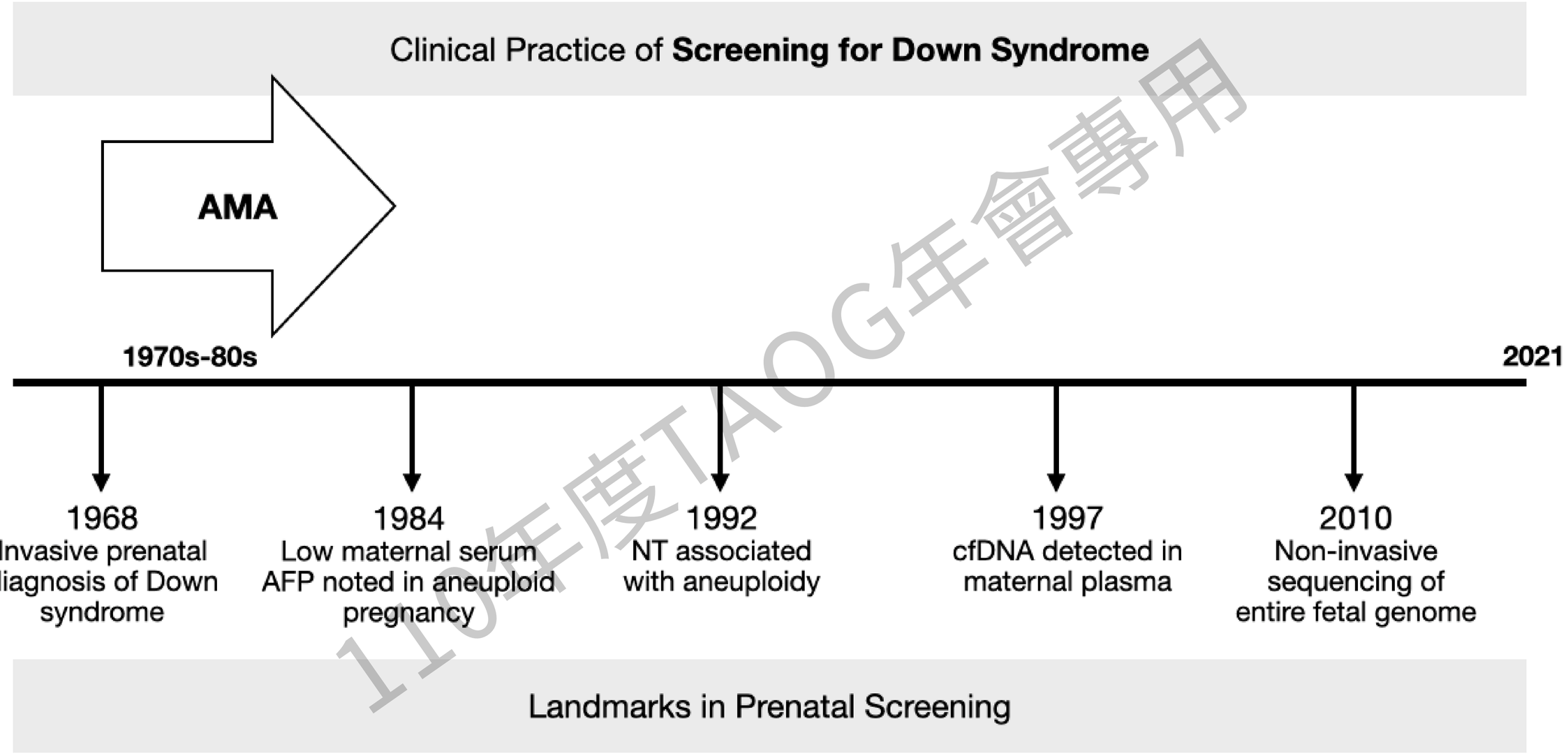
First Trimester Anatomical Screening

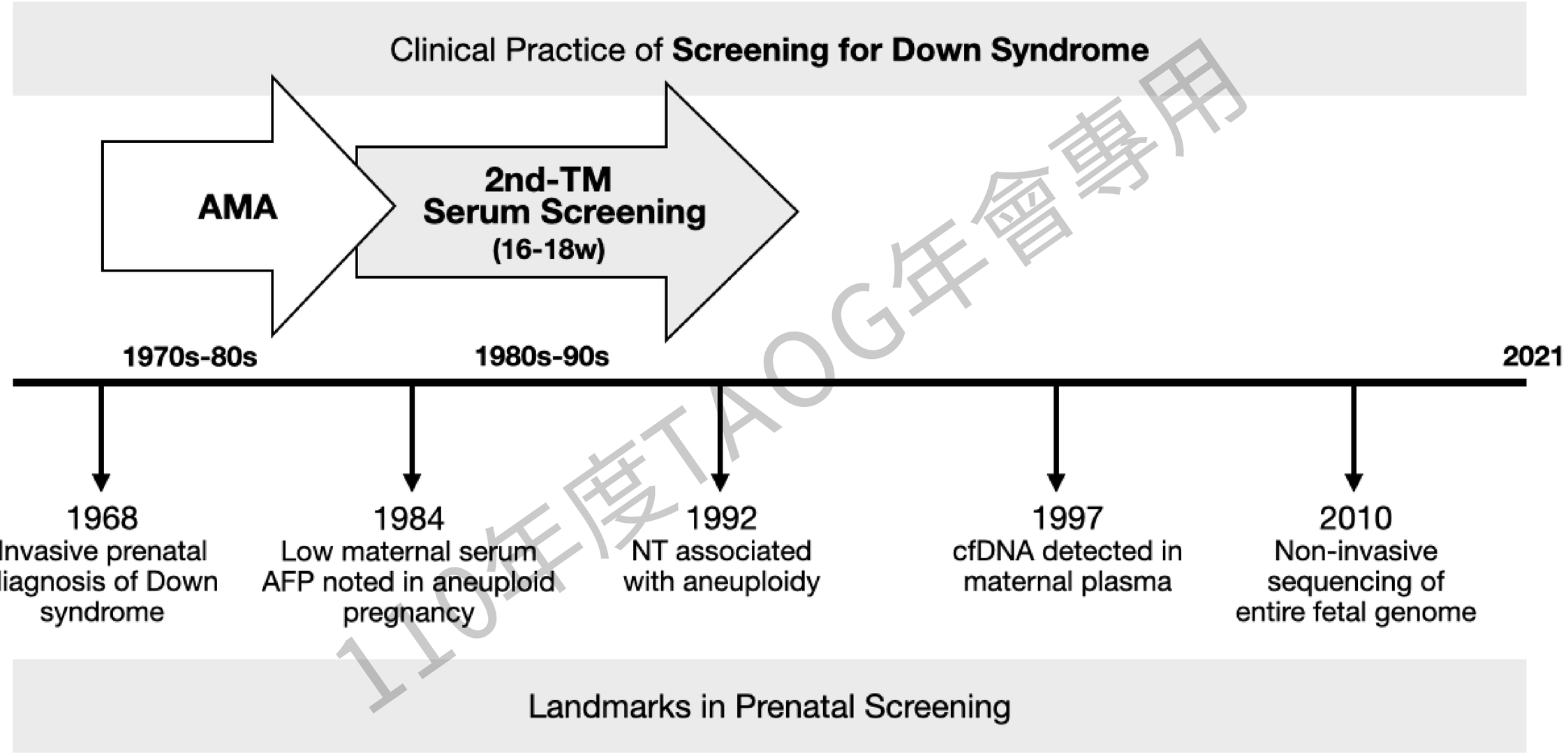
— Nuchal Translucency and Beyond

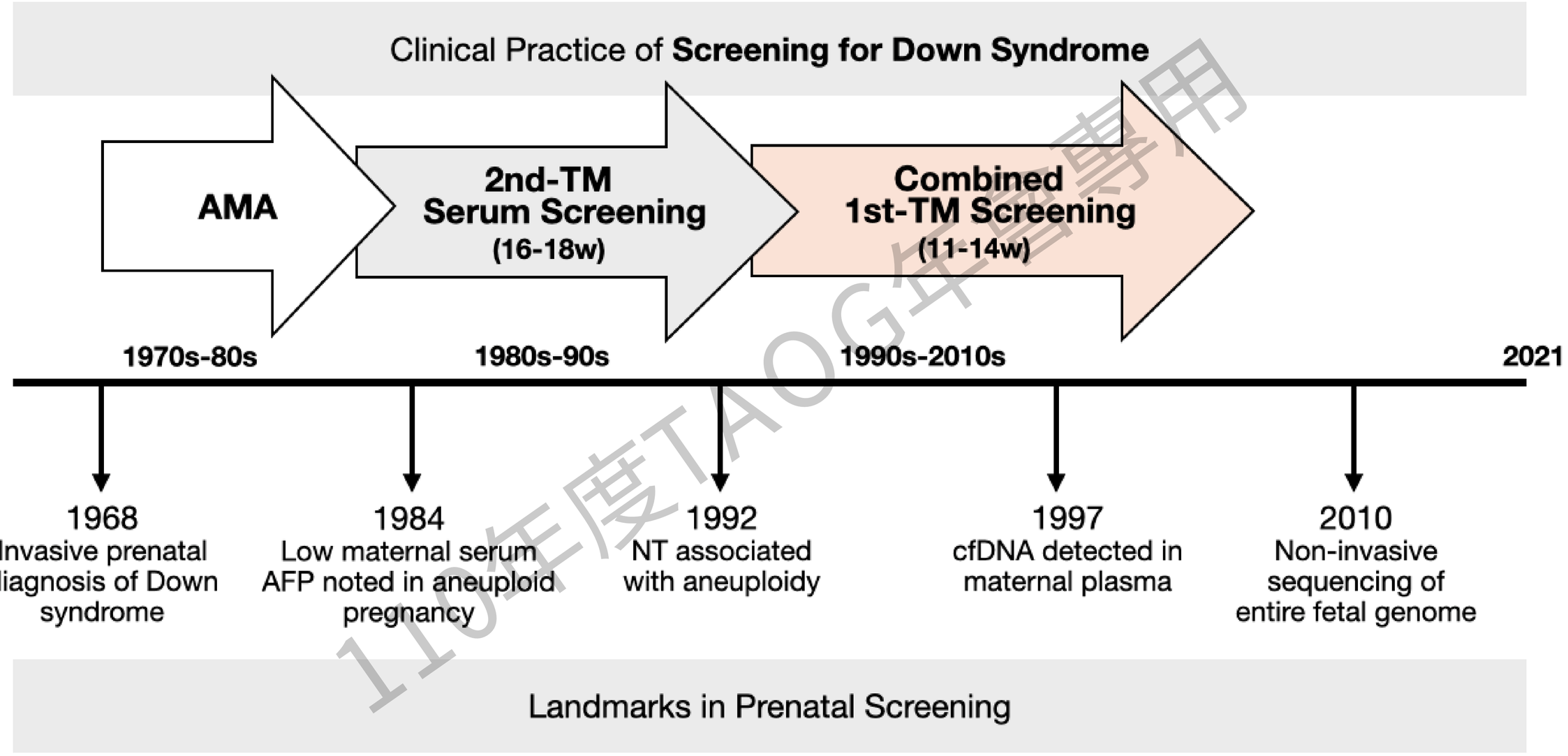
台兒診所/台兒中山集英聯合診所 吳佩臻醫師

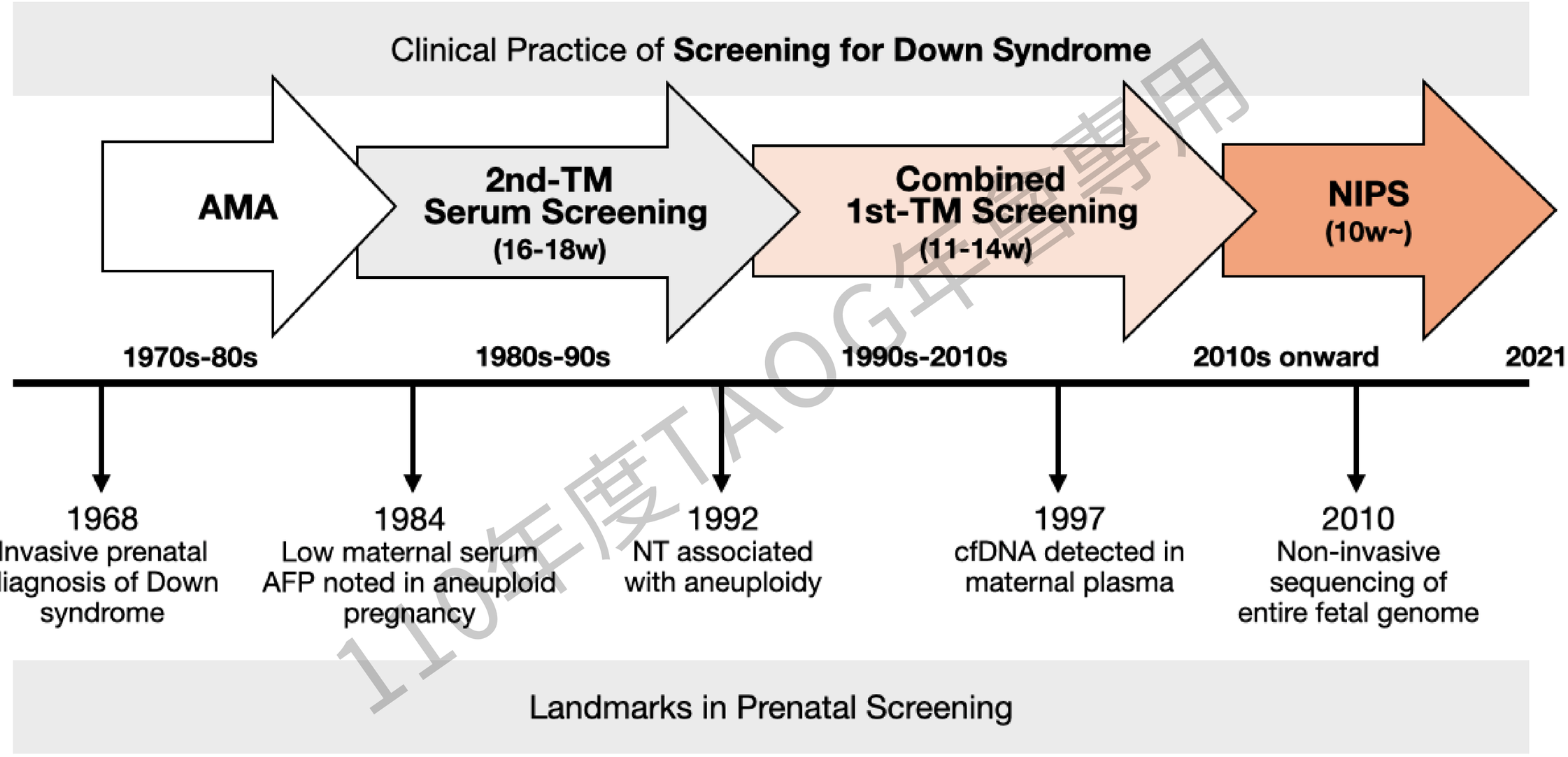
In the Era of cfDNA Testing…

1. The Role of Nuchal Translucency
2. The Value of First Trimester Ultrasound

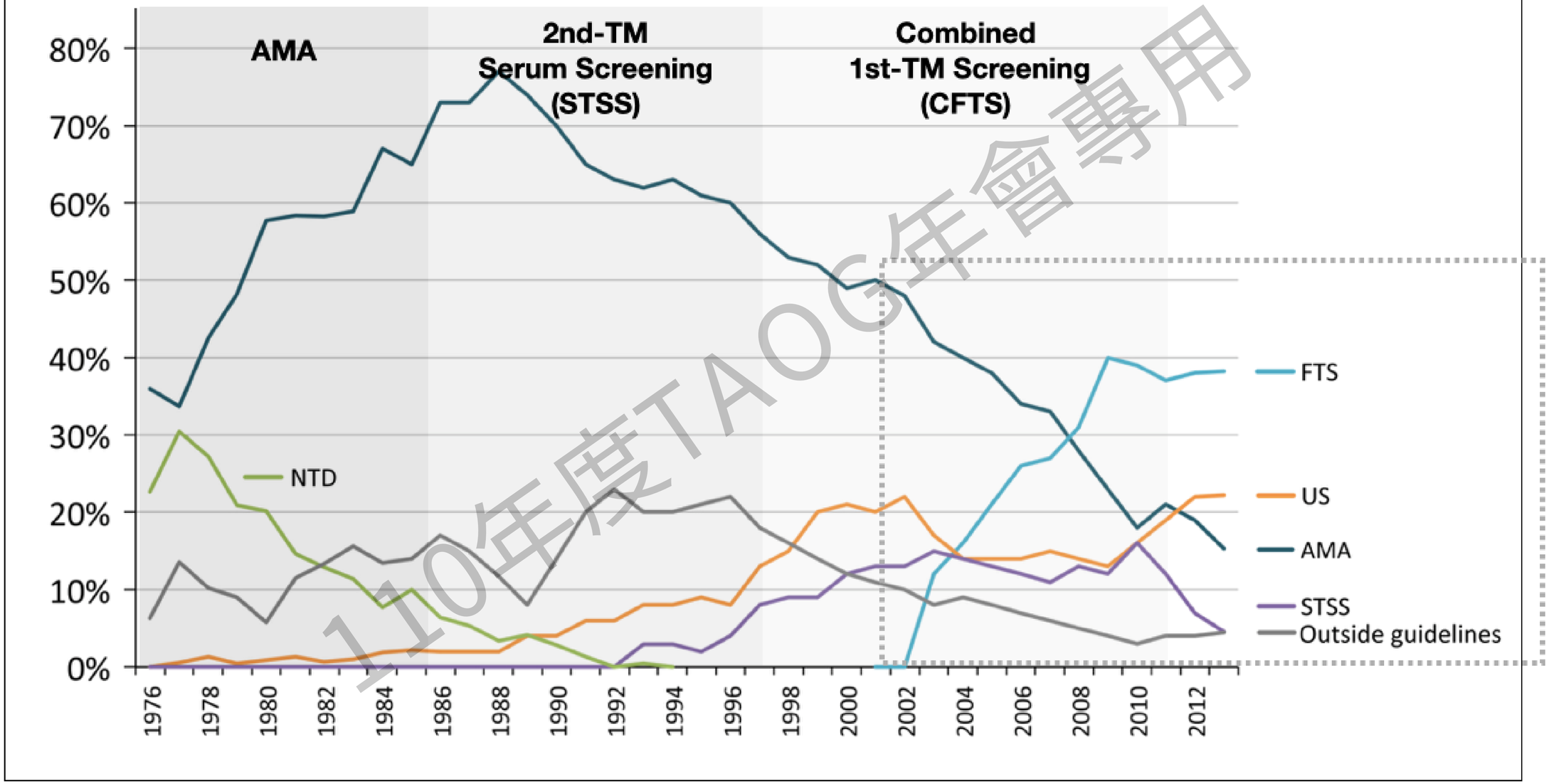








Indications for Invasive Prenatal Diagnosis as % of Total Tests



BJOG 2016;123:90-97

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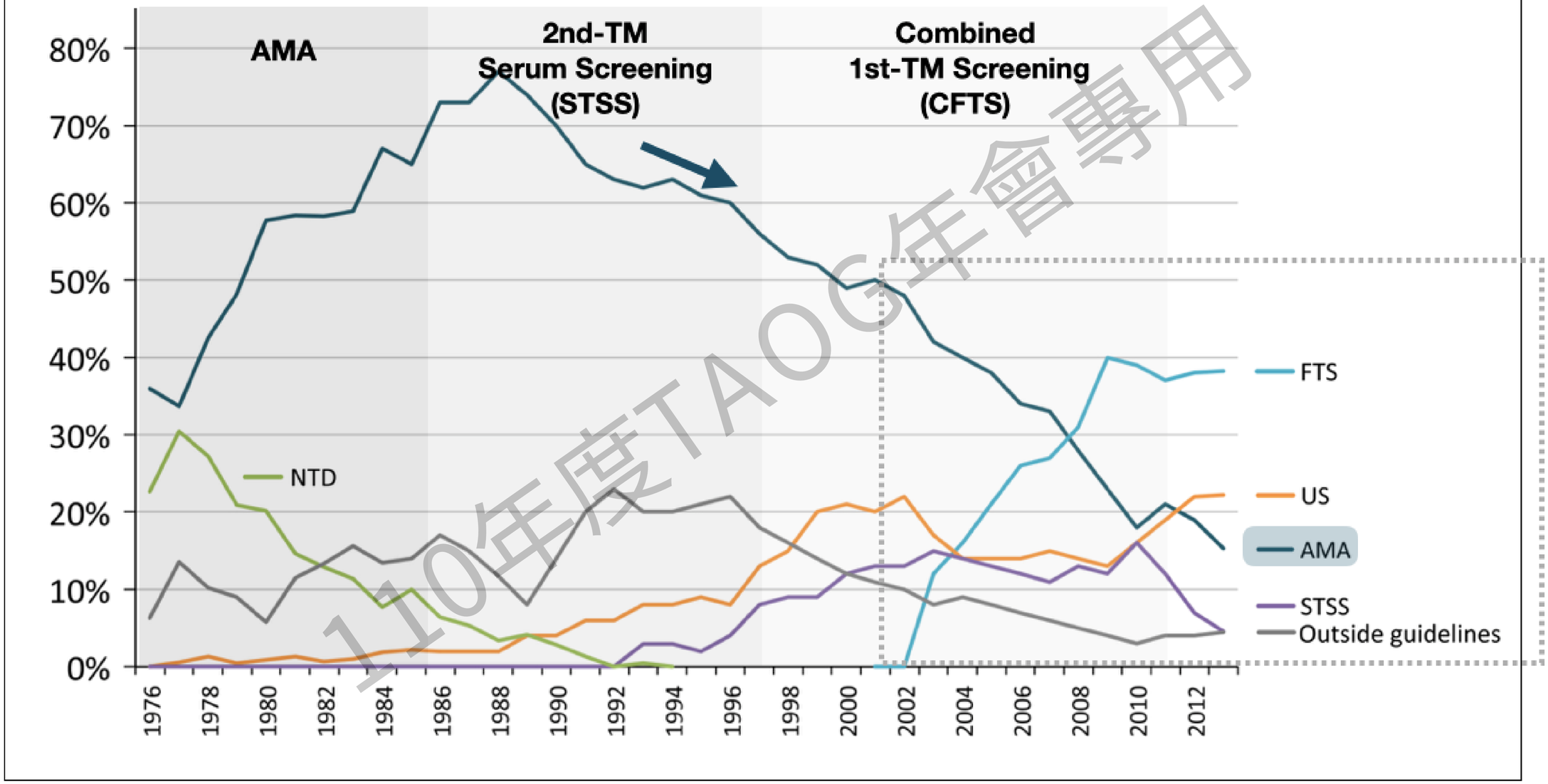

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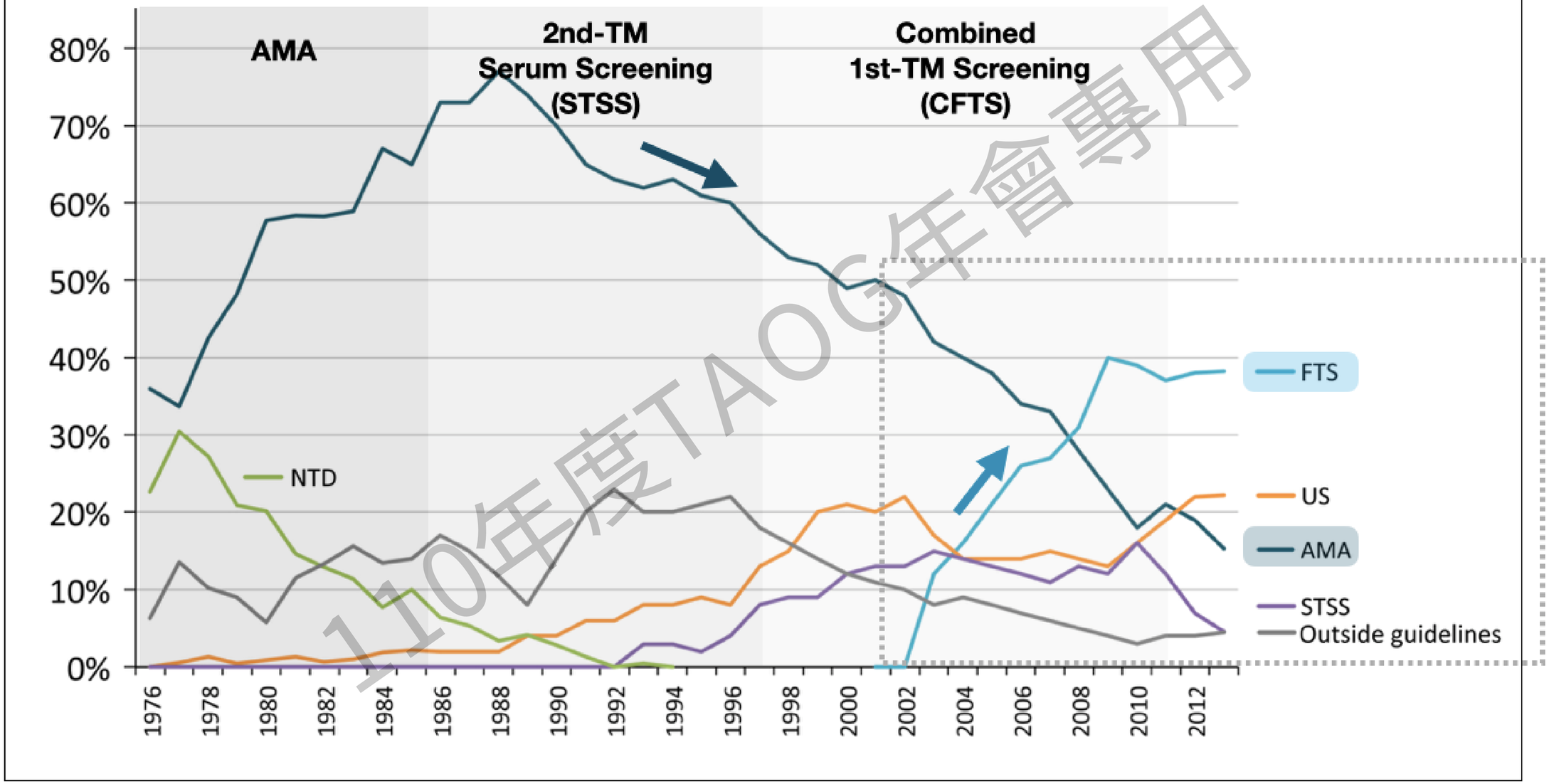

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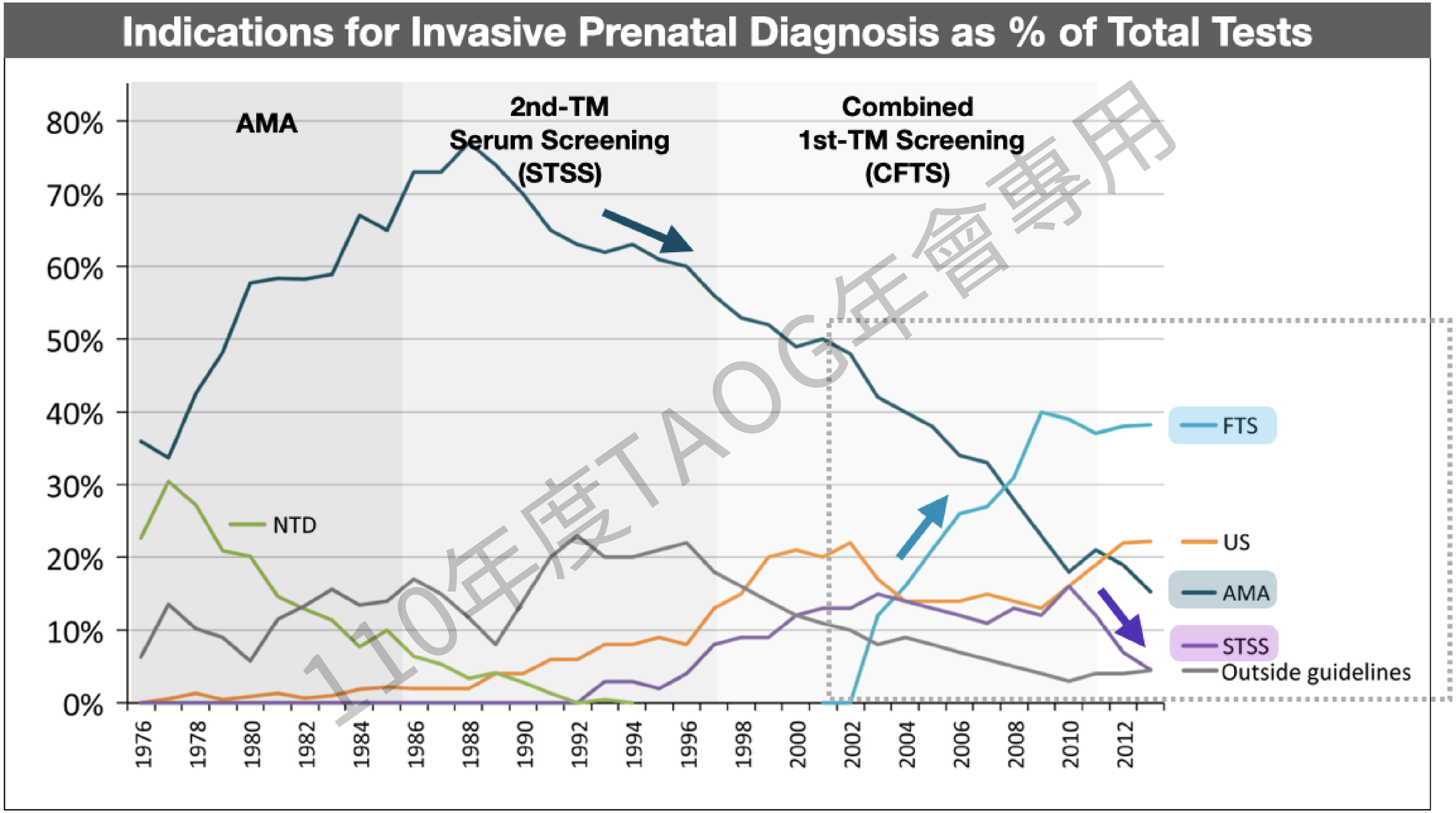
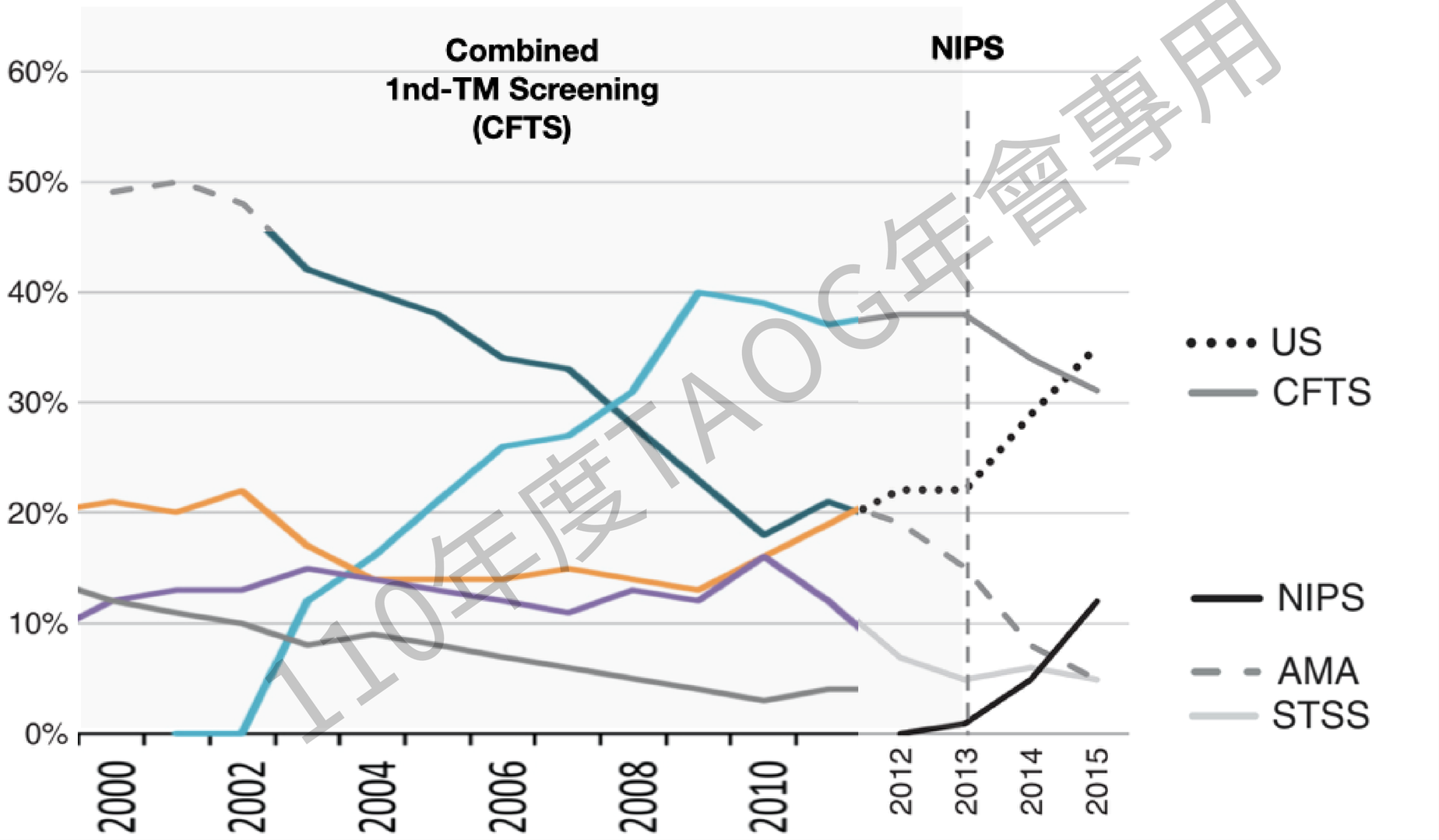


Genet Med. 2017 Dec;19(12): 13838-1345

Indications for Invasive Prenatal Testing as % of All Tests



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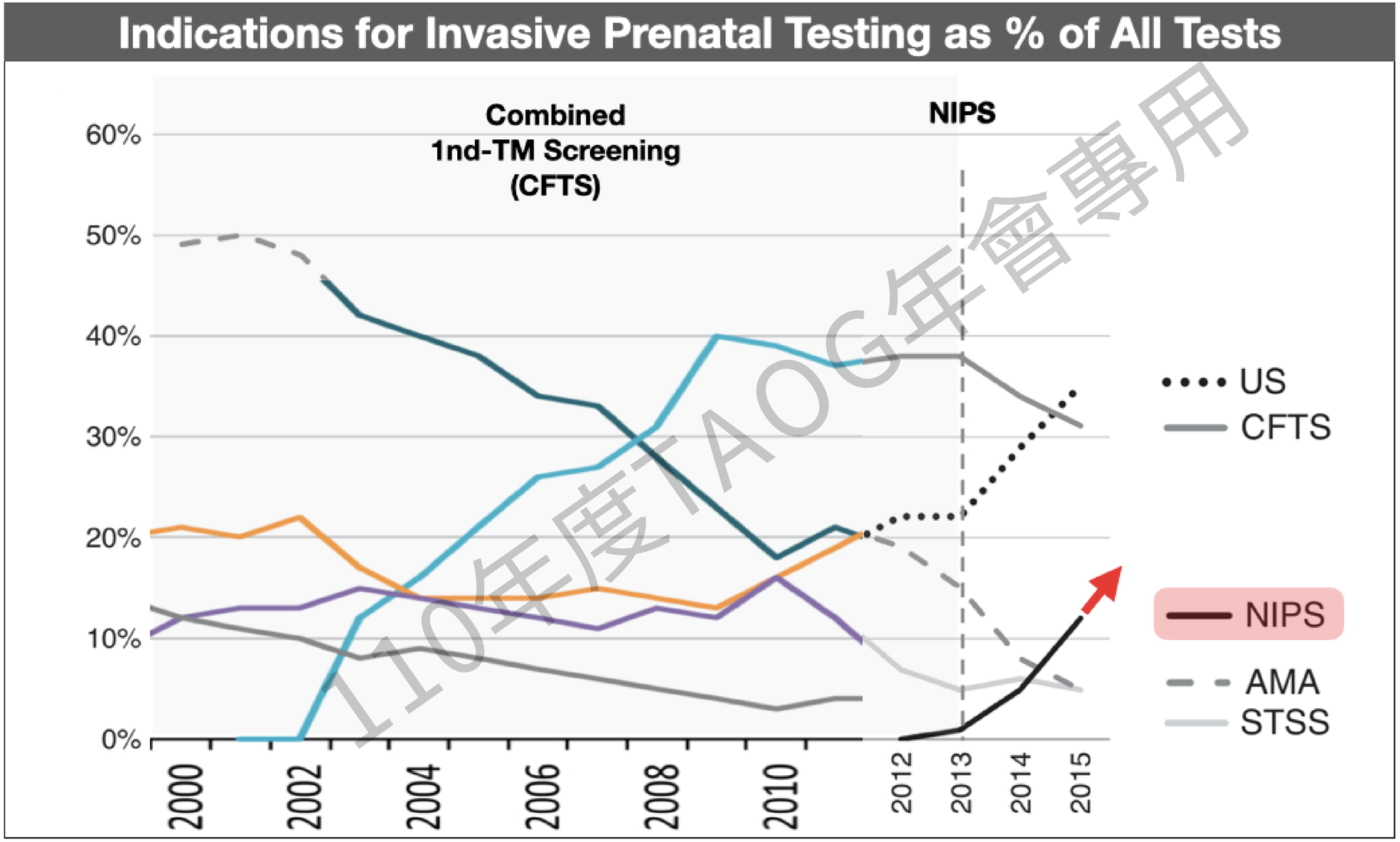

