sites analysis found that *Sm*LAPs contained substrate binding sites, zinc ion binding sites and peptide binding sites. In the prediction of B cell epitopes, *Sm*LAPa and *Sm*LAPc had 13 B cell epitopes, while *Sm*LAPb had 14

81560332 814298

1. 571199
2. 570311
E mail liangpeilp2012@163.com

B cell epitopes. The T cell epitope prediction showed that *Sm*LAPa, *Sm*LAPb and *Sm*

http //www.cbs.dtu.dk/services/ B
T

1.2.2 RNA

EP
0.1% DEPC
121 30 min

15~30 min
1 min 2 min
5 min

RNA

RNA NanoDrop 2000c

1.2.3 cDNA

RNA DNA 5×gDNA Buf
fer 2 μL Total RNA 0.5 μg RNAase Free ddH_2O
10 μL A
42 3 min

10×Fast
RT Buffer 2 μL RT Enzyme Mix 1 μL FQ RT
Primer Mix 2 μL RNase Free ddH_2O 10 μL
Mix gDNA
A
42 15 min 95 3 min
cDNA -80

1.2.4 PCR

*Sm*LAPs PCR 1

DNA club Primer Premier 3

1.2.5 Real time PCR

cDNA

cDNA

SYBR Premix Ex Taq 2× 10.0 μL Forward prim
er 0.2 μL 10 pmol/L Reverse primer 0.2 μL 10
pmol/L cDNA 2.0 μL ddH_2O 7.6 μL
Agilent Technologies strata
gene Mx3005P 3
3
Ct $2^{-\Delta\Delta Ct}$ *Sm*LAPa *Sm*LAPb *Sm*LAPc

[9]

2

2.1 SmLAPs

*Sm*LAPs
*Sm*LAPa *Sm*LAPb *Sm*LAPc
38% *Sm*LAPb *Sm*LAPc
44% 3
*Sm*LAPb
B *Sm*-
LAPa *Sm*LAPc 13 B
*Sm*LAPb 14 B *Sm*LAPa
*Sm*LAPb 8 B *Sm*LAPc
9 B *Sm*LAPb *Sm*LAPc 13
B T *Sm*LA
Pa 12 T *Sm*LAPb 13 T
*Sm*LAPc 14 T 3 *Sm*LAPs

1 *Sm*LAPs

Table 1 Primer sequences of *Sm*LAPs for real time PCR

SmLAPa	5′ TAGCGTTAGCTGCGTTTGCC 3′
	5′ GAGCCCAGTTCCCACATCGT 3′
SmLAPb	5′ AGTACGATGTTCTCGGAATTGTTG 3′
	5′ CAGTTTATGGTAGTATGCCTTGACG 3′
SmLAPc	5′ ACGGACGAGGAGCGAATGAC 3′
	5′ ACAGCACGGTTGACGGAAGC 3′
Smβ actin	5′ CATCTACGAGGGTTACGCACTG 3′
	5′ GCTCATCTCCTGCTCAAAGTCC 3′

2 T 2

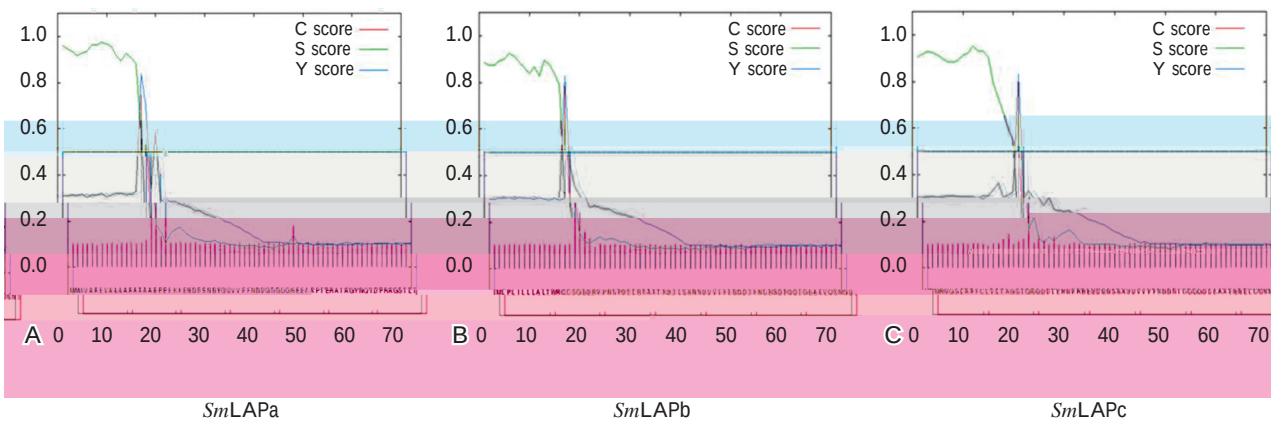
JAP_ *Sm*LAPa *Sm*LAPc 2

*Sm*LAPa *Sm*LAPb 4

2.2 SmLAPs

amino 3 *Sm*LAPs

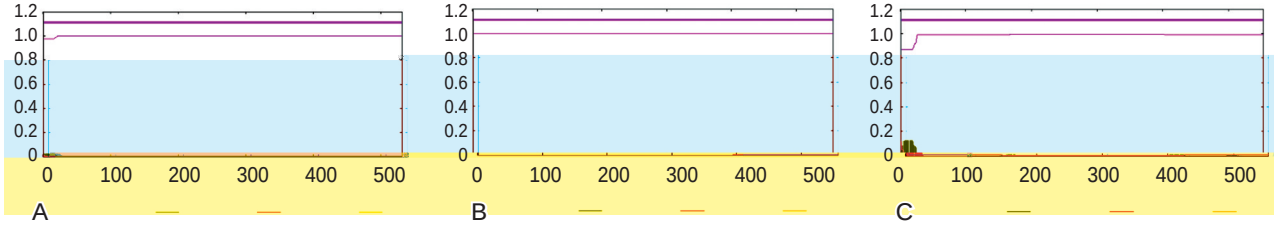
LAPc 18 amino acid aa *Sm*LAPb 1



A SmLAPa B SmLAPb C SmLAPc

2

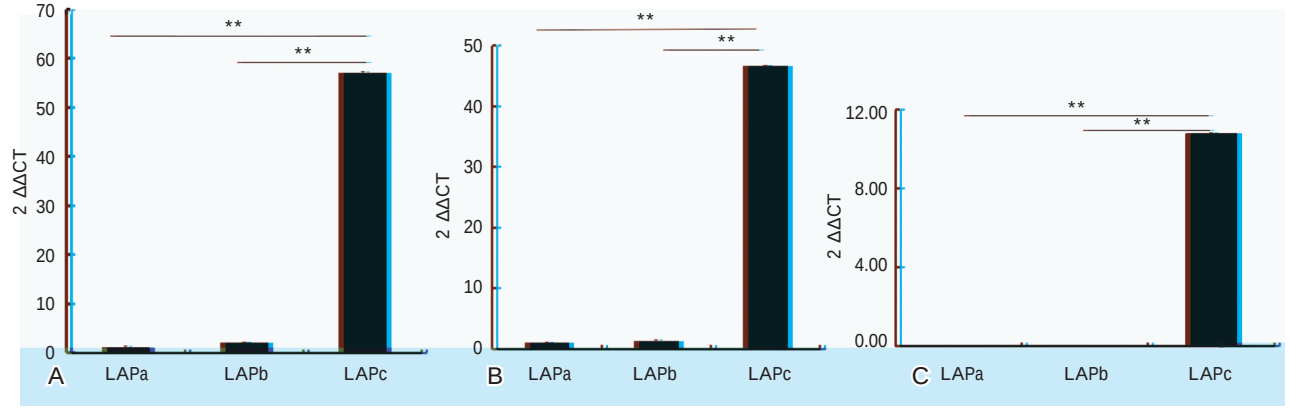
Figure 2 Signal peptide pepdiction of *Sm*LAPs sequences



