



# Webinars #13

## Non-invasive Fetal Autopsy

Tuesday 19th March from 5pm  
Raquel Gouveia Silva, Silvia Kalantari  
ESHG-Young committee.



# Welcome – Technical points

- **We are please to be numerous ~ 140 registration**
- **Webinar being recorded, 1h30**
  
- **Thank you for**
  - Turn off your microphone and disconnect your camera
  - Raise your hand at the time of the questions and discussions
  - We will answer the questions sent in the registration form
  - A satisfaction survey will be sent to you :
  
- **Webinars # will be available on ITHACA's Website + Pdf presentation**
- **<https://ern-ithaca.eu/documentation/educational-resources/>**
  
- Anne Hugon Project Manager - [anne.hugon@aphp.fr](mailto:anne.hugon@aphp.fr)

March 19, 2024

# Welcome and Introduction

- Public: Clinical Geneticists, Obstetricians, Pediatricians, Neonatologists (specialists and trainees).
- *This webinar will address the importance of autopsy and post-mortem evaluation in cases of miscarriage, stillbirth and neonatal loss. Traditional invasive autopsy is the most common strategy to provide detailed phenotyping and cause of death in these instances, however many parents find the procedure too invasive and some decline on that basis. The process of postmortem evaluation and alternatives to traditional autopsy will be discussed, including less invasive autopsy, postmortem MRI, micro-CT and the “genetic autopsy”. Furthermore, patients’ perspective regarding postmortem evaluation will be considered.*
- Chaired by **Raquel Gouveia Silva** and **Silvia Kalantari** on behalf of the **ESHG-Young committee**.

March 19, 2024

# Agenda

- **Welcome and Introduction – (5.00-5.05 pm)**
  - Raquel Gouveia Silva; Silvia Kalantari; ESHG-Young Committee
- **Rationale for less invasive autopsy and tissue-based diagnostics**
  - Dr Ciaran Hutchinson; UCL Great Ormond Street Institute of Child Health, London, UK
- **What imaging can offer for less invasive autopsy**
  - Dr Ian Simcock; UCL Great Ormond Street Institute of Child Health, London, UK
- **Parental and wider views on less invasive autopsy**
  - Dr Celine Lewis; UCL Great Ormond Street Institute of Child Health, London, UK
- **Discussion time**

March 19, 2024



# ESHG-Young



ESHG-Y Committee mission is to represent and support the young European geneticists by developing strategies and programs that aim for a better education.



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**Ileana-Delia Sabau, MD**  
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**Ana Raquel Silva, MD**  
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**Silvia Kalantari, MD**  
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**Ana Rita Matos,**  
Phd student  
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**Magdalena Mroczek, MD**  
Spokesperson



**Juliana Miranda Cerqueira, MSc,**  
Phd student  
Consultant

Scientific events  
promotion

Equal access to  
opportunities

ESHG-Y Network

Young Leadership

<https://www.eshg.org/eshgy>

March 19, 2024

# Our Speakers – Short Bio



**Dr. Ian Simcock** is a clinical-academic radiographer at Great Ormond Street Hospital for Children and Senior Research Fellow at the Institute of Child Health, London, UK. He is funded by the National Institute of Health and Care Research and is Chair of the Society of Radiographers Research Group.



**NHS**

**Great Ormond Street  
Hospital for Children**

NHS Foundation Trust

**Dr. Ciaran Hutchinson** is a Consultant Paediatric Pathologist at Great Ormond Street Hospital. He completed a PhD in Minimally Invasive Perinatal Autopsy in 2019.



**Dr. Celine Lewis** is a behavioural scientist working in the field of genetic and genomic medicine and rare disease. Her work focuses on how patients and families relate to, communicate and make decisions around personal genetic information and diagnostic test results, and the subsequent behavioural, psychological and social outcomes.



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# Topic 1

## Rationale for less invasive autopsy and tissue-based diagnostics

Dr Ciaran Hutchinson; UCL Great Ormond Street Institute of Child Health, London, UK

March 19, 2024



# Minimally invasive paediatric and perinatal autopsy

## Rationale for less invasive autopsy and tissue-based diagnostics

Dr. J. Ciaran Hutchinson MBBS PhD FRCPath  
Consultant Paediatric Pathologist  
Great Ormond Street Hospital, London, UK  
[Ciaran.Hutchinson@nhs.net](mailto:Ciaran.Hutchinson@nhs.net)





# Disclosure

- Previous academic collaboration with Nikon Metrology (2015 – 2020).
  - I received no financial inducements (cash / shares / IP) at any point.
- No other potential conflict of interest.

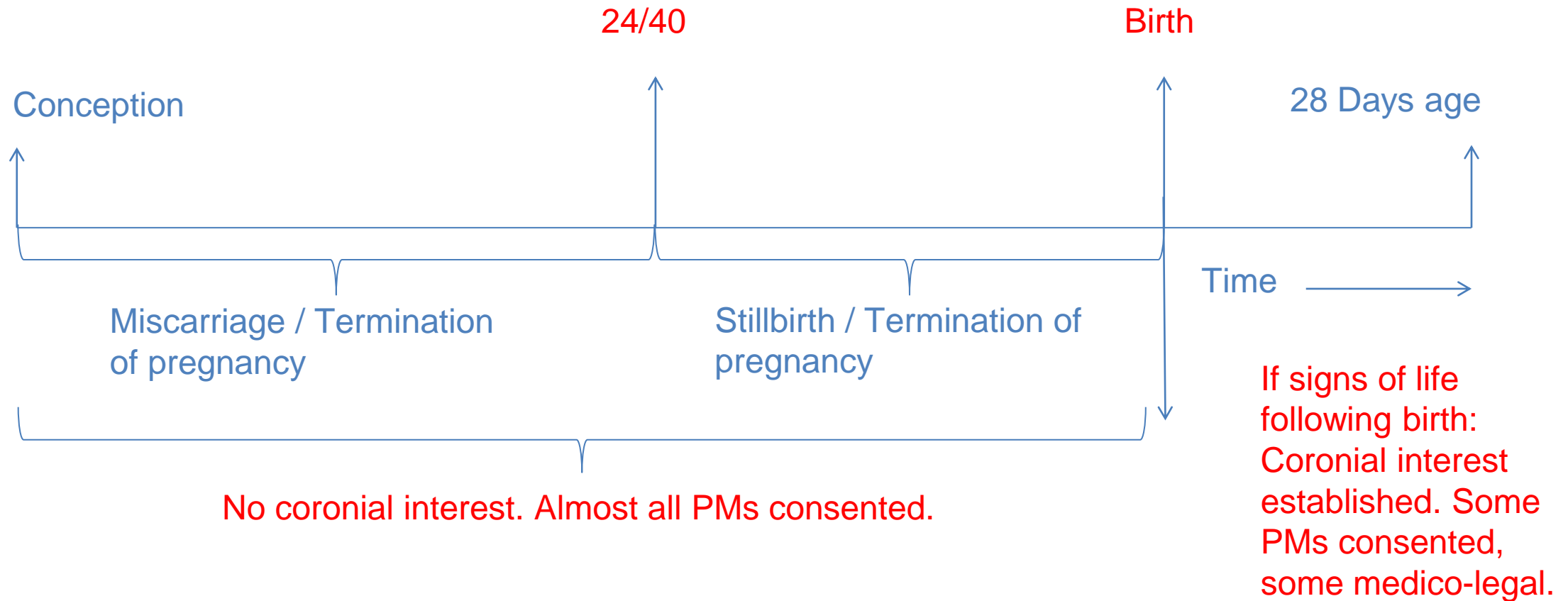
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# Specific objectives

1. To be able to explain the rationale for less invasive autopsy techniques.
2. To be able to approximate the yield (i.e. low yield or high yield) of routine histological examination of organ tissues in perinatal / pediatric autopsy by clinical context.

# Context

## Rationale for investigation after death in pediatric and perinatal cases - UK





# Potential benefits of autopsy

- Determine cause of loss.
- Inform future risk of recurrence.
- Provide genetic information to families.
- *Develop knowledge of rare disorders and syndromes.*
- *Provide epidemiological data.*
- *Audit antenatal findings.*

*“Clinically significant findings in 22– 70% of cases”*



BUT

# Acceptability is poor (AWPS 2013 data)

Post-mortem status	Intrauterine Death	Neonatal Death
Not offered	50 (1.6%)	137 (10.0%)
Offered but no consent	1503 (46.6%)	628 (45.7%)
Offered and full consent	1402 (43.5%)	372 (27.1%)

France: 39% perinatal autopsy acceptance (Sauvegrain P et al. *BMC Pregnancy Childbirth* 2019)



# Lots of barriers to PM uptake

Factors affecting uptake of postmortem examination in the prenatal, perinatal and paediatric setting

[C Lewis](#),<sup>1,2</sup> [M Hill](#),<sup>1,2</sup> [OJ Arthurs](#),<sup>2,3</sup> [C Hutchinson](#),<sup>2,4</sup> [LS Chitty](#),<sup>1,2</sup> and [NJ Sebire](#)<sup>2,4</sup>

*Parents wanted to 'protect' their baby or child from unnecessary harm.*

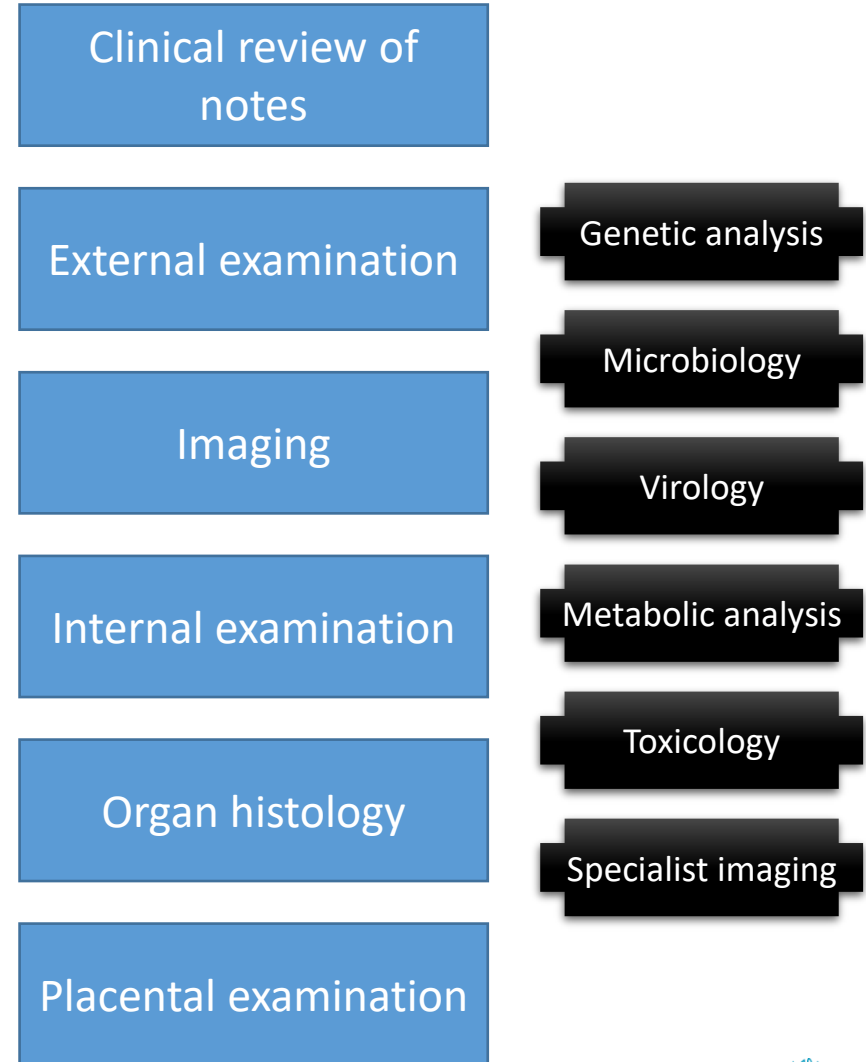
*Parents commented that their children had 'suffered enough', were 'fragile like dolls' and should be 'left in peace'.*

*“Clinically significant findings in 22–70% of cases”*

Autopsies are generally not really one examination.