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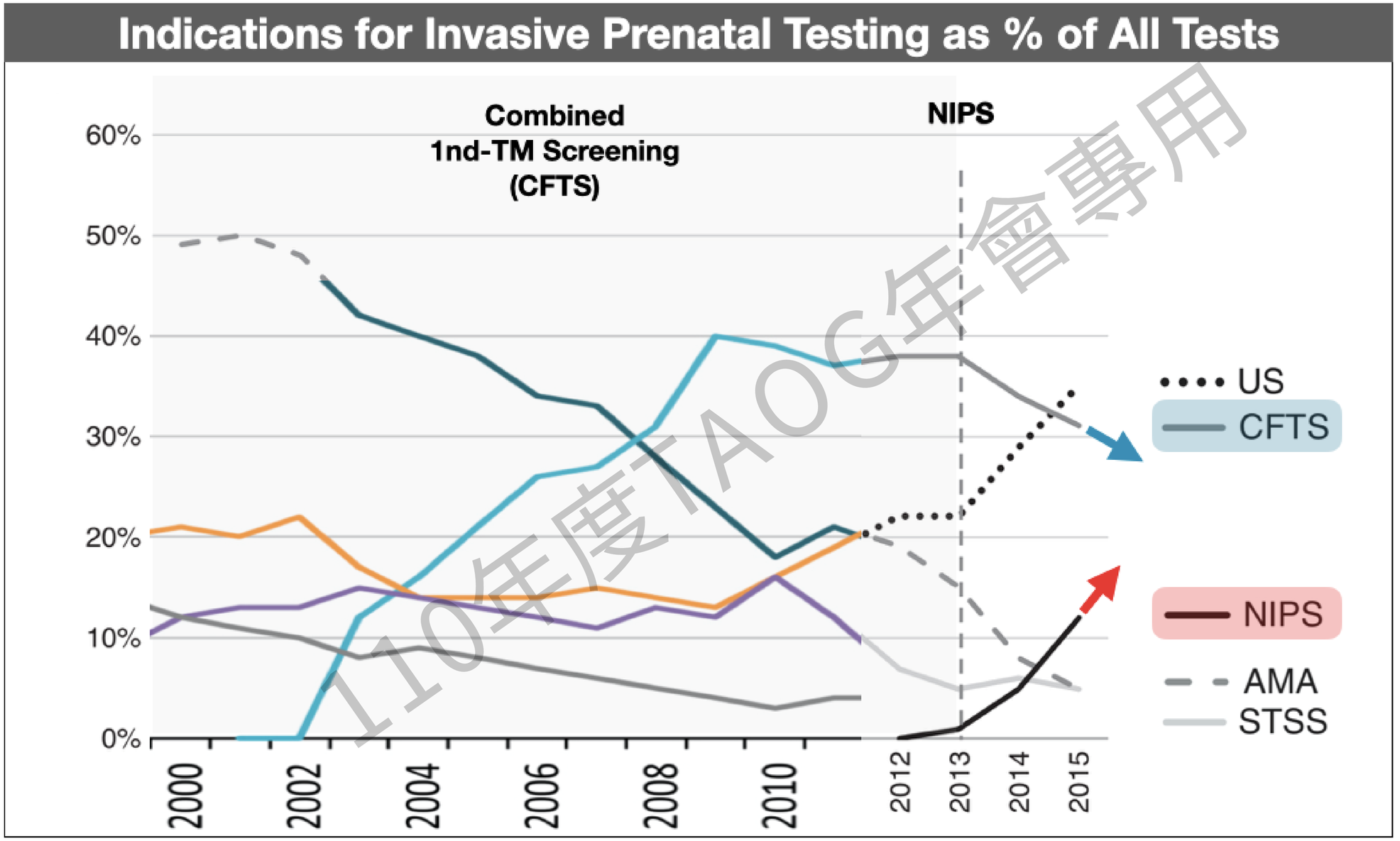

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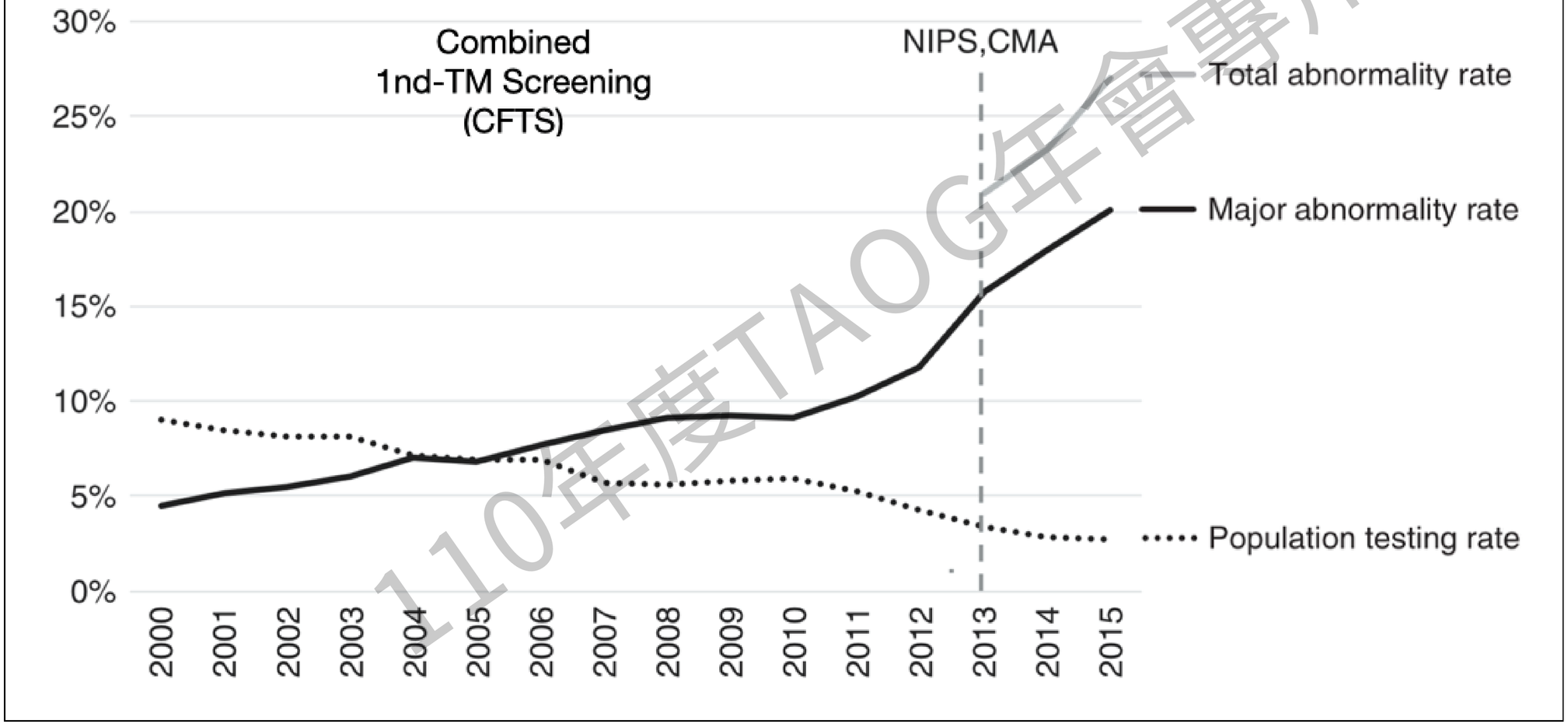
Statewide Trends in Prenatal Testing & Chromosome Abnormality Detection Rates



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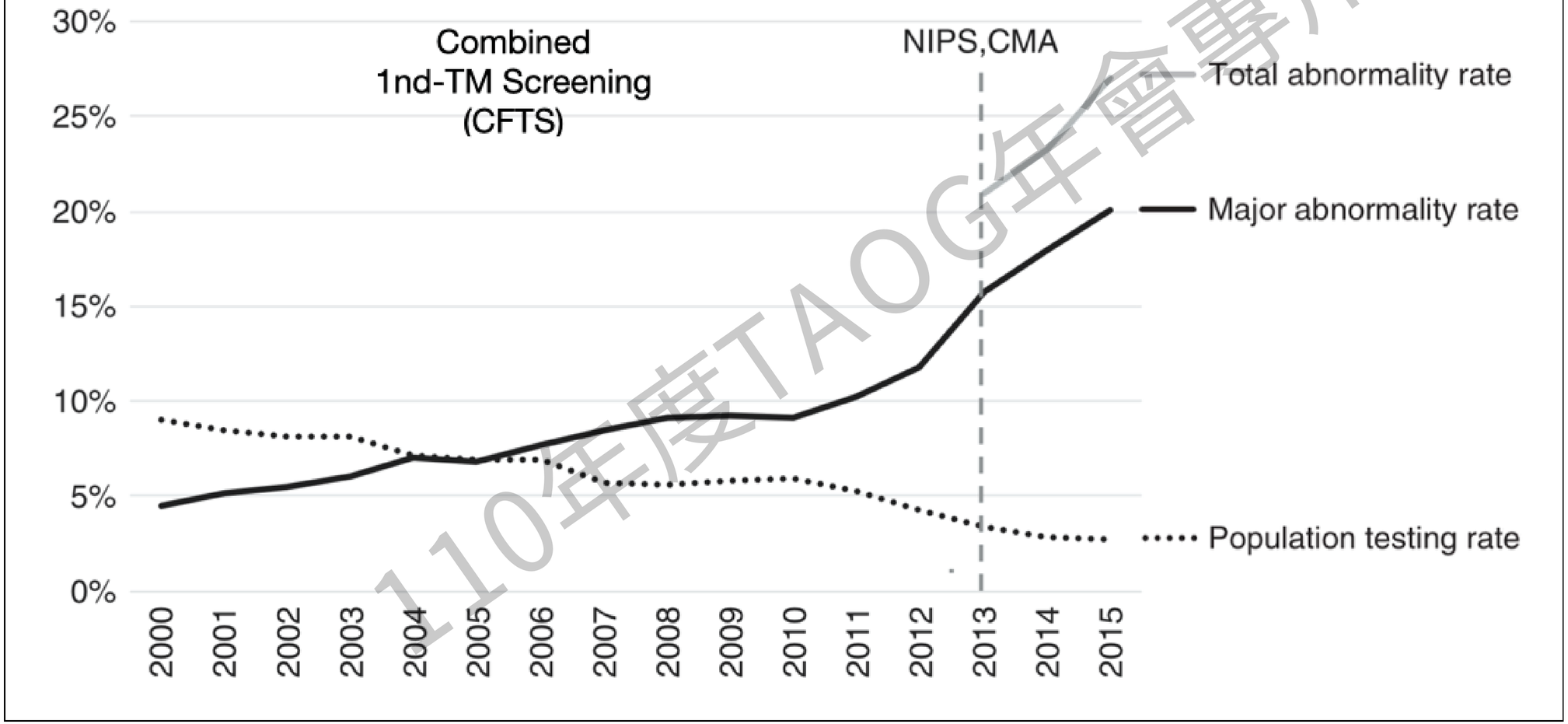

FPR

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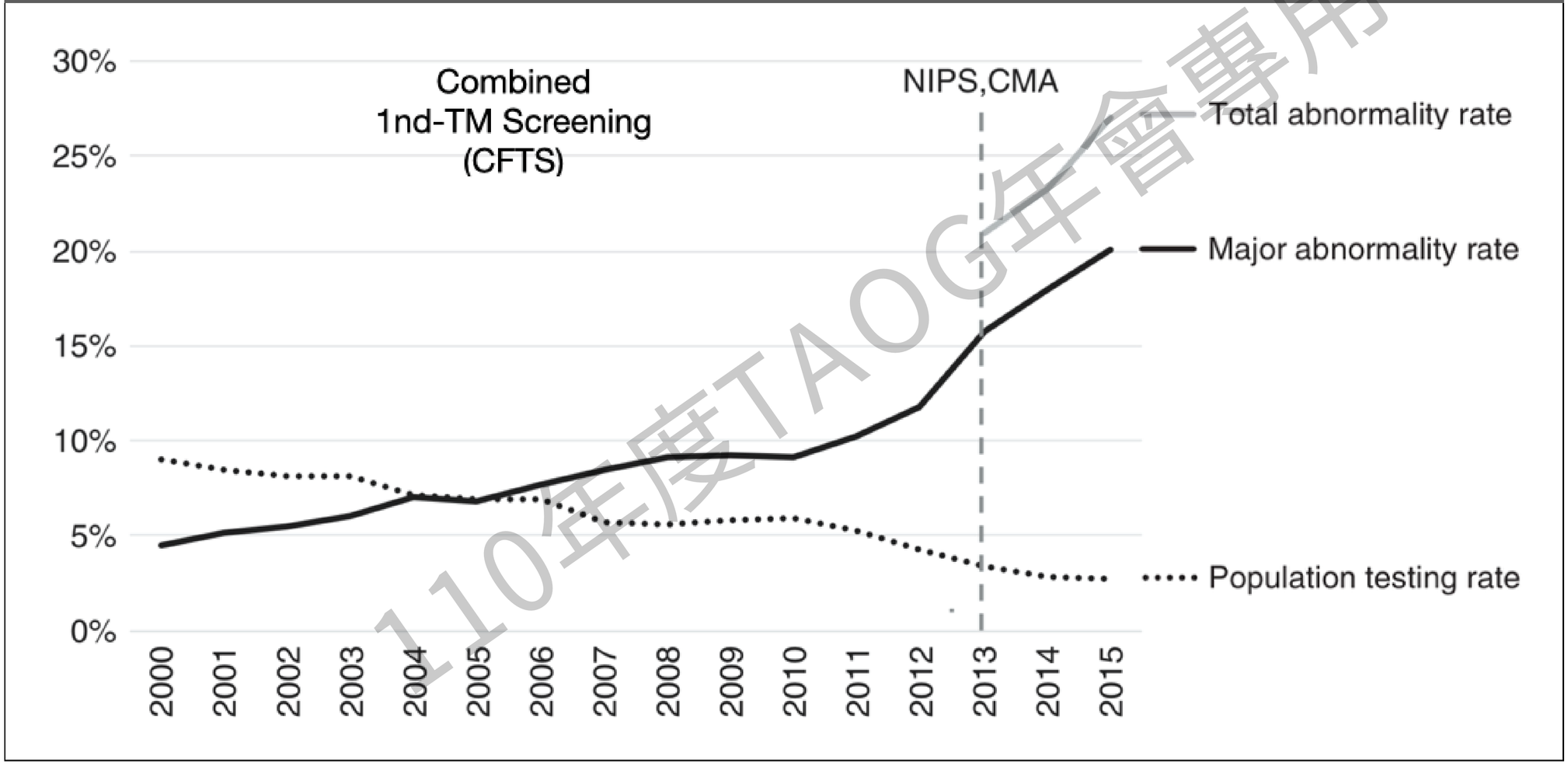
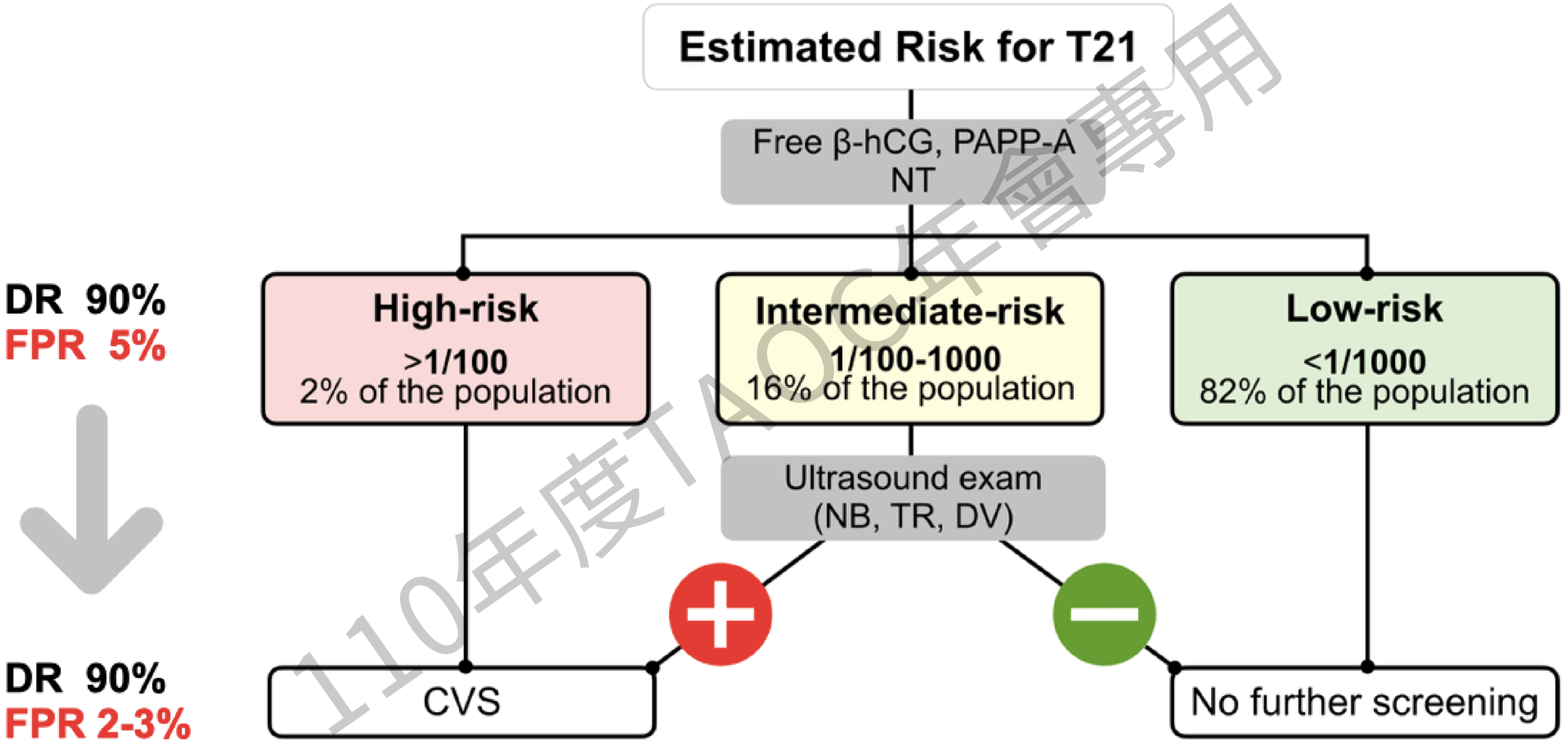


DR↑

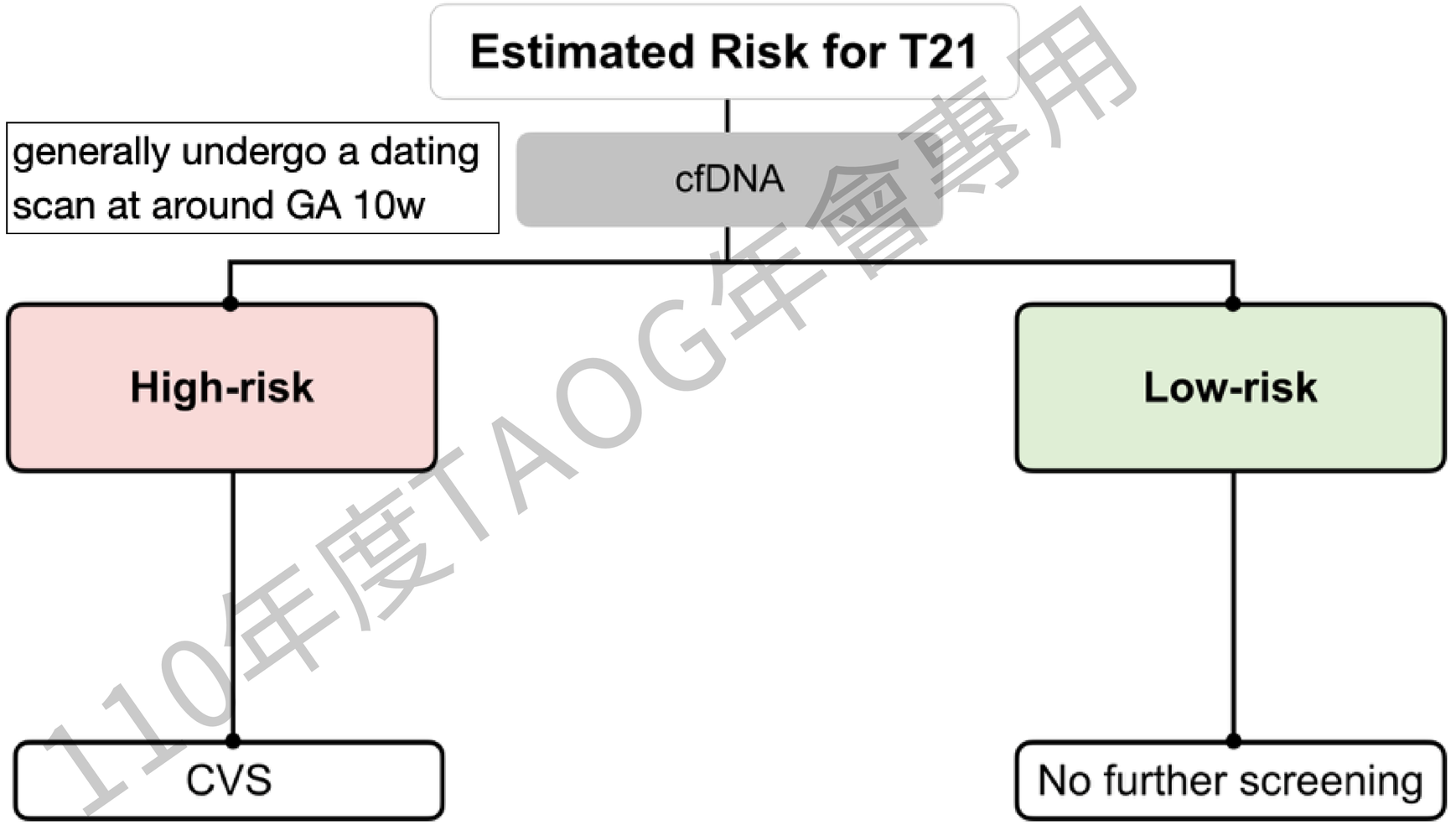
FPR↓

Ultrasound Obstet Gynecol 2005; 25: 221–226

CFTS (11w-13w6d)



NIPS (10w~)



DR >99%
FPR <1%

In the Era of cfDNA Testing…

1. The Role of Nuchal Translucency

CFTS (NT)

NIPS

CFTS (NT)

	Sensitivity	FPR
T21	90%	4%
T18	97%	4%
T13	92%	4%
45,X; **triploidy**	>95%	4%
Other aneuploidy	>50%	4%

Ultrasound Obstet Gynecol 2017; 49: 714-720

NIPS

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T21	**99.7%**	**0.04%**
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SCAs (other than 45,X)	**100%**	**0.004%**

Ultrasound Obstet Gynecol 2017; 50: 302-314

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Requires
1. high-quality ultrasound machines
2. specifically trained sonographers
3. continuous quality-control program in the ultrasound unit

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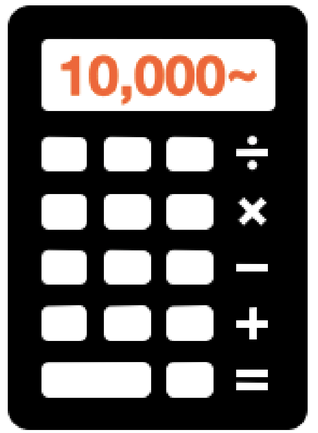

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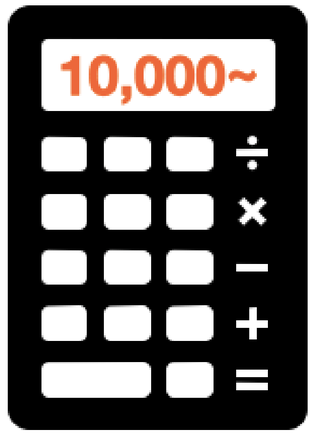

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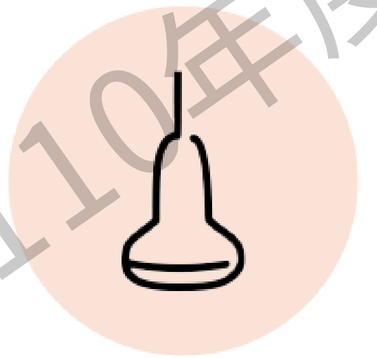

NT/NB, TR, DV

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NT/NB, TR, DV
Anatomical scan

大勝

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cfDNA

Taiji Case 1

Paternal family Hx of polydactyly

12w4d Taiji Clinic: (CFTS) **NT 6.2mm, high risk for chromosomal anomaly**

- NB (-)
- **Radial ray anomaly of left upper limb**

Risk: **T21(1/8); T18 (>1/4); T13 (1/10)**

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14w1d **CVS: T18**

Termination of pregnancy

Taiji Case 2

IVF; **NIPS: pending report**

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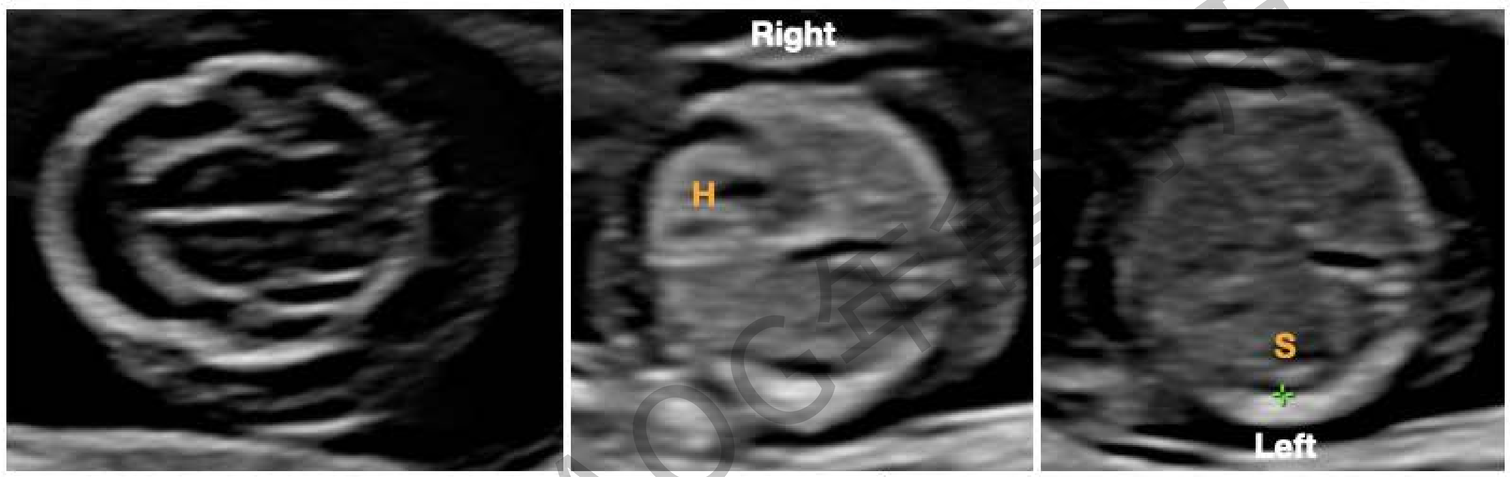
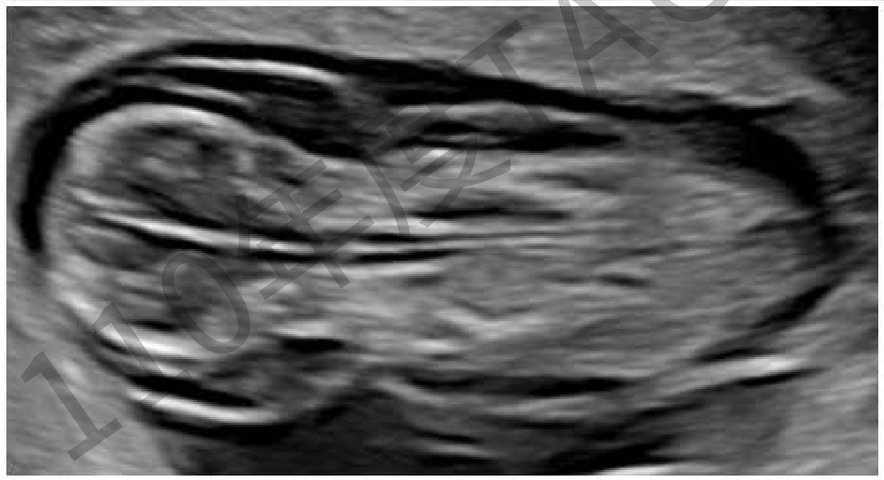
12w5d Taiji Clinic: **NT 5.2mm, alobar HPE, dextroposition of heart**





- NB (-), TR(+)
- general skin edema
- CRL <1st centile

Risk based on ultrasound findings:
T21(>1/4); T18 (1/6); T13 (1/22)

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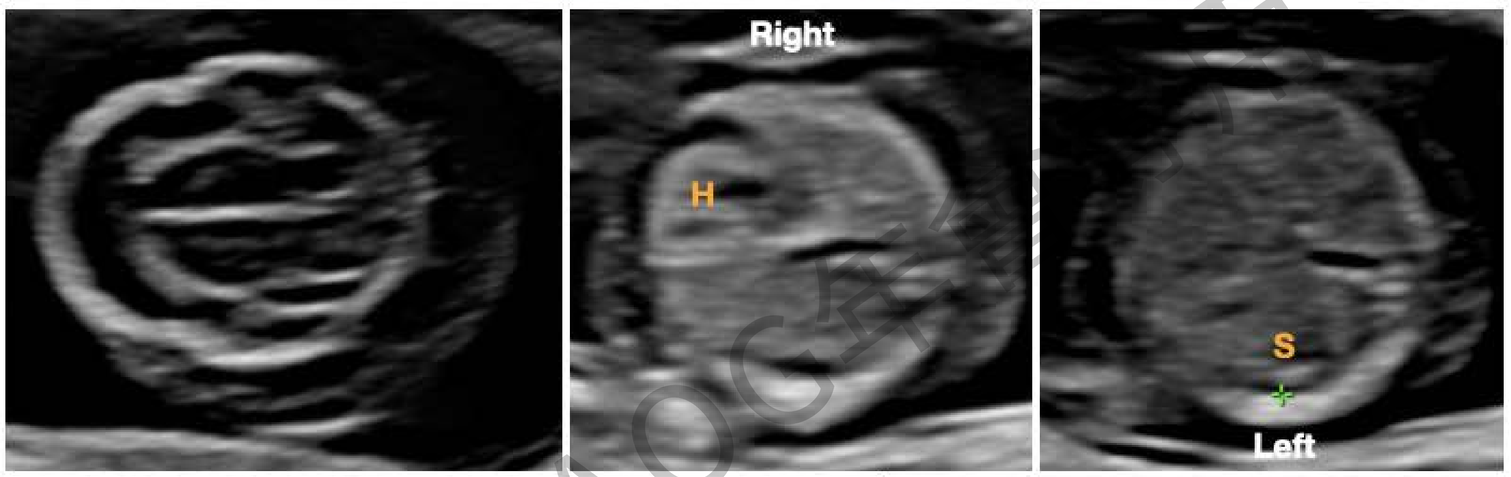

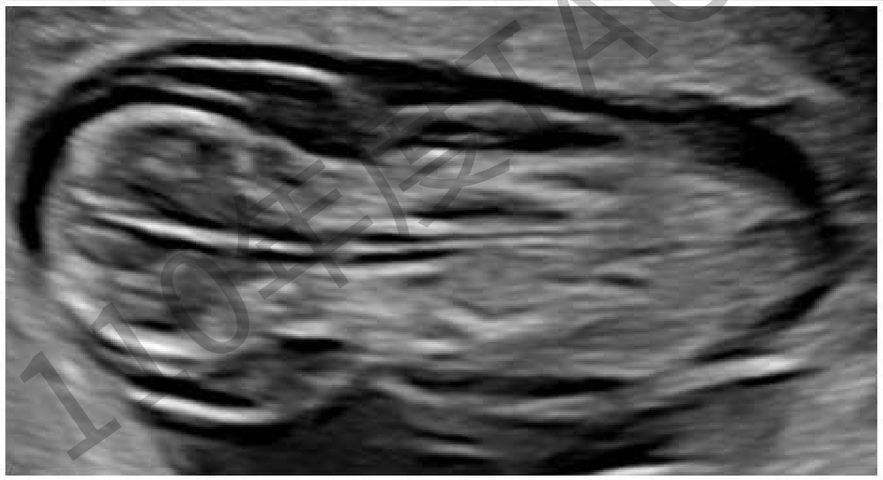

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14w5d **NIPS: high risk for T18**