A SmLAPa B SmLAPb C SmLAPc

3

Figure 3 Transmembrane helical prediction of *Sm*LAPs sequences



A 3 SmLAPs B 3 SmLAPs C 3 SmLAPs

4 *Sm*LAPs

Figure 4 Transcriptional level detection of *Sm*LAPs

B

13

3

3 14

330 kDa

11

enzyme linked immunosorbent 15 *Sm*LAPs

assay ELISA ¿ Ş η

12

*Sm*LAPs

Real time PCR 3

*Sm*LAPc

*Sm*LAPa

*Sm*LAPb *Sm*LAPc

*Sm*LAPc

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23 STR

1 2

23 STR
475 Huaxia™ Platinum
PCR DNA 23 23
STR 0.001 1~0.586 3 H 0.534 7~0.884 2
MP 0.015 8~0.233 7 DP 0.984 2~0.766 3
PE 0.219 7~0.763 3 CMP $3.158\ 4 \times 10^{-28}$
TDP 1 $3.158\ 4\times10^{28}$ CPE 1 $3.143\ 9\times10^{10}$ 23
STR
STR
STR

Genetic polymorphism of 23 STR Loci in Jiangmen Han population of Guang dong

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1. 510635
2. 512000
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short tandem repeats

SRT DNA micro satellite DNA

[1,2] STR

heterozygosity H match probability MP discrimination power DP probability of exclusion PE polymorphism information content PIC

23

Huaxia™ Platinum PCR STR

1

1.1 DNA

475

Chelex 100 DNA [4]

1.2 STR

Huaxia™ Platinum PCR Thermo Scientific 23 STR D1S1656 D2S441 D2S1338 D3S1358

1 23 STR n=475

Table 1 Allele frequencies of 23 STR loci in Jiangmen population of Guangdong n=475

CSF1PO			D1S1656			D2S441			D2S1338			D3S1358		
A	n	F	A	n	F	A	n	F	A	n	F	A	n	F
7	4	0.004 2	10	1	0.001 1	9	1	0.001 1	16	30	0.031 6	12	1	0.001 1
8	1	0.001 1	11	84	0.088 4	9.1	12	0.012 6	17	69	0.072 6	13	2	0.002 1
9	36	0.037 9	12	36	0.037 9	10	211	0.222 1	18	79	0.083 2	14	43	0.045 3
10	195	0.205 3	13	97	0.102 1	10.1	5	0.005 3	19	186	0.195 8	15	283	0.297 9
11	267	0.281 1	14	88	0.092 6	11	283	0.297 9	20	109	0.114 7	16	311	0.327 4
12	366	0.385 3	15	274	0.288 4	11.3	93	0.097 9	21	28	0.029 5	17	255	0.268 4
13	70	0.073 7	15.3	2	0.002 1	12	188	0.197 9	22	50	0.052 6	18	47	0.049 5
14	10	0.010 5	16	191	0.201 1	13	21	0.022 1	23	177	0.186 3	19	7	0.007 4
15	1	0.001 1	16.3	6	0.006 3	14	130	0.136 8	24	150	0.157 9	20	1	0.001 1
			17	70	0.073 7	14.1	1	0.001 1	25	64	0.067 4			
			17.3	57	0.060 0	15	3	0.003 2	26	6	0.006 3			
			18	9	0.009 5	16	2	0.002 1	28	2	0.002 1			
			18.3	32	0.033 7									
			19	1	0.001 1									
			19.3	2	0.002 1									

D5S818			D6S1043			D7S820			D8S1179			D10S1248		
A	n	F	A	n	F	A	n	F	A	n	F	A	n	F
7	39	0.041 1	9	3	0.003 2	7	6	0.006 3	10	156	0.164 2	8	3	0.003 2
8	1	0.001 1	10	35	0.036 8	8	140	0.147 4	11	134	0.141 1	11	6	0.006 3
9	69	0.072 6	11	111	0.116 8	9	56	0.058 9	12	108	0.113 7	12	67	0.070 5
10	203	0.213 7	12	143	0.150 5	9.1	7	0.007 4	13	157	0.165 3	13	326	0.343 2
11	266	0.280 0	13	108	0.113 7	10	172	0.181 1	14	142	0.149 5	14	217	0.228 4
12	218	0.229 5	14	125	0.131 6	11	321	0.337 9	15	166	0.174 7	15	220	0.231 6
13	145	0.152 6	15	16	0.016 8	12	211	0.222 1	16	66	0.069 5	16	87	0.091 6
14	9	0.009 5	16	4	0.004 2	13	33	0.034 7	17	18	0.018 9	17	22	0.023 2
			17	28	0.029 5	14	4	0.004 2	18	2	0.002 1	18	2	0.002 1
			18	145	0.152 6				19	1	0.001 1			
			18.2	1	0.001 1									
			19	156	0.164 2									
			20	66	0.069 5									
			21	7	0.007 4									
			21.3	1	0.001 1									
			22	1	0.001 1									

D12S391			D13S317			D16S539			D18S51			D19S433		
A	*n*	F	A	*n*	F	A	*n*	F	A	*n*	F	A	*n*	F
15	19	0.020 0	5	2	0.002 1	8	1	0.001 1	11	4	0.004 2	11	5	0.005 3
16	4	0.004 2	6	2	0.002 1	9	219	0.230 5	12	43	0.045 3	12	39	0.041 1
17	62	0.065 3	8	304	0.320 0	10	130	0.136 8	13	166	0.174 7	12.2	1	0.001 1
18	206	0.216 8	9	126	0.132 6	11	286	0.301 1	14	180	0.189 5	13	269	0.283 2
19	178	0.187 4	10	148	0.155 8	12	217	0.228 4	15	193	0.203 2	13.2	38	0.040 0
19.3	1	0.001 1	11	219	0.230 5	13	82	0.086 3	16	138	0.145 3	14	248	0.261 1
20	160	0.168 4	12	120	0.126 3	14	13	0.013 7	17	87	0.091 6	14.2	102	0.107 4
21	102	0.107 4	13	24	0.025 3	15	2	0.002 1	18	55	0.057 9	15	59	0.062 1
22	118	0.124 2	14	4	0.004 2				19	24	0.025 3	15.2	126	0.132 6
23	50	0.052 6	15	1	0.001 1				20	19	0.020 0	16	14	0.014 7
24	35	0.036 8							21	16	0.016 8	16.2	43	0.045 3
25	13	0.013 7							22	14	0.014 7	17	1	0.001 1
26	2	0.002 1							23	7	0.007 4	17.2	5	0.005 3
									24	4	0.004 2			

