

ICS 11.100.  
C 63

# SZDB/Z

SZDB/Z 122 2015

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2015 -01 -23

2015-02 -01

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SZDB/Z 122 2015

GB/T 1.1 2009

**1****2**

bp base pair

A Adenine

T Thymine

G Guanine

C Cytosine

U Uracil

DNA Deoxyribonucleic Acid

RNA Ribonucleic Acid

PCR Polymerase Chain Reaction

MALDI-TOF-MS / (Matrix Assisted Laser

Desorption/ionization Time-of-flight Mass Spectrometry)

dNTPs Deoxyribonucleoside Triphosphate

**3**

## 3.1

3.2

MALDI-TOF-MS

3.3

DNA RNA

A T G C U

3.4

3.5

3.6

3.7

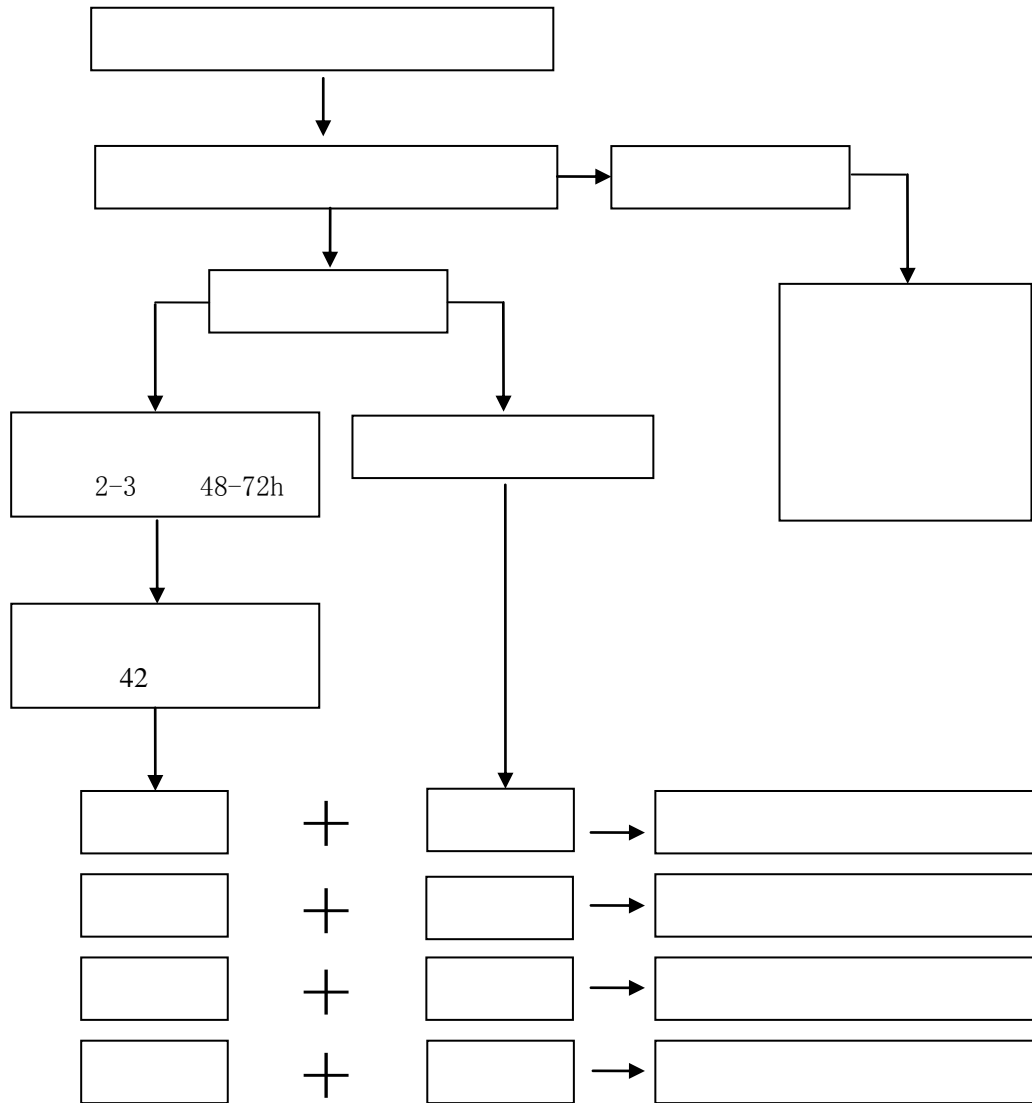
**4**

4.1

4.2

4.3

5



6





7.2.4.2 DNA

DNA

DNA

-20

DNA

4

-20

DNA

2

5

7.2.4.3 PCR

1)

PCR

2)

dNTPs

3)

7.2.4.4

1

2

/

MALDI-TOF-MS

7.2.4.5

MassARRAY Typer

7.2.4.6

D

7.3

7.3.1

7.3.2

7.3.3

7.3.4

**8**

8.1

3

E

8.2

8.3

3

6

1

6

8.4

8.5

8.5.1 12S rRNA

8.5.2

8.5.3

25%

8.5.4

## 9

9.1

9.2

A



C



D

TEOAE/DPOAE TEOAE/DPOAE AABR/ABR			
			/
GJB2	35del G	GG	
	167del T	TT	
	176_191del16 GCTGCAAGAACGTGTG	GCTGCAAGAACGTGTG. GCTGCAAGAACGTGTG	
	235del C	CC	
	299_300del AT	AT.AT	
GJB3	538 C>T	CC	
	547 G>A	GG	
SLC26A4 PDS	281 C>T	CC	
	589 G>A	GG	
	1174 A>T	AA	
	1226 G>A	GG	
	1229 C>T	CC	
	1975 G>C	GG	
	2027 T>A	TT	
	2162 C>T		

- 1.
- 2.
- 3.





[1] 64

[2] 2010

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