Some bits are more important than others.

CSF



## Nothing

No examination

## External only

- Clinical review of notes
- External examination
- Placental examination

## Full autopsy

- Clinical review of notes
- External examination
- Imaging
- Internal examination
- Organ histology
- Placental examination
- Genetic analysis
- Microbiology
- Virology
- Metabolic analysis
- Toxicology
- Specialist imaging

>50 % Genotype without phenotype???





The NEW ENGLAND  
JOURNAL of MEDICINE

ORIGINAL ARTICLE

## Causal Genetic Variants in Stillbirth

Kate E. Stanley, B.A., Jessica Giordano, M.S., C.G.C., Vanessa Thorsten, M.P.H., Christie Buchovecky, Ph.D., Amanda Thomas, Ph.D., Mythily Ganapathi, Ph.D., Jun Liao, Ph.D., Avinash V. Dharmadhikari, Ph.D., Anya Revah-Politi, M.S., C.G.C., Michelle Ernst, M.S., C.G.C., Natalie Lippa, M.S., C.G.C., Halie Holmes, M.S., C.G.C., *et al.*

Additional 6 - 7% diagnostic yield from whole exome sequencing in stillbirth cases.

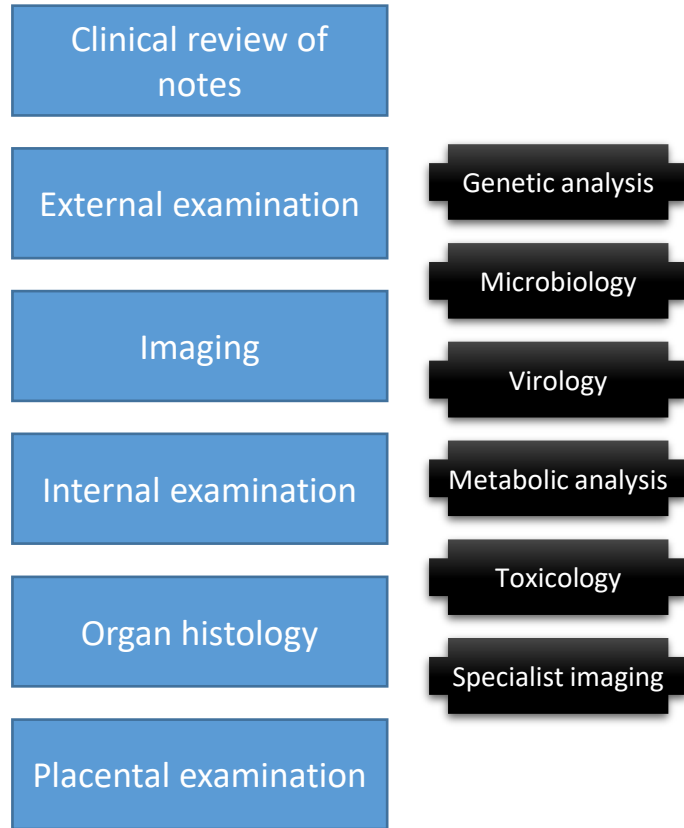
- 400 unselected fetal and children autopsies
- Blind reporting of the PMMRI & autopsy
- MRI data compared to full autopsy report.
- Retrospective review of MRI data:

*“in the cases for which a pathologist and radiologist jointly predicted that full autopsy was unnecessary, the concordance rate for cause of death or major pathology was almost 100%”*

Require tissue-based investigations to improve diagnostic yield

	Fetuses ≤24 weeks (n=185)	Fetuses >24 weeks (n=92)	Children (n=123)	Total (n=400)
<b>MRI alone vs conventional autopsy</b>				
Concordant	79 (42.7%, 35.8-49.9)	58 (63.0%, 52.8-72.2)	85 (69.1%, 60.5-76.6)	222 (55.5%, 50.6-60.3)
Non diagnostic	64 (35%)	4 (4%)	4 (3%)	72 (18%)
Discordant	42 (23%)	30 (33%)	34 (27%)	106 (27%)
Apparent false-positives	2 (1%)	2 (2%)	2 (2%)	6 (2%)
Callosal agenesis	1	0	0	1
Ischaemic brain injury	0	2	1	3
Lungs (drowning)	0	0	1	1
Dilated renal pelvis	1	0	0	1
Undetected abnormality	40 (22%)	28 (30%)	32 (26%)	100 (25%)
Genetic syndrome	2	4	0	6
Haematological	0	2	0	2
Lungs (aspiration)	0	0	1	1
Pulmonary haemorrhage	0	0	1	1
Metabolic (steatosis)	0	0	1	1
Athrogryposis	2	0	1	3
Fracture	0	0	1	1
Cleft palate	1	0	0	1
Skeletal dysplasia	1	0	0	1
Placental	34	21	3	58
Sepsis	0	1	24	25

# Minimally invasive vs. conventional autopsy



	Fetuses ≤24 weeks (n=185)	Fetuses >24 weeks (n=92)	Children (n=123)	Total (n=400)
<b>Minimally invasive autopsy vs conventional autopsy</b>				
Concordant	175 (94.6%, 90.3–97.0)	88 (95.7%, 89.3–98.3)	94 (76.4%, 68.2–83.1)	357 (89.3%, 85.8–91.9)
Non-diagnostic*	9 (5%)	1 (1%)	0	10 (3%)
Discordant	1 (<1%)	3 (3%)	29 (24%)	33 (8%)
Apparent false-positives	1 (<1%)	2 (2%)	3 (2%)	6 (2%)
Callosal agenesis	1	0	0	1
Ischaemic brain injury	0	2	1	3
Lungs (drowning)	0	0	1	1
Skull fracture†	0	0	1	1
Undetected abnormality	0	1 (1%)	26 (21%)	27 (7%)
Lungs (aspiration)	0	0	1	1
Pulmonary haemorrhage	0	0	1	1
Metabolic (steatosis)	0	0	1	1
Sepsis‡	0	1	23	24

# Key concepts

1. Imaging may be able to **substitute for internal examination** in some scenarios.
2. It should be possible to identify the **diagnostic yield** of various components of the autopsy, **by clinical indication**.
3. Less invasive options for autopsy may **improve acceptability** of autopsy for families and access to tissue and phenotype for medics and researchers.



# Key concepts

- ~~1. Imaging may be able to **substitute for internal examination** in some scenarios.~~
2. It should be possible to identify the **diagnostic yield** of various components of the autopsy, **by clinical indication**.
- ~~3. Less invasive options for autopsy may **improve acceptability** of autopsy for families and access to tissue and phenotype for medics and researchers.~~

# Internal examination & histology

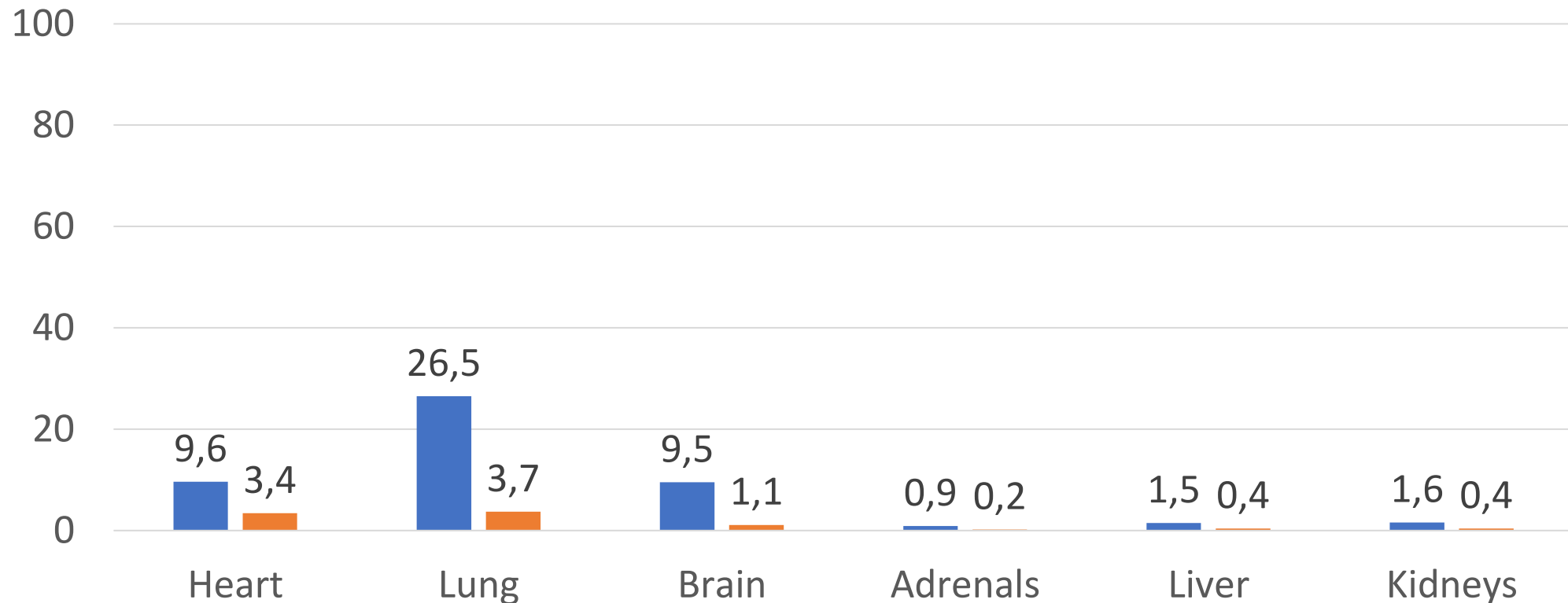
Next slides focus on the diagnostic yield of body histology by indication.