D21S11			D22S1045			FGA			Penta D			Penta E		
A	*n*	F	A	*n*	F	A	*n*	F	A	*n*	F	A	*n*	F
26	2	0.002 1	11	193	0.203 2	13	6	0.006 3	6	2	0.002 1	5	56	0.058 9
27	1	0.001 1	12	1	0.001 1	16	1	0.001 1	7	12	0.012 6	8	1	0.001 1
28	45	0.047 4	13	5	0.005 3	18	31	0.032 6	8	64	0.067 4	9	8	0.008 4
28.2	1	0.001 1	14	42	0.044 2	19	78	0.082 1	9	371	0.390 5	10	54	0.056 8
29	256	0.269 5	15	292	0.307 4	20	44	0.046 3	10	118	0.124 2	11	182	0.191 6
29.2	2	0.002 1	16	188	0.197 9	20.2	1	0.001 1	11	110	0.115 8	12	112	0.117 9
30	262	0.275 8	17	201	0.211 6	21	114	0.120 0	12	130	0.136 8	13	57	0.060 0
30.2	7	0.007 4	18	27	0.028 4	21.2	3	0.003 2	13	94	0.098 9	14	75	0.078 9
30.3	4	0.004 2	19	1	0.001 1	22	176	0.185 3	14	45	0.047 4	15	74	0.077 9
31	72	0.075 8				22.2	4	0.004 2	15	4	0.004 2	16	46	0.048 4
31.2	65	0.068 4				23	165	0.173 7				17	63	0.066 3
32	26	0.027 4				23.1	1	0.001 1				18	58	0.061 1
32.2	153	0.161 1				23.2	15	0.015 8				19	53	0.055 8
33	5	0.005 3				24	153	0.161 1				19.4	1	0.001 1
33.2	47	0.049 5				24.2	6	0.006 3				20	45	0.047 4
34	1	0.001 1				24.3	1	0.001 1				21	27	0.028 4
34.2	1	0.001 1				25	93	0.097 9				22	17	0.017 9
						25.2	2	0.002 1				23	12	0.012 6
						26	39	0.041 1				24	3	0.003 2
						26.2	3	0.003 2				25	3	0.003 2
						27	9	0.009 5				26	3	0.003 2
						27.2	2	0.002 1						
						28	3	0.003 2						

TH01			TPOX			Vwa		
A	*n*	F	A	*n*	F	A	*n*	F
6	132	0.138 9	7	1	0.001 1	14	261	0.274 7
7	272	0.286 3	8	557	0.586 3	15	34	0.035 8
8	40	0.042 1	9	72	0.075 8	16	135	0.142 1
9	428	0.450 5	10	36	0.037 9	17	230	0.242 1
9.3	26	0.027 4	11	257	0.270 5	18	193	0.203 2
10	52	0.054 7	12	27	0.028 4	19	81	0.085 3
						20	14	0.014 7
						21	2	0.002 1

A F *n*

3

STR

[1]

Thermo Scientific Huaxia ™ Platinum PCR 25 23 STR Amelogenin M Yindel 23 D3S1358 CSF1PO TPOX TH01 19 DP 0.9 H 0.7 PIC 0.7 [7 8] D3S1358 CSF1PO TPOX TH01 4 [9 12] D6S1043 FGA Penta E 3

D6S1043 H 0.896 8 DP 0.969 7 PIC 0.862 8 Penta E H 0.884 2 DP 0.984 2 PIC 0.903 1 D6S1043 Penta E 2 2

[9 13] Penta E D6S1043 Penta E STR

STR

combined paternity index CPI Huaxia ™ Platinum PCR PowerPlex®16 Identifiler® Plus

CPE 1 3.143 9×10 [10] [9] [10] 19 21 CPE

23

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Gap polymerase chain reaction Gap PCR

reverse dot blot RDB PCR+ 4

2 057 14 α 19 β

α β 13 α SEA β

IVS 654M 41/42M

The analysis of gene spectrum of thalassemia in Chaoshan area Guangdong Province

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[ABSTRACT] Objective To investigate the type of gene mutation of thalassemia and constituent ratio in the Chaoshan region, and provide the theoretical basis for gene diagnosis of thalassemia. Methods Thalassemia genotypes of Chaoshan area (including Shantou, Chaozhou, Jieyang, Shanwei) were identified by Gap PCR reverse dot blot (RDB), PCR and flow through hybridization. Results Among the 2 057 samples which were confirmed for thalassemia, 14 types of α thalassemia (α thal) alleles and 19 types of β thalassemia (β thal) alleles were identified. Moreover, 13 kinds of compound α thal and β thal were found. The most common α thal was SEA and the most common β thalassemia was IVS 654M, followed by 41/42M. Conclusion The gene types for thalassemia were diversely distributed in the Chaoshan region. The results would provide reference data for diagnosis and research of thalassemia in this area.

[KEY WORDS] Thalassemia; genetype; Chaoshan region

1 2

521021

E mail yangleeyee@sina.com

11

1 000 [3]

10 918.5

1 500

[4 5] 4

2 057

1

1.1

4

70

1 441

ς C 8 ű ⱪ ξ

1

Table 1 The genotype and composition ratio of thalassemia in Chaoshan region

α
SEA
$\alpha^{3.7}$
$\alpha^{4.2}$
$\alpha^{CS}\alpha$
$\alpha^{QS}\alpha$
$\alpha^{WS}\alpha$
$\alpha^{3.7}/\ \alpha^{3.7}$
$\alpha^{3.7}/\ \alpha^{4.2}$
$\alpha^{4.2}/$ SEA
$\alpha^{3.7}/$ SEA
$\alpha^{QS}\alpha/$ SEA
$\alpha^{QS}\alpha/$ SEA
$\alpha^{WS}\alpha/$ SEA
$\alpha^{3.7}/\alpha^{QS}\alpha$
42
MfS II
β
17M
28M
B
41/42M
IVS II 654M
CAPM
29M
71/72M
27 28M
β^{EM}
IntM
IVS I 5M
43M
41 41M/17M
41 42M/ IVS II 654M
41 42M/ 28M
M
IVS II
41 42M/β^{EM}
IVS II 654M /654

B2
454

B B4 ç ů

2 %

α							β							
SEA	$\alpha^{3.7}$	$\alpha^{4.2}$	$\alpha^{CS}\alpha$	$\alpha^{QS}\alpha$	$\alpha^{WS}\alpha$		17M	28M	41/42M	IVS II 654M	29M	71/72M	β^{EM}	
52.73	30.91	9.09	3.64	3.64	-	-	13.33	13.33	20.00	46.67	-	-	-	6.67
76.53	10.38	6.69	0.78	0.29	-	5.33	10.65	10.65	30.39	40.26	0.26	1.56	0.78	5.45
62.71	25.99	6.78	-	1.69	1.13	1.70	9.68	14.52	41.94	19.35	3.23	3.23	6.45	1.60
81.41	7.04	4.52	1.01	2.01	-	4.01	8.08	9.09	31.31	43.43	3.03	1.01	3.03	1.02