Termination of pregnancy

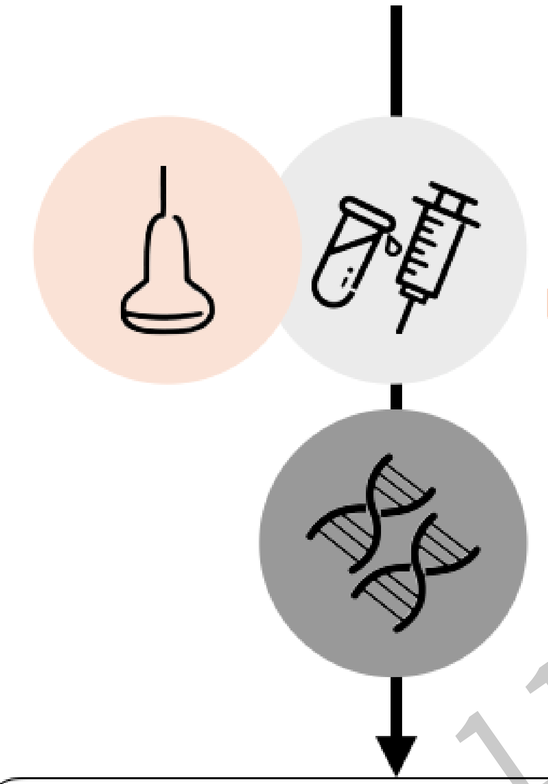
Case 1

Case 2

Trisomy 18, termination of pregnancy

Case 1

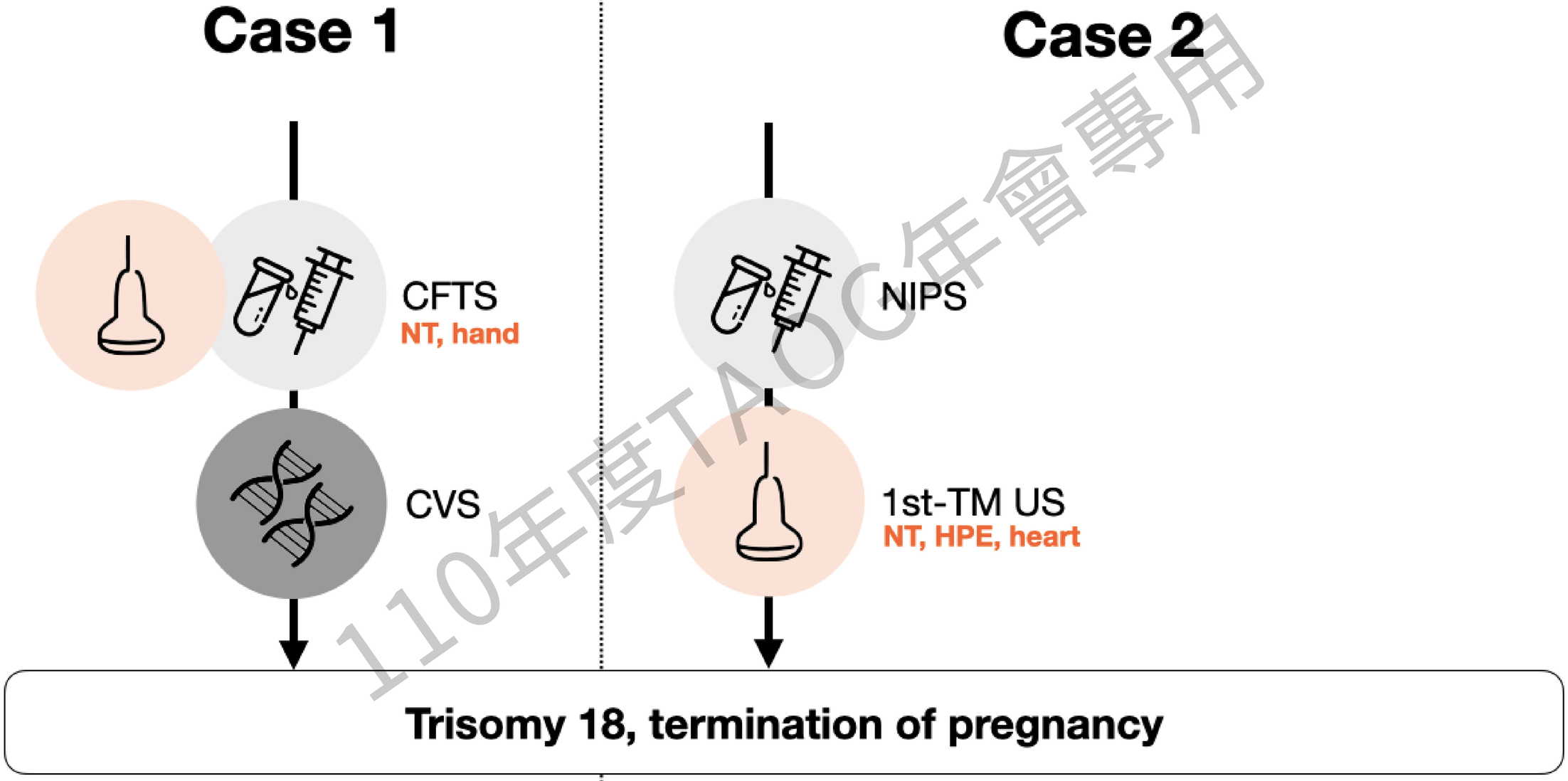
Case 2

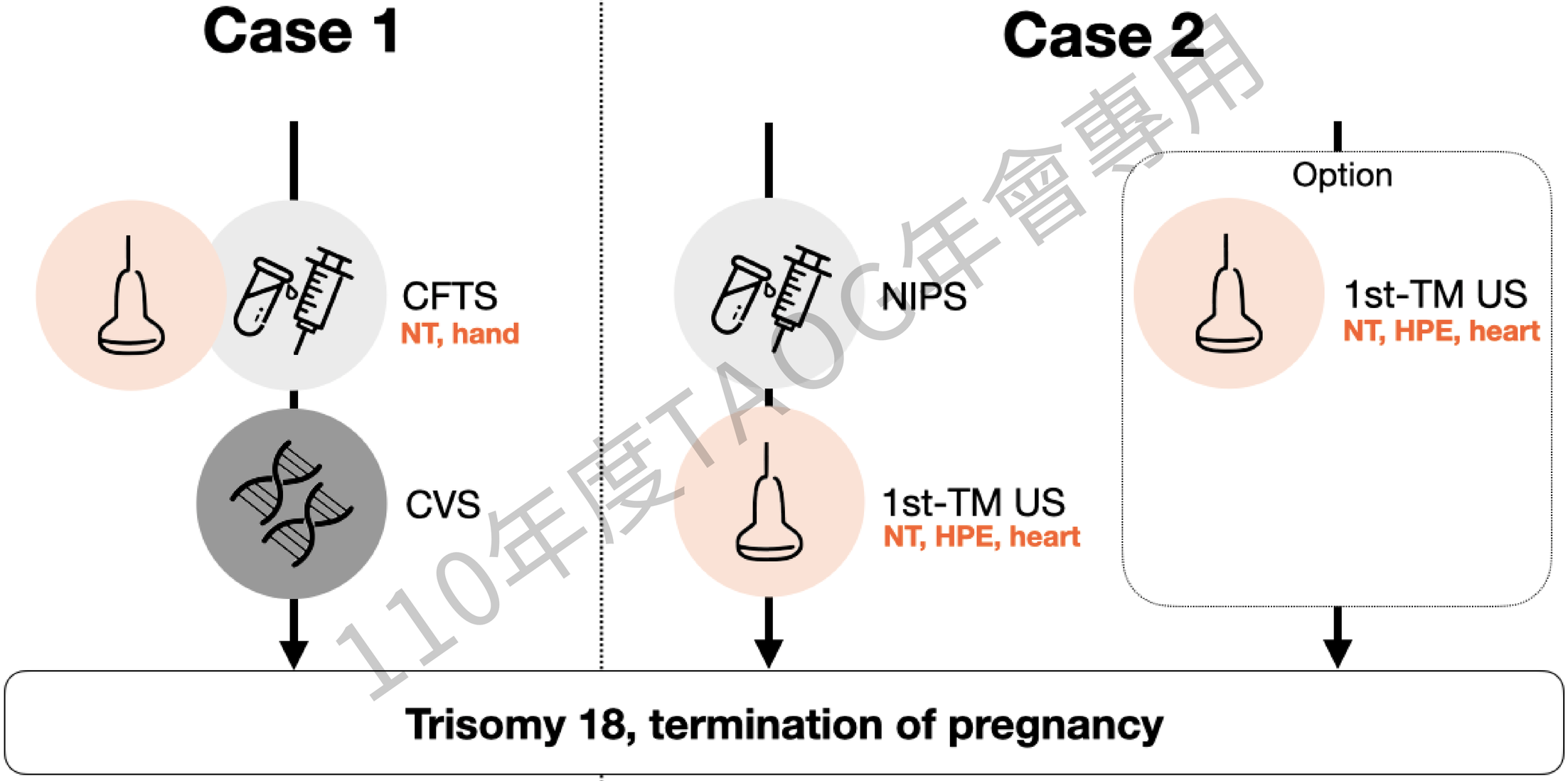


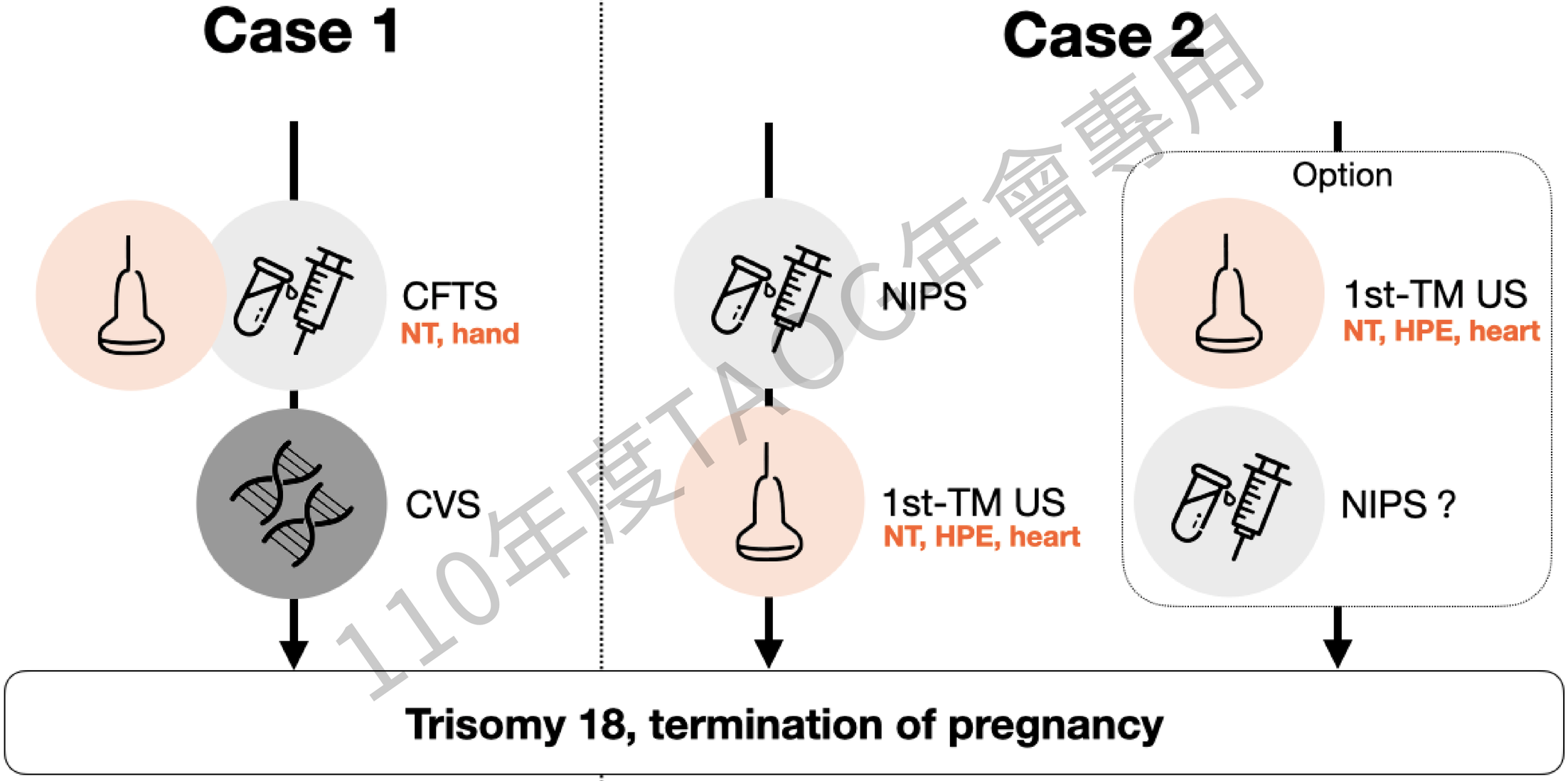
CFTS
NT, hand

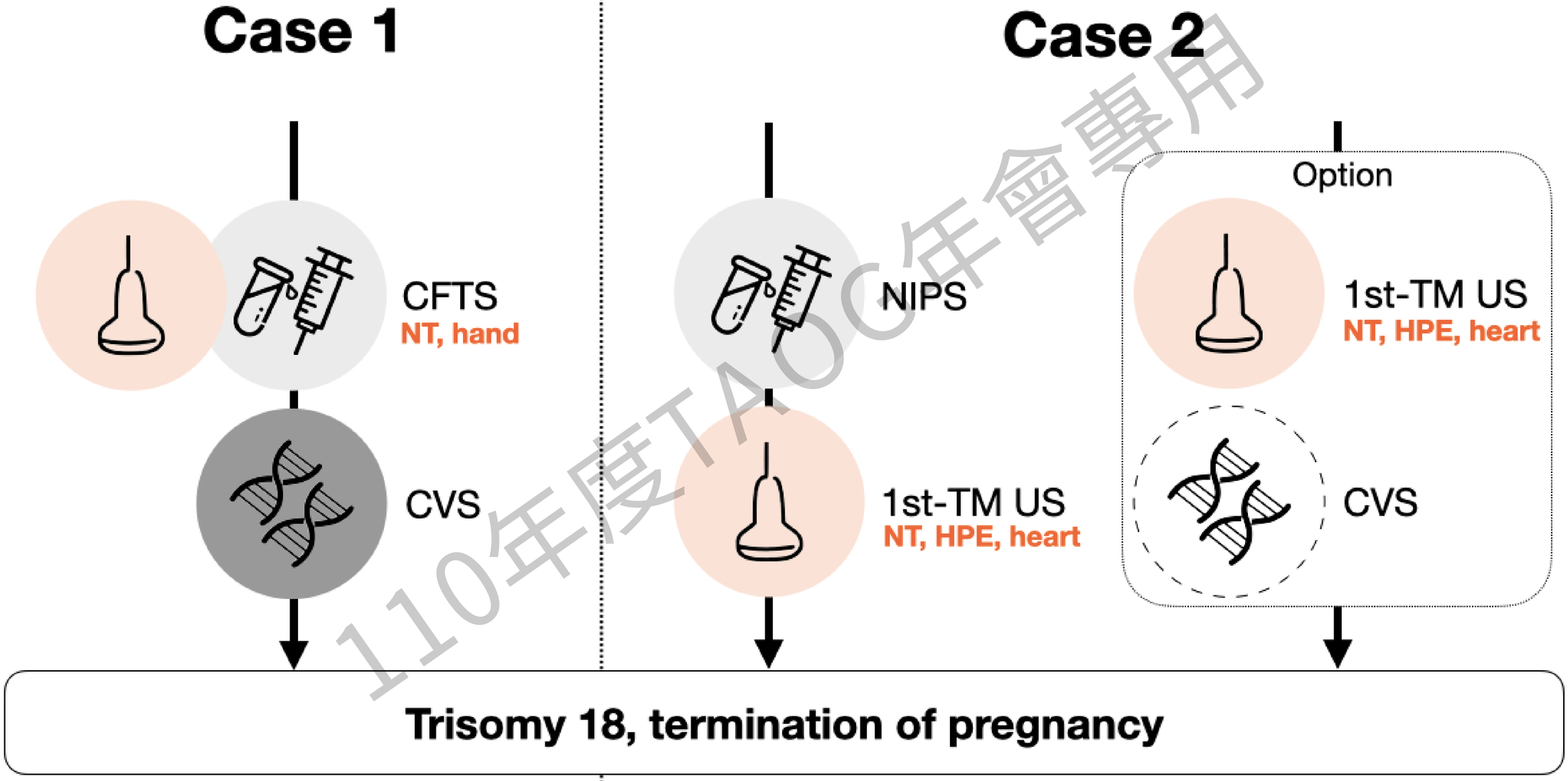
CVS

Trisomy 18, termination of pregnancy



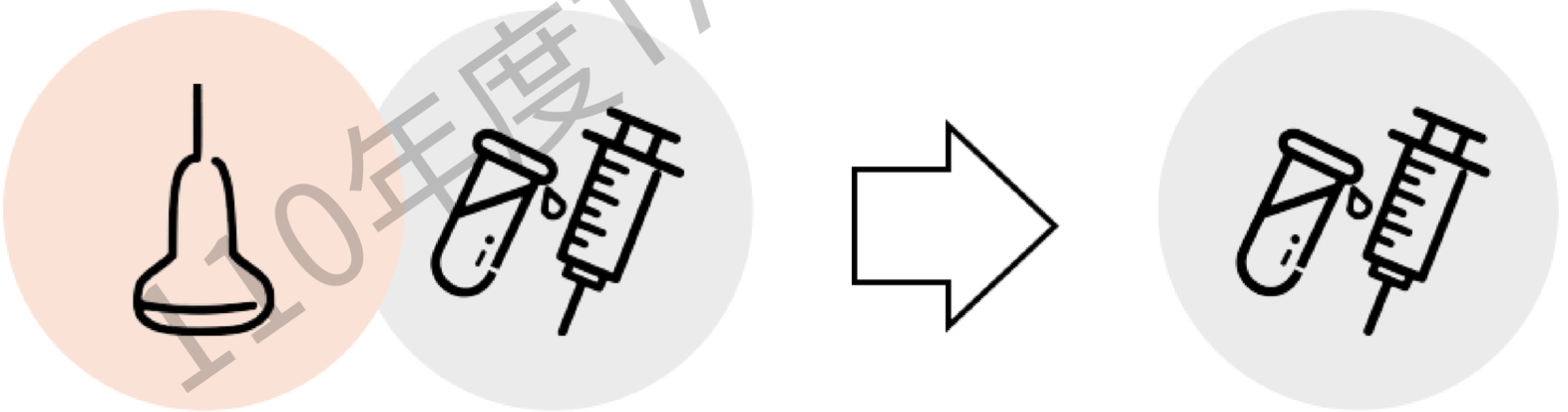






??? ①

What would be missed in the first trimester
if NT measurement is replaced by cfDNA screening?



CFTS

NIPS

Ultrasound Obstet Gynecol 2020; 55: 645–651

Should cfDNA testing be used in pregnancy with increased fetal NT?

I.MIRANDA et al. (Spain)

- cfDNA does not appear to be the appropriate genetic test in **fetuses with NT >99th centile**, given that it would miss **12-19% of genetic anomalies** in this group.
- In fetuses with **increased NT**, **genetic testing should be performed by CMA rather than karyotyping**, because it provides approximately a 5% incremental yield of pathogenic copy number variants above karyotyping.

??? ②

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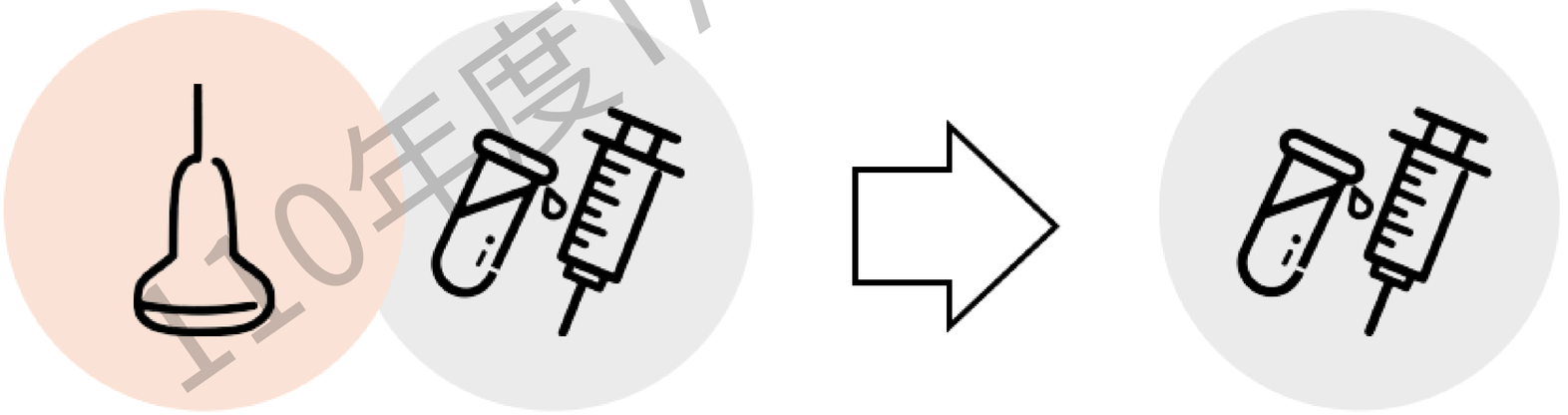
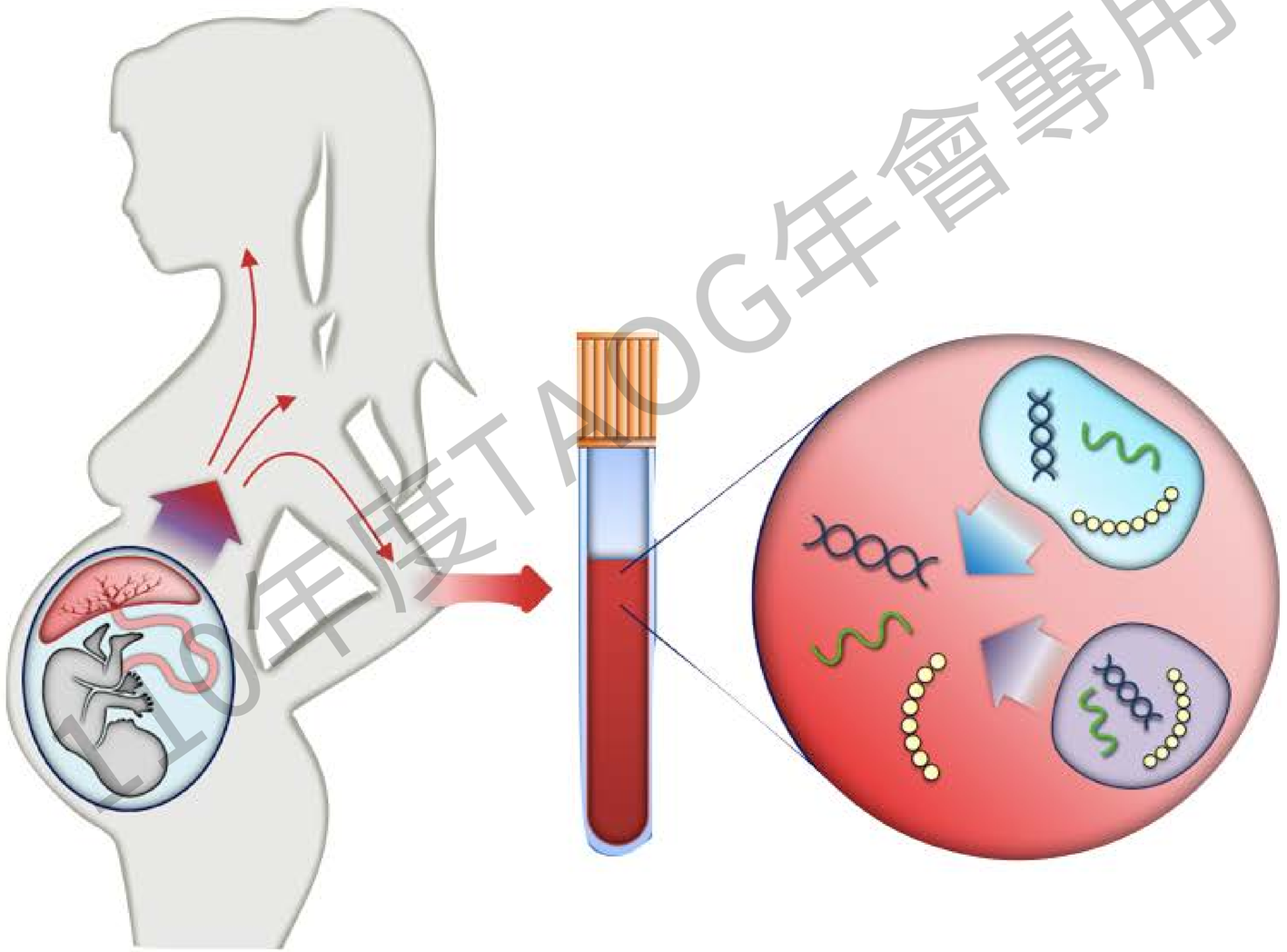


CFTS

NIPS

PLoS One. 2016 Jan 15;11(1):0146794

Noninvasive Prenatal Screening (NIPS)



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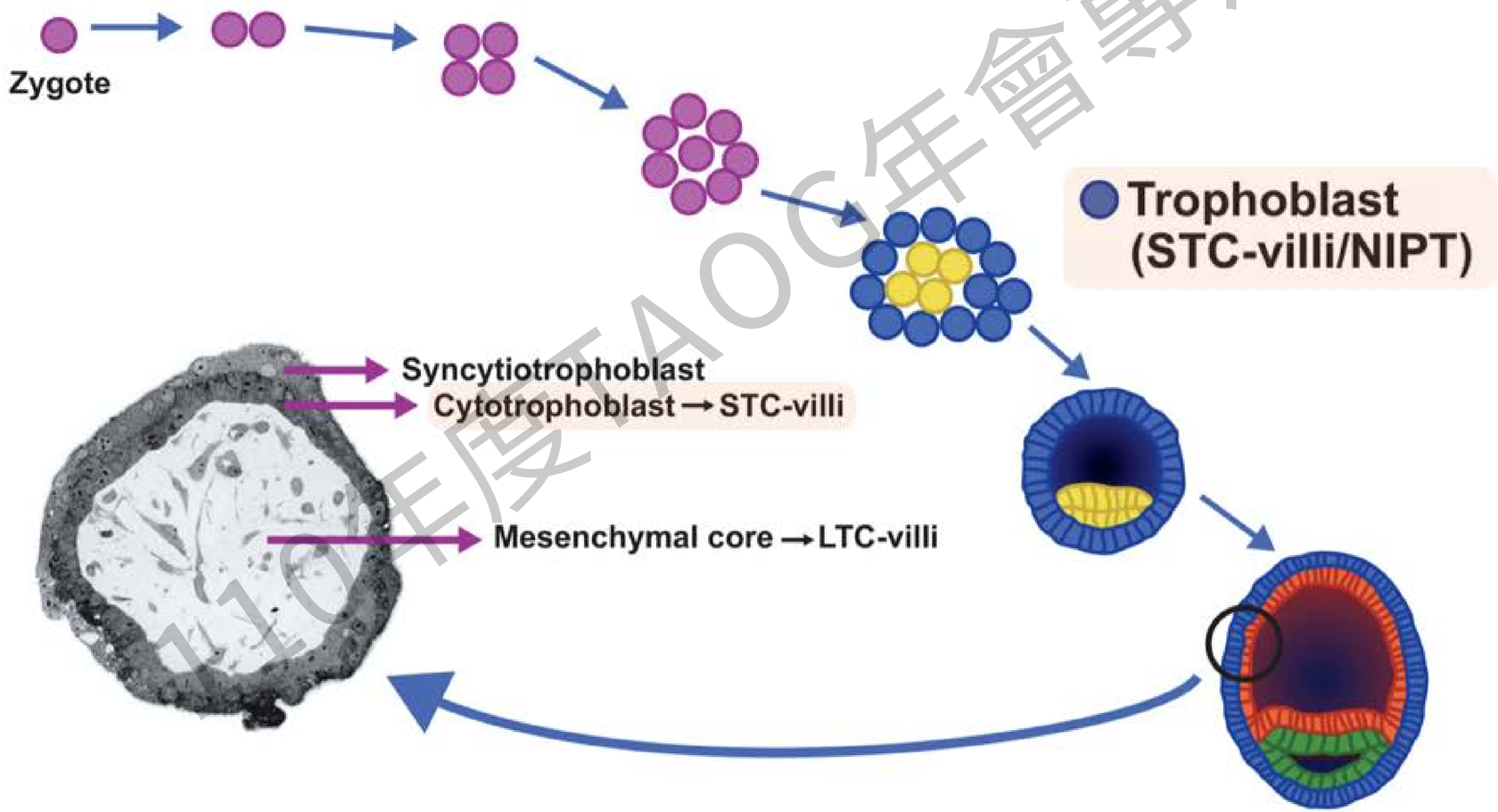
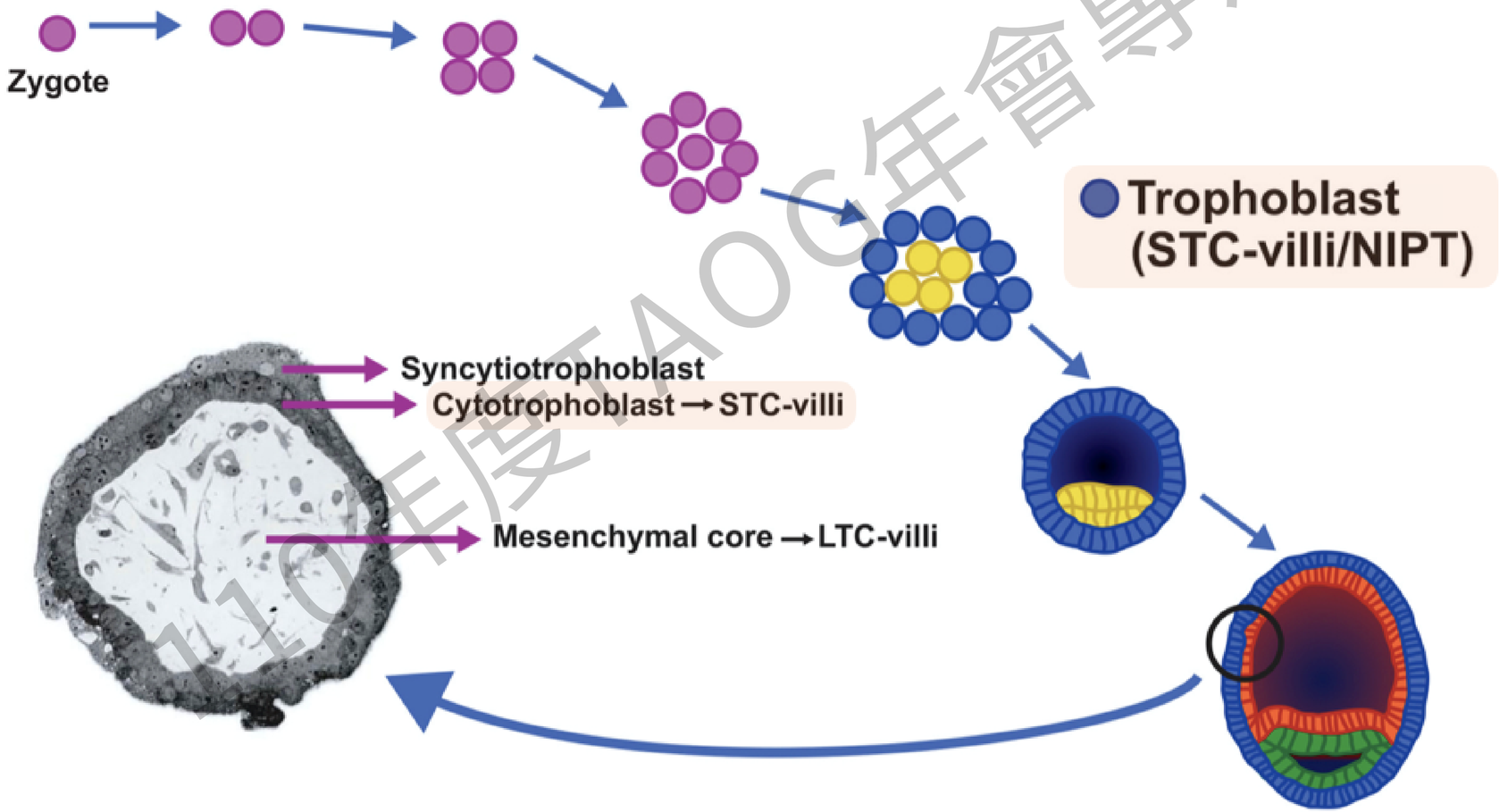


Noninvasive Prenatal Screening (NIPS)

✗ Noninvasive prenatal ~~Diagnosis (NIPD)~~



Noninvasive Prenatal Screening (NIPS)

“No Call”

Low Fetal Fraction

- GA <10w
- Maternal obesity
- Maternal malignancy
- Maternal transplant

Noninvasive Prenatal Screening (NIPS)

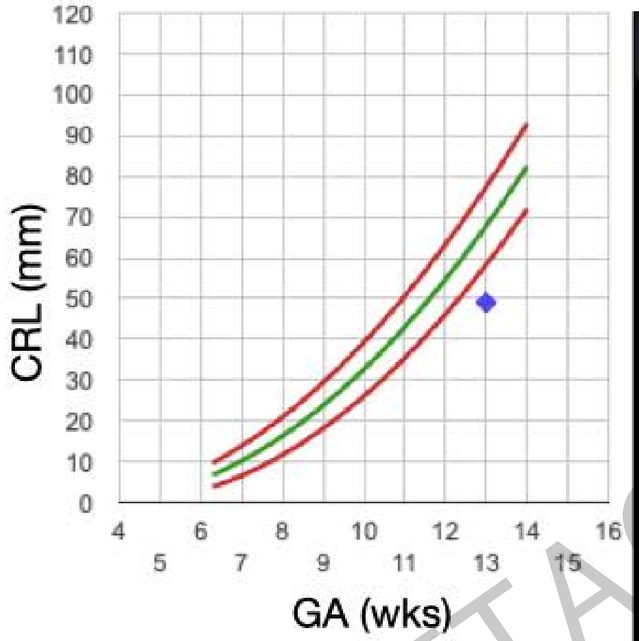
"No Call"	FP
Low Fetal Fraction • GA <10w • Maternal obesity • Maternal malignancy • Maternal transplant	**CPM** (Confined placental mosaicism) **Co-twin demise** (co-twin with aneuploidy)

Noninvasive Prenatal Screening (NIPS)

"No Call"	FP	FN
Low Fetal Fraction • GA <10w • Maternal obesity • Maternal malignancy • Maternal transplant	**CPM** (Confined placental mosaicism) **Co-twin demise** (co-twin with aneuploidy)	**Small placenta** • low β-hCG, PAPP-A e.g. T18, T13, digynic triploidy... **Placental mosaicism** **DC twin**

Taiji Case

13w0d Taiji Clinic: **digynic triploidy**



- asymmetric FGR
- relative large head

Termination of pregnancy

Taiji Case

NIPS: low risk

Taiji Case

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22w0d Taiji Clinic: **multiple anomalies**

- bilateral CPCs
- open 4th ventricle
- large VSD
- trivial TR
- suspected CoA
- T12 hemivertebra
- equivocal anal pit

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22w4d Amniocentesis (rapid test): **T18**

Termination of pregnancy

Taiji Case

IVF; **NIPS: low risk**

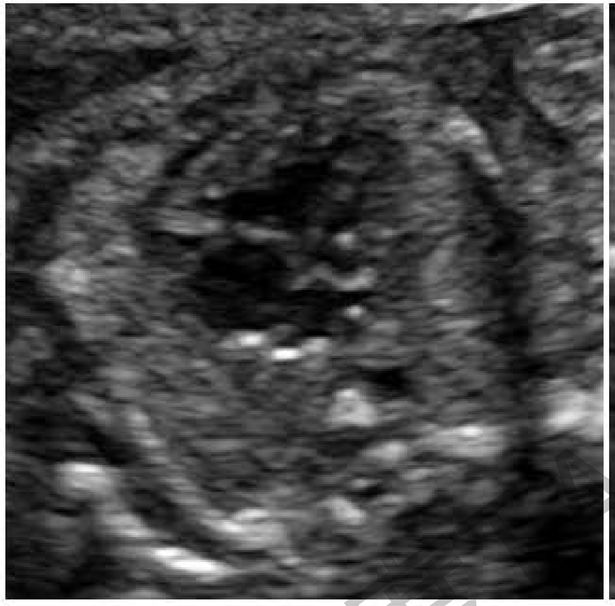
Cri-du-Chat syndrome/貓哭症	5p15	-0.37	未檢出異常

Taiji Case

IVF; **NIPS: low risk**

Cri-du-Chat syndrome/猫哭症	5p15	-0.37	未検出異常

22w3d Taiji Clinic: **CHD, severe IUGR** (z-score of HC, AC, FL, TCD: all below -2 to -3)



- hypoplastic LV
- VSD
- CoA
- PLSVC
- PRUV
- low conus medularis

Taiji Case

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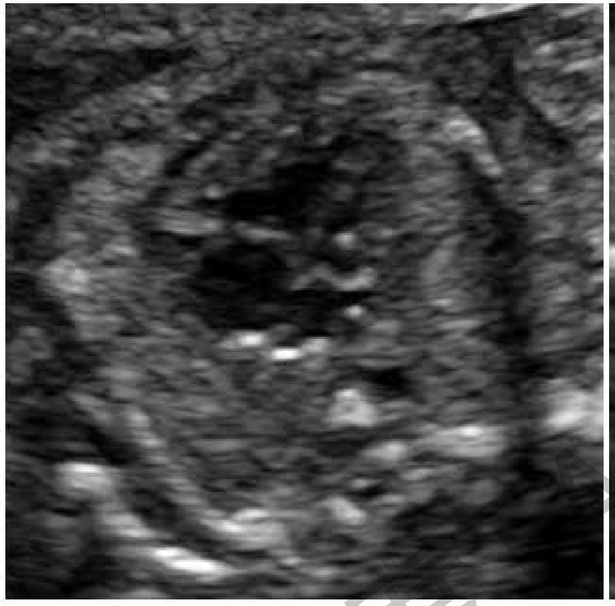


- hypoplastic LV
- VSD
- CoA
- PLSVC
- PRUV
- low conus medularis

24w4d **Karyotyping**: **46,XY,del(5)(p13)**

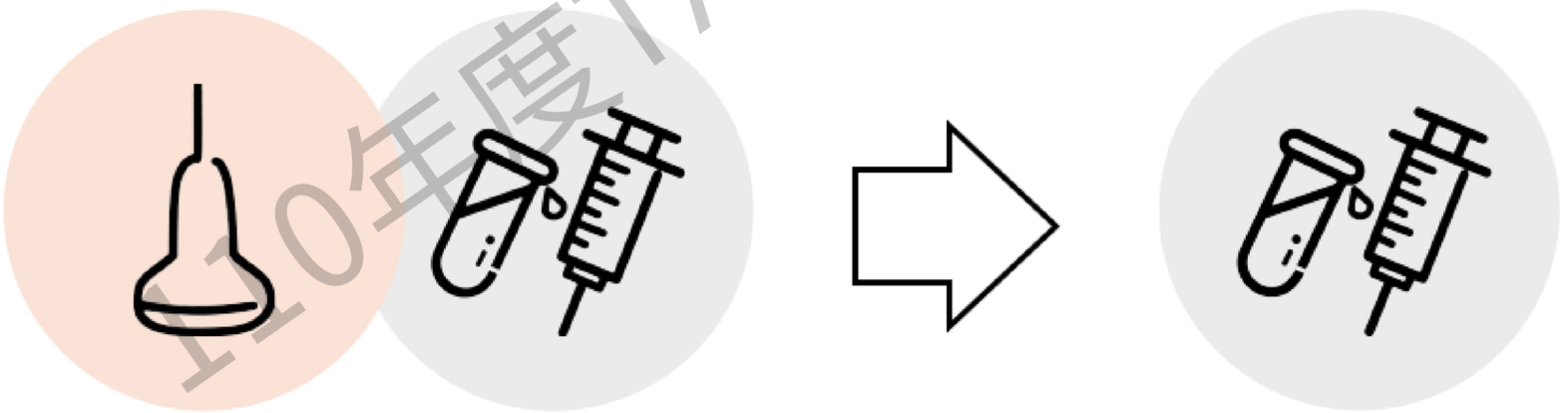
CMA: **Cri-du-chat syndrome**

5p15.33p13.2: **35.1Mb** deletions
5p13.2: 833kb deletions

25w6d Termination of pregnancy

??? ③

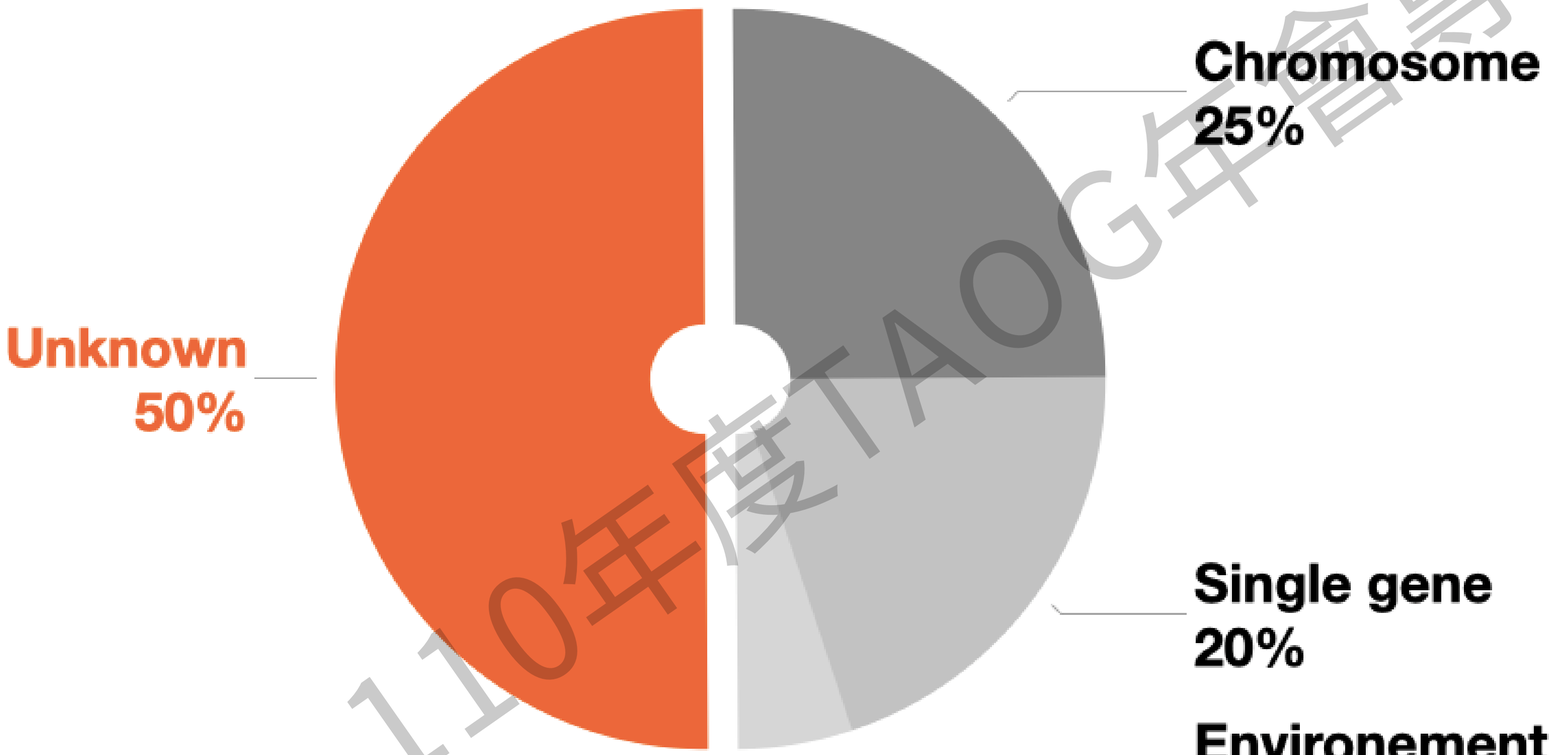
What would be missed in the first trimester
if NT measurement is replaced by cfDNA screening?