# Internal examination & histology

- GOSH Autopsy Database updated (2005 – 2016)
- Cases split by indication
  - SUDC, SUDI, IUFD, ToP
- Histology and internal examination analysed for each major organ and each indication
- Findings in each organ coded objectively
  - Definitely, Possibly or Non-contributory to cause of death

# Internal examination & histology

Definite cause of death (%) obtained in Sudden Unexpected Death in Childhood (SUDC) by histology. [n = 824]

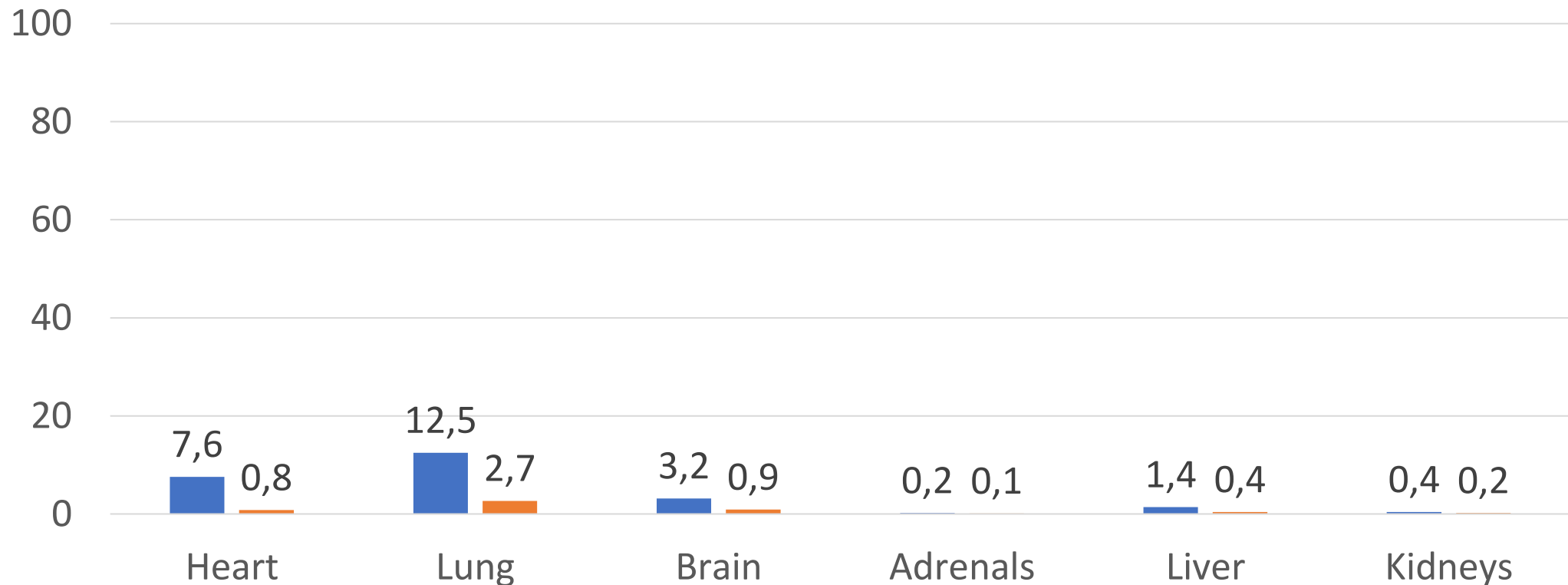


- Cause of death, abnormal macroscopic appearance
- Cause of death, normal macroscopic appearance



# Internal examination & histology

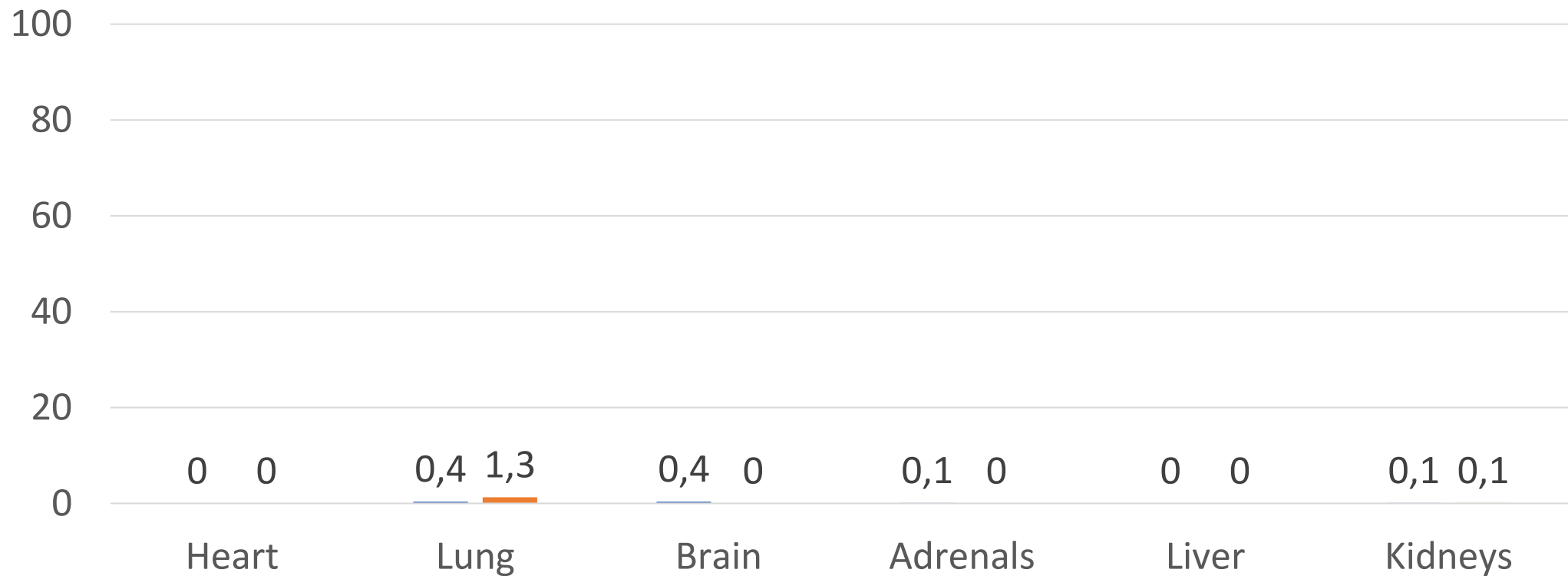
Definite cause of death (%) obtained in Sudden Unexpected Death in Infancy (SUDI) by histology [n = 1,739]



- Cause of death, abnormal macroscopic appearance
- Cause of death, normal macroscopic appearance

# Internal examination & histology

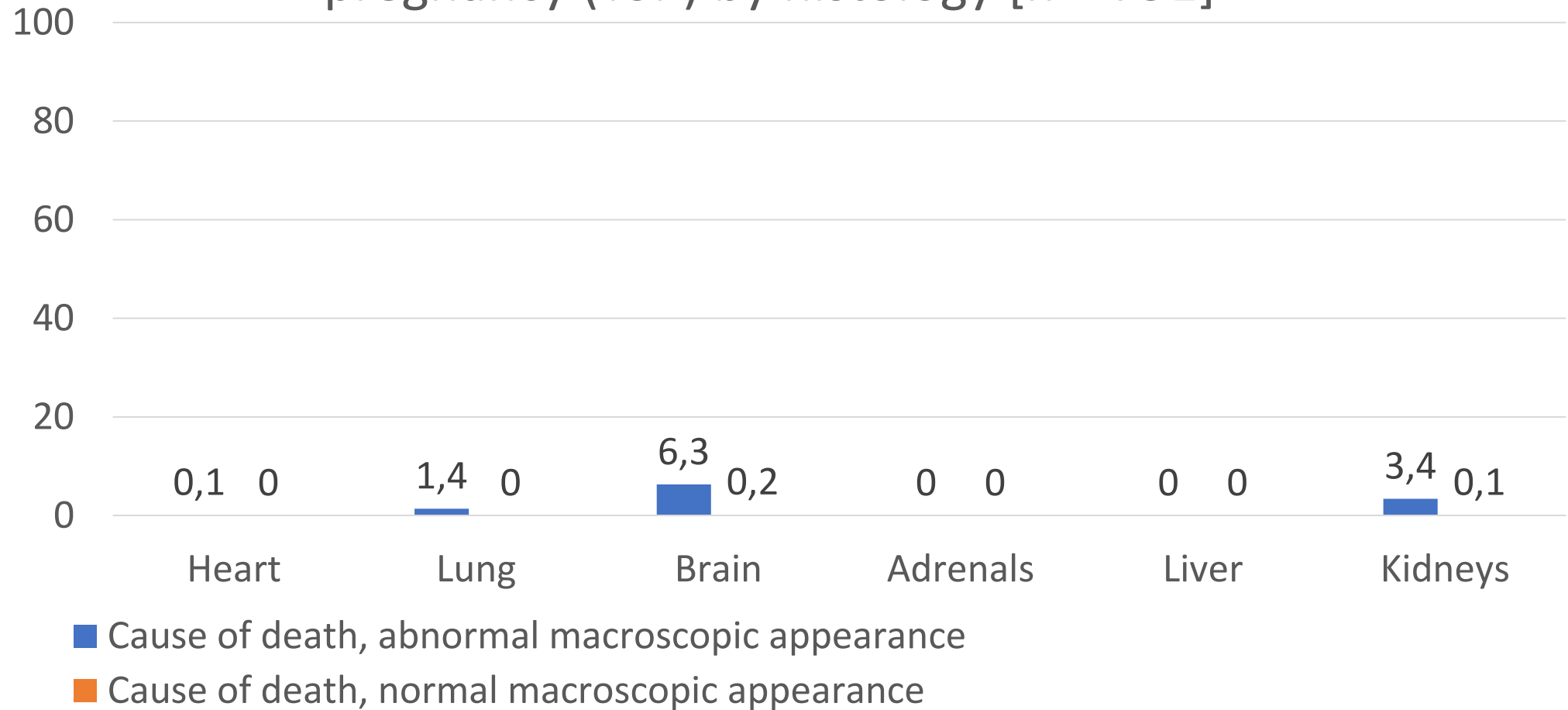
Definite cause of death (%) obtained in Intrauterine fetal death (IUFD) by histology [n = 1,957]



- Cause of death, abnormal macroscopic appearance
- Cause of death, normal macroscopic appearance

# Internal examination & histology

Cause of death (%) obtained in termination of pregnancy (ToP) by histology [n = 791]



# Key messages

- In SUDI and SUDC, thorough sampling of macroscopically normal organs is necessary due to occult disease, regardless of imaging findings.
- In IUFD and ToP, there is a low diagnostic yield from routine organ histology of macroscopically normal organs. Histology is not routinely necessary.
- In IUFD and ToP, further examination of the organ by a pathologist is much more likely to be helpful if there is clinical or radiological suspicion of an abnormality relevant to death.



# Tissue for adjunct investigations

**Tissue remains important for adjunct investigations:**

**How will we get it?**

Clinical review of notes

External examination

Imaging

Internal examination

Organ histology

Placental examination

Genetic analysis

Microbiology

Virology

Metabolic analysis

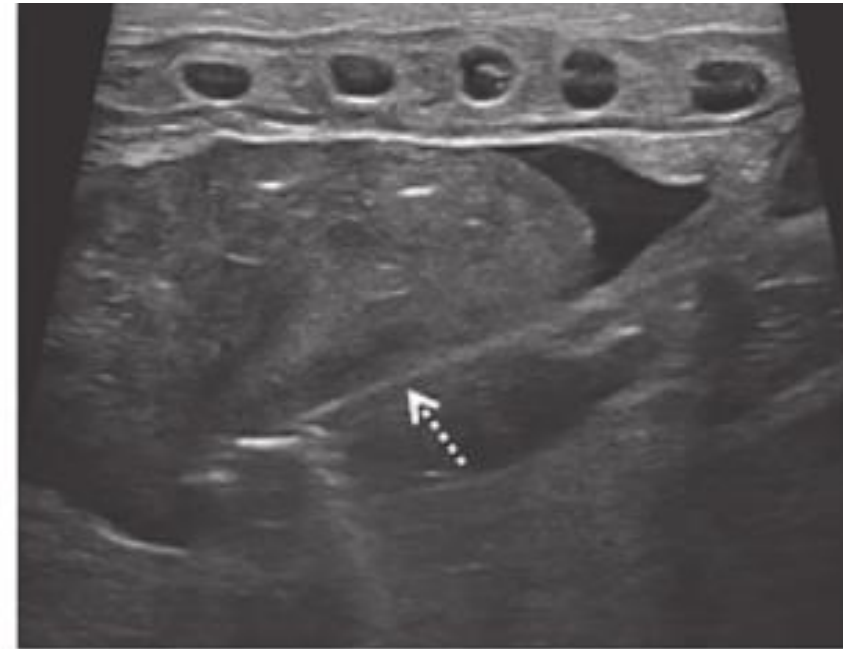
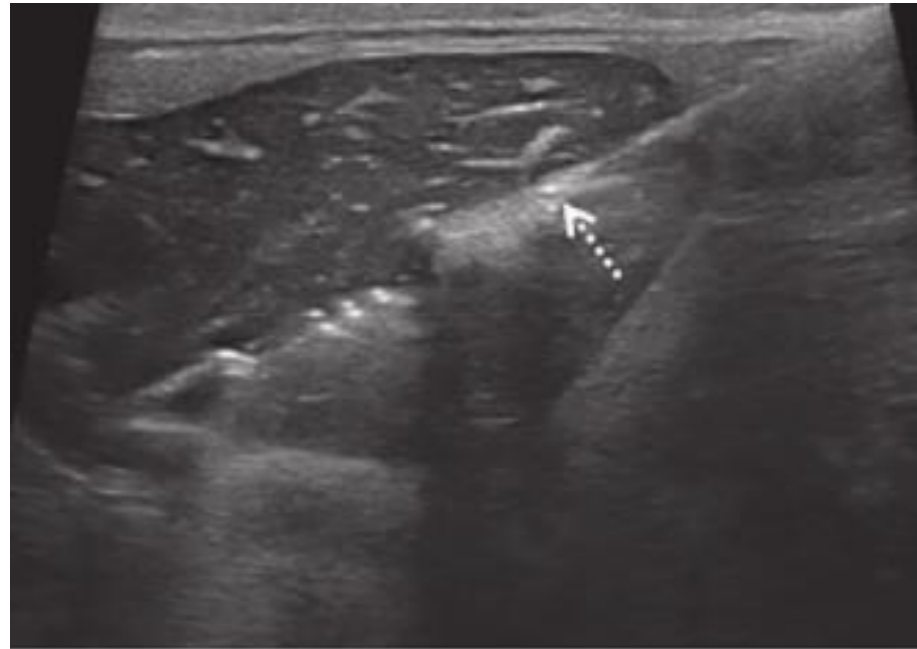
Toxicology