“ ” 0

3

41/42M 17M IVS II 654M
41/42M IVS II 654M [8 9]

5 SEA

SEA Hb Bart s

3

α

11

β 7

β^0/β^0

41 41M/17M β^+/β^+ IVS II 654M/ 28M IVS II 654M/ IVS II 654M β^0/β^+ 41 41M/IVS II 654M 41 41M/ 28M IVS II 654M/17M

β

α

SEA $\alpha^{3.7}$ 7

β

IVS II 654M 41/42M 41/42M 28M [7] β

β

[10 11] β

41/42M [12]

β 41/42M [13 14]

41/42M 42.47% [8]

β

IVS II 654M [10 12] Lin [6]

IVS II 654M CD41/42M

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gap PCR ^{SEA}/

αα $\alpha^{3.7}$/αα $\alpha^{4.2}$/αα 3 α $\alpha^{QS}\alpha$ $\alpha^{CS}\alpha$ $\alpha^{WS}\alpha$ 3

α β 17 29 28 17 βE 41 42 43 71 72

654 Int 14 15 27/28 1 5 31 30 CAP 32 α 1 359

(490 ь 36.06% 490/1359 ^{SEA}/αα

2015AB000532

528200

E mail xynan718@163.com

[1]

2.5%

20%

15.20% [2] 21.09% [3]

2/5

[4]

2015

1

1.1

2015 1 2015 12

mean corpuscular volum MCV <82 fL

mean corpuscular hemoglobin MCH <27 pg

1.2

2 mL EDTA K2

DNA

4 7 d

1.3

α gap PCR ^{SEA}/αα $\alpha^{3.7}$/αα $\alpha^{4.2}$/αα 3

α

$\alpha^{CS}\alpha$ $\alpha^{QS}\alpha$ $\alpha^{WS}\alpha$ 3

β

β 17

29 28 17 βE 41 42 43 71 72 654 Int 14 15 27/28 1 5 31 30 CAP 32

1.4

1.5

1.5.1 α

1 826 bp

2

1 826 bp

1.5.2 α

3 QSN CSN WSN

1.5.3 β

7 41 42N 654N 28N 71 72N 17N βEN 31N

1.5.4 α β

α / α β

2

2.1 α α β

α 1 359 α

Ư 1 210 β

Ư1 073 1

2.2 α

490 α

$\alpha^{4.2}$/αα HKαα 2

2.3 α

35 α

$\alpha^{CS}\alpha$ $\alpha^{QS}\alpha$ $\alpha^{WS}\alpha$ 3

3

2.4 β

268 ç

1 α α β

Table 1 The detection of α thalassemia α thalassemia non missing β thalassemia

				%
α	1 359	490	36.06	61.79% 490/793
α	1 210	35	2.89	4.41% 35/793
β	1 073	268	24.98	33.80% 268/793
	3 642	793	63.93	100.00%

2 490 α

Table 2 The detection of 490 cases of α thalassemia gene deletion type

		%
^{SEA}/αα	333	67.96
$\alpha^{3.7}$/αα	84	17.15
$\alpha^{4.2}$/αα	45	9.19
^{SEA}/ $\alpha^{3.7}$	16	3.27
^{SEA}/ $\alpha^{4.2}$	6	1.22
$\alpha^{3.7}$/ $\alpha^{4.2}$	1	0.20
$\alpha^{3.7}$/ $\alpha^{3.7}$	2	0.41
$\alpha^{4.2}$/ $\alpha^{4.2}$	1	0.20
^{SEA}/ ^{SEA}	1	0.20
HKαα	1	0.20
	490	100.00

α α 4 α β 35

3 35 α

Table 3 The detection of 35 cases of α thalassemia non missing gene deletion type

		%
α^{QS}α/αα	12	34.29
α^{CS}α/αα	10	28.57
α^{WS}α/αα	12	34.29
α^{WS}α/α^{WS}α	1	2.85
	35	100.00

α α 4 α β 2

α / α

[5]

α β δβ δ

α β

α α

2

3 α ^{SEA}/αα $\alpha^{3.7}$/αα $\alpha^{4.2}$/αα 3 α

4 268 β

Table 4 The detection of 268 cases of β thalassemia gene deletion type

		%
β^{41-42}/β^{N}	119	44.40
β^{654}/β^{N}	51	19.03
β^{17}/β^{N}	34	12.69
β^{-28}/β^{N}	30	11.19
$\beta^{\beta E}/\beta^{N}$	12	4.48
β^{71-72}/β^{N}	7	2.61
β^{43}/β^{N}	7	2.61
β^{-29}/β^{N}	3	1.12
$\beta^{IVS-I-1}/\beta^{N}$	2	0.75
$\beta^{14/15}/\beta^{N}$	2	0.75
$\beta^{27/28}/\beta^{N}$	1	0.37
	268	100.00

β α / 37

α^{CS}α α^{QS}α α^{WS}α α 525 490 35

α ^{SEA}/αα 67.96% $\alpha^{3.7}$/αα 17.14% $\alpha^{4.2}$/αα 9.18% 3 94.28%

[6 7]

α α^{QS}α/αα 34.29% α^{WS}α/αα 28.57% α^{CS}α/αα 34.29% 3

[8]

HKαα

HKαα $\alpha^{3.7}$ αα$\alpha^{anti4.2}$ HKαα/αα $\alpha^{3.7}$/αα [9]

β β

β 200 48 46 [5]

β 11 β $\beta^{41\ 42}$/

βN 44.40% β654/βN 19.03% β17/βN 12.69% β 28/
βN 11.19% 4 β
87.31%
2
β β654/βN
41.13% β41 42/βN 29.00% β17/βN 13.42%
3 β β41 42/βN
46.89% β17/βN 20.20% β654/βN 14.97%
β

1 β
CD17
MCV
MCV
α3.7/αα α4.2/αα
α/ β
14
15 1 514
α 3.04%
3.7 4.2 WS 3
16 1 392
5.39%
α

2007
2016 1
MCV<82 fL
MCH<27 pg
α
β

6
α
β α

1 .

invasive cribriform carcinoma ICC
ductal carcinoma in situ DCIS 12 12

CK5/6 CK 34βE12 P63 Calponin ER PR HER2 E cad Ki67 ICC
DCIS ICC
ICC
DCIS ICC DCIS

Comparative analysis based on clinicopathology and mmunohistochemistry of invasive cribriform carcinoma of the breast and cribriform structuer of ductal carcinoma in situ

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[ABSTRACT] Objective To investigate the clinicopathologic characteristics and the immunophenotyp ic markers of invasive cribriform carcinoma (ICC) in comparison to cribriform structuer of ductal carcinoma in situ(DCIS). Methods 12 cases of ICC and 12 cases of cribriform structure of ductal carcinoma in situ were collected for this analysis, in which the morphologic features and immunohistochemical expressions were mea sured and compared. The antibody CK5/6, CK(34βE12), P63, Calponin, ER, PR, HER2, E cad, and Ki67 were included in this analysis. Results The median age of onset of ICC was much older referring to the data of the involved cases. The ICC patients were found with palpable tumors, whereas axillary lymph node metastases were rare and the prognoses were satisfied. All cases showed similar cribriform structure in microscopic exami nations, the immunostaining for myoepithelial cells clear expressed negative to 12 ICC patients, but showed posi tive in 12 cases of cribriform structure of ductal carcinoma in situ. Conclusions The histological features of ICC were similar to those found in cribriform structure of ductal carcinoma in situ, the immunohistochemistry was of great significance in diagnosing ICC from the other.