CFTS

NIPS

The causes of over 50% of birth defects are UNKNOWN.



Seminar in Fetal & Neonatal Medicine (2013) 1-10
AJOG (2005) 192, 1005-21

Relation between NT Thickness and Prevalence of CHD and Major Fetal Abnormalities

Nuchal translucency	Chromosomal defects	Fetal death	Major fetal abnormalities	Congenital heart disease	Alive and well
(<95th centile)	0.2%	1.3%	1.6%	1.36%	97%
95th-99th centiles	3.7%	1.3%	2.5%	1.82%	93%
3.5-4.4 mm	21.1%	2.7%	**10%**	**3.52%**	70%
4.5-5.4 mm	33.3%	3.4%	**18.5%**	**6.44%**	50%
5.5-6.4 mm	50.5%	10.1%	**24.2%**	**12.67%**	30%
>6.5 mm	64.5%	19%	**46.2%**		**15%**
	H	H	H	H	L

chromosomally normal fetuses

Ultrasound Obstet Gynecol 2020; 55: 637–644

Diagnosis of major heart defects by routine 1st-TM ultrasound examination: association with increased NT, TR and abnormal in DV

… K.H. Nicolaides (UK)

Timing of diagnosis of major heart defects and their association with increased NT, TR and abnormal flow in DV at 11–13-week scan

	Cases (n)	Timing of Dx				Increased NT		TR	Abnormal DV flow	Any marker
		1st-TM	2nd-TM	3rd-TM	Postnatal	≥ p95	≥ p99			
Major CHD	211	113 (53.6)	82 (38.9)	10 (4.7)	6 (2.8)	77 (36.5)	45 (21.3)	61 (28.9)	58 (27.5)	117 (55.5)
Normal live birth	92998	-	-	-	-	5678 (6.1)	857 (0.9)	1136 (1.2)	1644 (1.8)	8166 (8.8)

… highlight the continuing importance of the markers in screening for heart defects

Prenatal Diagnosis. 2020;40:197–205

Is there still a role for NT measurement in the changing paradigm of first trimester screening?

Francesca Bardi et al. (the Netherlands)

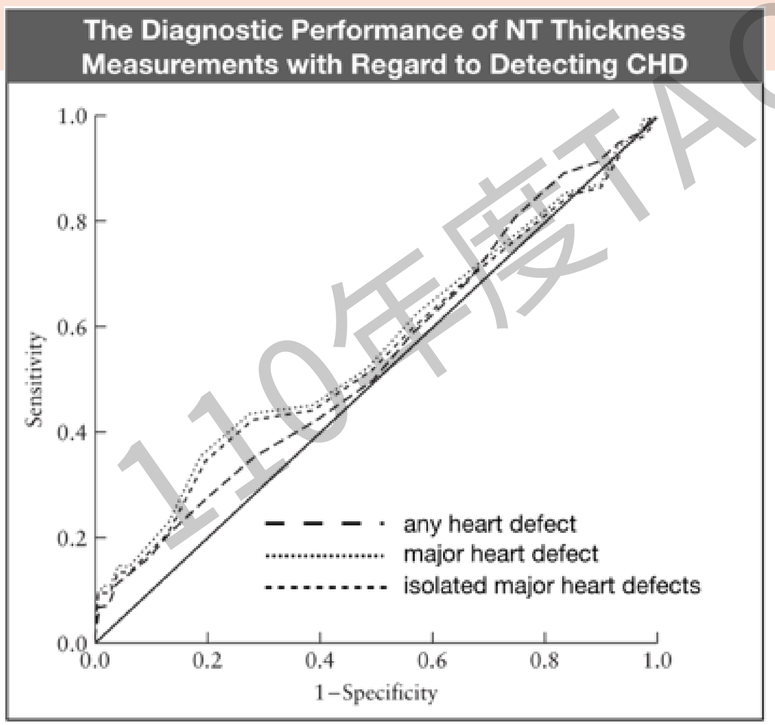
Congenital abnormalities associated with increased NT (>95th centile)

		Congenital abnormality *n* (%)						
			Detected genetic abnormality (*n*=636, 33.3%)					
			Chromosomal (*n*=560, 29.4%)					(isolated) Structural (*n*=178, 9.3%)
NT (mm)	All fetuses	All abnormal fetuses	Total	T21-18-13*	Other¶	Submicroscopic‡	Single-gene disorders§	
p95-p99	894 (47)	190 (21.3)	124 (13.8)	112 (12.5)	12 (1.3)	8 (0.9)	5 (0.6)	53 (5.9)
≥p99	1007 (53)	624 (62)	436 (43.2)	344 (34)	92 (9.1)	30 (3)	33 (3.3)	125 (12.4)

If cfDNA were used as the only 1st TM screening test, 34% of fetal congenital abnormalities would be missed in the 1st TM of pregnancy.

Nuchal translucency cannot be relied on as the sole of major screening tool for CHD.

Ultrasound Obstet Gynecol 2006; 27: 613–618



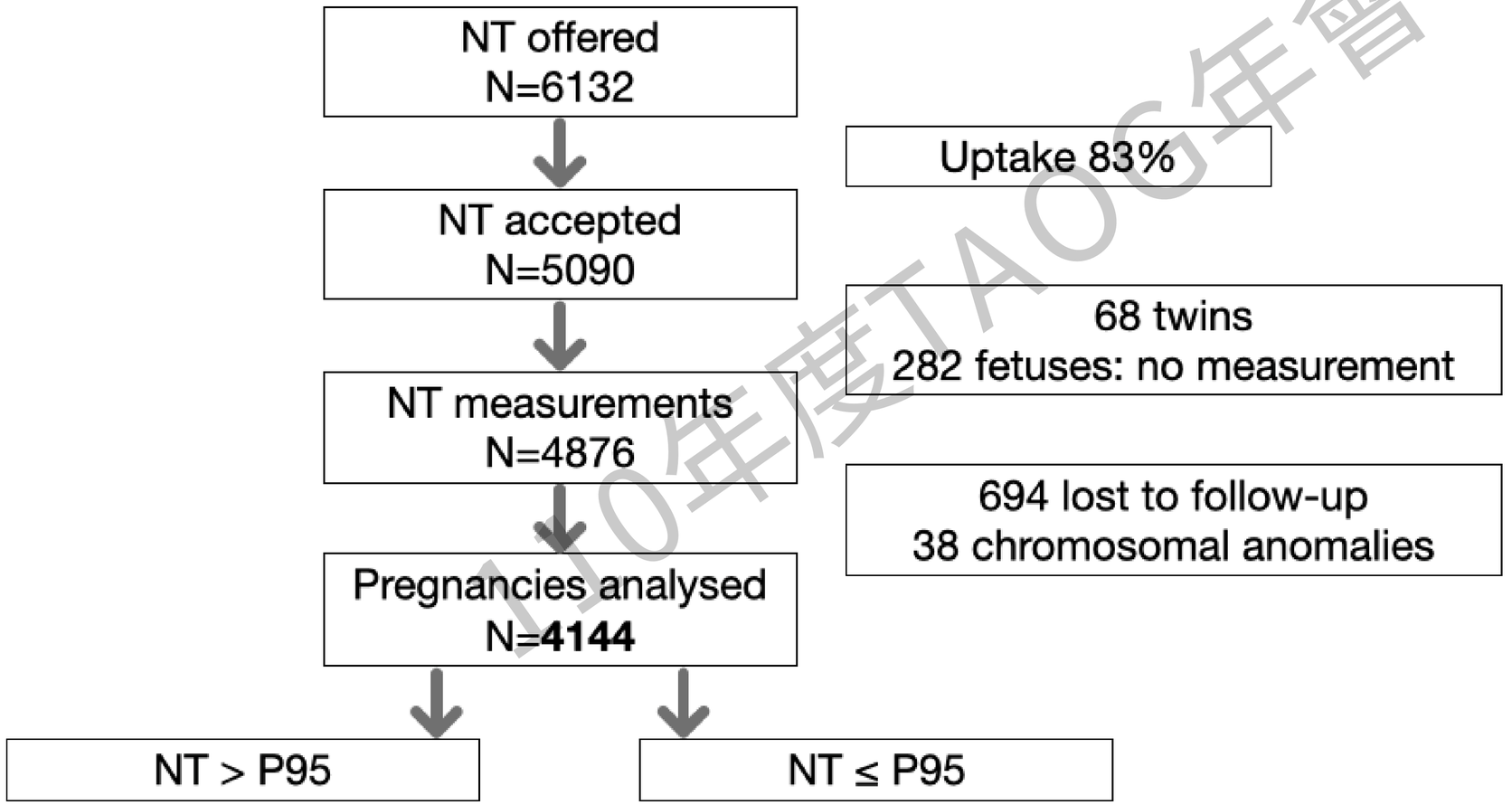
Most cases (67.1%) with major abnormalities presented with normal nuchal translucency.

Ultrasound Obstet Gynecol 2013; 42: 300–309

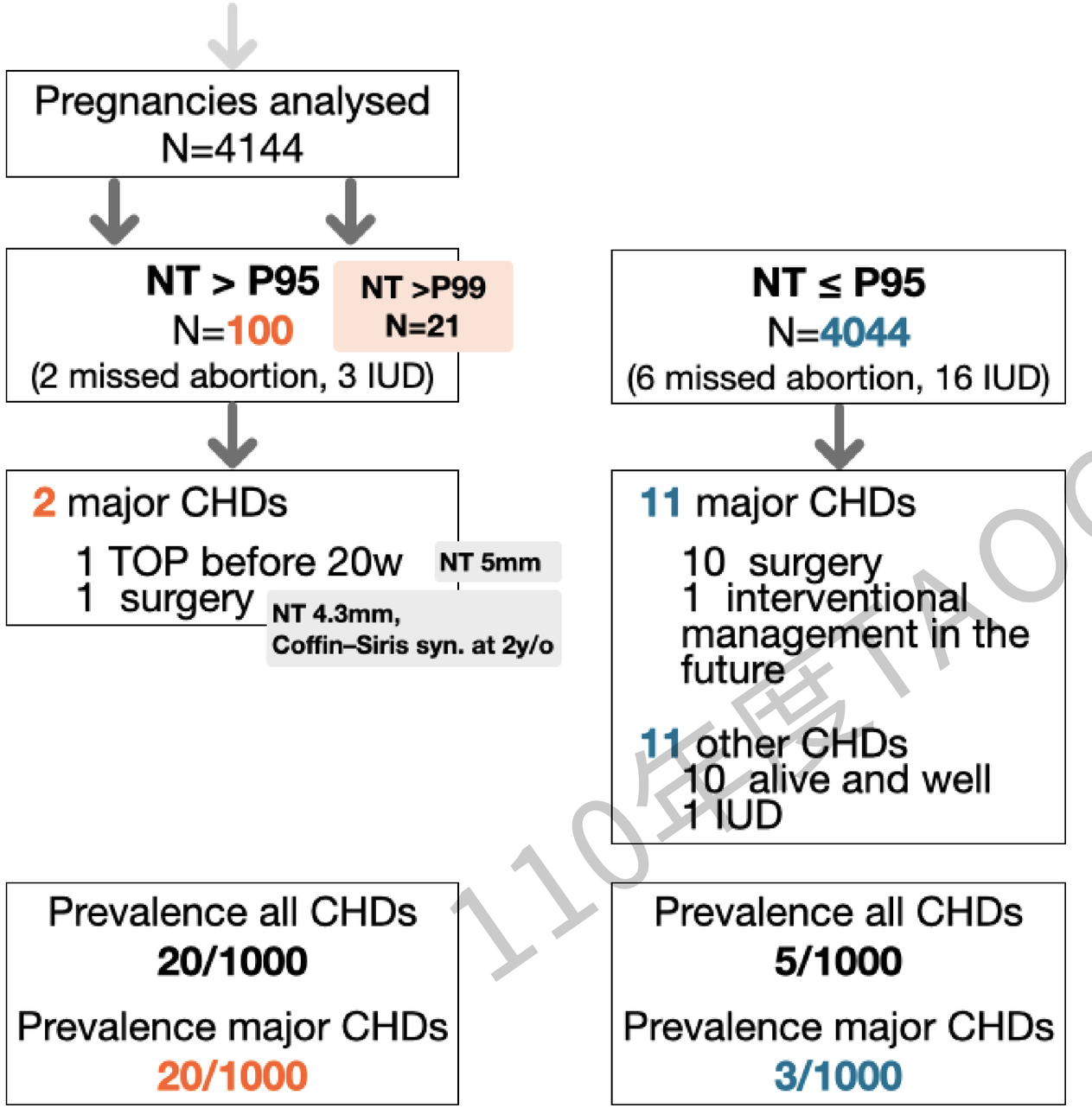
Prenat Diagn 2007; 27: 164–169

Nuchal translucency measurement and congenital heart defects: modest association in low-risk pregnancies

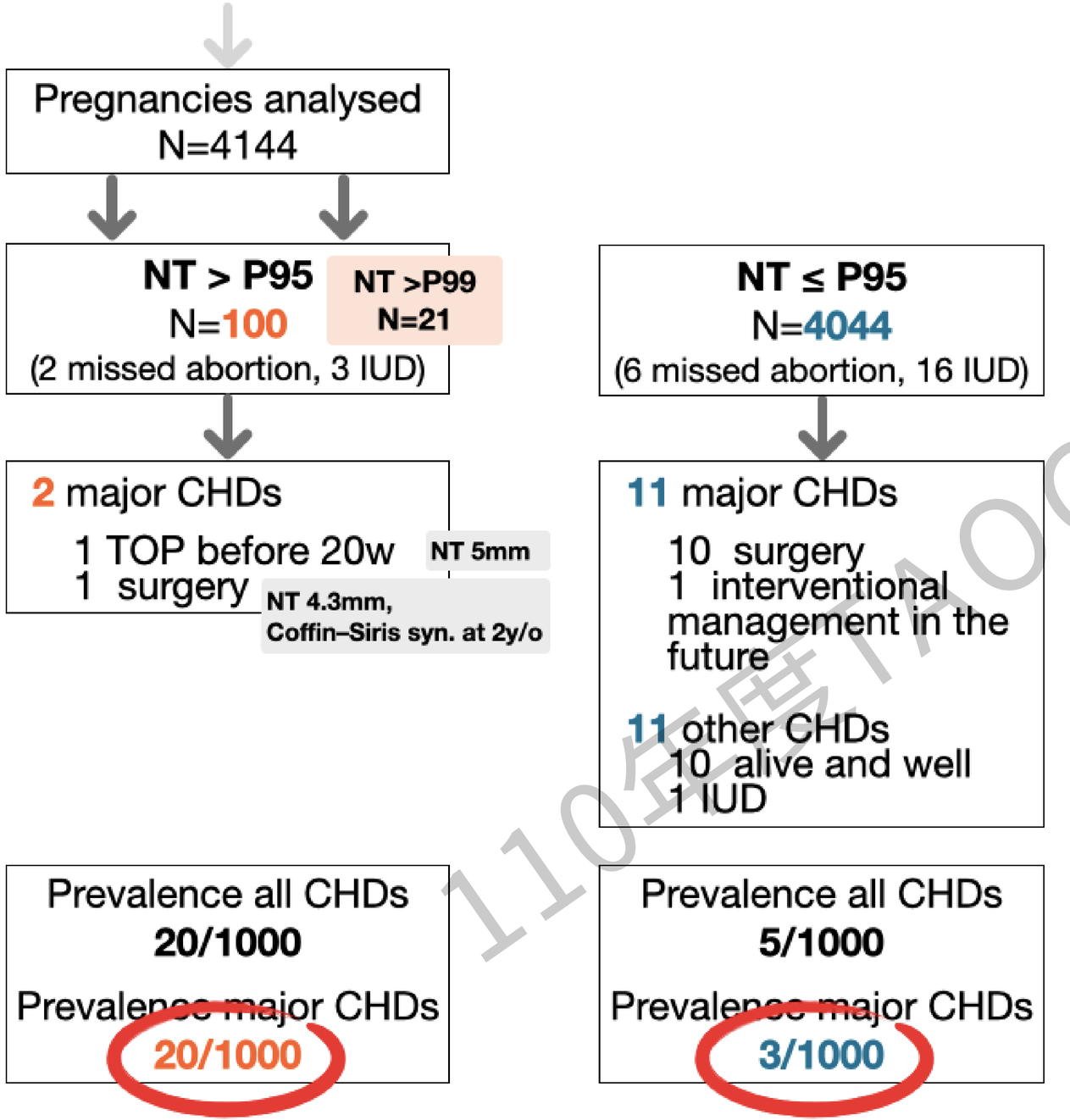
M.A.Muller et al. (The Netherlands)



Prenat Diagn 2007; 27: 164–169

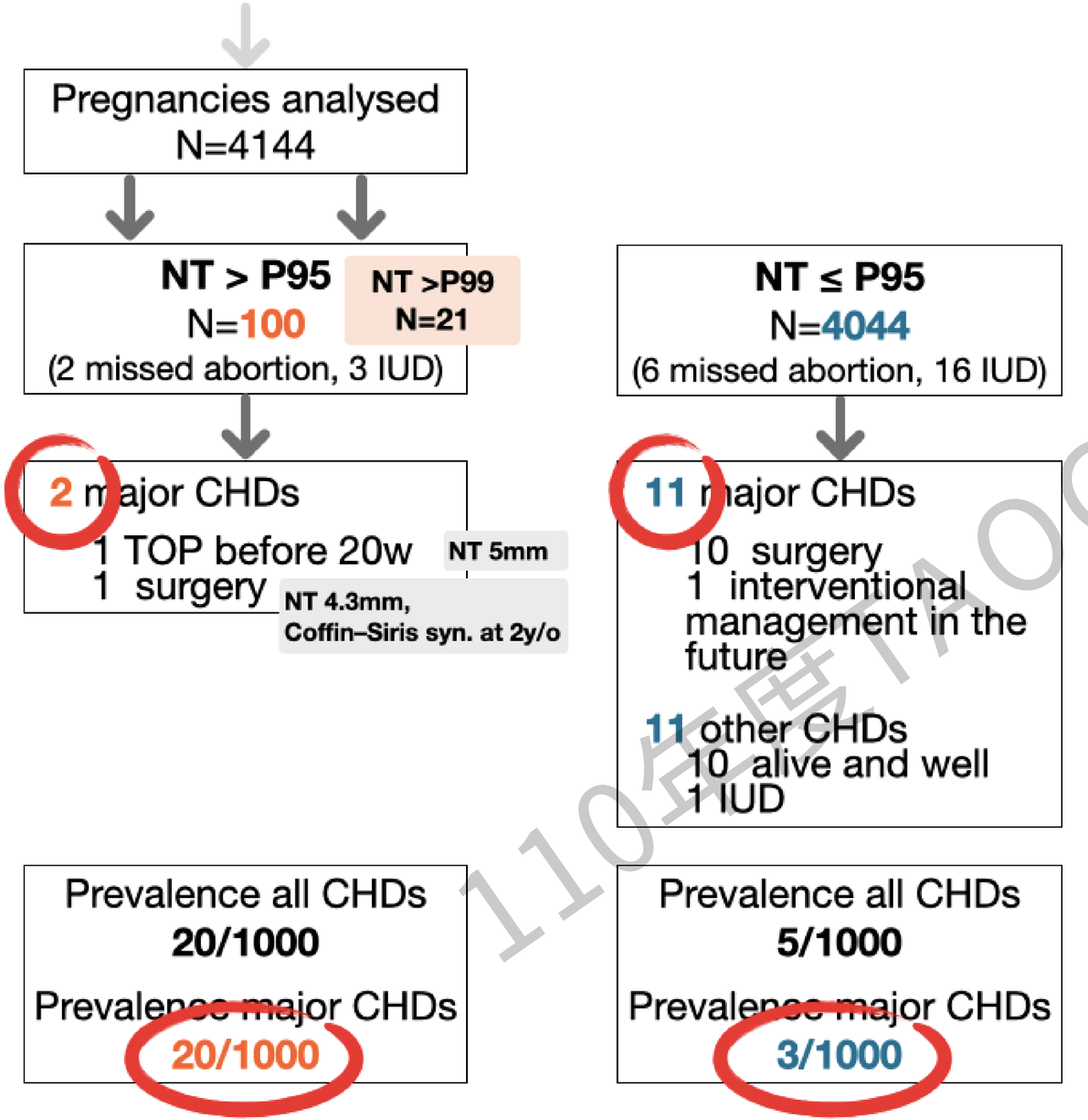


Prenat Diagn 2007; 27: 164–169



- NT above the 95th or 99th percentile is a strong indication for specialised echocardiography.

Prenat Diagn 2007; 27: 164–169



- NT above the 95th or 99th percentile is a strong indication for specialised echocardiography.
- **Most cardiac anomalies occur in low-risk pregnancies.**

Lancet 348(9031):854-857

Ultrasound Obstet Gynecol 2020; 55: 637–644

Diagnosis of major heart defects by routine 1st-TM ultrasound examination: association with increased NT, TR and abnormal in DV

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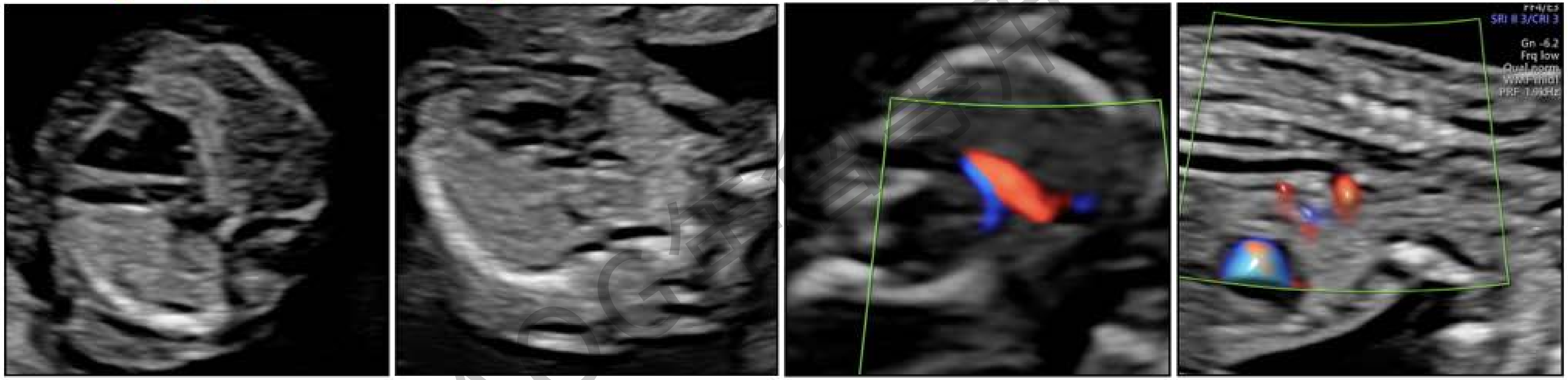
Normal live birth	92998	-	-	-	-	5678 (6.1)	857 (0.9)	1136 (1.2)	1644 (1.8)	8166 (8.8)

More than half of major heart defects can be detected by routine ultrasound exam at 11-13 weeks' gestation.

Taiji Case

12w2d Taiji Clinic: (CFTS) **T21 risk: 1/15876**

14w1d **CHD (suspected HLHS)**



- unbalanced 4CV
- small LVOT
- small AAo with reversed flow
- hypoplastic Ao arch
- reversed flow of foramen ovale

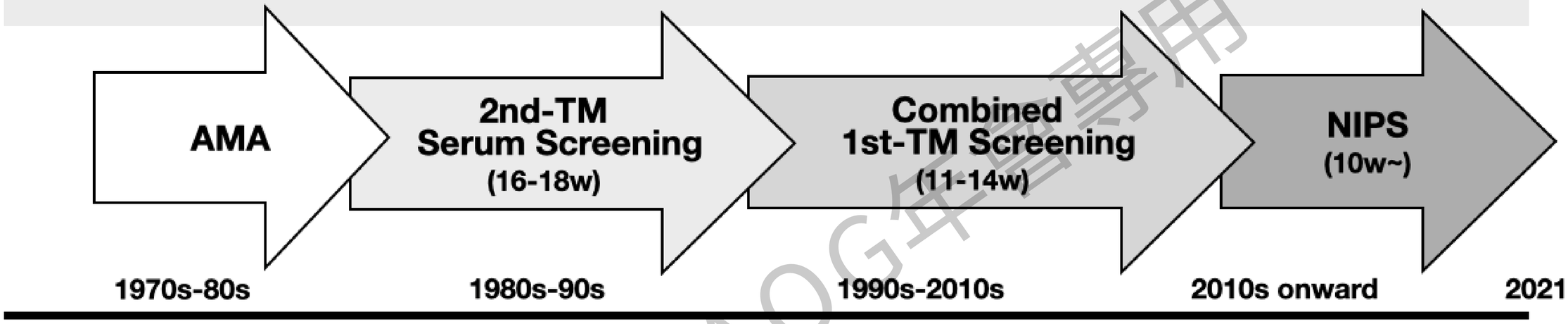
Karyotyping & CMA are indicated: not done

Termination of pregnancy

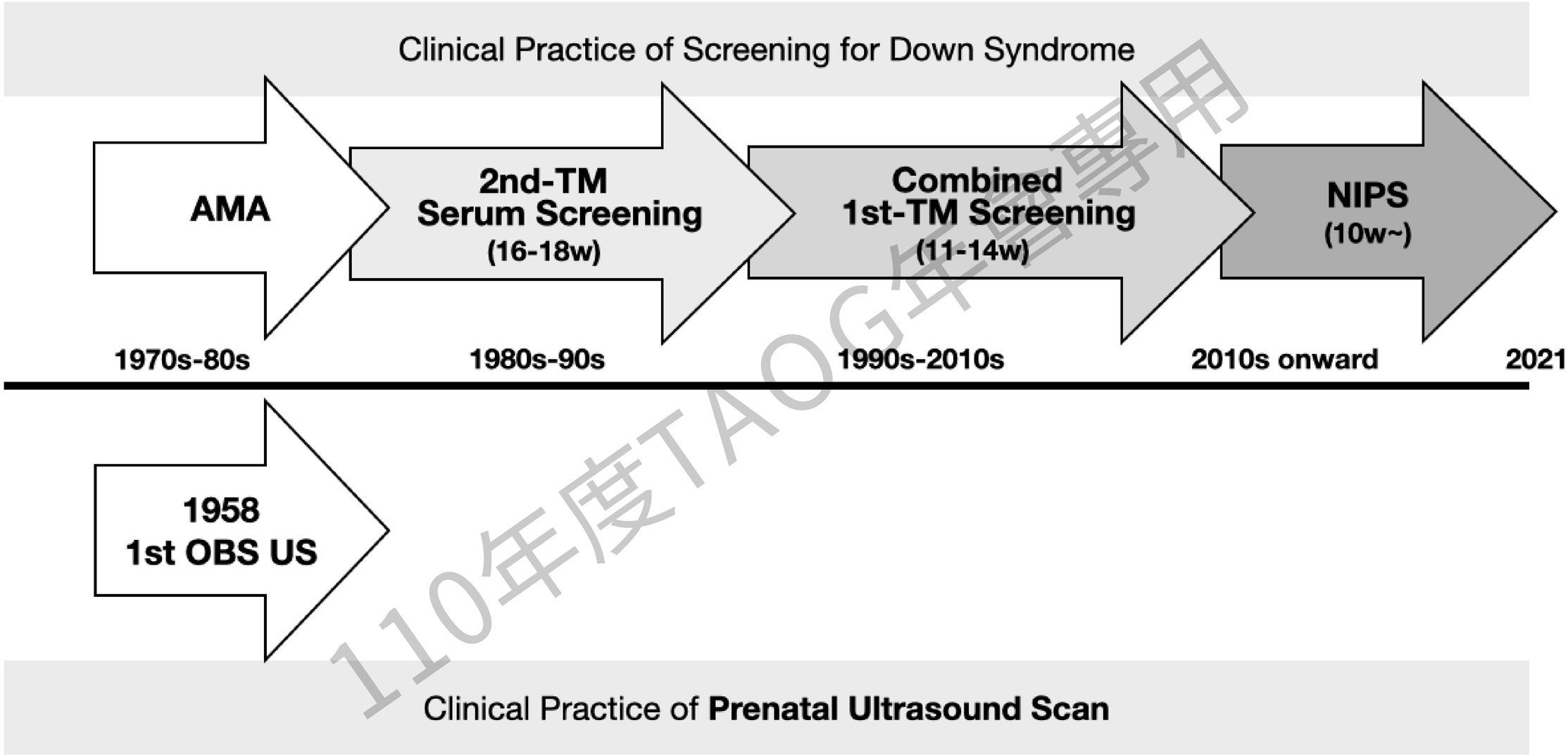
In the Era of cfDNA Testing…

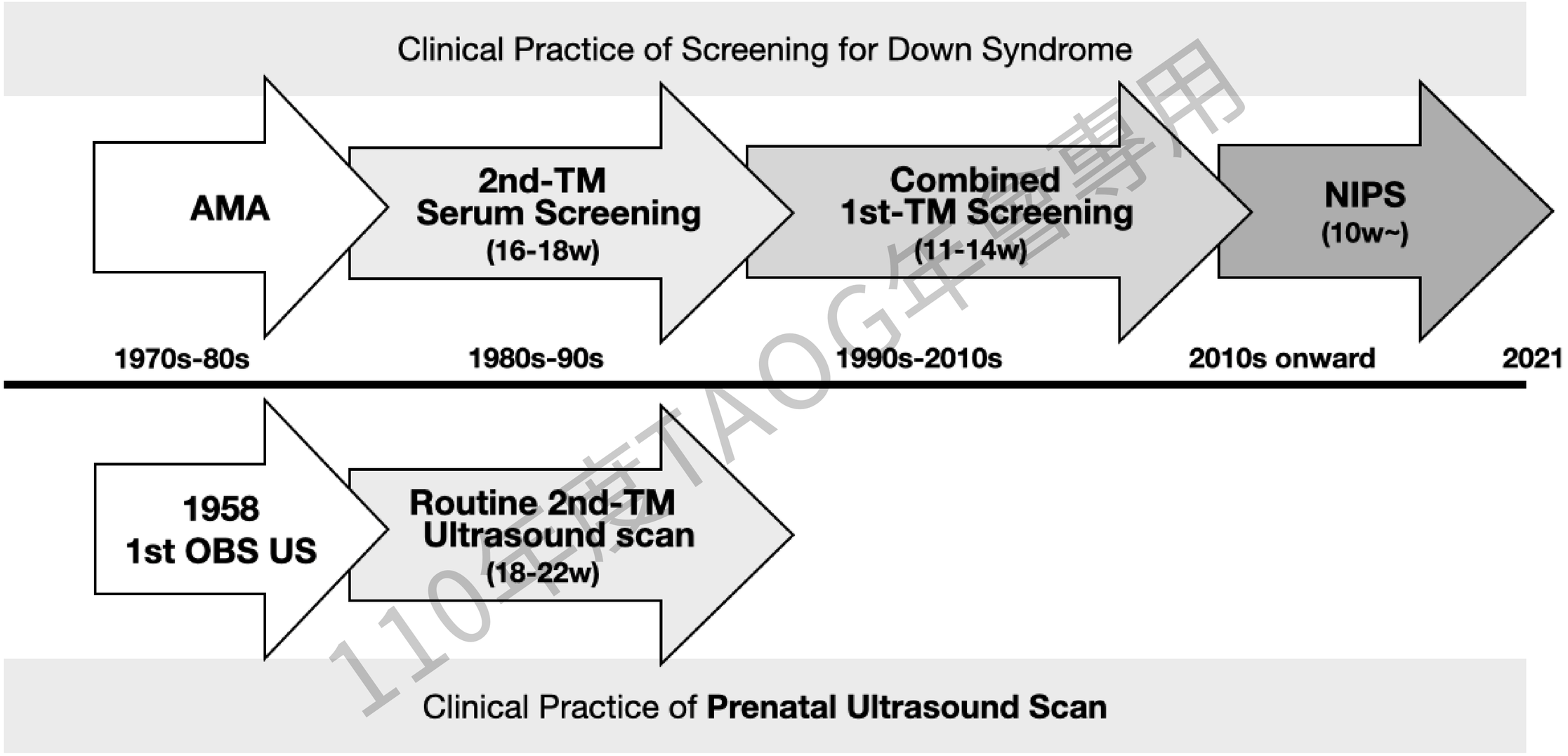
2. The Value of First Trimester Ultrasound