Specialist imaging

# Minimally invasive tissue sampling

## INTACT




### (INcisionless TArgeted Core Tissue biopsy)




# Minimally invasive tissue sampling

**ULTRASOUND**  
in Obstetrics & Gynecology



Original Paper |  Open Access |  

**Feasibility of INTACT (INCisionless TArgeted Core Tissue) biopsy procedure for perinatal autopsy**

S. C. Shelmerdine , J. C. Hutchinson, L. Ward, T. Sekar, M. T. Ashworth, S. Levine, N. J. Sebire, O. J. Arthurs,

- USS guided via the umbilical vein.
- USS confirmation of biopsy sampling site.
- Benefits of PM-USS examination with guided sampling.
- No incisions or needle puncture marks left on the body.
- Overall sampling success >75%.
  - Spleen was particularly poor




# Minimally invasive tissue sampling

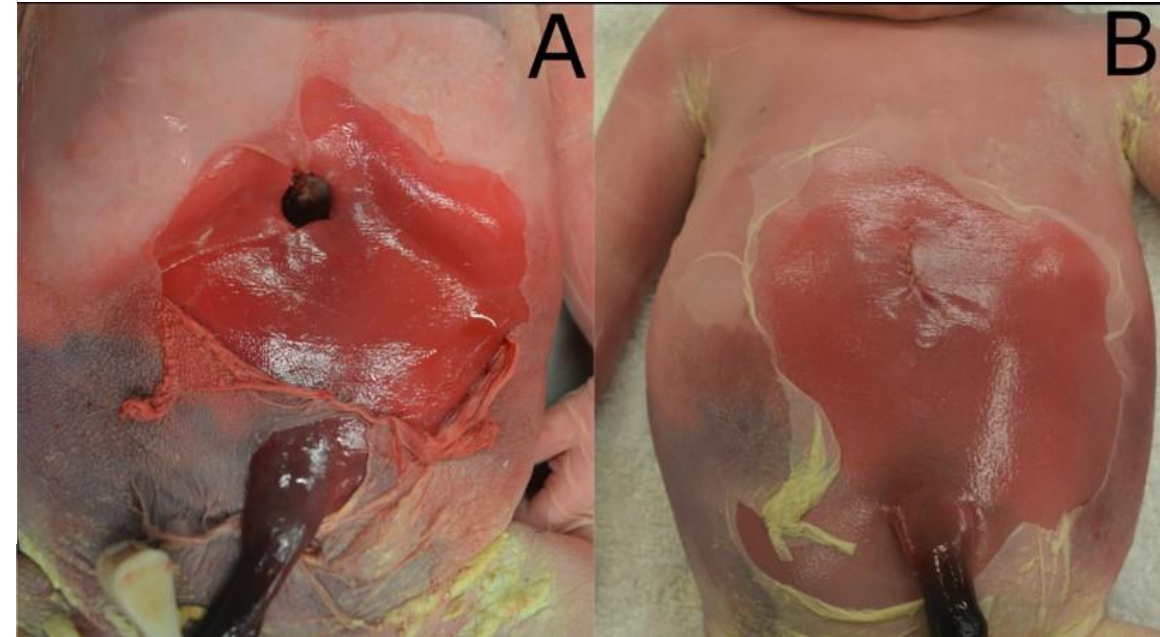
**ULTRASOUND**  
in Obstetrics & Gynecology



Original Paper | [Free Access](#)

Minimally invasive perinatal and pediatric autopsy with laparoscopically assisted tissue sampling: feasibility and experience of the MinImAL procedure

J. C. Hutchinson, S. C. Shelmerdine, C. Lewis, J. Parmenter, I. C. Simcock, L. Ward, M. T. Ashworth, L. S. Chitty, O. J. Arthurs, N. J. Sebire 

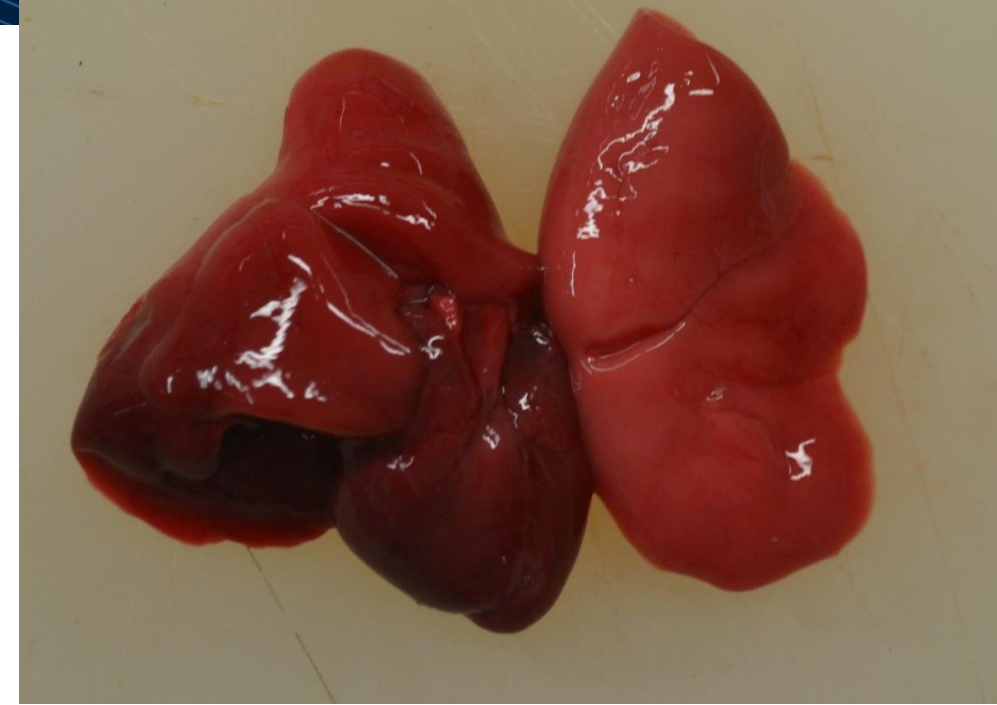


- 1 – 3 cm incision sub-xiphisternally or in left hypochondrium.
- No need to open the head with normal MRI/US (high NPV)



# Minimally invasive tissue sampling

- En-bloc resections possible
- Good tissue sampling success rates in heart, lungs, kidneys (all 100%), liver (97%) and adrenals (89%).
- Skilled operator required.




**ULTRASOUND**  
in Obstetrics & Gynecology



Original Paper | [Free Access](#)

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# Topic 2 (v2)

# What imaging can offer for less invasive autopsy

Dr Ian Simcock; UCL Great Ormond Street Institute of Child Health, London, UK

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## Non-invasive fetal autopsy

“What imaging can offer for less invasive autopsy”

- Dr Ian Simcock
- Clinical Academic Radiographer
  - UCL GOSH ICH BRC
  - Twitter @ian\_simcock



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Great Ormond Street  
Hospital Biomedical  
Research Centre



# Non-invasive fetal post-mortem imaging

- What imaging modalities are available?
  - X-ray
  - CT
  - MRI
  - Ultrasound
  - Micro-CT
- When should we use them?
- Where is the clinical service now?
- What does the future hold?



# Radiographs

- Most widely used PM radiology technique
- Overview of bone structure + abnormalities – skeletal dysplasias

Pediatr Radiol (2014) 44:252–257  
DOI 10.1007/s00247-013-2804-0

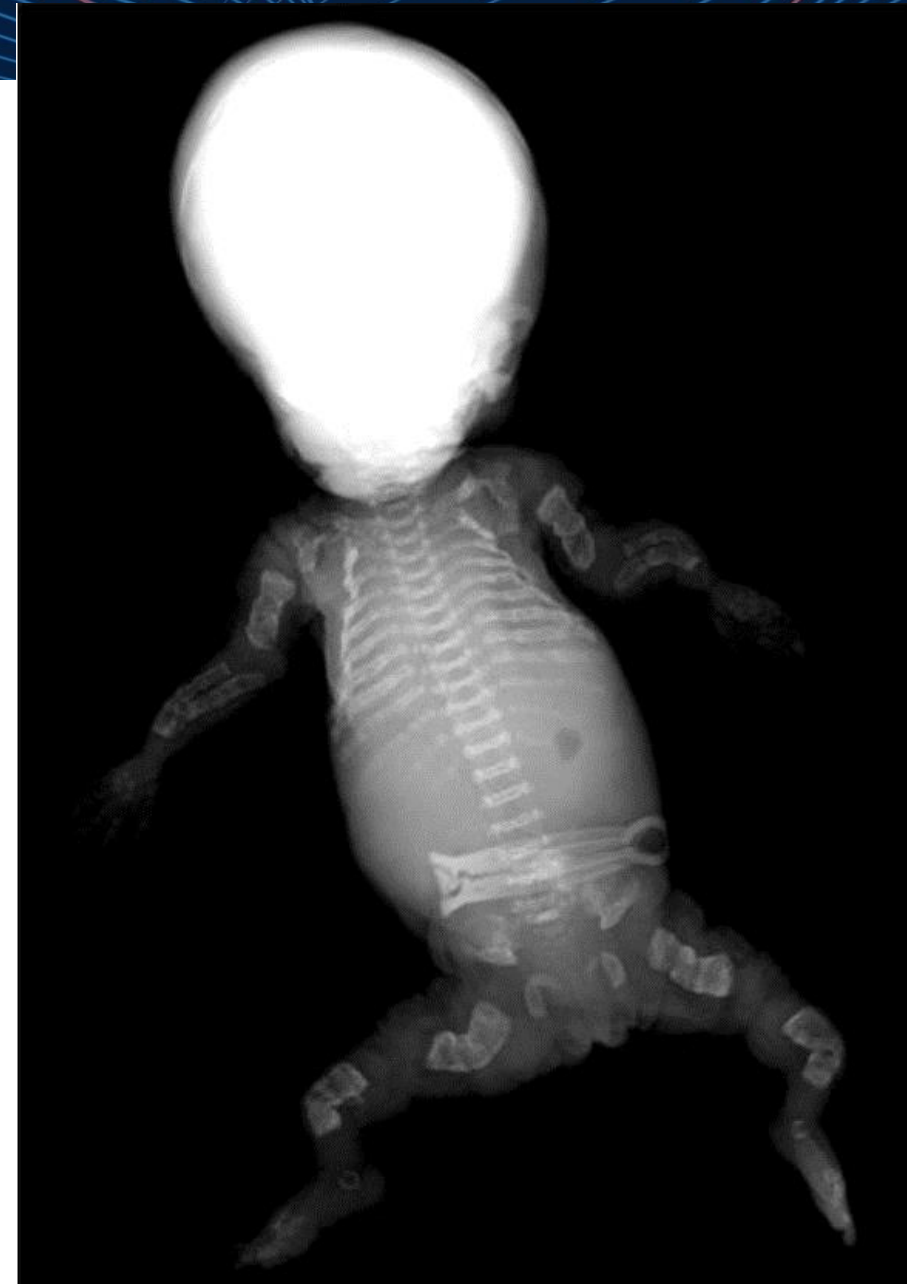
ORIGINAL ARTICLE

## **Routine perinatal and paediatric post-mortem radiography: detection rates and implications for practice**

Owen J. Arthurs • Alistair D. Calder • Liina Kiho •  
Andrew M. Taylor • Neil J. Sebire

Crumpled long bones and ribs with multiple fractures from osteogenesis imperfecta type II, terminated fetus

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# Cost of workload?