[KEY WORDS] Breast; Pathology; Invasive cribriform carcinoma of the breast; Cribriform structuer; Ductal carcinoma in situ; Immunohistochemistry

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[1]
invasive cribriform carcinoma ICC
ductal carcinoma in situ DCIS
[2]
50% ICC
10% 49%
ICC DCIS

12 12

1

1.1

2005 1 2015 1

12 - hematoxylin eosin staining HE 12
6

27 74
54 12

1.2

10%
3 μm HE
CK5/6 CK 34βE12 P63 Calponin ER PR HER2 Ki67 E cad ICC
12 fluorescence *in sits* *ER*

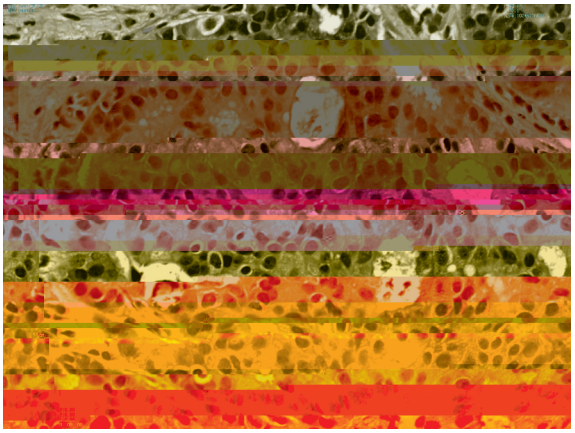
2 DCIS

Table 2 Clinical data of cribriform structure of ductal carcinoma in situ

			(cm)	
*1	30	1/4	0.8	3a
*2	28	1	2.2	3a
*3	36	1/2	3.0	4a
*4	62	6	1.5	4a
*5	44	14	1.0	3b
*6	43	24	2.0	3b
*7	32	1/4	0.6	3a
*8	41	1	2.0	3a
*9	26	1/2	1.2	3a
*10	24	6	1.5	3b
*11	36	14	1.0	3b
*12	43	24	2.0	3a

1 ICC HE ×200

Figure 1 Microscopic morphology of ICC HE ×200



2 ICC HE ×400

Figure 2 Microscopic morphology of ICC HE ×400

P=0.028 P 1

0.127

2.2

2.2.1 2

6

ICC 6

3

2.2.2

2.2.3 100% Ki67 20% ER PR 3 4 ICC 100% HER2 3+ CK5/6 P63 3 Cal FISH ponin CK 34βE12 83% ER 100% 4 PR 91% HER2 0 2+ FISH ICC HER2 1 Ki67 Page [3] 1983 ICC DCIS CK5/6 0.3% 0.8% 4% P63 Calponin CK 34βE12 53 58 [4] [5]

	CK5/6	CK(34βE12)	P63	Calponin	ER
ICC 1		+			+
ICC 2		+			+
ICC 3		+			+
ICC 4					
ICC 5		+			
ICC 6		+			
ICC 7					
ICC 8		+			
ICC 9		+			
ICC 10		+			
ICC 11		+			
ICC 12		+			

ICC

[13]

ICC ICC

>50 ICC 5

100 50%

ICC 88% [2] ICC 14.13

3 [14]

E cad

[15] Saleh [16]

CD44vB v6

v4

ICC DCIS

2

ER PR

ICC DCIS

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1 2 1 1

human papillomavirus HPV
2016 1 2016 12 15
1 373 935 438
21 HPV 1 373
HPV 192 13.98 192/1 373 19 HPV HPV 5
HPV52 3.35 46/1 373 HPV51 1.97 27/1 373 HPV58 1.97 27/1 373
HPVcp8304 1.75 24/1 373 HPV53 1.60 22/1 373 HPV
P<0.05 HPV 32.35% 11/34 24.63% 33/
134 HPV HPV 14.12 132/
803 13.70 60/378 P 0.05
HPV HPV HPV

Study on the prevalence and subtype distributions of human papillomavirus in low income women in Hainan

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[ABSTRACT] Objective To evaluate the prevalence and subtype distributions of human papillomavirus (HPV) in low income women in Hainan. Methods 1 373 low income women from 15 cities of Hainan were organized to conduct a physical examination by the Hainan Provincial Federation of trade unions and the city and county trade unions from January 2016 to December 2016. Among them, 935 people were of Han nationality and 438 were of non Han nationality. The prevalence of 21 subtypes of HPV in the cervical cells of these women was detected using flow through hybridization and gene chip technology. Results Among the 1 373 women studied, HPV infection was found in 192 cases, with a total infection rate of 13.98% (192 / 1 373). 19 subtypes of HPV were found in these samples, and the top 5 subtypes of HPV infection from high to low were HPV 52 (3.35%, 46/1 373), HPV 51 (1.97%, 27/1 373), HPV 58 (1.97%, 27/1 373), HPV cp8304 (1.75%, 24/1 373) and HPV 53 (1.60%, 22/1 373). There is a significant difference in the infection rate of HPV in different cities and counties (P 0.05). The highest rate of HPV infection was found in Haikou with an infection rate of 32.35% (11/34), followed by Changjiang with an infection rate of 24.63% (33/134). The dominant subtype of HPV infection was also different among different cities and counties. No significant

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difference was found in the infection rate between women of Han nationality 14.12 (132/803) and non Han nationality 13.70 (60/378) (P 0.05). Conclusion Compared to the domestic general population, the prevalence of HPV infection in low income women in Hainan province is relatively low, and the infection rate of HPV and the distribution of infected subtypes vary in different cities and counties. So the goal will be to strengthen the prevention and control of cervical cancer in the area with high HPV prevalence.