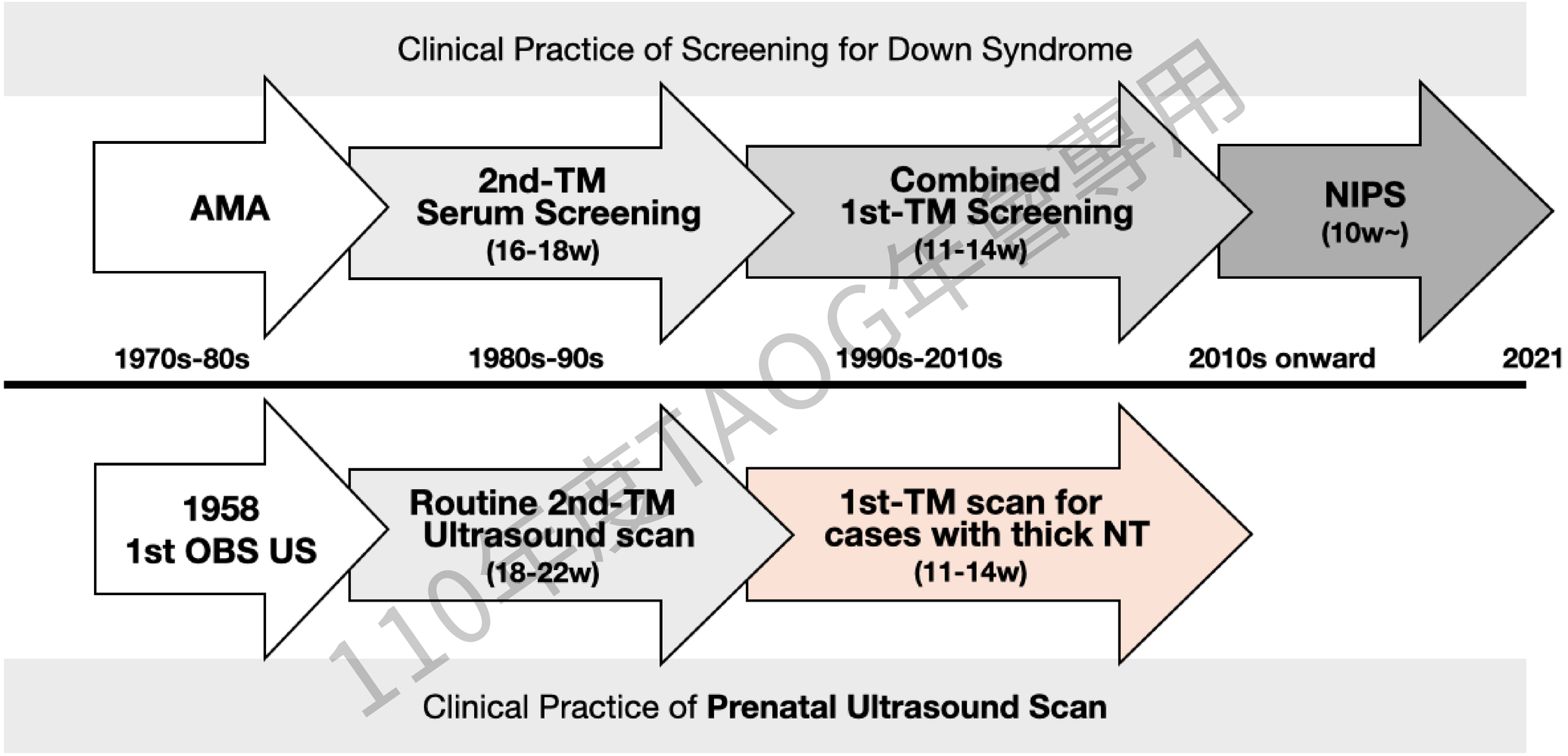
Clinical Practice of Screening for Down Syndrome

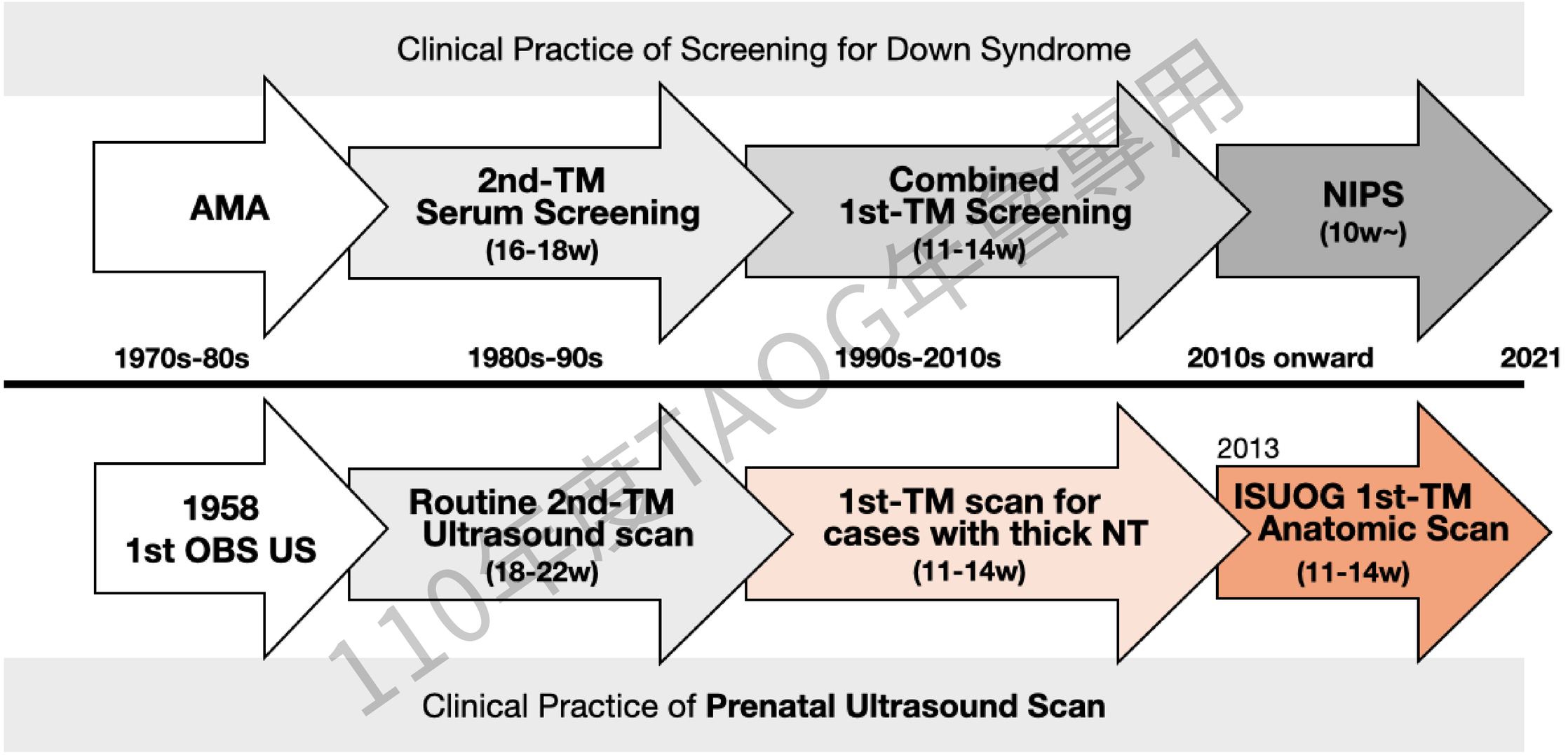


Clinical Practice of **Prenatal Ultrasound Scan**



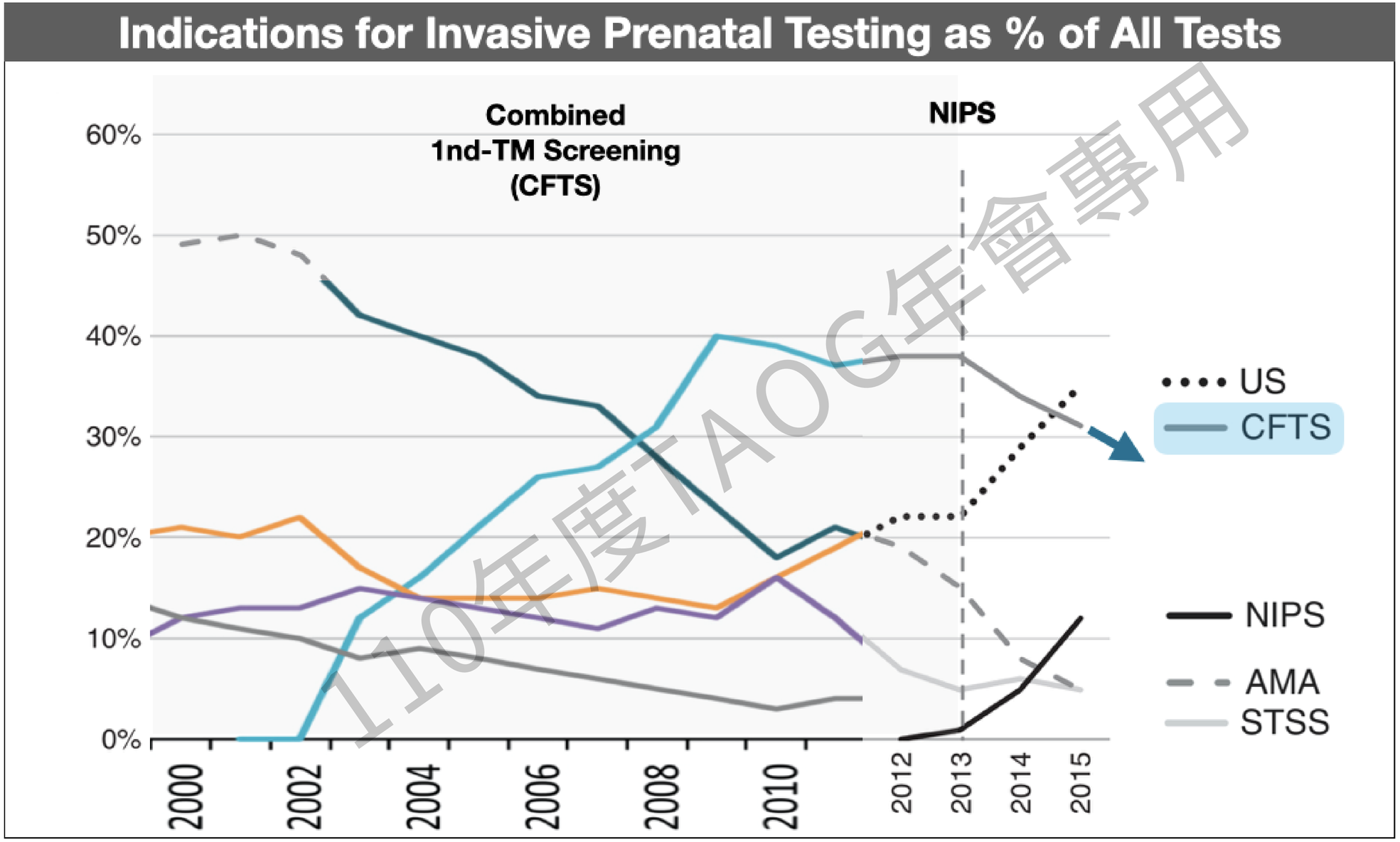






Genet Med. 2017 Dec;19(12): 13838-1345

Indications for Invasive Prenatal Testing as % of All Tests



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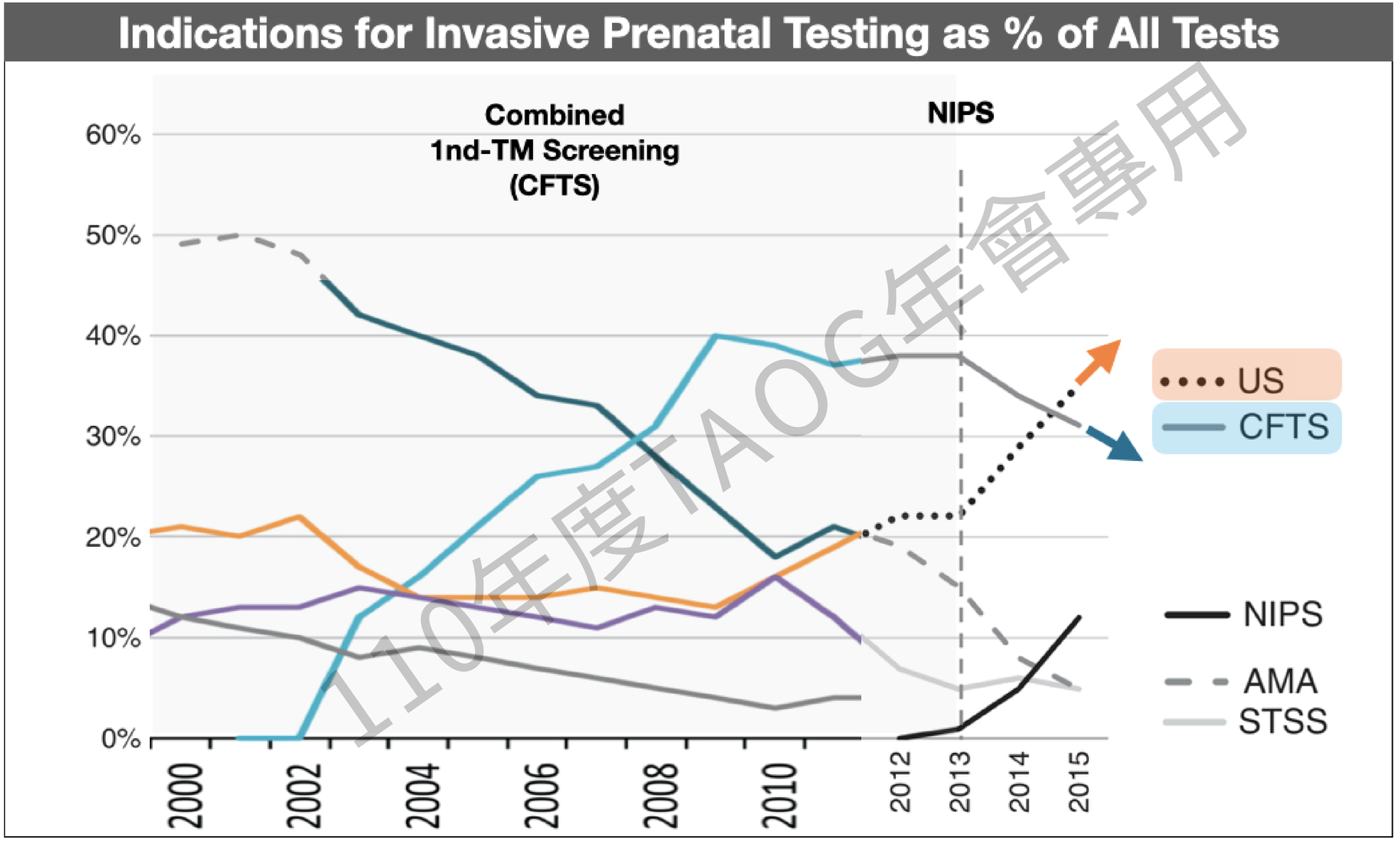


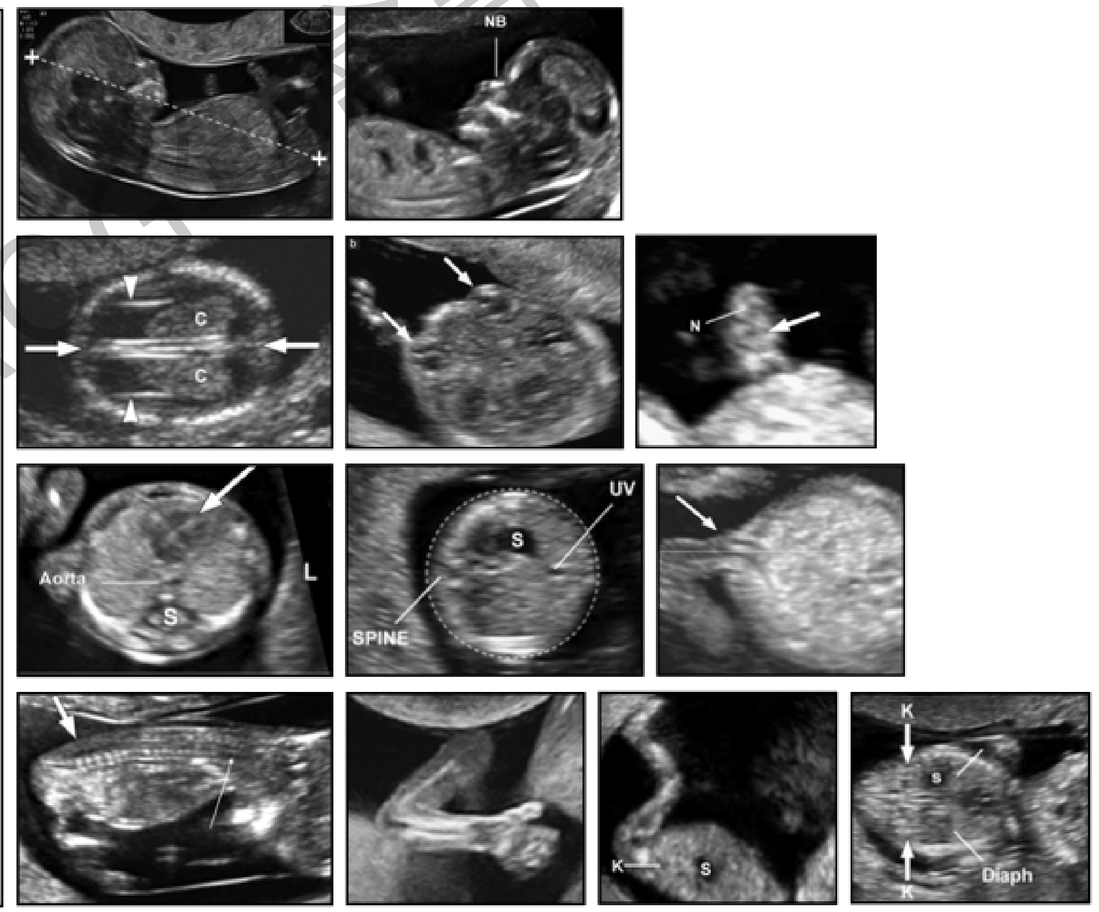
Ultrasound Obstet Gynecol 2013; 41: 102-113

ISUOG Practice Guidelines:
Performance of first-trimester fetal ultrasound scan

isuog.org GUIDELINES

11w-13w6d

Organ/anatomical area	*Present and/or normal?*
Head	Present
	Cranial bones
	Midline falx
	Choroid-plexus-filled ventricles
Neck	Normal appearance
	Nuchal translucency thickness (if accepted after informed consent and trained/certified operator available)*
Face	Eyes with lens*
	Nasal bone*
	Normal profile/mandible*
	Intact lips*
Spine	Vertebrae (longitudinal and axial)*
	Intact overlying skin*
Chest	Symmetrical lung fields
	No effusions or masses
Heart	Cardiac regular activity
	Four symmetrical chambers*
Abdomen	Stomach present in left upper quadrant
	Bladder*
	Kidneys*
Abdominal wall	Normal cord insertion
	No umbilical defects
Extremities	Four limbs each with three segments
	Hands and feet with normal orientation*
Placenta	Size and texture
Cord	Three-vessel cord*



Ultrasound Obstet Gynecol 2017; 49: 815–816

ISUOG updated consensus statement on the impact of cfDNA aneuploidy testing on screening policies and prenatal ultrasound practice

isuog.org
CONSENSUS STATEMENT

All women should be offered a first-trimester ultrasound scan according to ISUOG guidelines, regardless of their intention to undergo cfDNA testing.

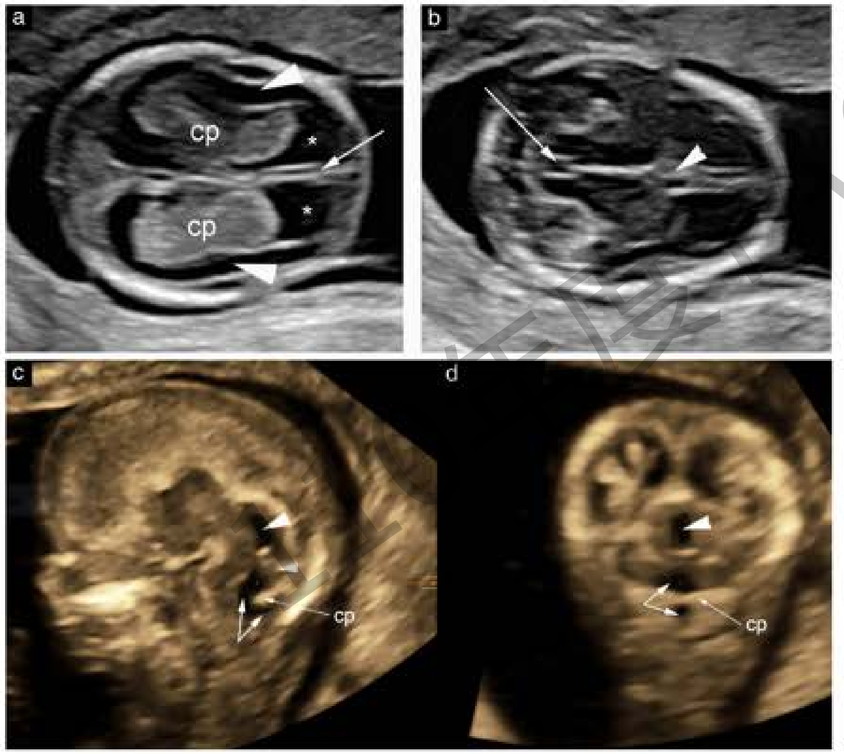
Ultrasound Obstet Gynecol 2021 Apr;57(4):661-671

ISUOG Practice Guidelines (updated): Sonographic examination of the fetal central nervous system. Part 2: performance of targeted neurosonography

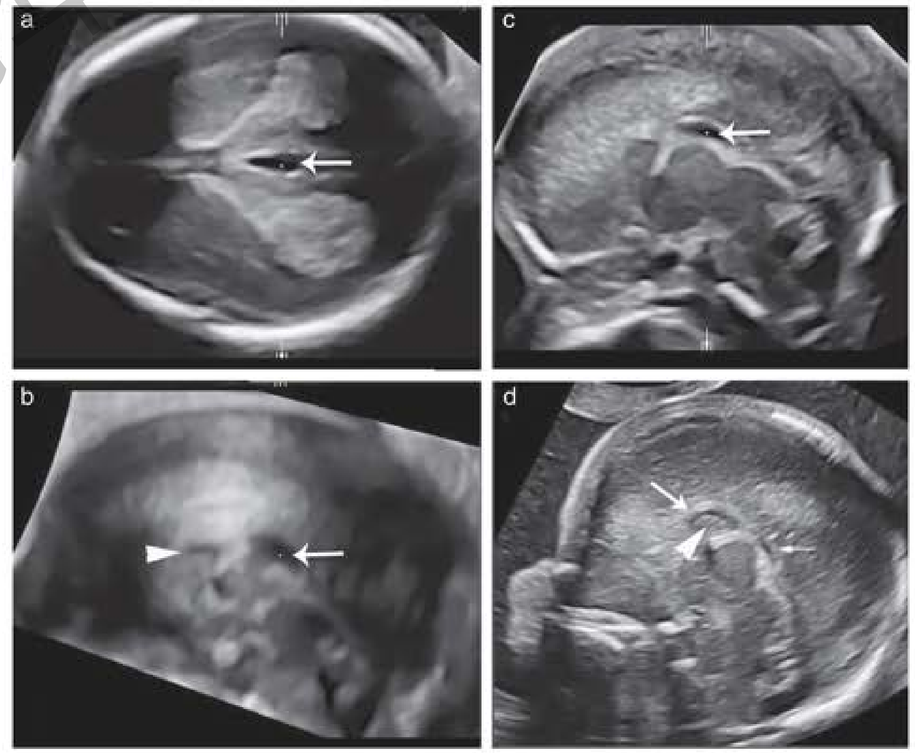
isuog.org GUIDELINES

Neurosonography at 13-17 gestational weeks:

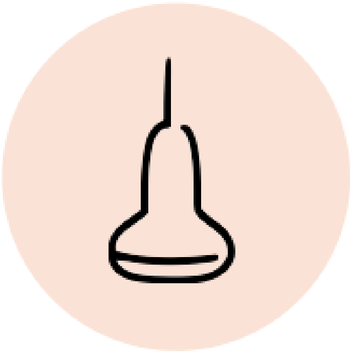
13wks



15wks



AJOG 2005 Sep;193(3 Pt 2):1260-5
AJOG 2004 Oct;191(4):1247-52



High frequency transducers

Knowledge of embryology

- Normal developmental milestones

Operator's experience

- Qualified and trained sonographers

Ultrasound Obstet Gynecol 2017; 50: 429-441

Systematic review of 1st-TM ultrasound screening for detection of fetal structural anomalies and factors that affect screening performance

J.N.Karim et al. (UK)

The use of a **standardized anatomic protocol** was the **most crucial factor** to **improve the sensitivity** of 1st-TM ultrasound screening for all anomalies and major anomalies **in various patient risk groups.**

Am J Obstet Gynecol 2021;224:396.e1-15

Routine first-trimester ultrasound screening using a standardized anatomical protocol

Yime I Liao et al. (China)

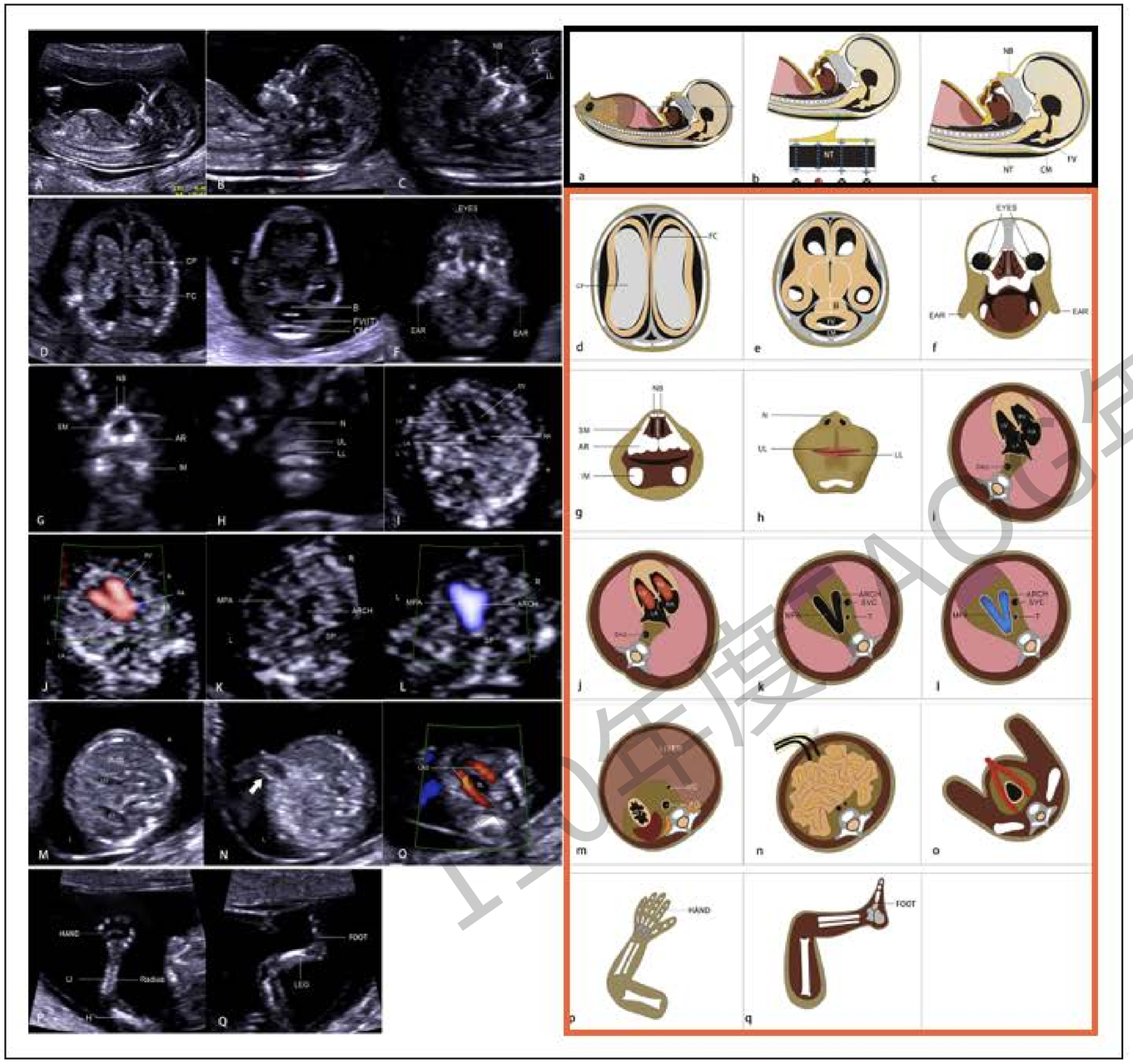
Retrospective study (**2008-2015**)

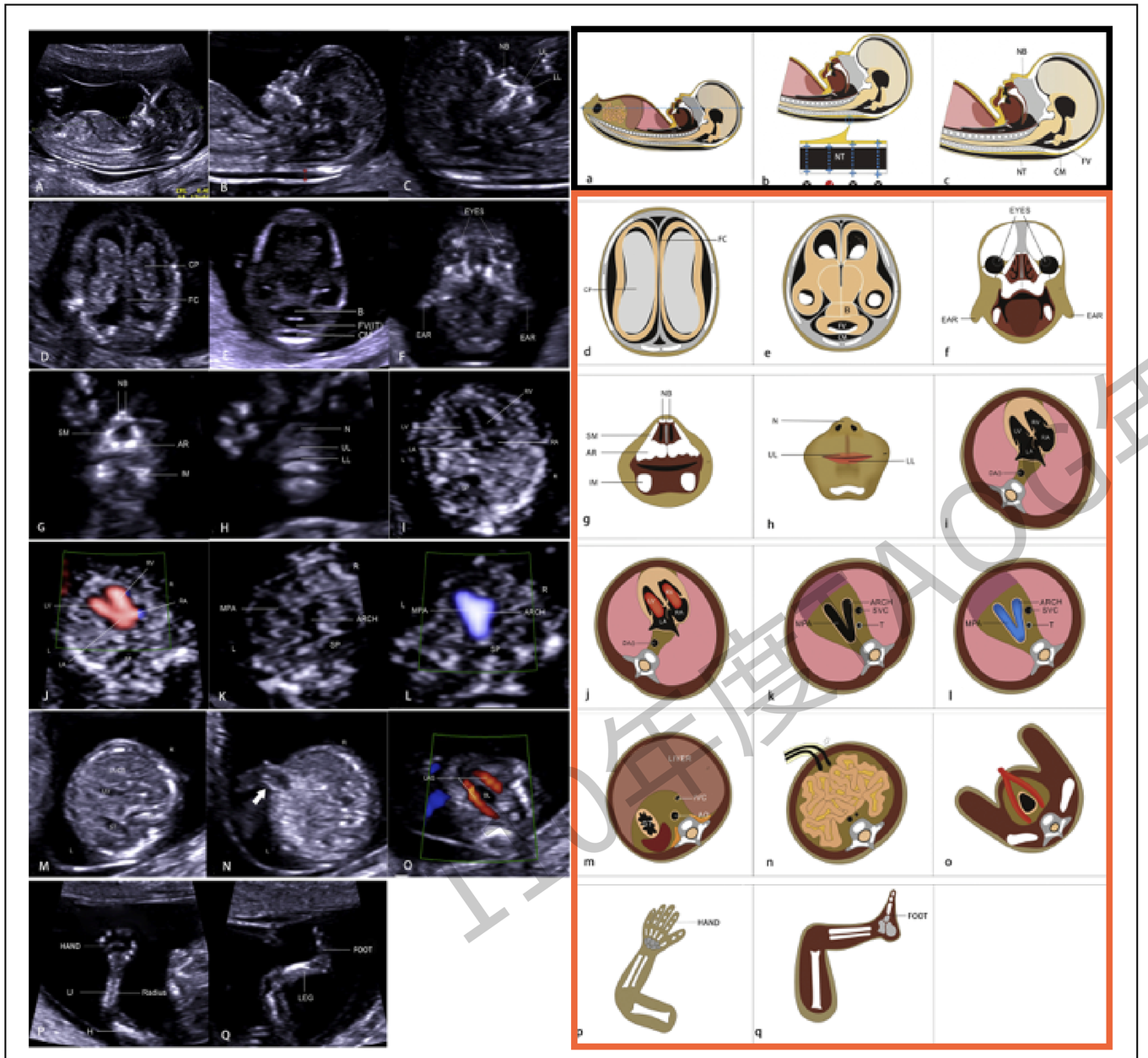
59,063 sequential **unselected** pregnancies

Scans at 11w-13w6d

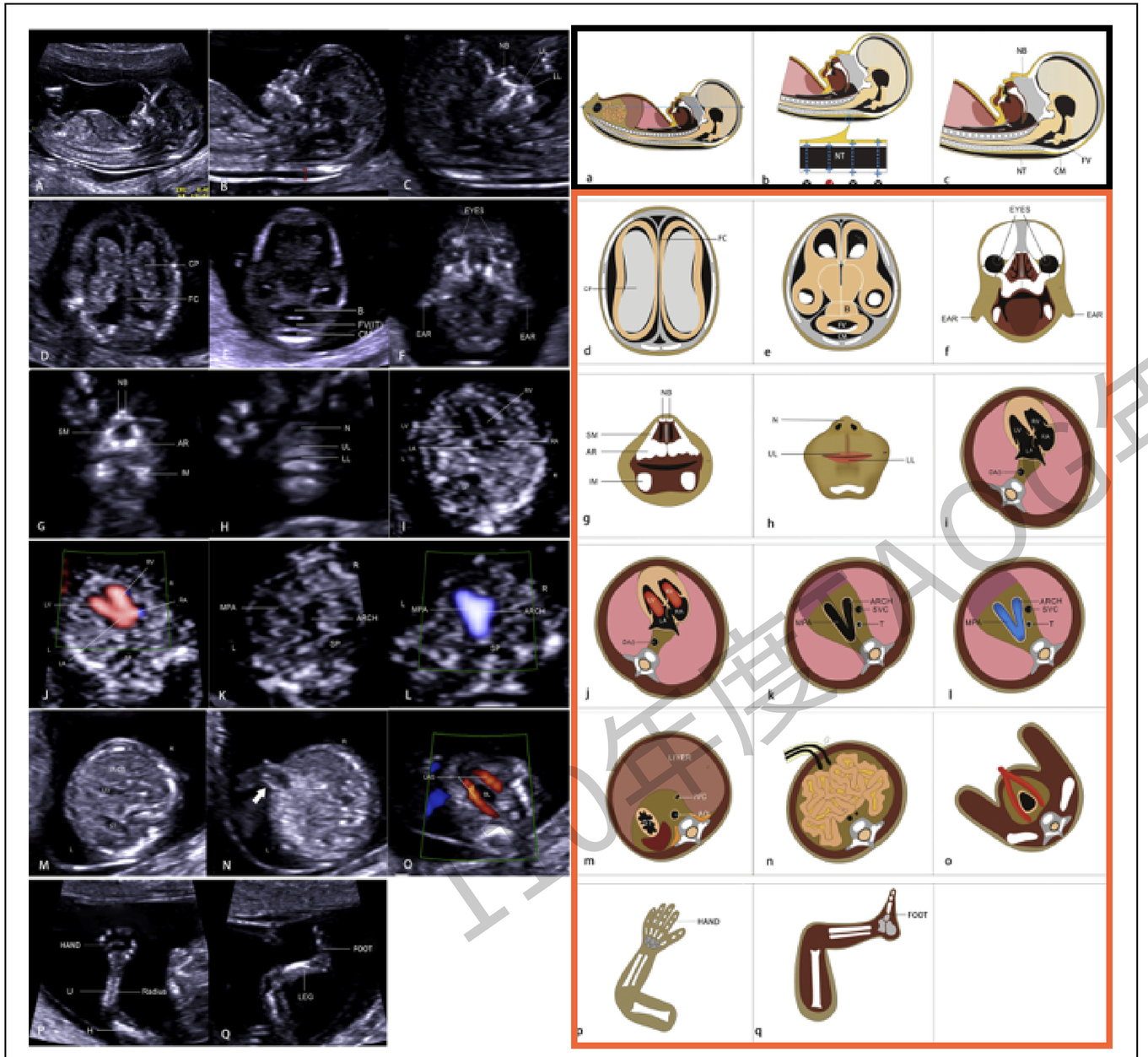
Midsagittal view and 14 standard sections