- Low overall yield when imaging fetuses
- Only 0.5% contribution to diagnosis in non-indicated cases
- Yield higher when imaging those fetuses in whom an abnormality was suspected – to confirm diagnosis.
- Could use CT instead?
  - Provides bony information

Pediatr Radiol (2014) 44:252–257  
DOI 10.1007/s00247-013-2804-0

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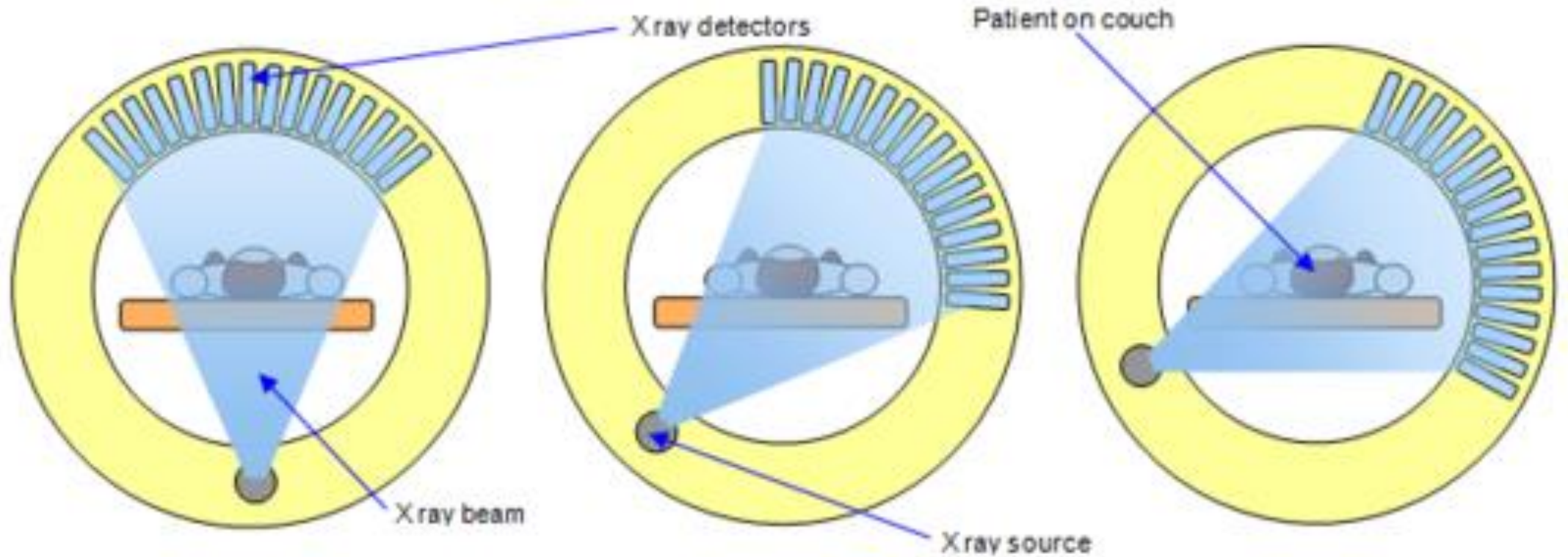
# CT scanners



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# CT scanners

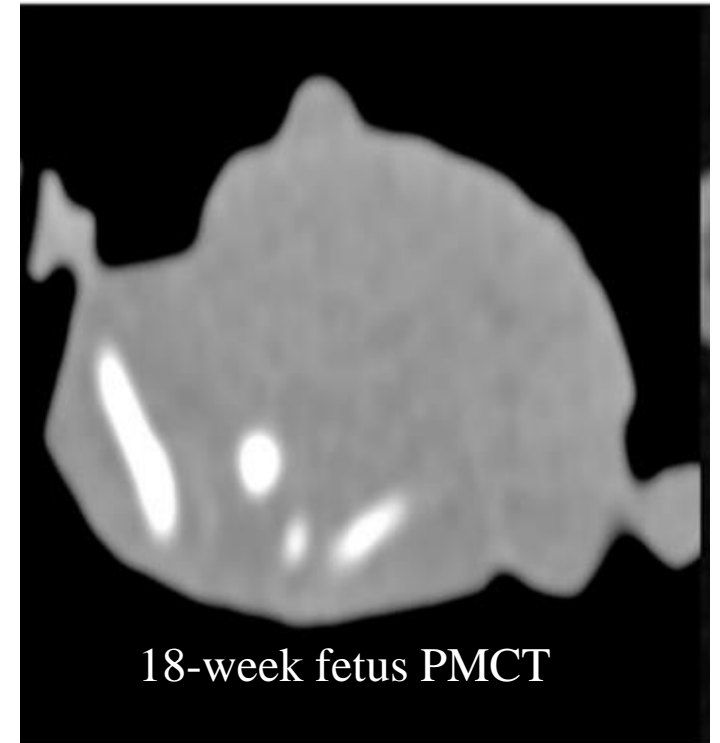


CT scanner with the X ray source and detectors shown in three positions



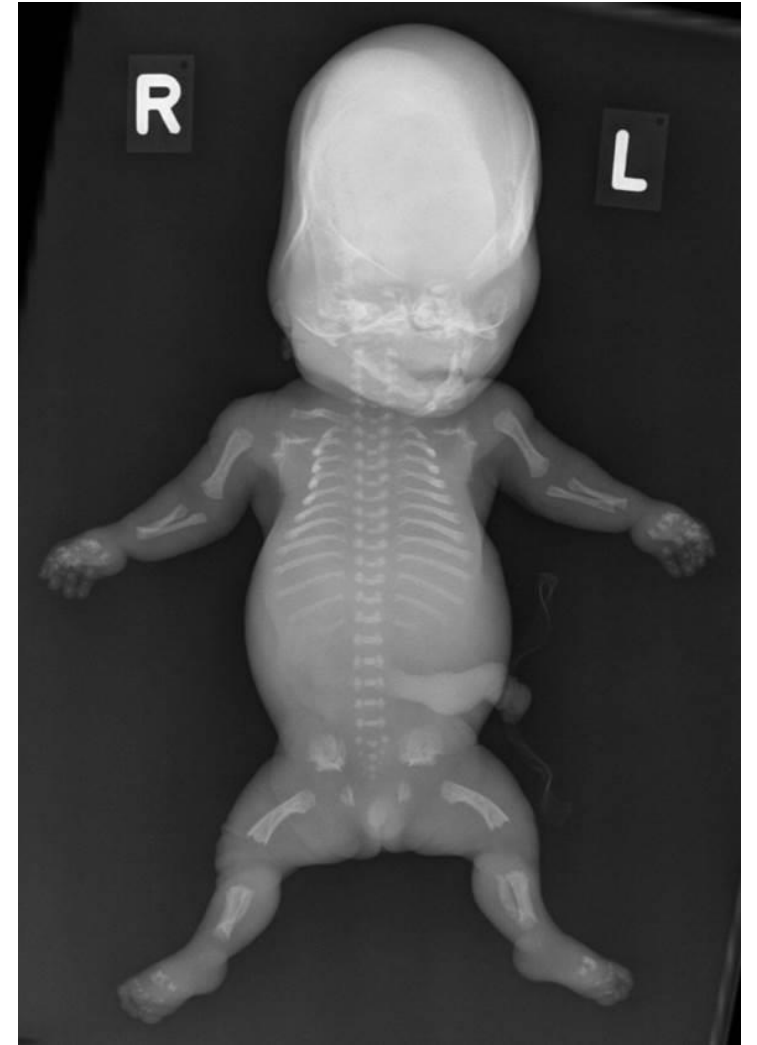
# Post-mortem CT

- PM CT is fast, readily inexpensive and widely available.
- Provides bone detail and can detect presence of air
- Vascular contrast agent used in adults
  - Difficult vascular access in fetuses
- Reduced soft tissue contrast – little subcutaneous fat around organs
- Challenging thoracic + abdominal diagnosis
- **Fetal**
- Lower resolution due to small size of fetuses
- CT largely undiagnostic in perinatal autopsy