[KEY WORDS] Low income women; Subtype; Papillomavirus; Cervical tumor

2

80

[1 2]

human papillomavirus HPV

90%

HPV HPV

[3 4]

HPV HPV

HPV [5 6]

HPV

HPV

2016 1

2016 12

1 373 HPV

1

1.1

2016 1 2016 12

15

1 373

100

23 60

935 438

1.2

3

HPV DNA

1.3

HPV

HybriMax

16 18 31 33 35 39 45

5l 52 56 58 59 66 68 6 11

42 43 44 53 81 CP8304 21 HPV

HPV

HPV 2 HPV

HPV HPV

HPV HPV

HPV HPV

1.4

SPSS 21.0

HPV HPV

F P<0.05

2

2.1 HPV

15 1 373 HPV

192 13.98 192/1 373

144 75 48 25

19 HPV HPV 5

HPV52 3.35 HPV51

1.97 HPV58 1.97 HPV cp8304 1.75

HPV53 1.60 HPV

HPV52 3.35 HPV51 1.97 HPV58

1.97 HPV18 1.53 HPV39 1.46

HPV 16 1.38 HPV68 1.02 HPV66 0.80

HPV33 0.44 31 0.29 HPV45 0.15

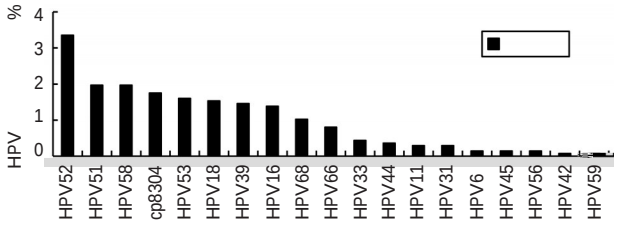
HPV56 0.15 HPV59 0.07

HPV43 HPV35 HPV

HPVcp8304 1.75 HPV53 1.60

HPV44 0.36 HPV11 0.29 HPV6 0.15

HPV42 0.07 1



1 HPV

Figure 1 The prevalence of HPV subtype in low income women in Hainan

HPV 32.35% 11/34 24.63% 33/134 HPV P 0.05 HPV 14.12% 132/935 13.70% 60/438 P 0.05 1 4 1 HPV52

2.2 HPV HPVcp8304 HPV

1 HPV

Table 1 The dominant subtypes of HPV in women from different cities and ethnic populations

/		HPV	P	HPV			
		n %		%	%		%
	935	132 14.12	0.835	52 3.42	51 2.14	58 1.71	53 1.60
	438	60 13.7		52 3.20	58 2.51	cp8304 2.05	53 1.60
	94	10 10.64	0.003	cp8304 3.19	16 2.13	66 1.06	11 1.06
	134	33 24.63		52 4.69	58 4.69	18 2.34	53 2.34
	59	5 8.47		52 3.39	68 1.69	33 1.69	cp8304 1.69
	70	7 10.00		52 5.71	58 1.43	16 1.43	51 1.43
	49	5 10.20		52 2.04	58 2.04	18 2.04	51 2.04
	151	24 15.89		18 4.64	51 3.97	58 2.65	cp8304 2.65
	34	11 32.35		52 8.82	39 8.82	68 5.88	53 5.88
	131	20 15.27		51 4.58	39 3.05	16 2.29	53 2.29
	72	6 8.33		52 1.39	53 1.39	66 1.39	11 1.39
	25	3 12.00		cp8304 8.0	56 4.0	68 4.0	53 4.0
	191	26 13.61		52 5.76	51 2.09	16 1.57	53 1.57
	87	10 11.49		53 3.45	51 2.30	39 2.30	cp8304 2.30
	76	11 14.47		52 3.95	16 2.63	53 2.63	39 2.63
	54	7 12.96		52 7.41	51 3.70	18 3.70	39 3.70
	146	14 9.59		58 3.42	52 2.74	18 2.05	53 1.73

2.3 HPV HPV 2 1 373 44 23 60 30 39 3 30 9.09% 15 30 HPV HPV P>0.05 13.98 192/1 373 HPV

2 HPV

Table 2 The age distribution of HPV positive women

	n	*n*	%	*F*	*P*	%	%	%
30	20	2	9.09	-	-	9.09	0	0
30~39	262	46	14.94	0.564	0.453	12.01	2.60	1.30
40~49	801	128	13.78	0.400	0.527	11.19	2.58	1.61
49	98	16	14.04	0.393	0.531	11.40	2.63	1.75

HPV52 HPV51 HPV58 HPVcp8304 HPV53

[7]

[8]

HPV 16.1

HPV52 HPV58

[9 11] [12]

HPV

HPV

HPV HPV

HPV [5]

HPV

15 HPV

32.35% %

USA the UK and Australia an international survey J . Infections Sexually Transmitted 2014 90 3 201 207.

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• •

17α

1 2 1 1

3 17α
2015 2 10 120 120
3 17α
3 R^2 0.82 0.76 0.77
100% 100% 99.2% 97.5% 98.3% 98.3% 98.8% 99.2%
98.8% 3 P>0.05 3 17α
17α

The clinical comparison of three methods for 17α OH progesterone assay kit

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[ABSTRACT] Objective To compare the correlation and consistency of results of 17α OH progesterone assay kits from 3 different manufactures. Methods From February to October 2015, 17α OH progesterone in serum samples collected from 120 patients with congenital adrenal hyperplasia and 120 healthy subjects were detected by 3 different manufactures. The results of 17α OH progesterone were statistically analyzed. Results The R^2 of results detected by the 3 kits were 0.82, 0.76 and 0.77. The positive coincident rate were 100%, 100% and 99.2%. The negative coincident rate were 97.5%, 98.3% and 98.3%. The total coincident rate were 98.8%, 99.2% and 98.8%. The value of 3 kits were without statistical difference (P>0.05). Conclusion The performance of the 3 kinds of 17α OH progesterone assay kit might be equivalent, all of which could meet the requirements of clinical application.

[KEY WORDS] 17α OH progesterone; Immuno radiametric assay(RIA); Enzyme linked immunosorbent assay (ELISA); Chemiluminescent microparticle immunoassay (CMIA)

congenital adrenal CAH 90% ~ 95% [2] CAH
hyperplasia CAH
nonclassic 21 hydroxylase deficiency NCCAH
[1] 21 17 10/10
3 11 21 l0
21 hydroxylase deficiency 21 OHD [3]

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21

17α 17α OH progesterone 17α OHP
17α OHP CAH
NCCAH

17α OHP CAH

17α OHP

3

1 3

Table 1 The test procedure of 3 kits

	min
A	60
B	30
C	100

3 3

Table 3 The expected normal values of 3 kits

		A ng/mL	B ng/mL	C ng/mL
		0.61 3.34	0.31 2.01	0.5 2.1
		0.40 1.02	0.05 1.02	0.1 0.8
		1.26 4.28	0.3 2.34	0.6 2.3
		0.14 1.11	0.1 1.4	0.3 1.4
	Post ACTH			< 3.2
		0.23 1.36	< 0.93	0.13 0.51
			2.28 9.24	2.0 12
1 13		0.07 1.53	< 2.32	80 0.01 1.7
1 1		1.06 40.41	0.82 16.63	0 16.8

P

9 3

Table 9 The result of variation sample in 3 kits

A		B		C		
ng/mL		ng/mL		ng/mL		
15.88	+	24.72	+	24.54	+	+
3.80	+	10.21	+	7.50	+	+
14.71	+	12.78	+	26.45	+	+
22.86	+	12.08	+	13.42	+	+
3.69	+	6.77	+	4.28	+	+
2.66	-	2.65	-	0.31	-	-
0.13	-	0.29	-	3.35	+	-
7.12	+	12.03	+	4.20	+	+
4.85	+	7.57	+	5.60	+	+
6.73	+	13.08	+	7.69	+	+
17.92	+	25.69	+	17.65	+	+
21.24	+	28.43	+	18.33	+	+
1.50	-	5.40	+	9.42	+	-