Duration of exam: normal fetus within 30 mins; abnormal fetus within 1hr





3% of fetuses had at least one structural abnormality



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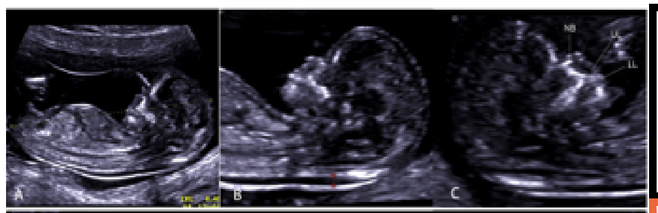
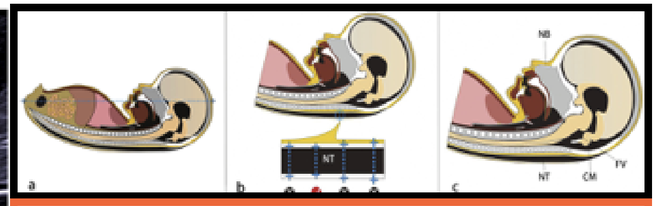
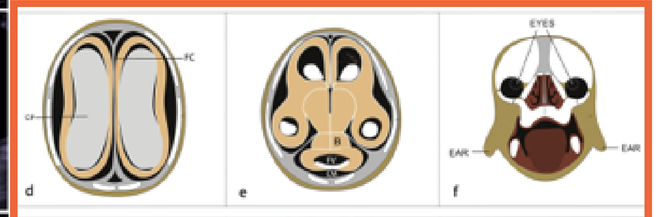
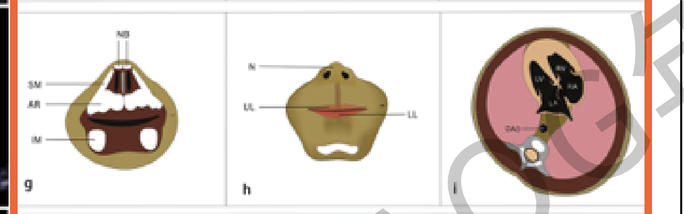
DR of fetal anomalies:

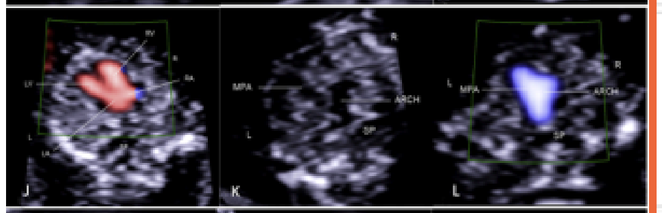
1st-TM: 43.1%
2nd-TM: 30.9%
3rd-TM: 7.1%
Postnatal: 18.9%

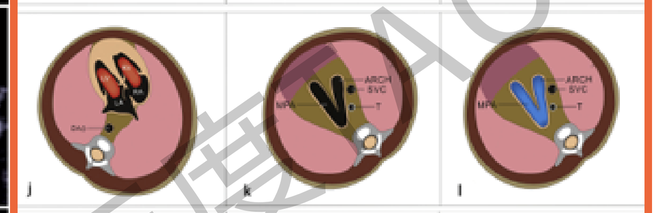


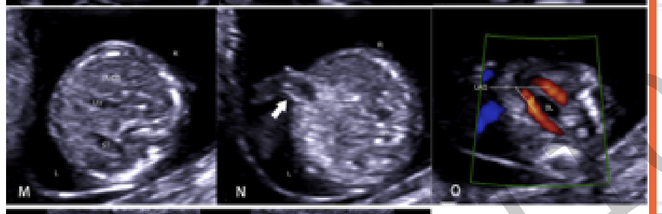


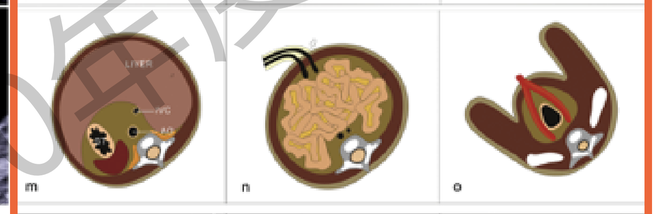


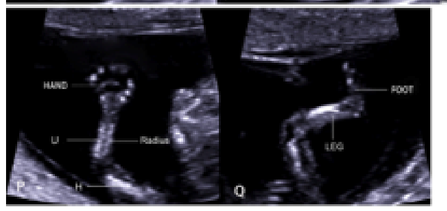


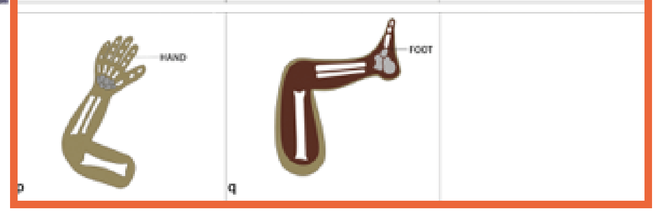












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2nd-TM: 30.9%
3rd-TM: 7.1%
Postnatal: 18.9%

High DR for:

anencephaly
exencephaly
cephalocele
holoprosencephaly
exomphalos
gastroschisis
Pentalogy of Cantrell
sirenomelia
body stalk anomaly

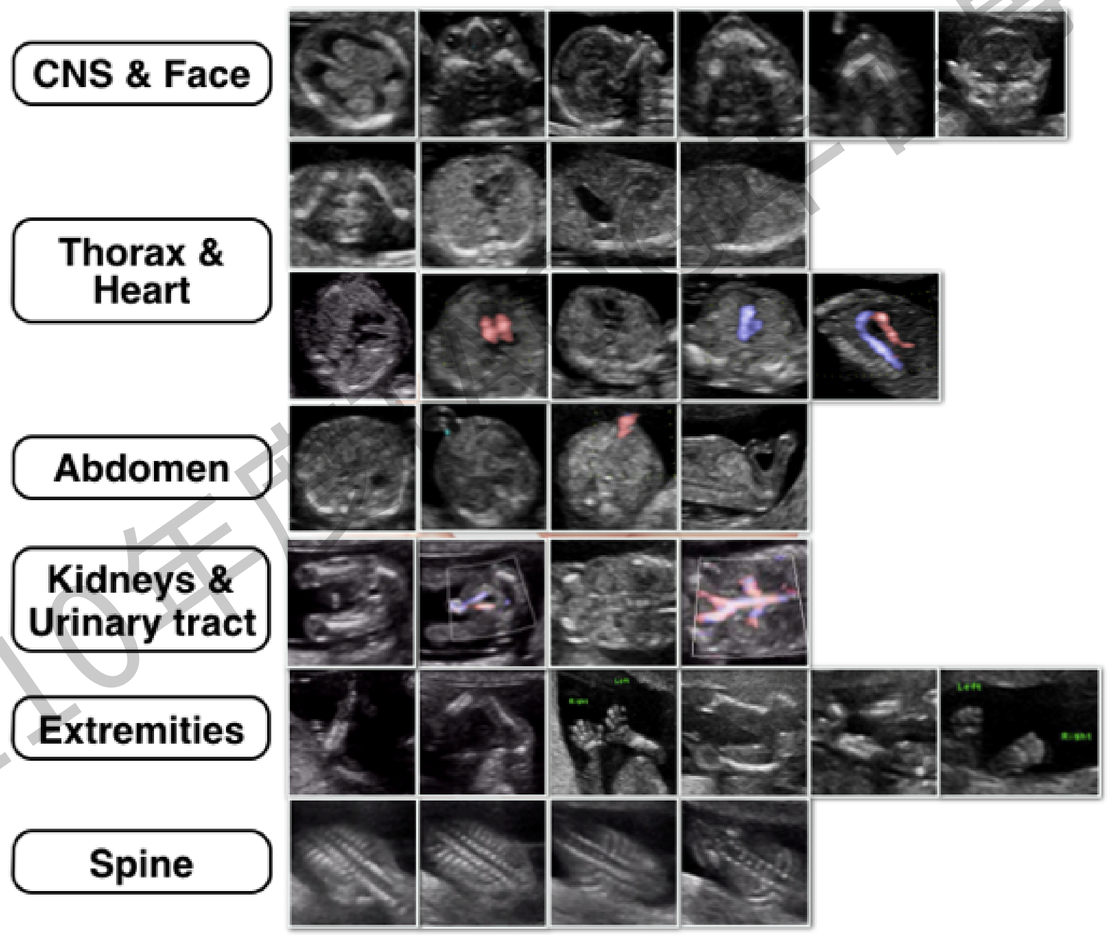
中華民國醫用超音波學會 2015 年學術研討會

Commonly Detected Abnormalities During First Trimester

台兒診所、台兒中山集英聯合診所
台兒臨床超音波暨胎兒醫學訓練所
胎兒醫學專業、產前超音波診斷專業、超音波唐氏症篩檢

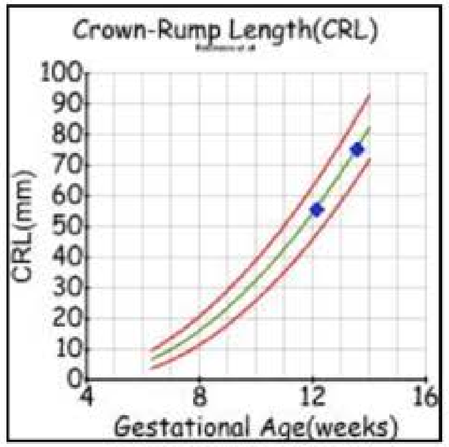
吳佩臻醫師

② A systematic approach to first-trimester fetal anatomic ultrasound should be used. [Best Prac Res Clin Obstet Gynecol 26 (2012) 561-573]

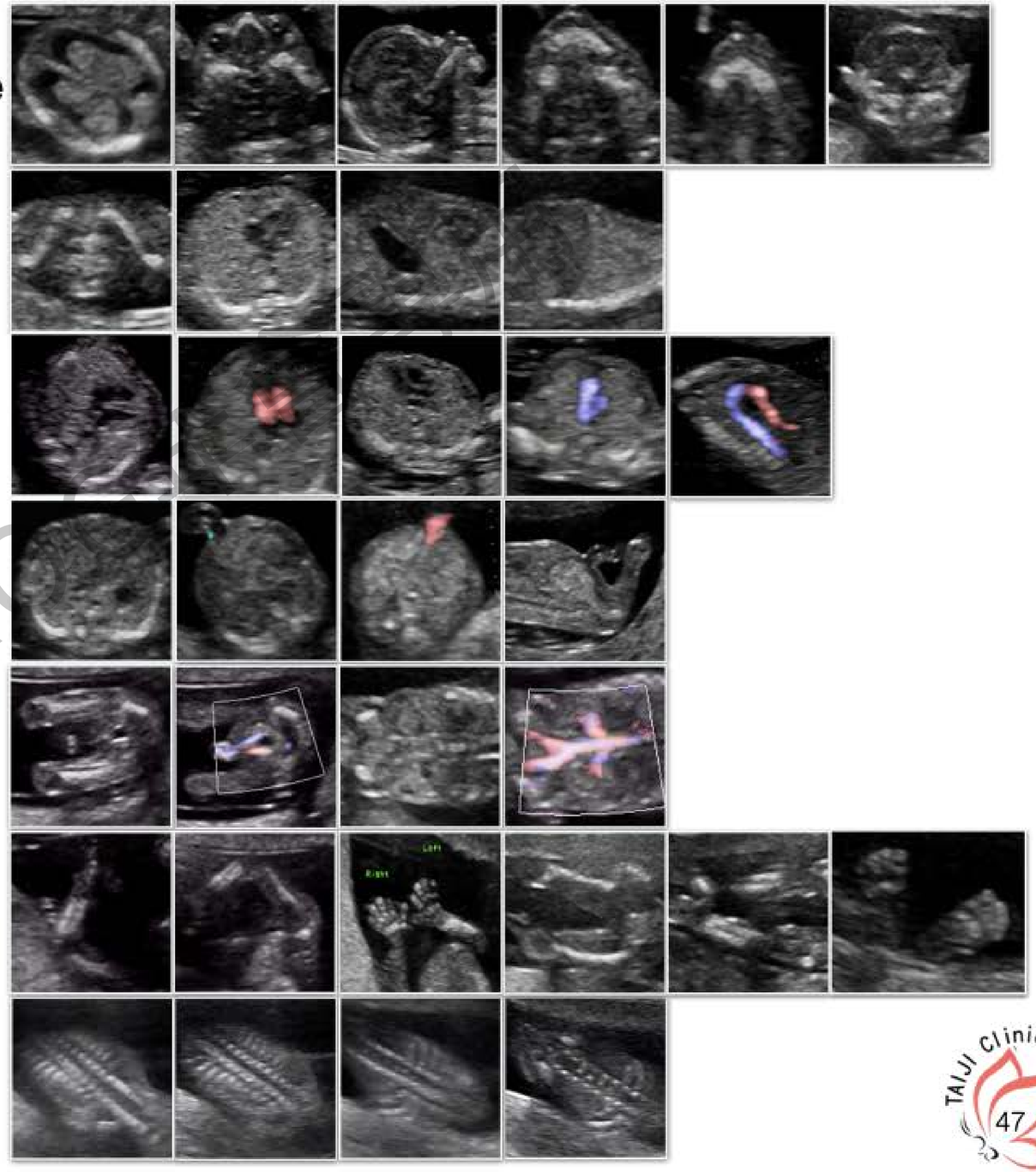


台兒早期胎兒結構檢查內容

- **Growth**



- **Anatomy**
  - **CNS, Face**
  - **Chest**
  - **Heart**
  - **Abdomen**
  - **GU**
  - **Limbs**
  - **Spine**

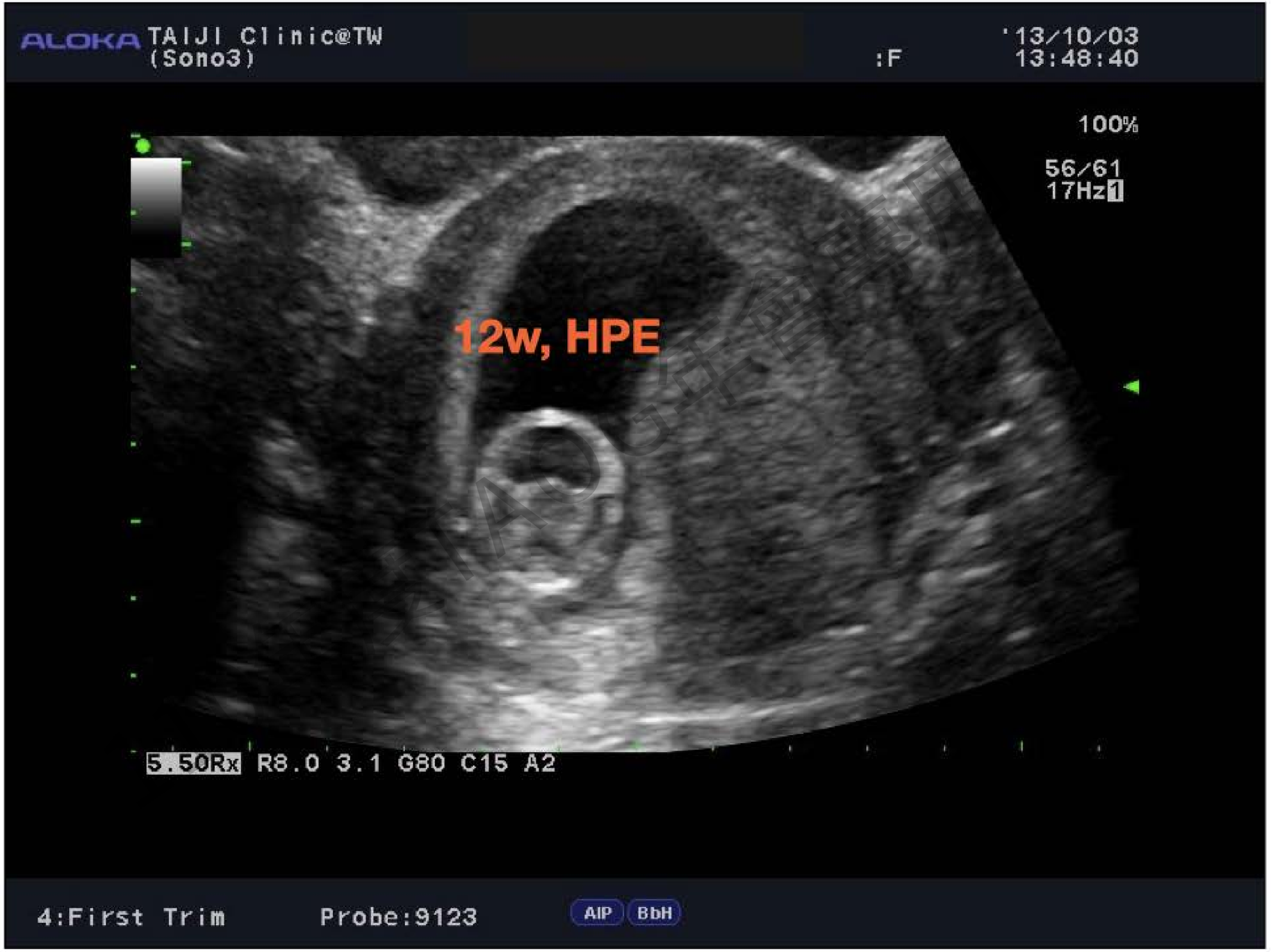


Systematic approach
1st-TM anatomical scan
since 2012

Taiji Case


ALOKA TAIJI Clinic@TW
(Sono3)
Y
:F
'13/03/06
16:46:32
100%
68/69
22Hz 1
12w, acrania
5.50Rx R7.0 G73 C15 A2
Dist: cm
4:First Trim
Probe:9123
AIP
BbH

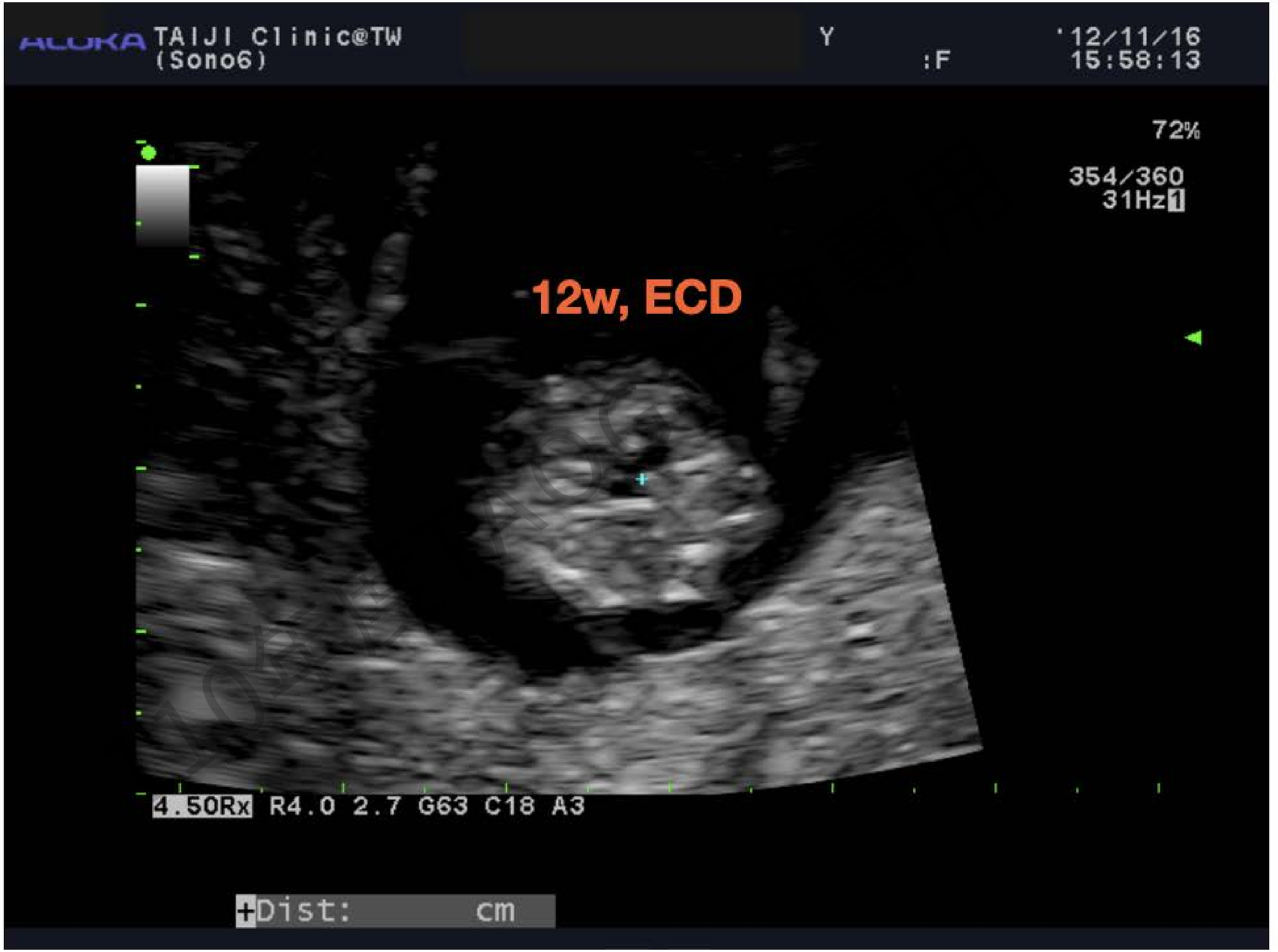
Taiji Case


ALOKA TAIJI Clinic@TW
(Sono3)
:F
'13/10/03
13:48:40
100%
56/61
17Hz1
12w, HPE
5.50Rx R8.0 3.1 G80 C15 A2
4:First Trim
Probe:9123
AIP
BbH

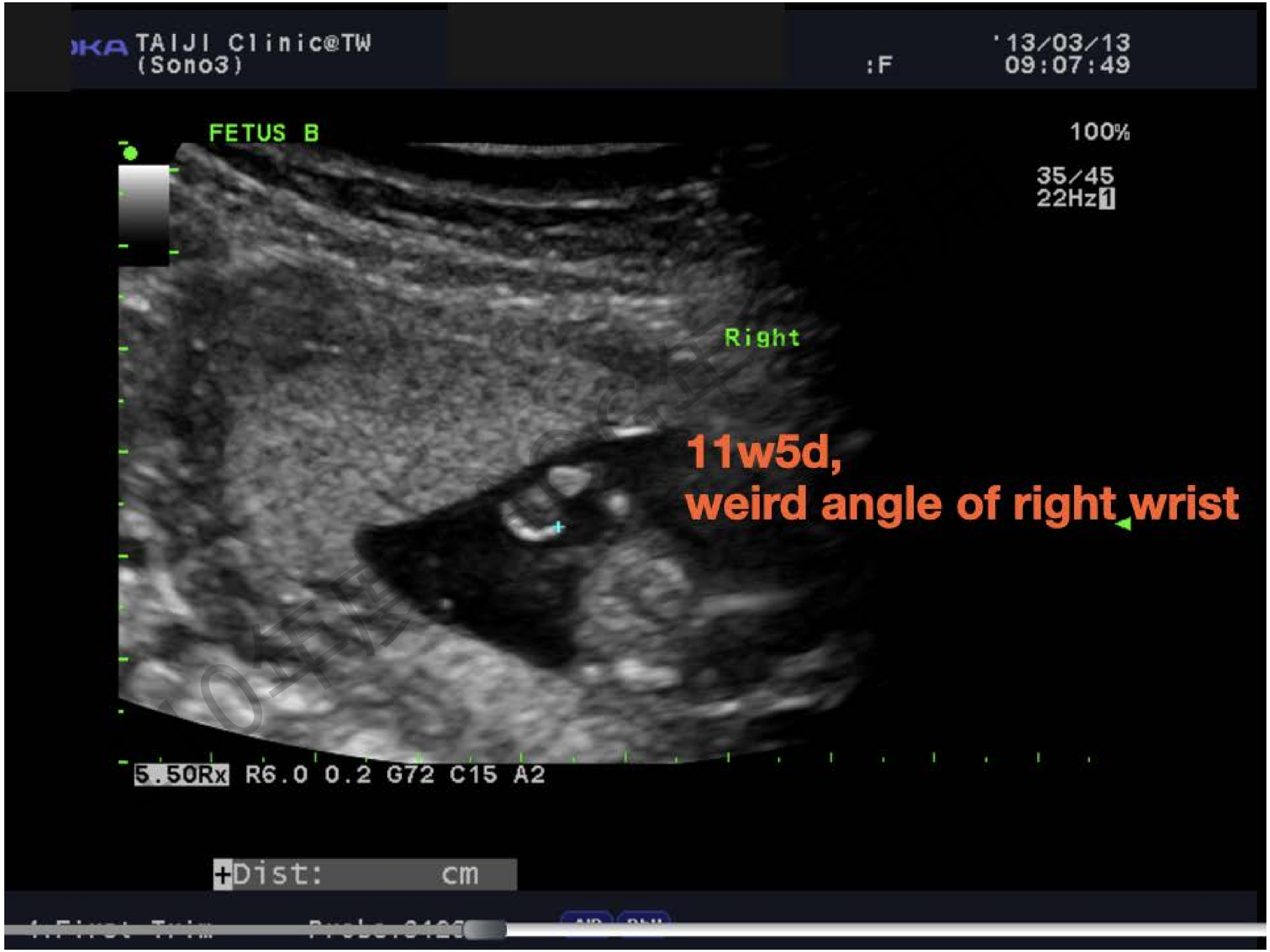
Taiji Case


ALOKA TAIJI Clinic@TW
(Sono5)
Y
:F
'13/04/01
13:55:40
TWIN B
72%
44/48
31Hz
14w5d, CDH
H
S
4.50Rx R5.0 3.1 G72 C18 A3
+Dist: cm
5:1trim Heart
Probe:9127
AIP
BbH

Taiji Case

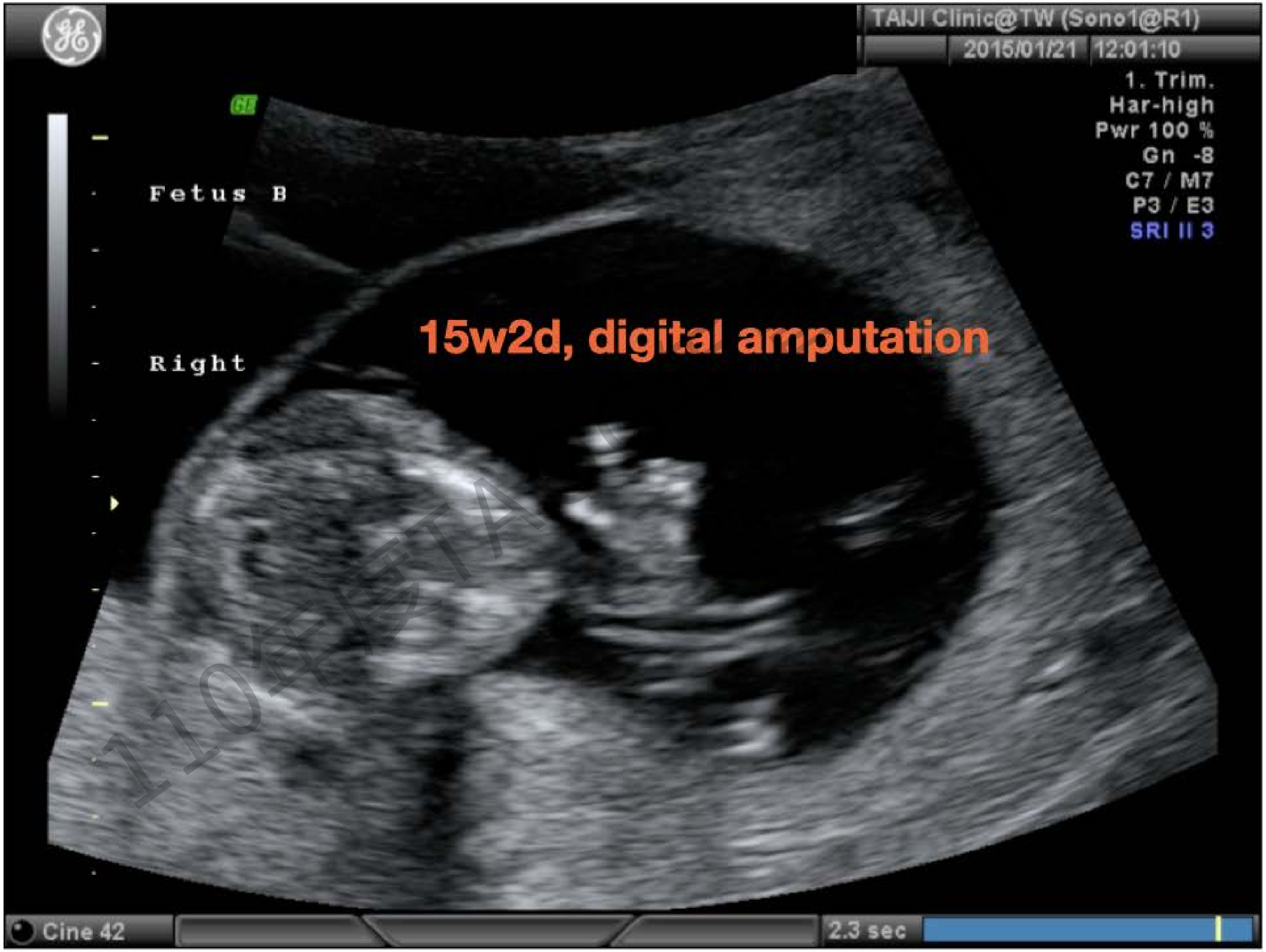

ALOKA TAIJI Clinic@TW
(Sono6)
Y
:F
'12/11/16
15:58:13
72%
354/360
31Hz1
12w, ECD
4.5ORx R4.0 2.7 G63 C18 A3
+Dist: cm

Taiji Case

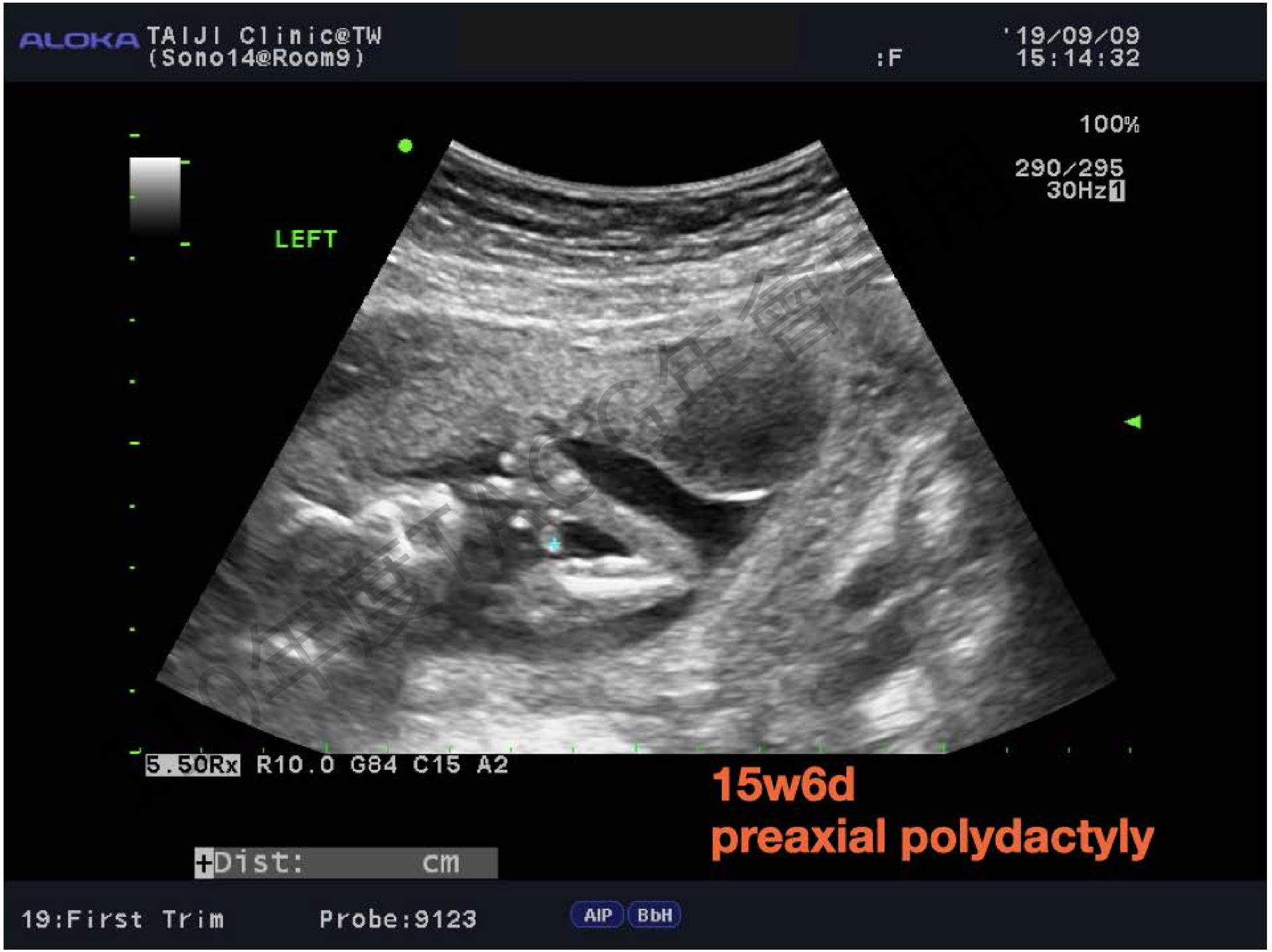

TAIJI Clinic@TW
(Sono3)
'13/03/13
09:07:49
:F
FETUS B
100%
35/45
22Hz
Right
11w5d,
weird angle of right wrist
5.50Rx R6.0 0.2 G72 C15 A2
+Dist: cm

Taiji Case

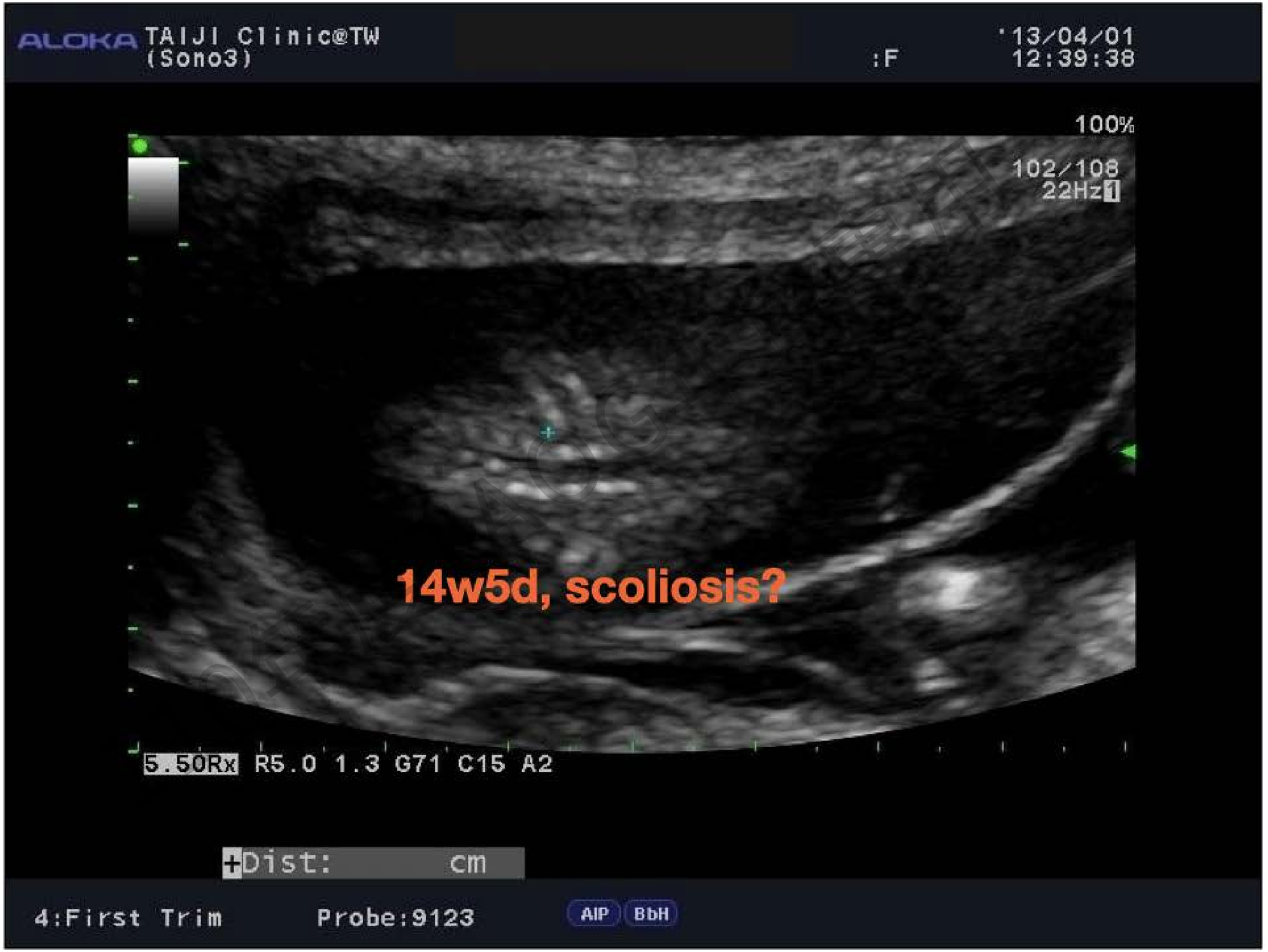
Taiji Case


TAIJI Clinic@TW (Sono1@R1)
2015/01/21 12:01:10
1. Trim.
Har-high
Pwr 100 %
Gn -8
C7 / M7
P3 / E3
SRI II 3
Fetus B
Right
15w2d, digital amputation
Cine 42
2.3 sec