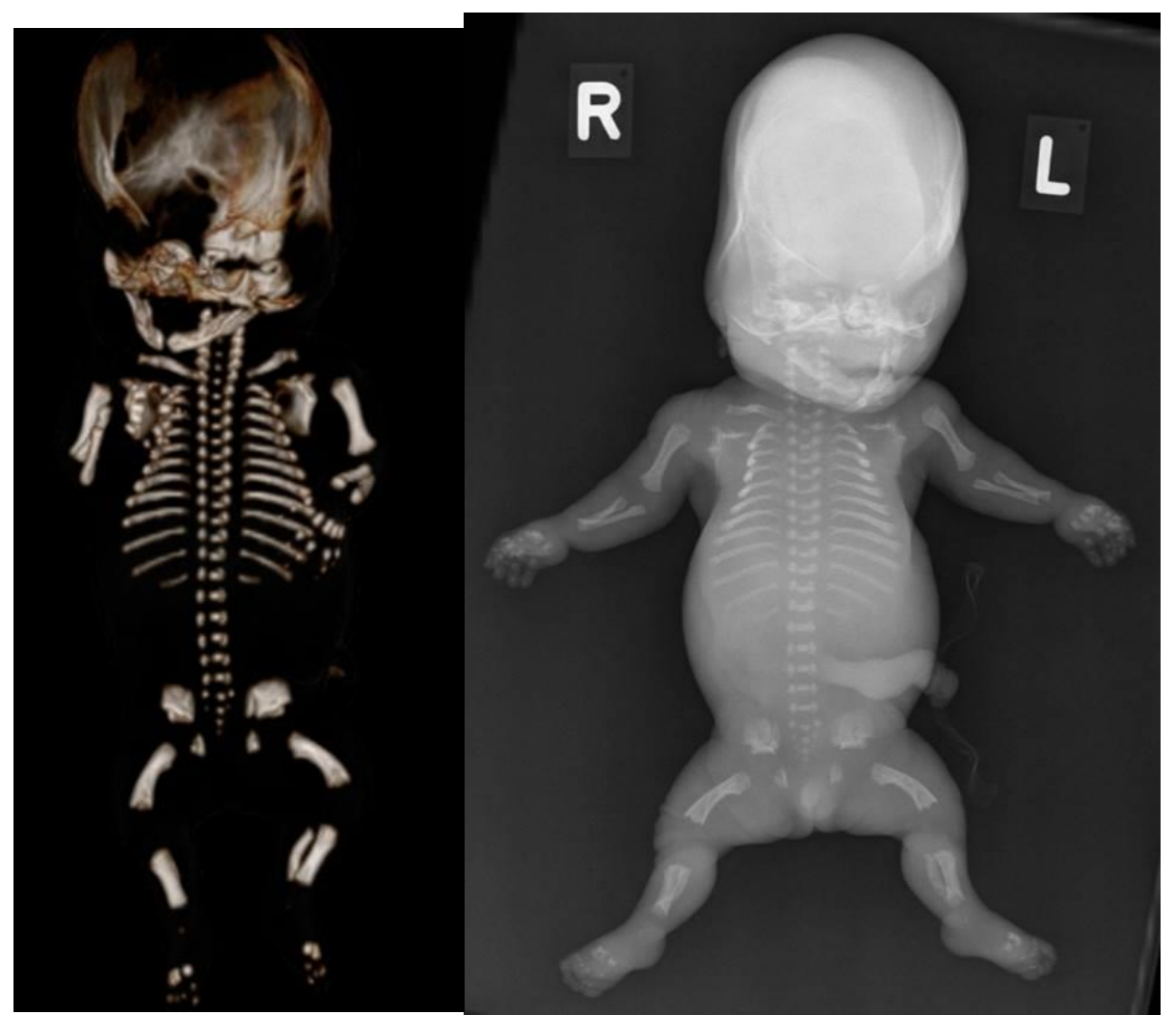
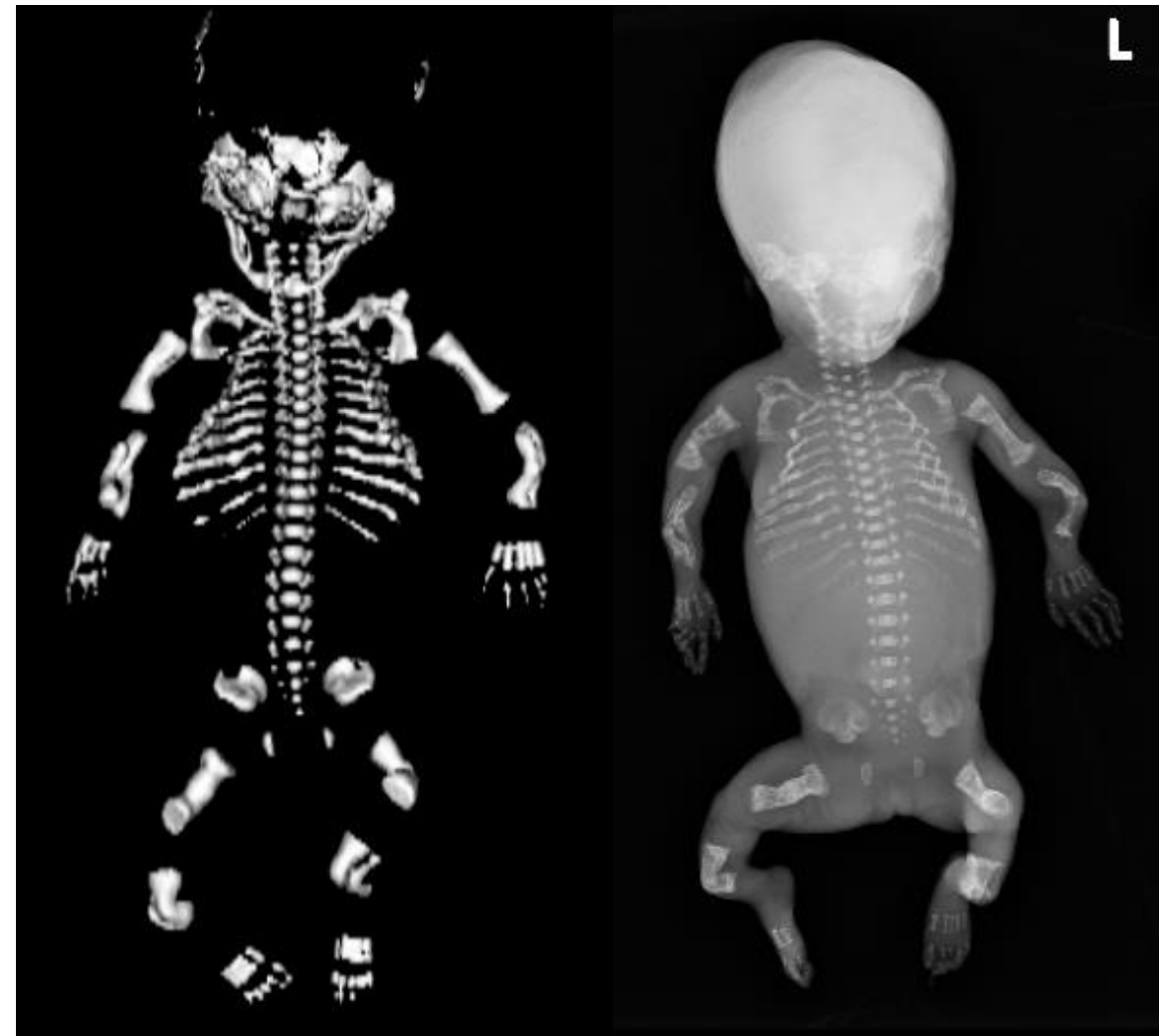


Ref: Arthurs OJ et al., 2017  
Forensic Sci Med Pathol

# Techniques: X-ray vs CT



# Techniques: X-ray vs CT

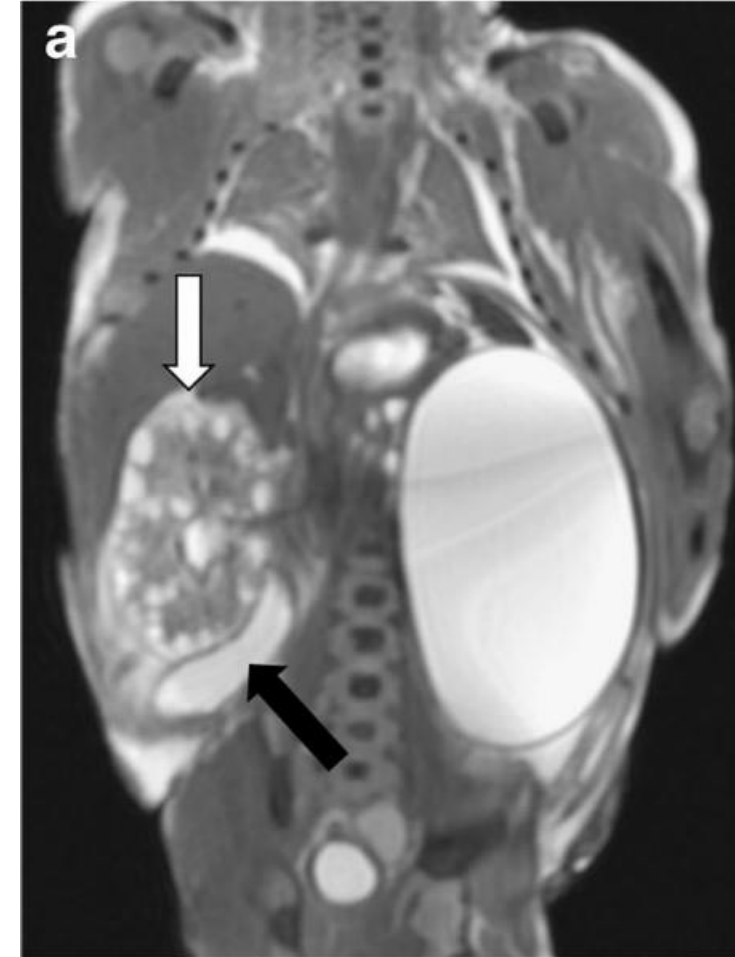




# Post-mortem MRI

- MRI is more expensive, can be challenging to access
- Multiple field strengths (1.5T, 3T and up to 9.4T)
- Excellent for soft tissue without contrast agent
- **1.5T**
  - Technical issues relating to small fetal size and image resolution
  - Increased fetal weight = increased diagnostic yield
  - >500g body weight = >90% diagnostic
  - <200g body weight = <50% diagnostic

Jawad N et al., 2016 UOG  
Arthurs O et al., 2015 Pediatr Radiol



22-week gestation fetus – multi-cystic right kidney and obstructed left kidney – bladder outflow obstruction



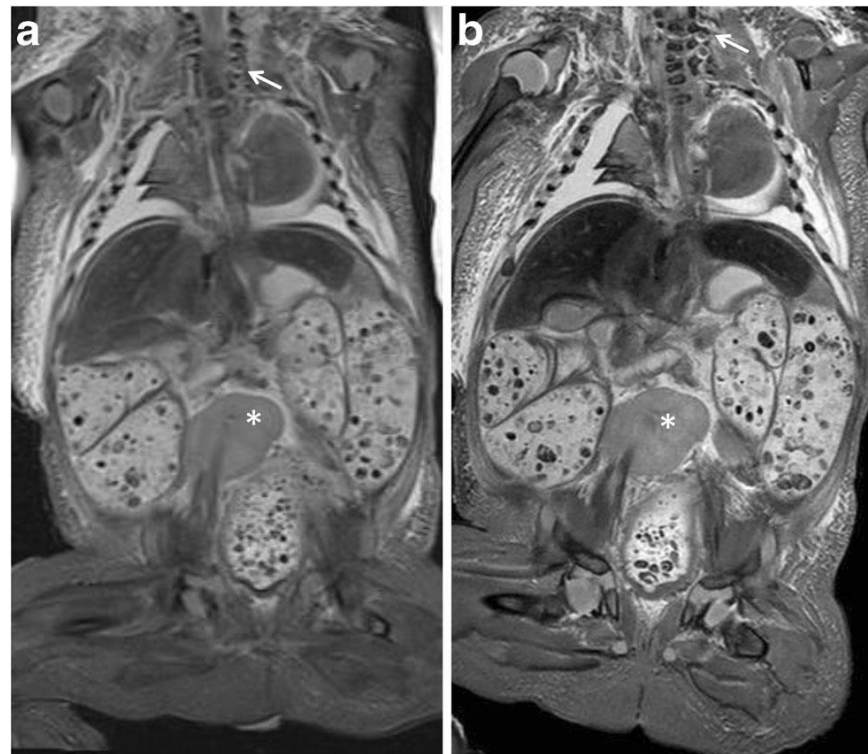
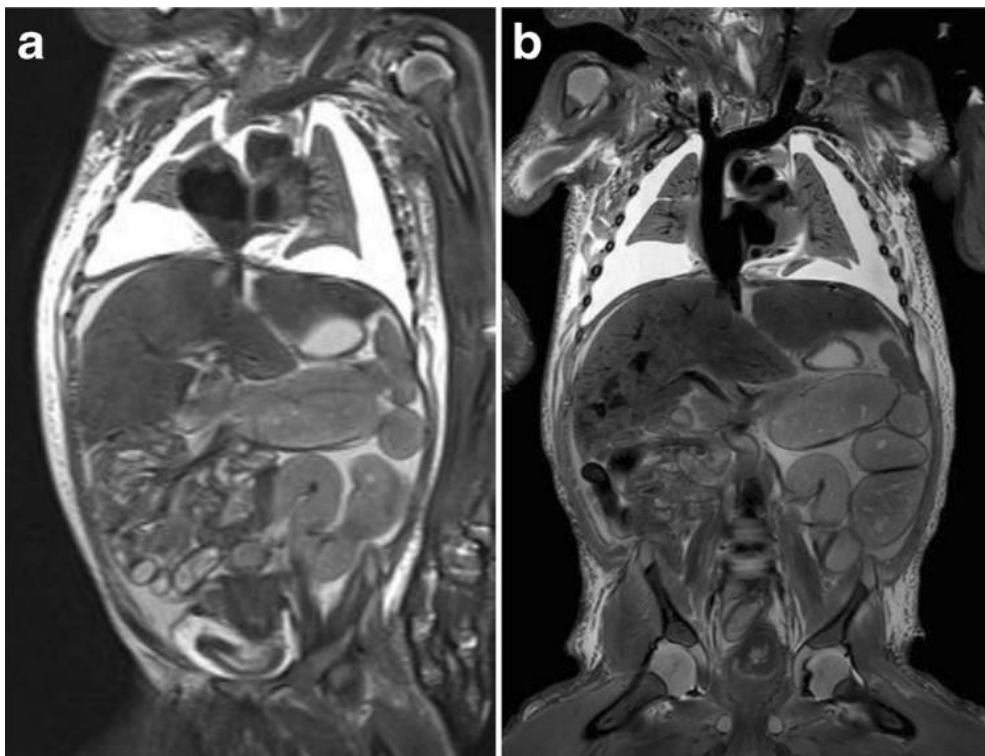
# 3T MRI