3

CAH

CAH

1977 17α OHP

21 [5] 3

NCCAH

[6 7] 17α

OHP NCCAH

[8 9]

17α OHP

21

[10 11] 17α OHP

17α 3

OHP 3

>98% 3

R^2 0.9

[12]

3

[13]

17α OHP

[14 15]

QUEST

Labcorp

3

17α OHP

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1 2 1

thanatophoric dysplasia TD
TD I TD II 2 *FGFR3* TD I
c.742C>T/p.R248C autosomal dominant AD
p.R248C
p.R248C
“ *FGFR3* Fibroblast growth factor receptor 3 FGFR3 Fibroblast growth factor receptor 3 TD I p.R248C
” *FGFR3*
Ig Ig
α

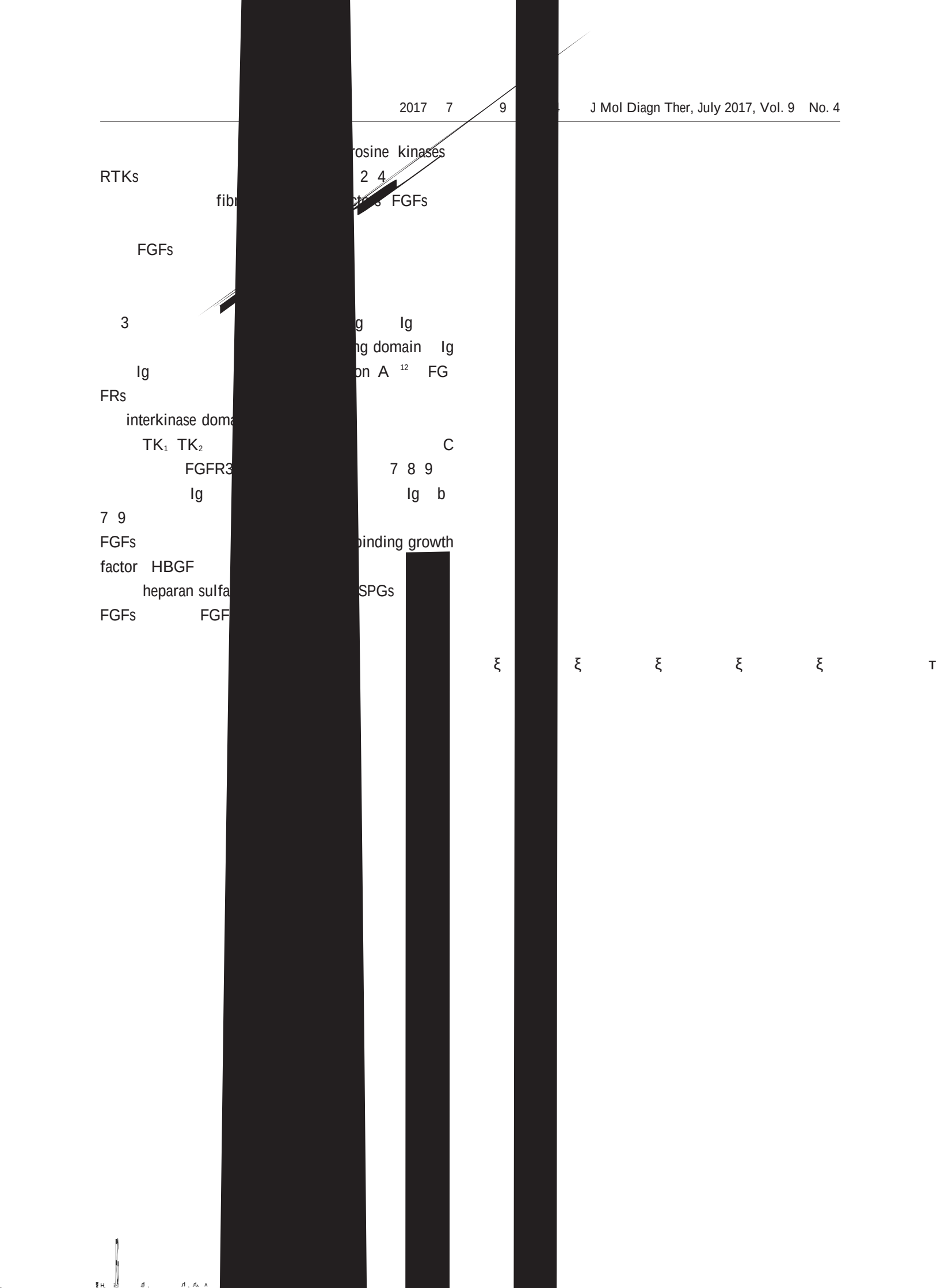
c.742C>T/p.R248C

I

rosine kinases
RTKs 2 4
fibr ctors FGFs

FGFs

3 g Ig
ng domain Ig
Ig on A [12] FG
FRs
interkinase doma
TK_1 TK_2 C
FGFR3 7 8 9
Ig Ig b
7 9
FGFs binding growth
factor HBGF
heparan sulfa SPGs
FGFs FGF



X807C X807G X807L X807S X807W 7
141 TD
[19] 248
p.R248C TD
100 55% [20]
FGFR3 E9
AGSVYAGILSYGVGFFL
FILVVAAVTLC 2

GPLYVLVEYAAKGNLRE FGFR3
[21]
dimer [5]

DNA [22]

FGFR3 [23]

FGFs ligand α
receptor *FG-*
FR3
unli
gand constitutive activa
tion *FGFR3*
FGFR3