Taiji Case



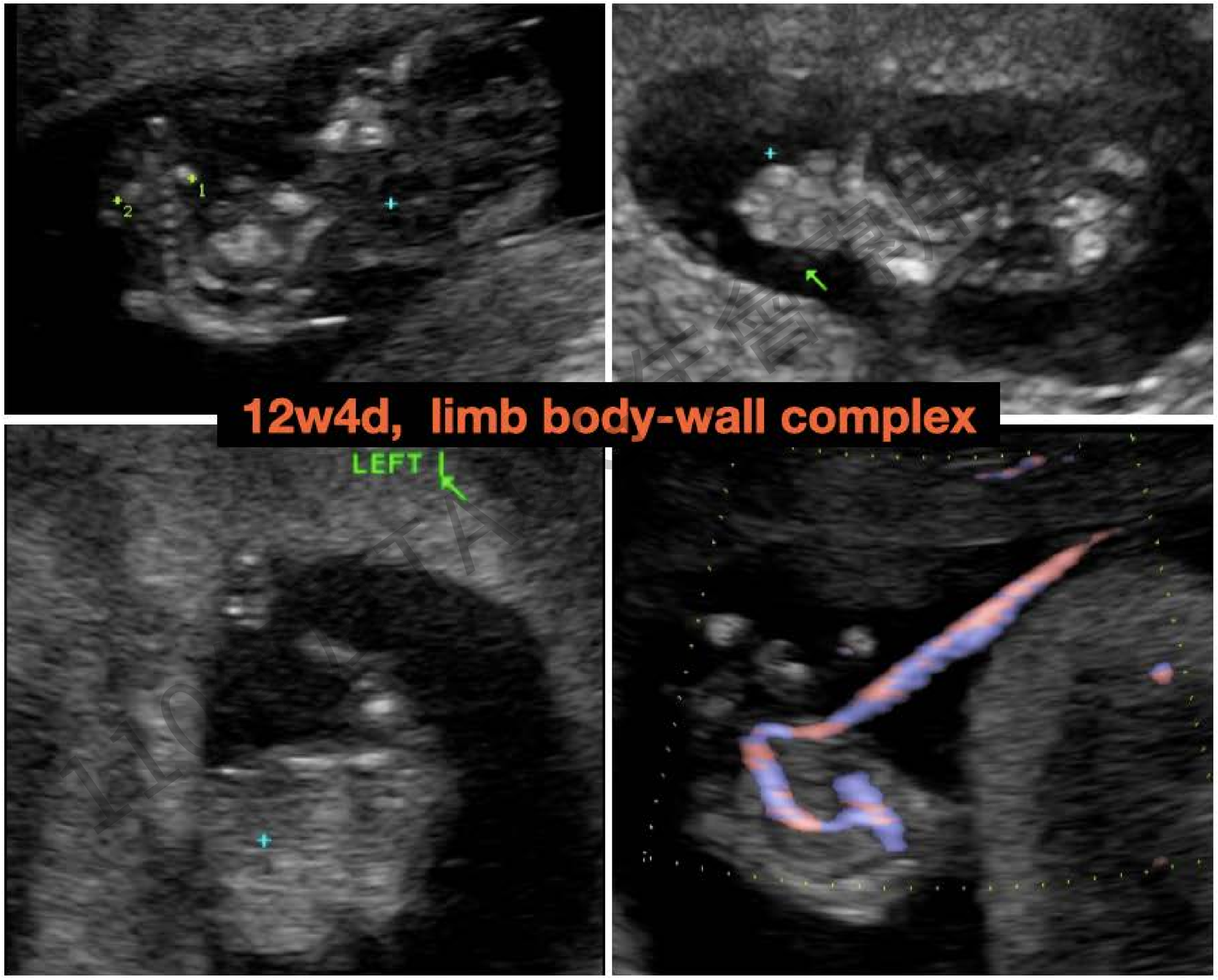
Taiji Case


ALOKA TAIJI Clinic@TW
(Sono3)
:F
'13/04/01
12:39:38
100%
102/108
22Hz
14w5d, scoliosis?
5.50Rx R5.0 1.3 G71 C15 A2
+Dist: cm
4:First Trim
Probe:9123
AIP
BbH

Taiji Case


TAIJI Clinic@TW
(Sono3)
:F
'13/04/03
10:10:12
FETUS B
100%
144/151
22Hz
15w, scoliosis
5.5ORx R6.0 G71 C15 A2

Taiji Case



Curr Opin Obstet Gynecol 2015, 27:143–150
Obstet Gynecol 2013;122:1160–7

Which Anomalies Should Be Targeted in Early Pregnancy?

Nearly Always Detectable (approx. 90-100%)

Severe CNS anomalies
- Acrania, anencephaly
- Alobar HPE
- Encephalocele

Ectopia cordis

Abdominal wall defects
- Omphalocele
- Gastroschisis
- LBWC/Body stalk anomaly
- Megacystis

Curr Opin Obstet Gynecol 2015, 27:143–150
Obstet Gynecol 2013;122:1160–7

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Potentially Detectable (approx. 2-90%)

CDH

Major heart defects
- TGA
- DORV
- CoA
- HLHS
- Septal defects

Spina bifida

MCDK

Skeletal disorders
- Lethal skeletal dysplasia
- Limb reduction
- Polydactyly

Curr Opin Obstet Gynecol 2015, 27:143–150
Obstet Gynecol 2013;122:1160–7

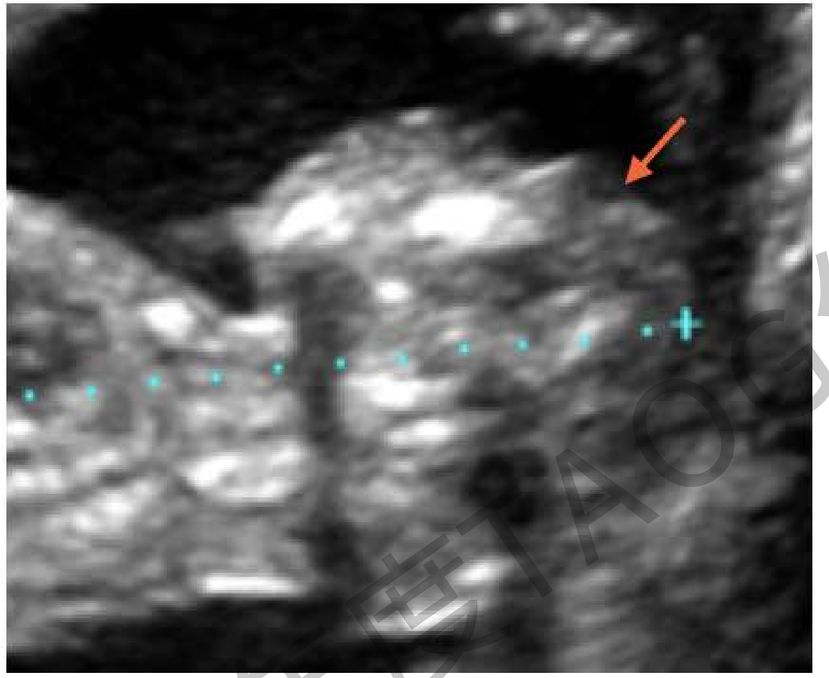
Which Anomalies Should Be Targeted in Early Pregnancy?

Nearly Always Detectable (approx. 90-100%)	Potentially Detectable (approx. 2-90%)	Virtually Undetectable (<2%)
Severe CNS anomalies Acrania, anencephaly Alobar HPE Encephalocele	**CDH**	**Cerebellar hypoplasia**
Ectopia cordis	**Major heart defects** TGA DORV CoA HLHS Septal defects	**ACC**
Abdominal wall defects Omphalocele Gastroschisis LBWC/Body stalk anomaly Megacystis	**Spina bifida**	**Echogenic lung lesions** CPAM Extralobar BPS
	MCDK	**GI disorders** Duodenal atresia Bowel obstruction Anal atresia
	Skeletal disorders Lethal skeletal dysplasia Limb reduction Polydactyly	**Mild renal anomalies** Duplex kidneys Hydronephrosis
		Ovarian cysts
		Fetal tumors

Taiji Case

NIPS: low risk

16w Taiji Clinic: **acrania, anencephaly**



17w Termination of pregnancy

Taiji Case

NIPS: low risk

14w6d Suspected brain, spine and skeletal anomalies

15w1d 2nd opinion: HPE, solitary kidney, abnormal FHB

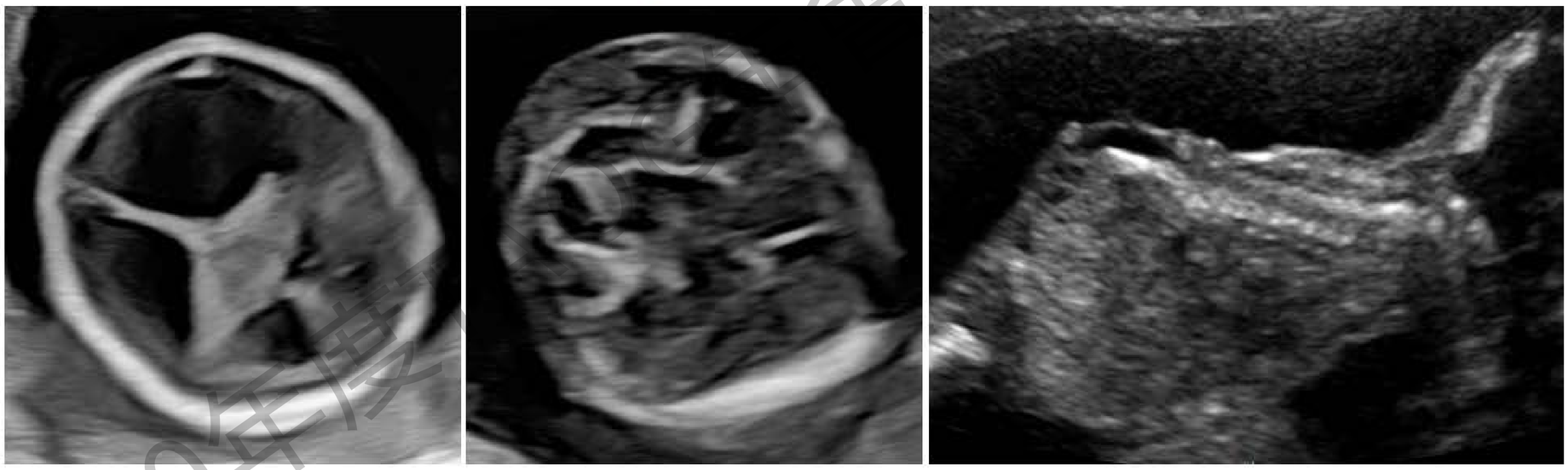
Taiji Case

NIPS: low risk

14w6d Suspected brain, spine and skeletal anomalies

15w1d 2nd opinion: HPE, solitary kidney, abnormal FHB

17w0d Taiji Clinic: **amniotic band sequence**



- severe bilateral ventriculomegaly
- compressed CM (banana sign)
- kyphoscoliosis
- deformed and limited motion of hands and feet

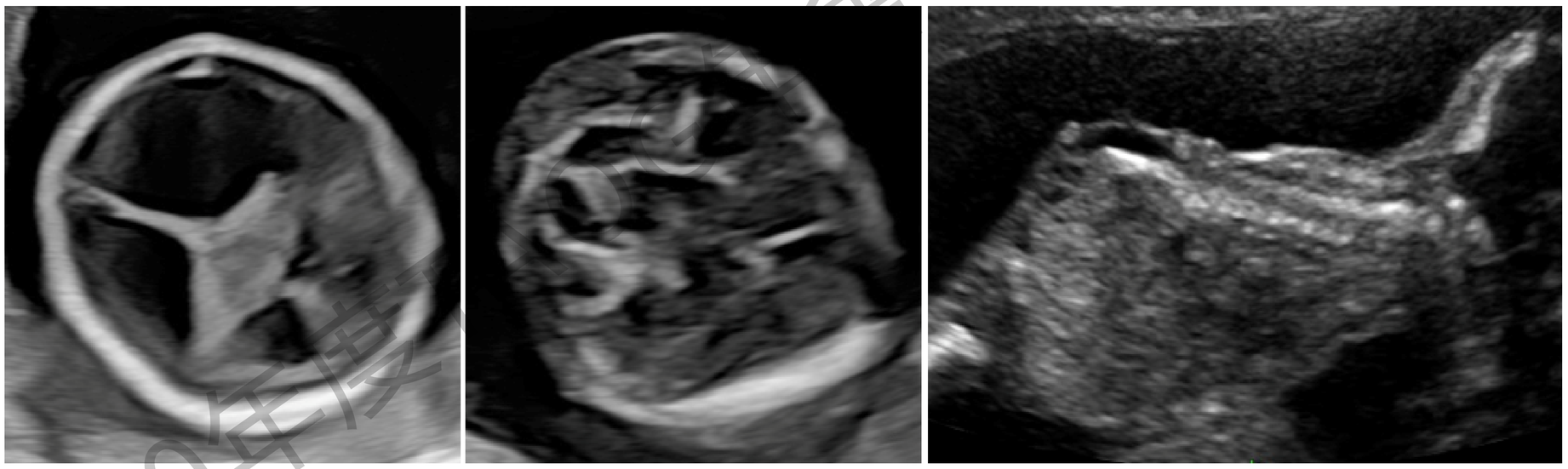
Taiji Case

NIPS: low risk

14w6d Suspected brain, spine and skeletal anomalies

15w1d 2nd opinion: HPE, solitary kidney, abnormal FHB

17w0d Taiji Clinic: **amniotic band sequence**



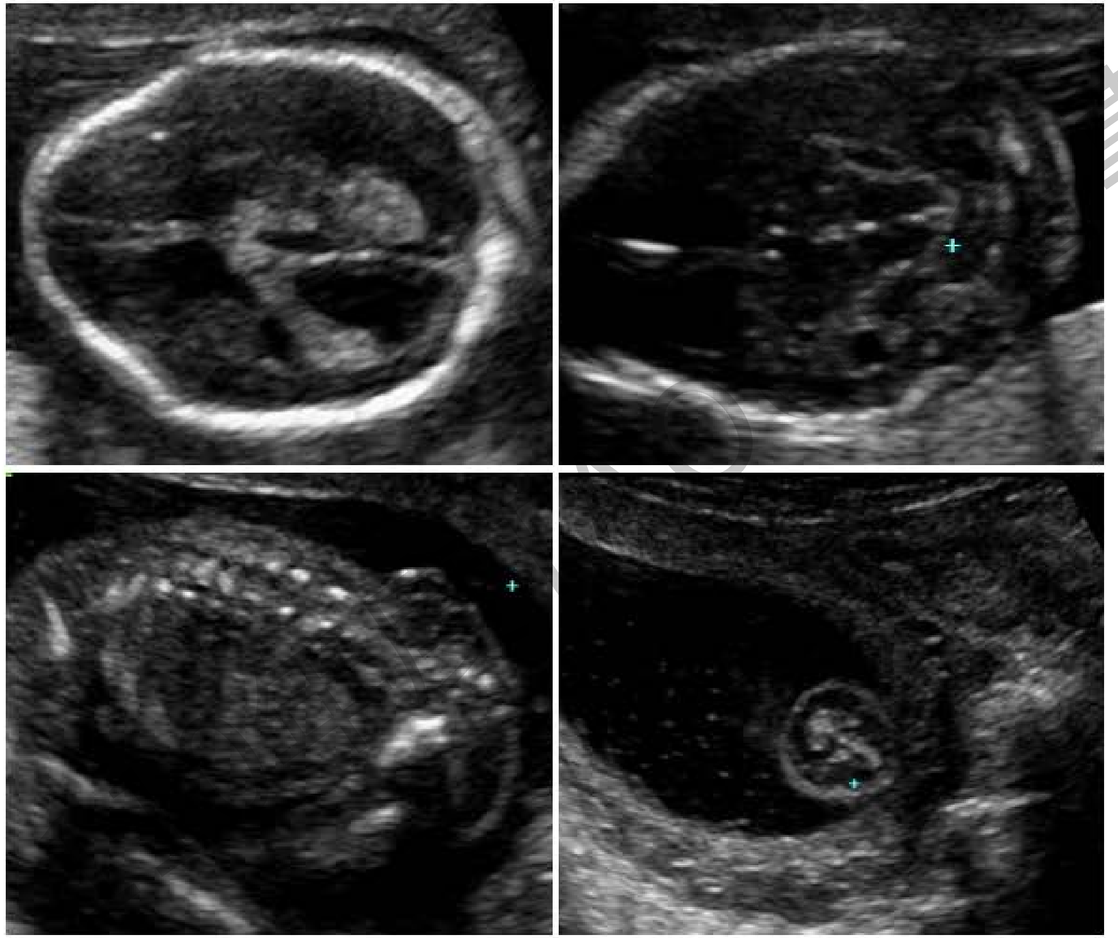
- severe bilateral ventriculomegaly
- compressed CM (banana sign)
- kyphoscoliosis
- deformed and limited motion of hands and feet

17w1d Termination of pregnancy

Taiji Case

IVF; **NIPS: pending report**

18w2d Taiji Clinic: **open spina bifida**



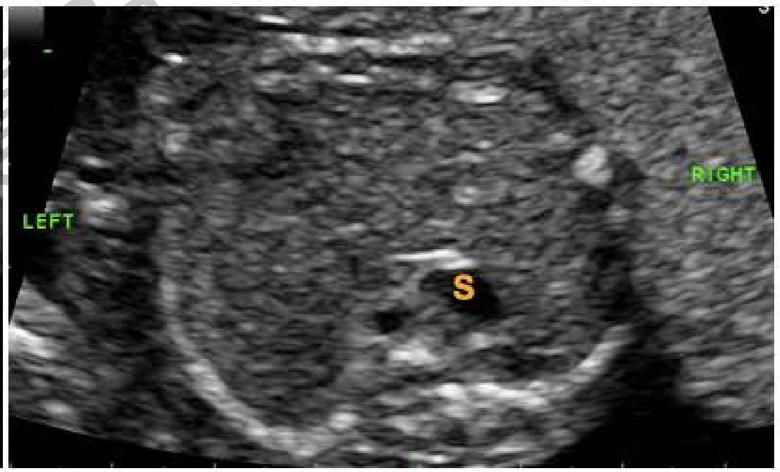
- bilateral ventriculomegaly
- **lemon sign**
- **banana sign**
- obliterated cisterns manga
- complex cystic lesion over sacrolumbar area

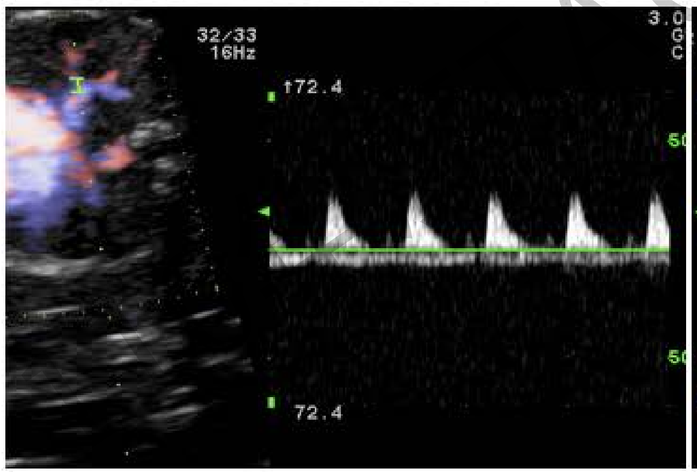
18w4d Termination of pregnancy

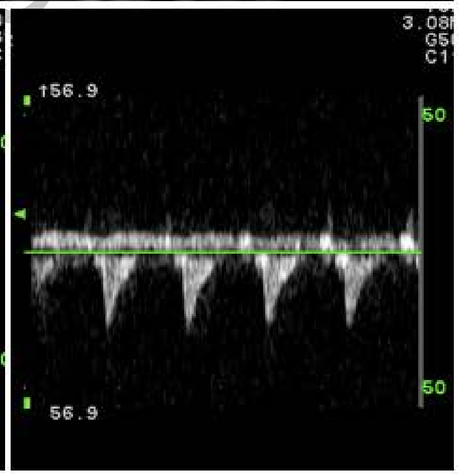
Taiji Case

NIPS: low risk

21w6d Taiji Clinic: **RAI, complex CHD**







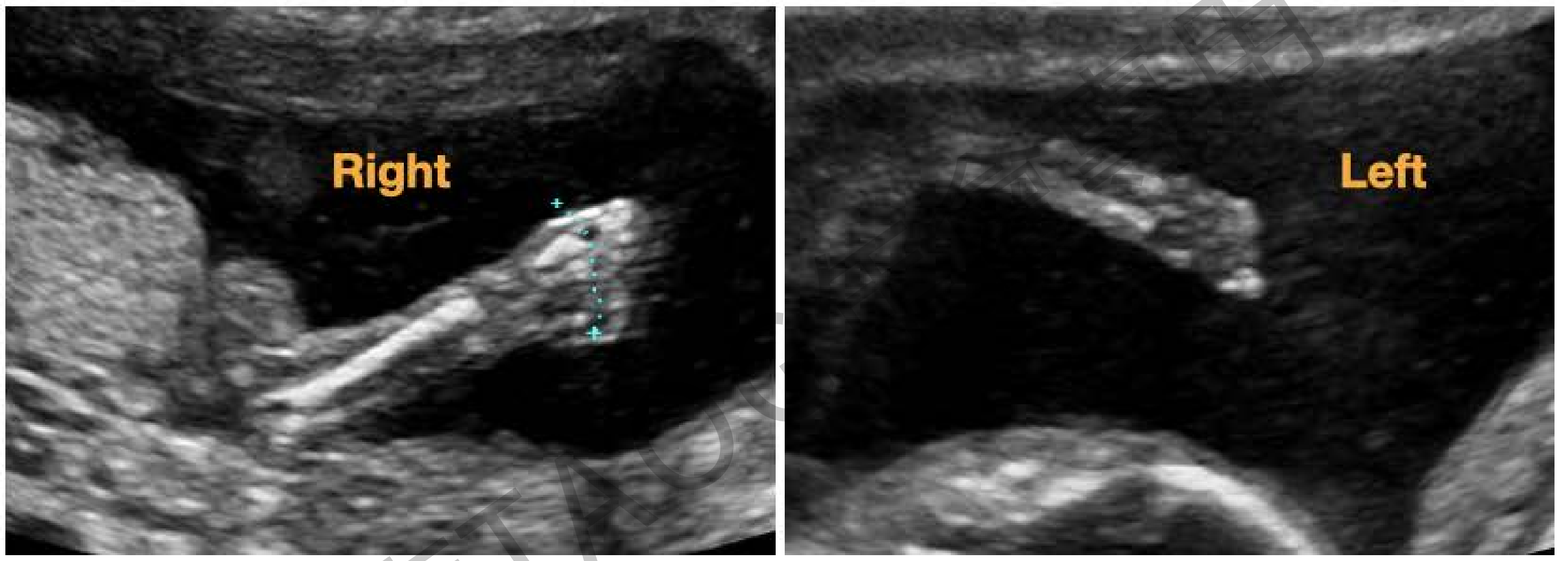
- **situs ambiguous::** heart on the left, stomach on the right
- unbalanced AVSD
- DORV with PA
- TAPVR

22w4d Termination of pregnancy

Taiji Case

NIPS: low risk

18w2d Taiji Clinic: **adactyly of left hand**



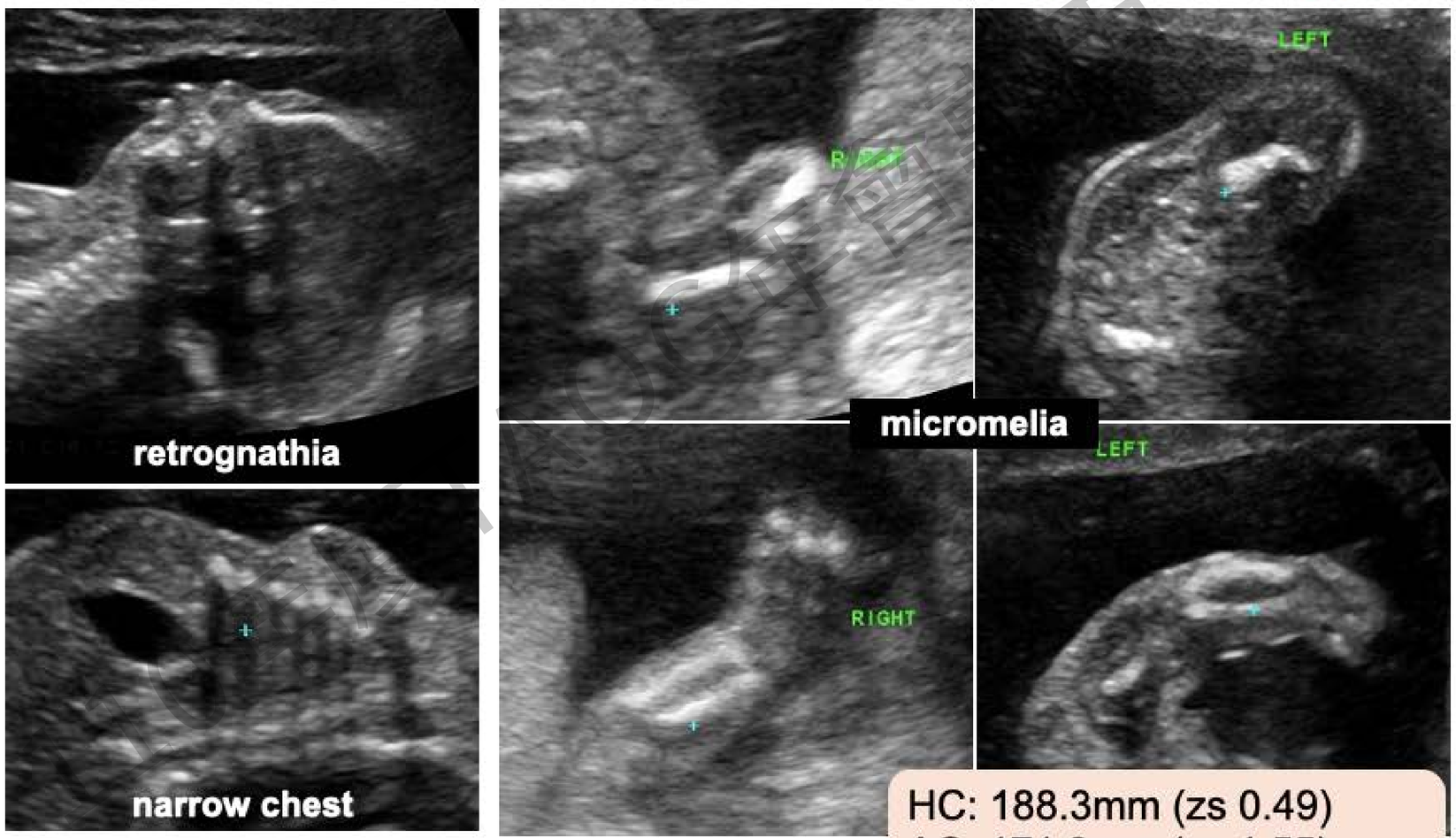
CMA: negative finding

19w2d Termination of pregnancy

Taiji Case

NIPS: low risk

21w0d **Short femur** noted, referred to Taiji Clinic: **suspected TD type 1**



21w1d Termination of pregnancy

1. Ultrasound scan is still an important component of 1st-TM screening.

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2. Patients who screen negative for aneuploidy by cfDNA remain at risk for fetal structural anomalies.

Prenatal Diagnosis 2016, 36, 260–265

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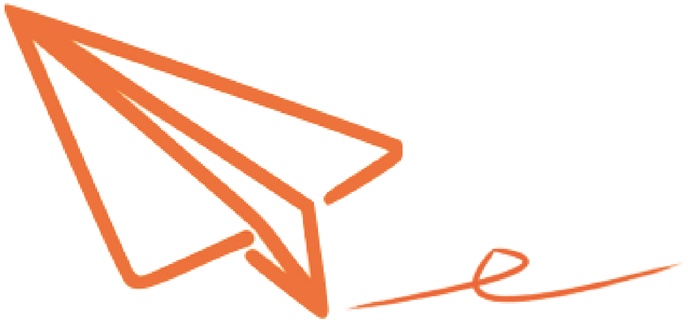
Prenatal Diagnosis 2016, 36, 260–265

3. Focusing only on screening for chromosomal aneuploidies in 1st TM and postponing the anatomic assessment to 2nd TM may result in delaying the detection of “major structural malformations”

J Perinat Med. 2019 Oct 25;47 (8): 871-878



The Era of cfDNA Testing & 1st-TM Ultrasound Exam…

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J Perinat Med. 2019 Oct 25;47 (8): 871-878

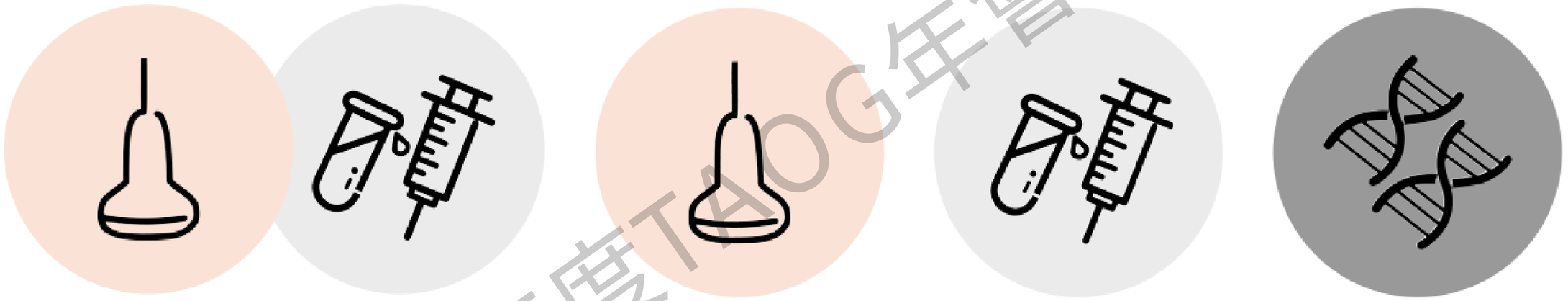
Indications for karyotyping & CMA

— High risk for atypical abnormal karyotypes:

① Maternal age >45y/o
② NT ≥3.5mm (3.0mm)
③ CFTS risk >1/100
④ Free β-hCG ≥5MoM or <0.2MoM
⑤ Serum PAPP-A <0.2MoM
⑥ Ultrasound-detected abnormality

Ultrasound Obstet Gynecol 2014; 43: 265–271

Proposal for a Revised Algorithm for Prenatal Diagnosis



CFTS

1st-TM US

NIPS

CVS
Amniocentesis

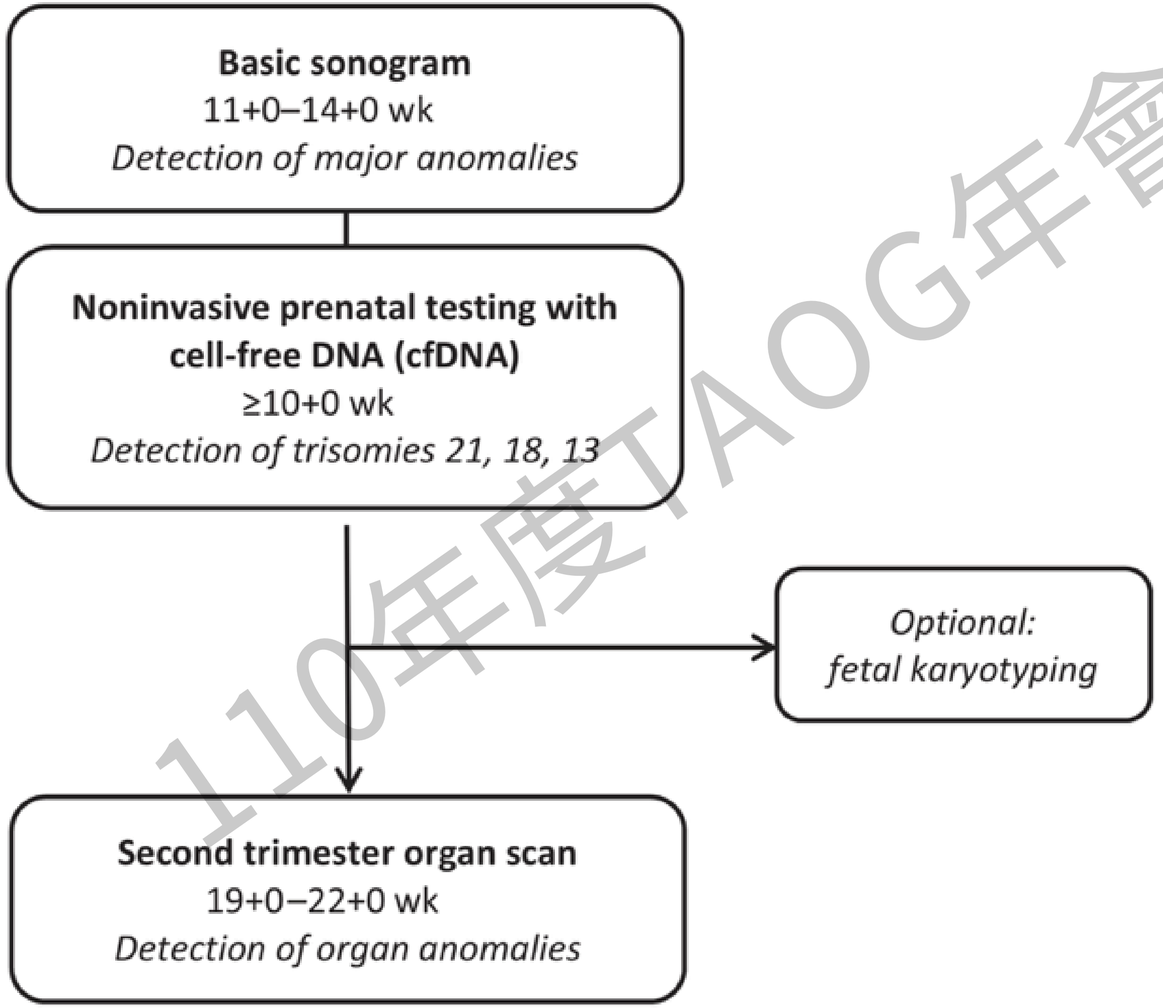
Proposal for a Revised Algorithm for Prenatal Diagnosis