- 3T MRI is less accessible than 1.5T, but is increasing
- Higher field strength = increased SNR, spatial resolution and more detailed imaging on smaller patients
- Increased accuracy, better image contrast, lower diagnostic error for thorax, heart and abdomen at 3T than 1.5T
- These increases are mainly seen in fetuses <20-week gestation

Ref: Kang X et al., 2017 Eur Radiol

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# 1.5T vs 3T



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# 7T to 9.4T

- Ultra-high field MRI even greater SNR and spatial resolution on smaller patients
- 9.4T can diagnose cardiac abnormalities regardless of gestation
- Solely research scanners due to limited clinical application, very expensive and long scanning times



13-week gestation fetus 7T thickened interventricular septum and left ventricle wall.



# MRI Fetal Imaging

- Larger fetuses, 3<sup>rd</sup> trimester
- Congenital abnormalities – brain, cardiac, abdominal
- Prematurity
- Perinatal events
- Infection
- Most useful imaging modality: MRI
  - US where MRI not available
  - Remember antenatal US findings !

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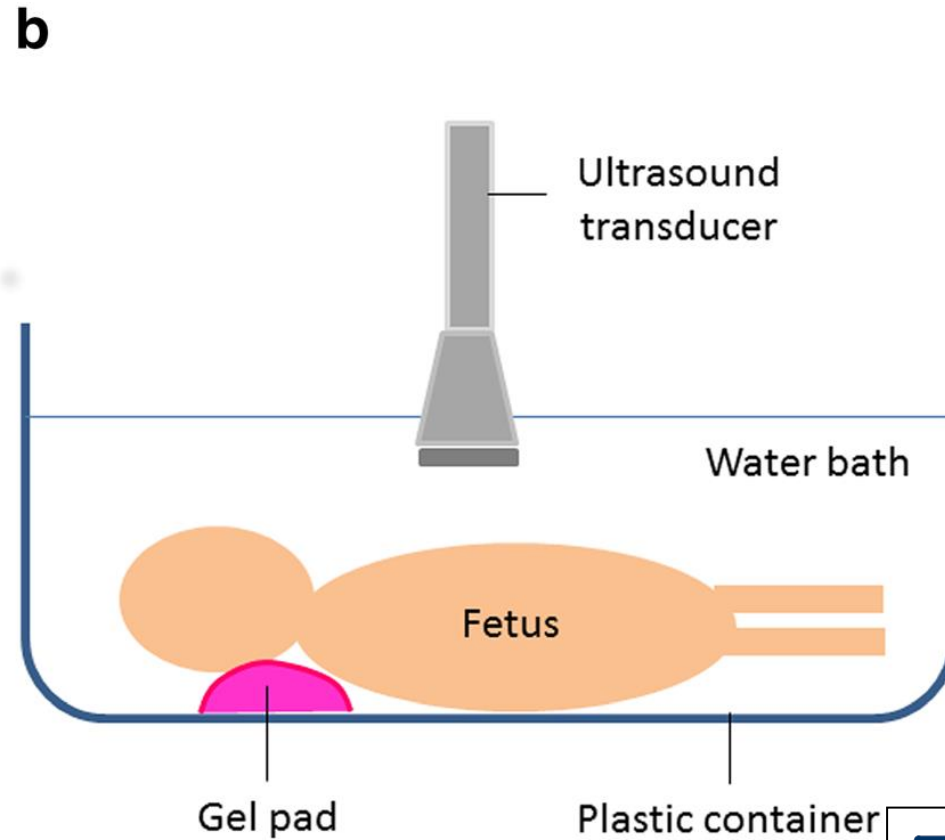
# PM ultrasound

- Cheap, accessible, portable
- Understandable to parents
- Easily translatable to PM imaging
- Paediatric Radiology, fetal medicine, obstetrician experience, sonographers
- Can be used to guide tissue biopsy



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# PM Ultrasound



EDUCATIONAL REVIEW

Open Access

Perinatal post mortem ultrasound (PMUS):  
a practical approach

Susan C. Shelmerdine<sup>1,2\*</sup>, Neil J. Sebire<sup>1,2</sup> and Owen J. Arthurs<sup>1,2</sup>



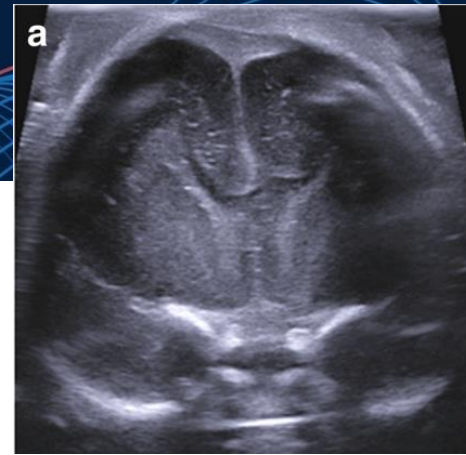
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# PM Ultrasound

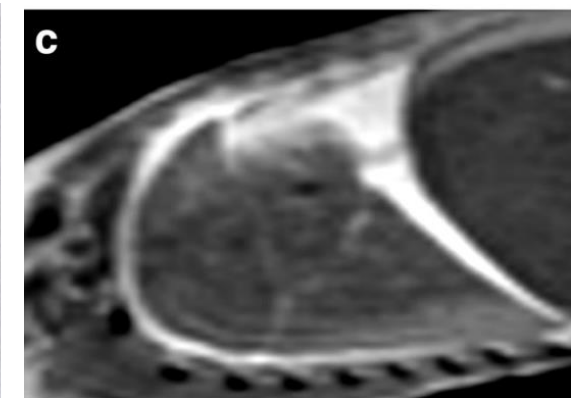
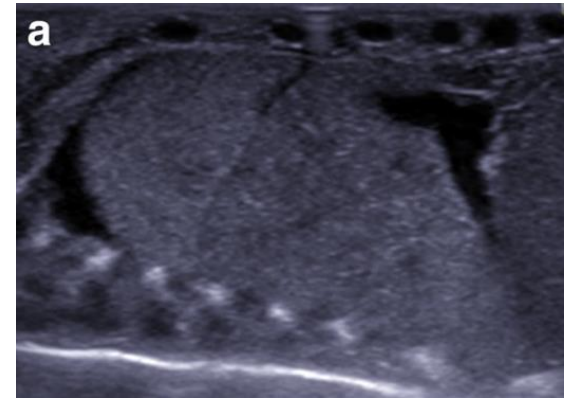
- Wide range of areas of the body
- Could be used as a first line “screening” – Which parents benefit from further imaging or biopsy techniques
- Maceration can degrade images and <20-week gestation
- Increased accuracy >20-week gestation
- Therefore, use for >20-week gestation and non-macerated

Ref: Shelmerdine SC et al., 2019 Insights Imaging

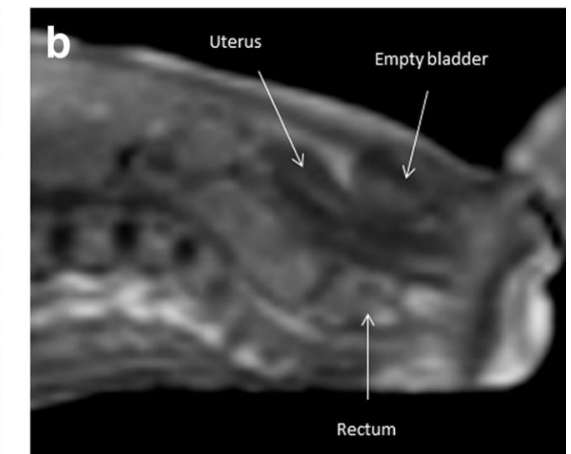
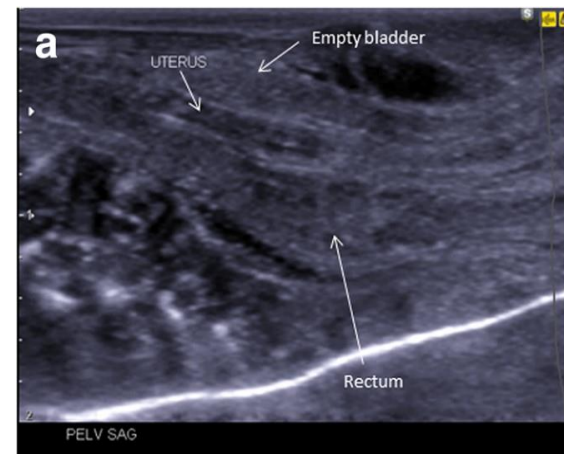
Neuro