[24] Ig Ig
E R P H R P

α

Cys
Ig Ig

TD *FGFR3* c.742C>T/
p.R248C *c-fos*
Fos

5 4 [25] p.R248A
DEDb
6 2 6 *c-fos*
Fos *c-fos*
DNA

COL1A1/COL1A2

osteogenesis im

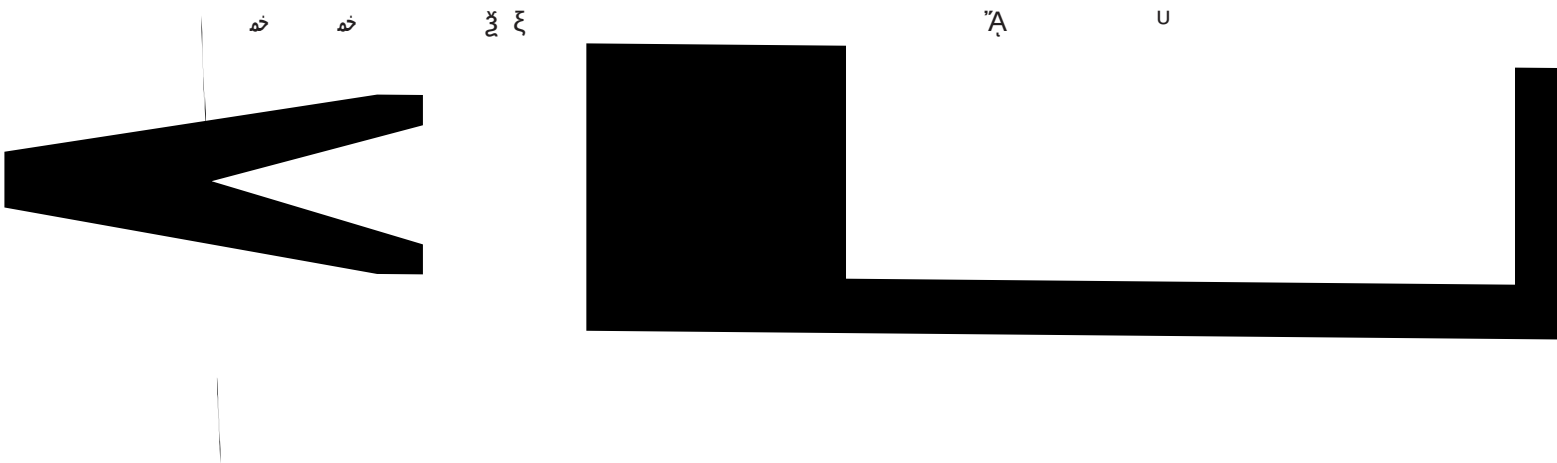
perfecta type ,OI

OI

28

TD

AD



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MiRNA 3p/5p

1 2 2

Pre miRNA Dicer 2 miRNA 3p miRNA 5p
miRNA 3p miRNA 5p miRNA 3p/5p
miRNA 3p/5p
miRNA 3p miRNA 5p

MiRNA 3p/5p in tumor development

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ABSTRACT Pre miRNA by the Dicer enzyme cuts produced 2 products, miRNA 3p and miRNA 5p, respectively, most of previous studies are dominated by a single miRNA 3p or miRNA 5p, and miRNA 3p/5p into the regulation of tumor development in pairs is rarely reported. In this paper recent development of miRNA 3p/5p and cancer will be summarized.

KEY WORDS MiRNA 3p MiRNA 5p Cancer Mechanism

RNA microRNA miRNA
RNA mRNA 3' miRNA 3p 5p 2
[3 7 8] miR
mRNA NA

1 miRNAs

miRNA precursor miRNA pre miRNA
5' miRNA miRNA
miRNA 5p 3' miRNA RNA 17~25
3p miRNA
miRNA* Dicer
3p 5p
[1 6] miRNA 2

NO KJ2015ZD29 KJ2016SD37 NO 1508085MH159
NO gxbjZD2016069 NO 20150309
NO Byycx1615

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2 miRNAs

2.1 miRNAs

miRNA
RNA II RNA ploymerase II RNA
III RNA ploymerase III miRNA pri
miRNA Pri miRNA Drosha Rnase
III 3′ 5′ 7
pre
miRNA pre miRNA
Dicer miRNA [9 11] Pre miRNA
Dicer 22
RNA RNA miRNA
miRNA*
RNA
miRNA miRNA* [12]
Dicer 3′ 5′ pre miRNA
3′ Dicer 3′ 21~25
miRNA 3p 5′
Dicer 5′ 22
22 bp miRNA 5p [13] miR
NA Ruby [14]
miRNA Mirtron
Drosha miRNA
pre miRNA miR
NA

? h ´ ? » ? ?

2.2 miRNAs

miRNA
miRNA lin 4 let 7
miRNA miR
mir
mmu hsa
rno hsa miR 155 mmu miR 155
miRNA
a b c…… hsa miR 125a hsa miR 125b
pre miRNA 70~80 nt
miRNA
miRNA *
miRNA
miRNA* [1 3 5]
miRNA*
miRNA* miR
NA [6 8 15 16]
miRNA
miRNA “ 5p ” “ 3p ”
5′ 3′ hsa miR
155 3p has miR 155 5p
DNA
miRNA hsa
miR 521 1 hsa miR 521 2 [3]

3 miRNA*

miRNA* miRNA
22 RNA
miRNA*
miRNA miRNA
AGO1 RISC RNA
miRNA* RISC
[13 17 18] miR
NA* miRNA
[1 19] miRNA*
miRNA 3′ 5′
miRNA [2 3] miRNA 3′
miRNA* 5′
miRNA
RISC miRNA* [4 5 20 21]
miRNA*
miRNA
miRNA* miRNA

miR 142 3p miR 142 5p [26] miRNA miRNA* [3 6 8 22 26] miRNA miRNA* hsa miR 28 5p hsa miR 28 3p [27] miR 96 5p miR 96 3p [28]

4 miRNA miRNA*

miRNA*s miRNA* miRNA miRNA* mRNA [15 29] miRNA* miRNA miRNA miRNA* miR199a 3p/5p miR 297b 3p/5p mRNA [23 24] miRNA miRNA* [6 16 30] miRNA miRNA* Kuchenbauer [31] miR 223 miR 233 miR 223* 1 / 3 miR 223* miR 233 miR 223*

5 miRNA 3p/5p

5.1 miR 590 5p/3p

YANG [22] miRNA qRT PCR HepG2 Hep3B Huh7 L O2 qRT PCR hepatocellular carcinoma HCC miR 590 3p miR 590 5p qRT PCR HepG2 Hep3B Huh7 3 HCC L O2 miR 590 3p miR 590 5p Western Blot miR 590 3p miR 590 5p PTEN 10 programmed cell death 4 PDCD4 PI3K AKT AKT1 S473 HCC miR 590 2 miRNA HCC 2

5.2 miR 582 3p/5p

Keita [32] UM UC 3 5637 J82 TCC SUP T24 HT1376 RT4 29 53 28 qRT PCR miR 582 3p/5p miR 582 3p miR 582 5p

miR 582 3p/5p *PGGT1B LRRK2 DIX-DC1* miR 582 3p/5p siRNA miR 582 3p/5p [32] miR 582 3p/5p

5.3 miR 96 3p/5p

Zhang [23] 28 34 16 miR 96 5p HCC HCC miR 96 5p high grade dysplastic nodule HGDN 2.83 HCC HCC miR 96 5p miR 96 3p

HCC

[23] miR 96 5p HCC

47.1 79

miR 96 5p HCC

miR 96 3p HCC

88.2 84.2 miR 96 3p

HGDN HCC

GPC3 HCC

67.7 100 miR 96

HCC

5.4 miR 409 3p/5p

Sajni [24]

miR 409 3p

miR 409 5p Gleason

Glea

son miR 409 3p miR 409 5p

miR 409

3p/5p miR 409

5p

miR 409 3p/5p

RSU1 RSU1

Ras/MAPK

integrin linked kinase ILK [33 35]

miR 409 5p *STAG2* *NPRL2*

miR 409 3p/5p

miR 409

6

miRNA

miRNA

miRNA* miRNA

miRNA*

miRNA

miRNA*

miRNAs miRNAs

miRNA 3p/5p

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1.1

1.4

20

Wu [13]

iTRAQ

2 659

141

Liang [14]

93%

Yun [15] H460

H460

8

4 *these-pai-2 nomo2 klc4*

plod3

NSCLC

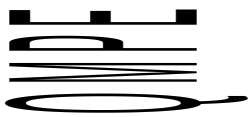
1 PAI 1

[16]

p53 HIF 1α Smad3

PAI 1 caspase 3

PAI 1



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4.1

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