1st-TM US

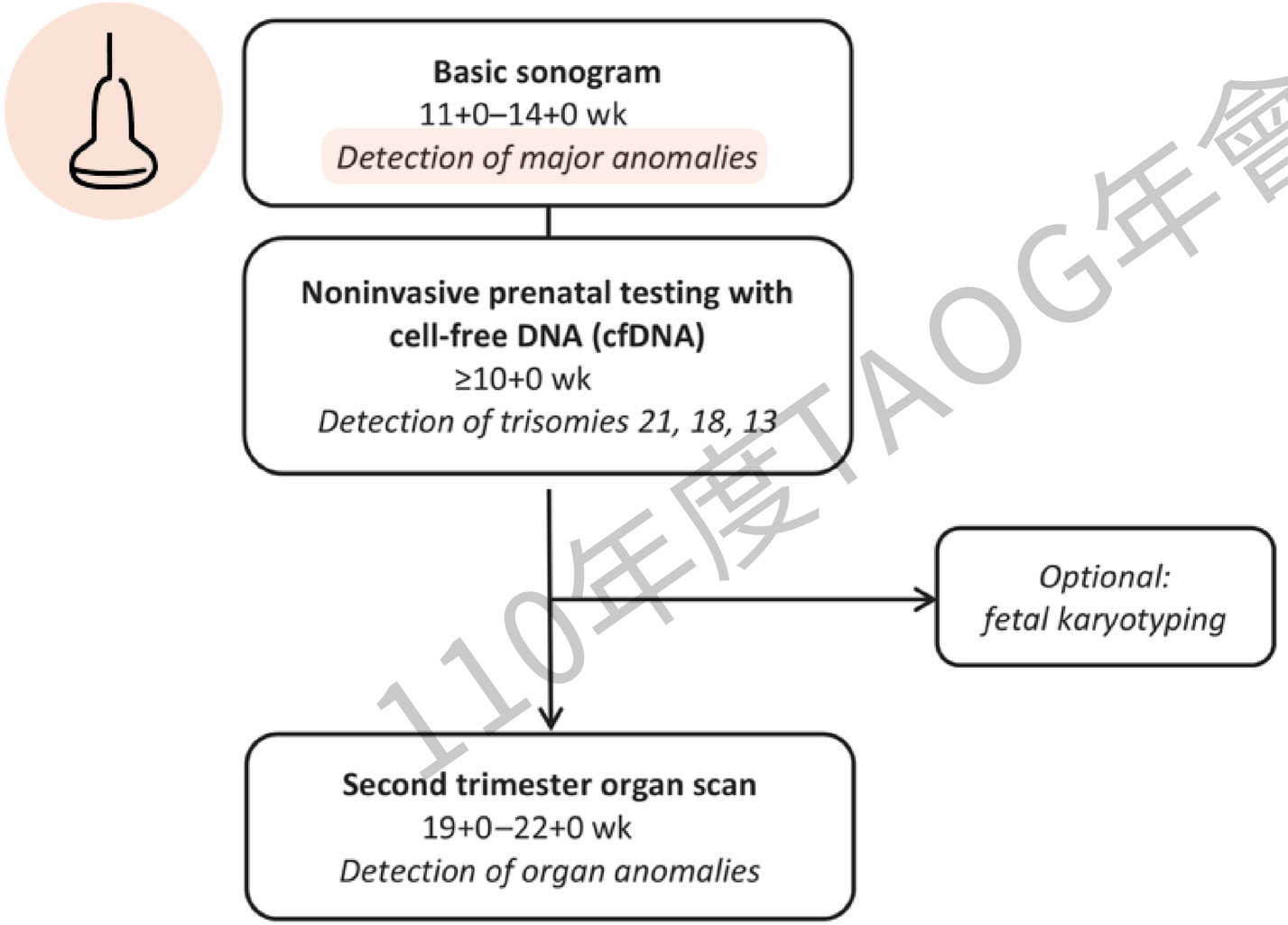
NIPS

CVS
Amniocentesis

Proposal for a Revised Algorithm for Prenatal Diagnosis



Proposal for a Revised Algorithm for Prenatal Diagnosis

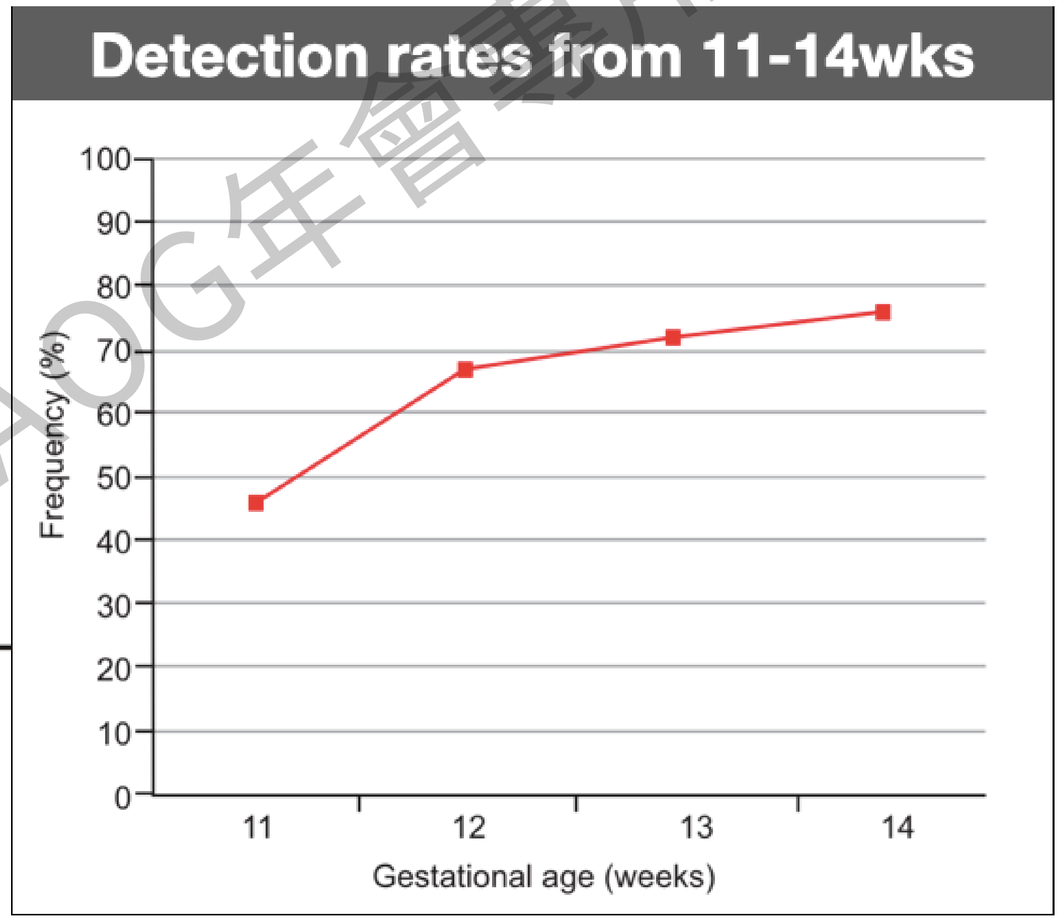


Proposal for a Revised Algorithm for Prenatal Diagnosis

Basic sonogram
11+0–14+0 wk
Detection of major anomalies

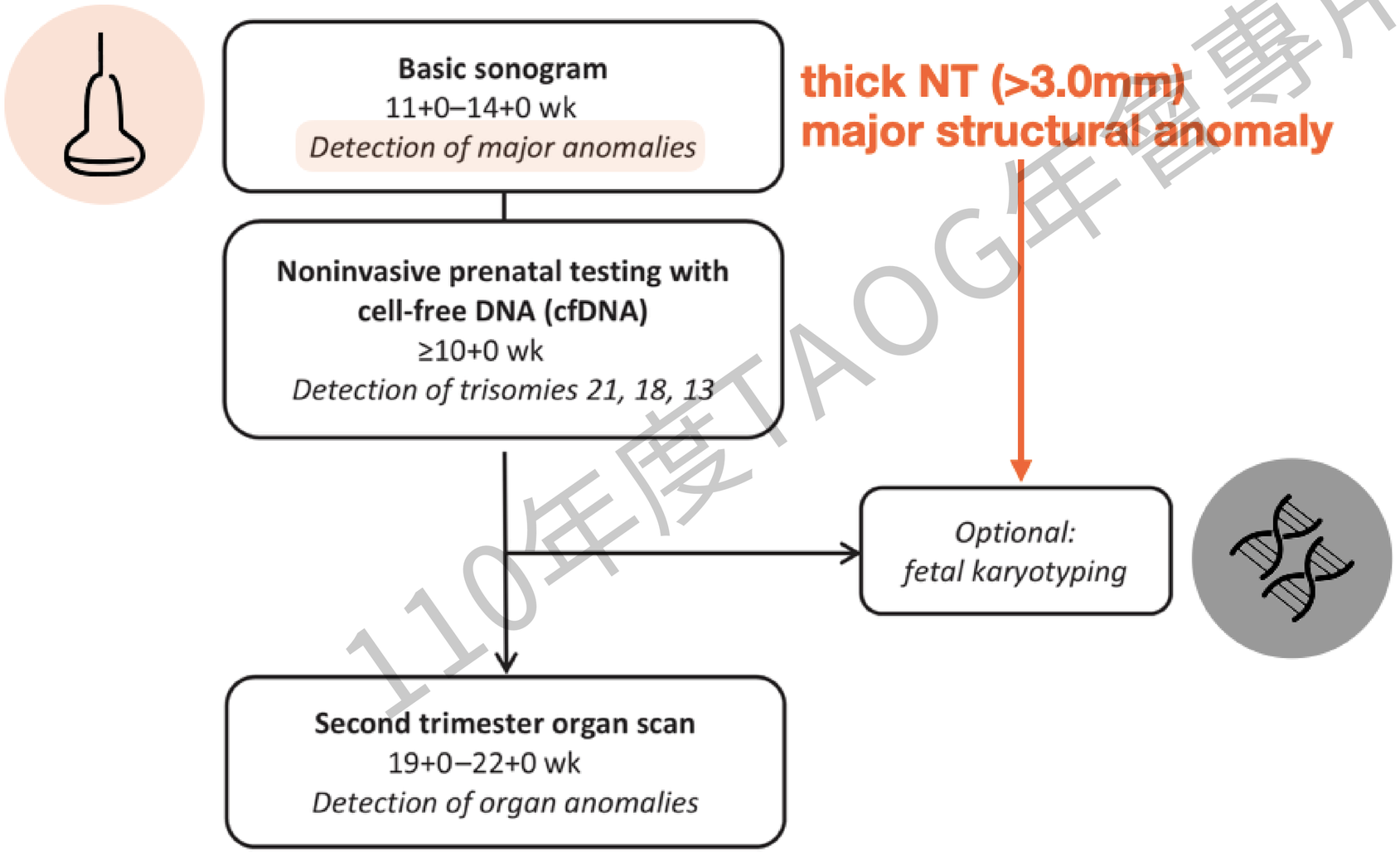
Noninvasive prenatal testing with cell-free DNA (cfDNA)
≥10+0 wk
Detection of trisomies 21, 18, 13

Second trimester organ scan
19+0–22+0 wk
Detection of organ anomalies

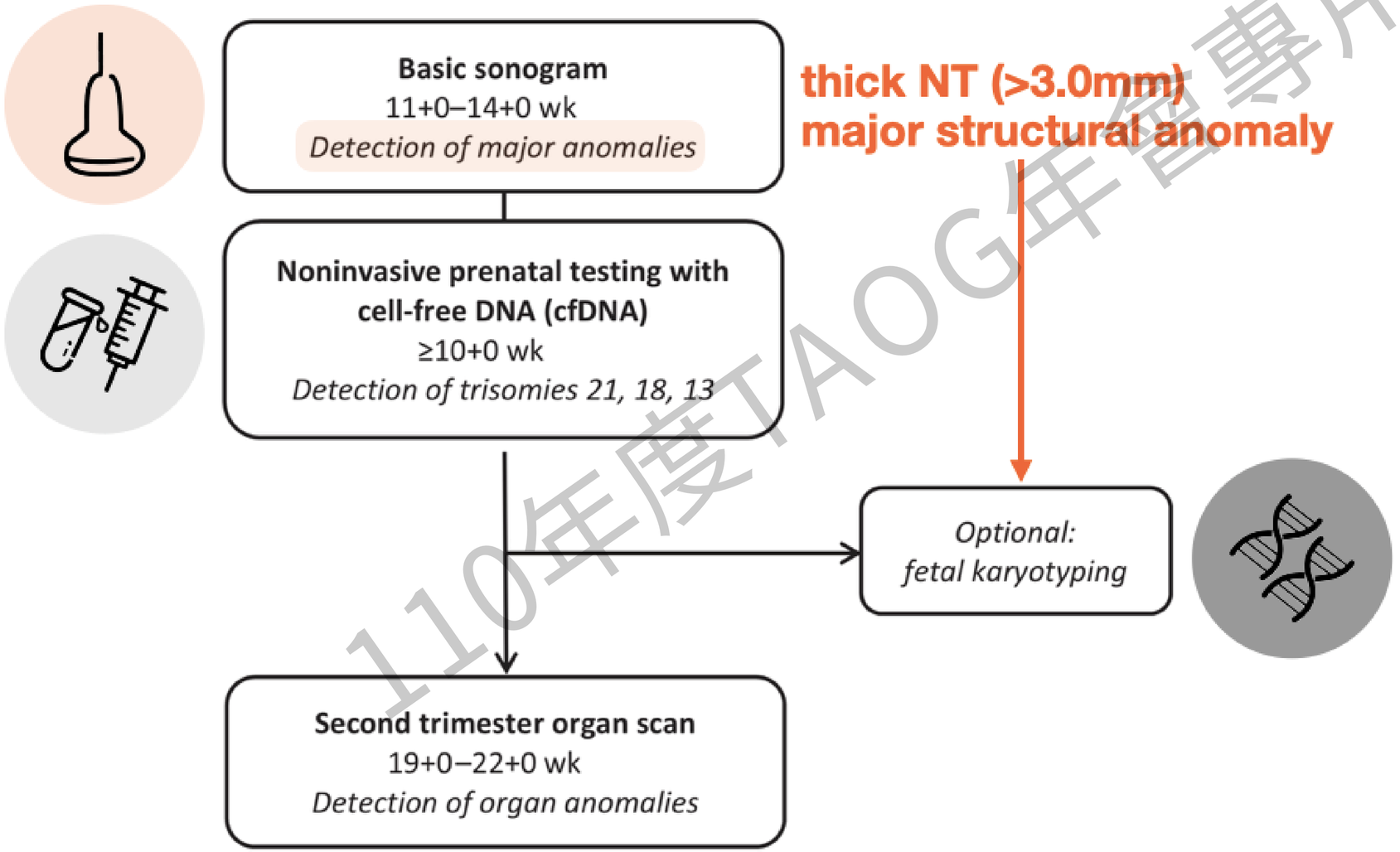


Obstet Gynecol 2013;122:1160–7

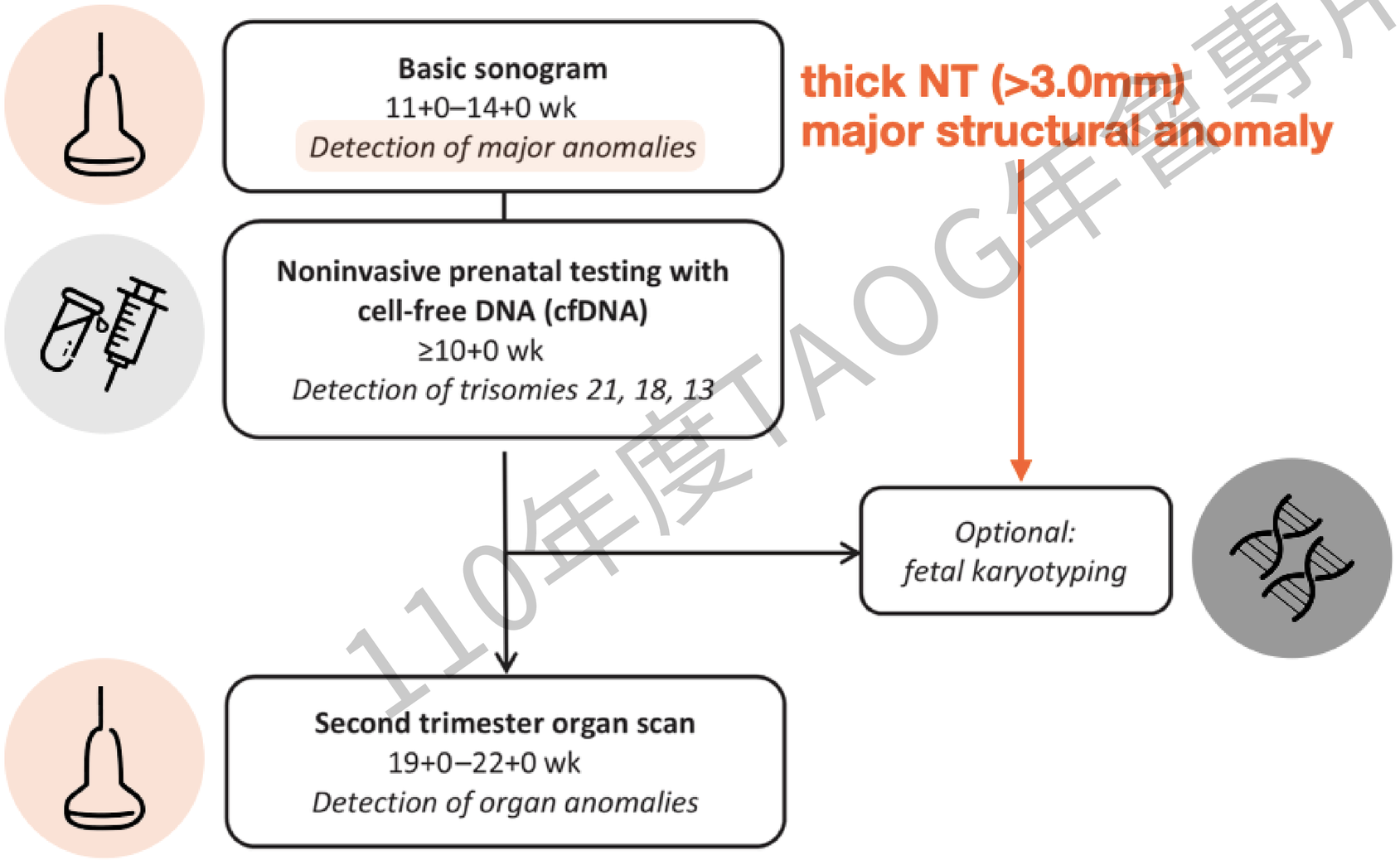
Proposal for a Revised Algorithm for Prenatal Diagnosis



Proposal for a Revised Algorithm for Prenatal Diagnosis



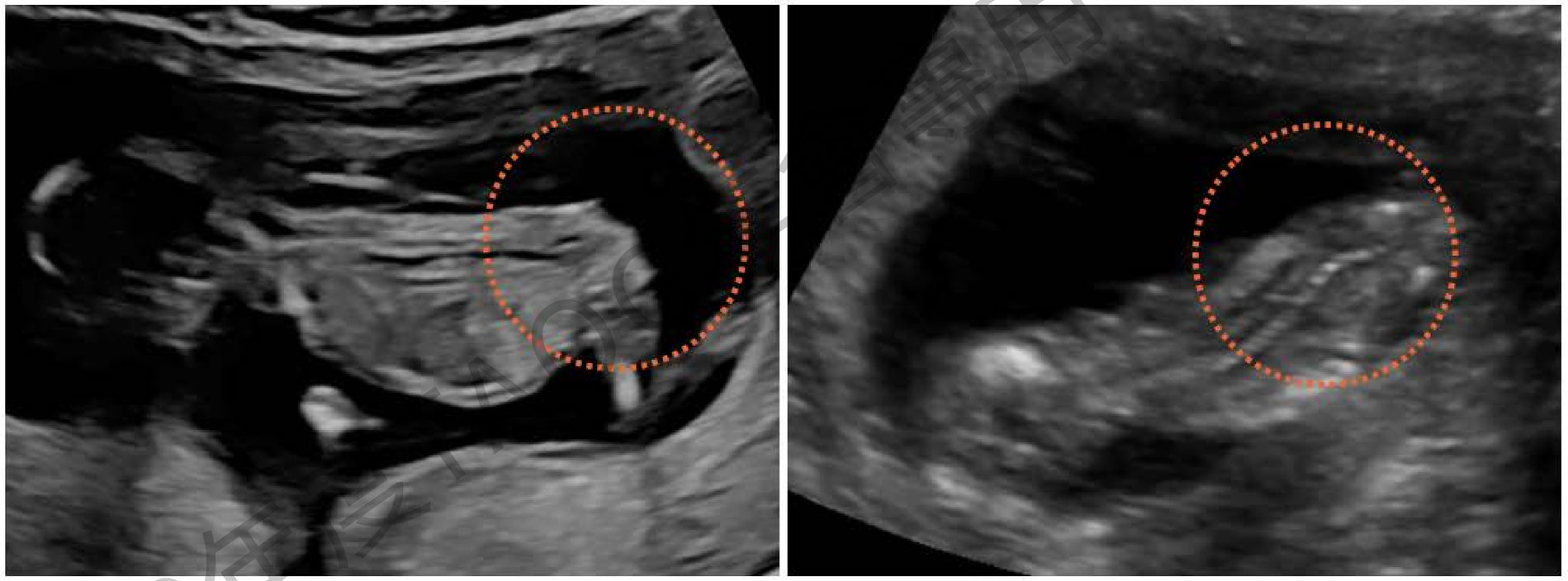
Proposal for a Revised Algorithm for Prenatal Diagnosis



Taiji Case

AMA, IVF, maternal type 2 DM (on insulin); **NIPS: low risk**

13w0d Taiji Clinic: **equivocal appearance of lumbosacral area**



Taiji Case

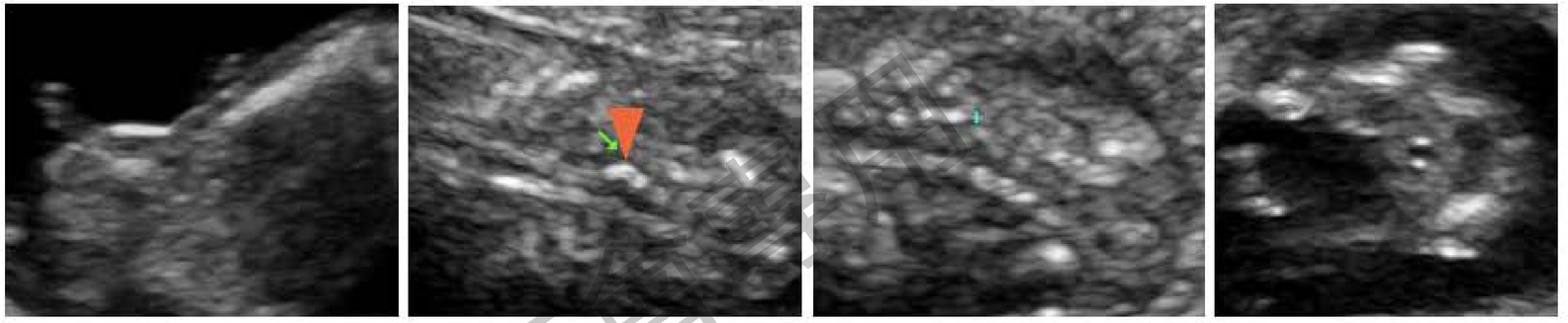
AMA, IVF, maternal type 2 DM (on insulin); **NIPS: low risk**

13w0d Taiji Clinic: **normal appearance of IT**

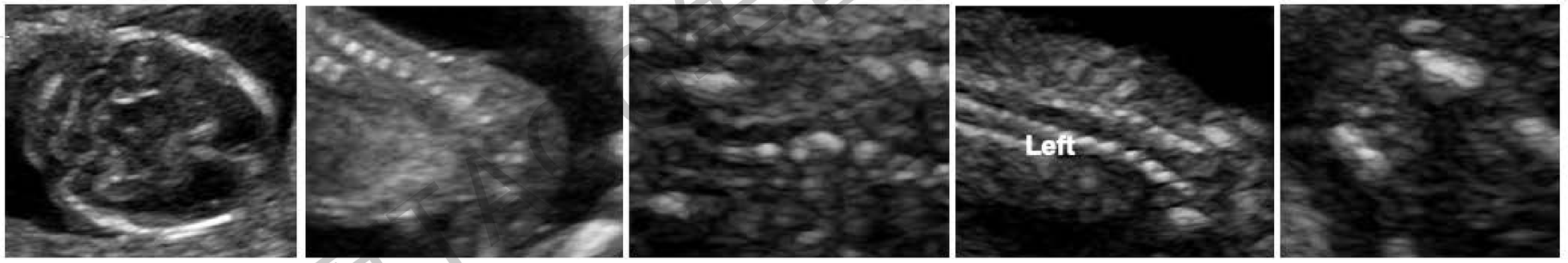
Taiji Case

F/U: normal movements of lower limbs

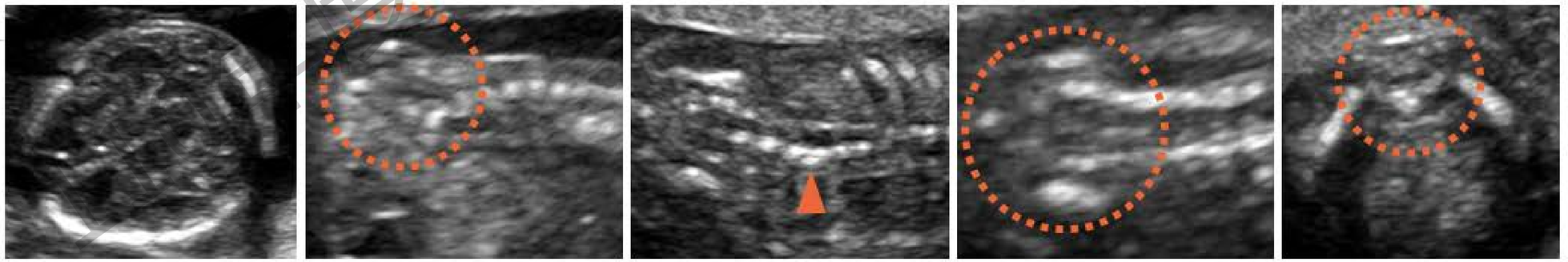
13w6d



15w3d

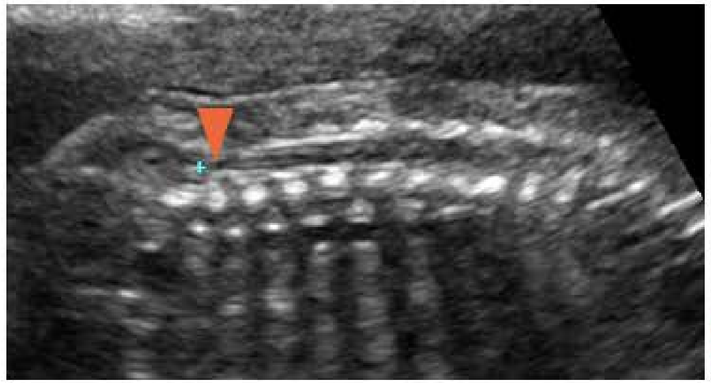


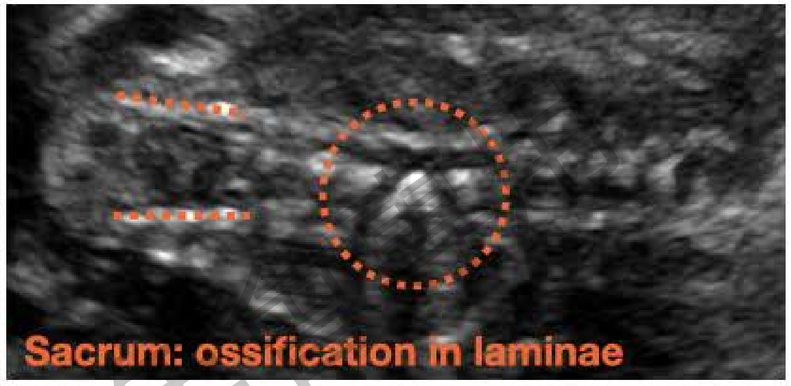
17w3d

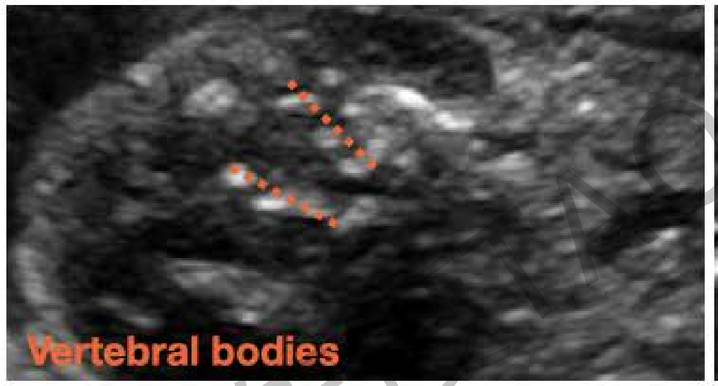


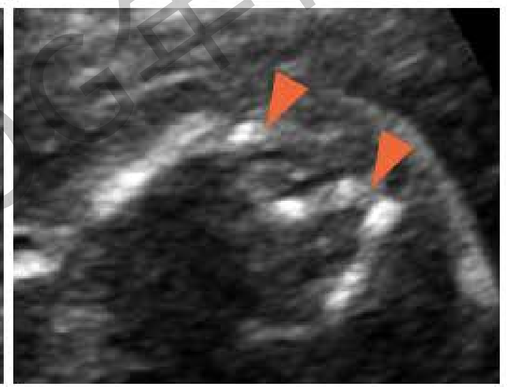
Taiji Case

22w3d Level II scan: spina bifida occultation, with lipoma; L2 hemivertebra

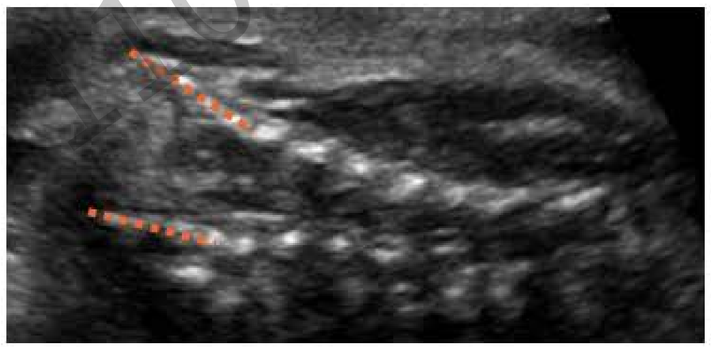








25w3d F/U

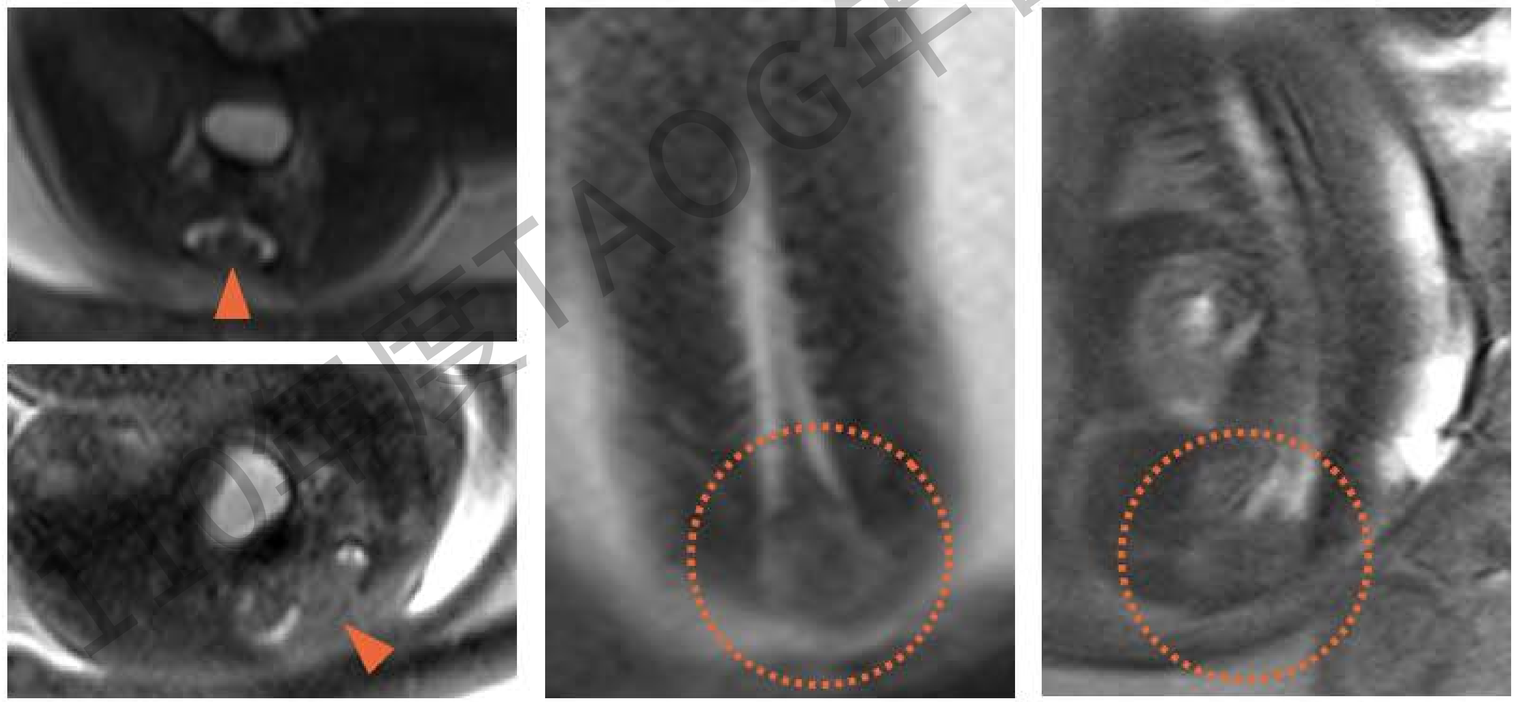


Taiji Case

22w3d — **MRI** at Taipei VGH

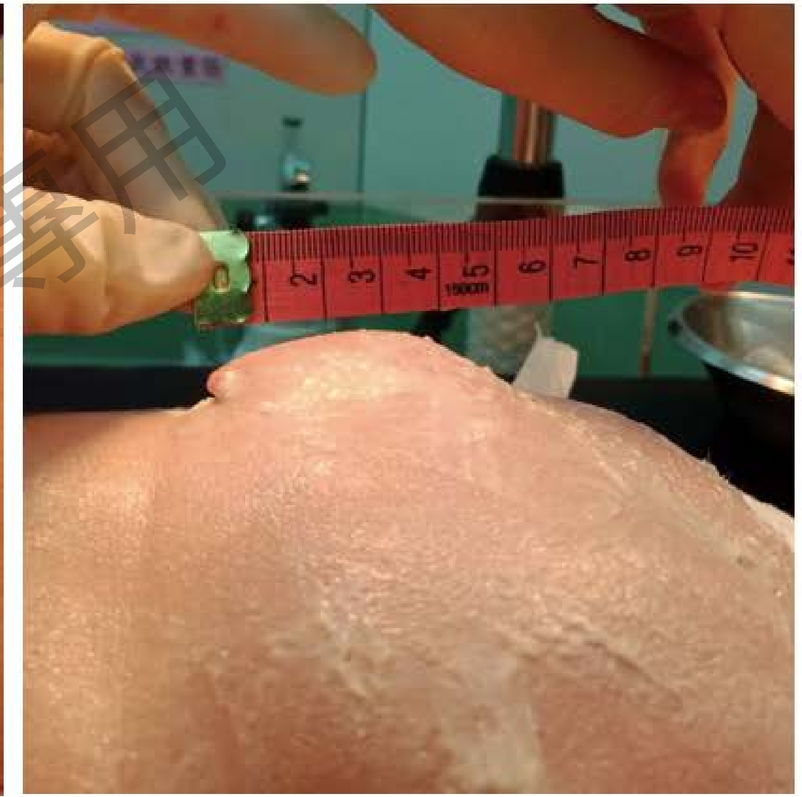
- **Closed-type NTD** (LMMC or lipomyelocele) with low-lying spinal cord below S1)
- Hemivertebra at L2

28w6d — **MRI f/u** at Taipei VGH; **multidisciplinary conference**



Taiji Case

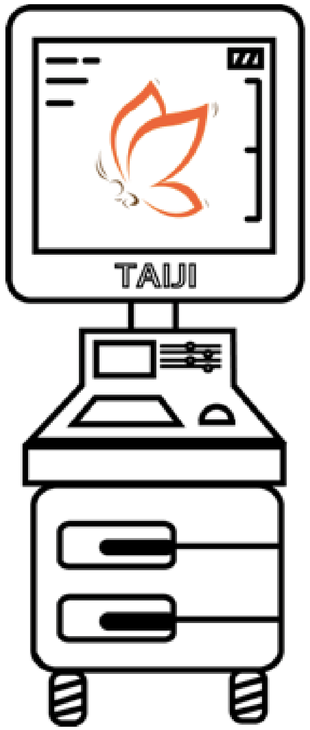
Term pregnancy, VD



台北榮總兒童脊髓整合門診（兒童神經外科、兒童外科、兒童腎臟科、復健）

Main goal of prenatal screening is to provide accurate information that will facilitate the delivery of optimized antenatal care, with the best possible outcome for both mother and fetus.

Ultrasound Obstet Gynecol 2017; 49: 815–816



Thank You for Listening