Thoracic



Pelvic





# Micro-CT scanners



Clinical CT scanner

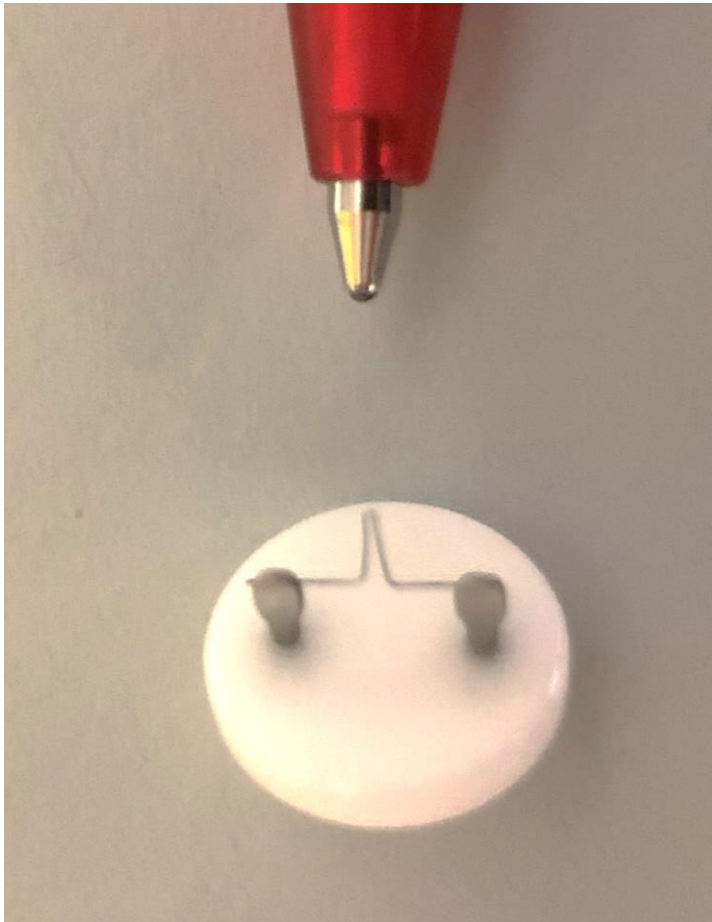


Micro-CT scanner

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# Micro-CT

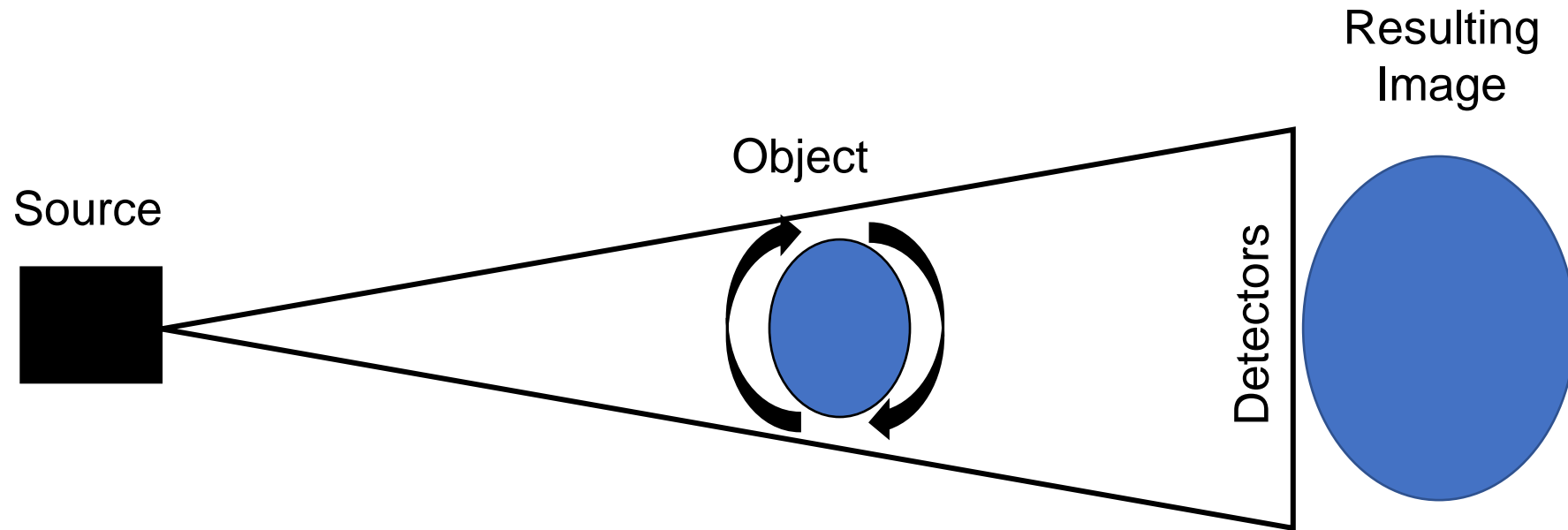


Higher resolution due to smaller filament



# Micro-CT setup

- Varied object position
- Closer to source = increased resolution
- Smaller the specimen, higher the resolution



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# Micro-CT protocol

- Requires contrast agent
- Immersed in a bath of potassium tri-iodide ( $I_2KI$ )
- Difficult to access due to availability

nature  
protocols

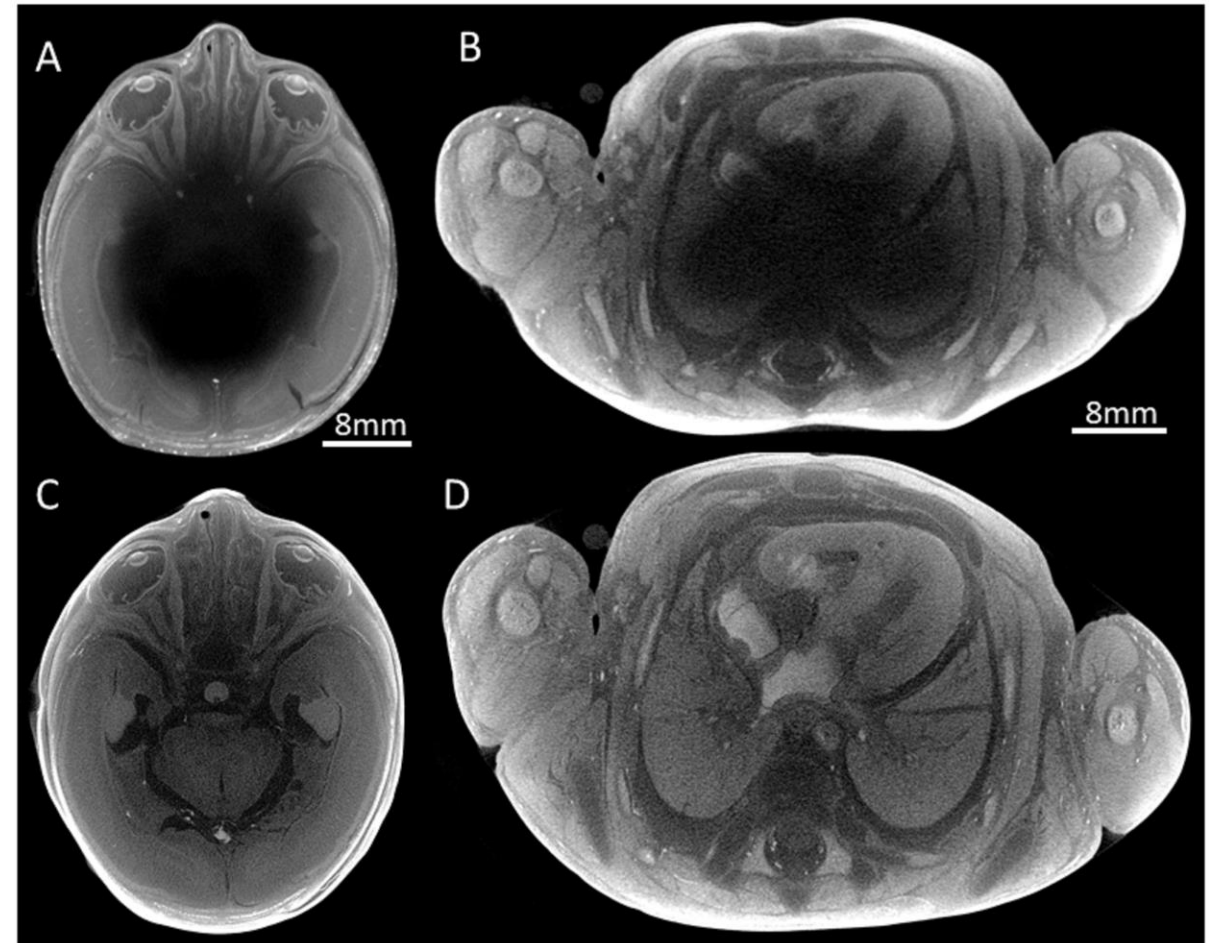
PROTOCOL

<https://doi.org/10.1038/s41596-021-00512-6>



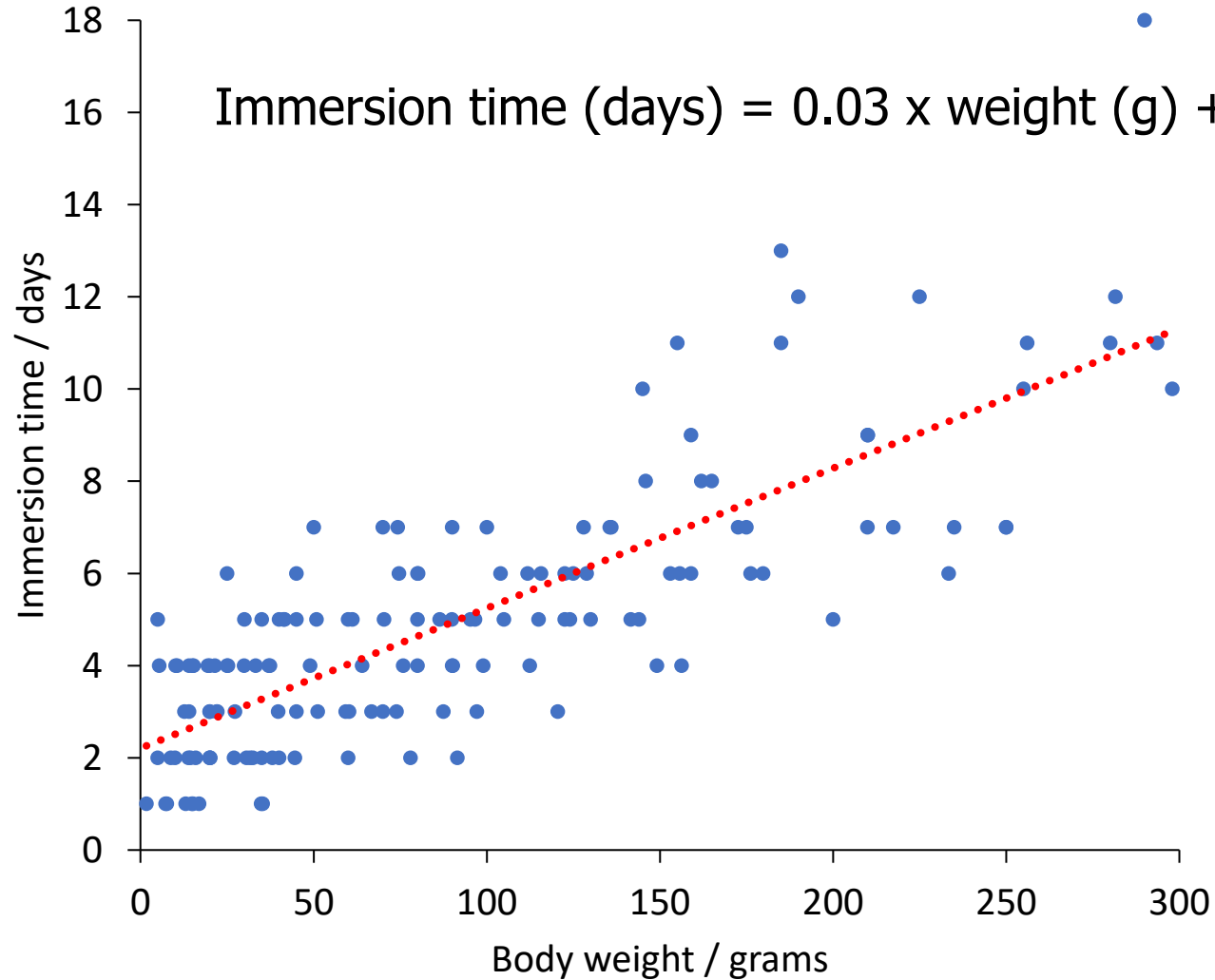
## Human fetal whole-body postmortem microfocus computed tomographic imaging

Ian C. Simcock<sup>1,2,3</sup>, Susan C. Shelmerdine<sup>1,2,3</sup>, J. Ciaran Hutchinson<sup>2,3,4</sup>, Neil J. Sebire<sup>2,3,4</sup> and Owen J. Arthurs<sup>1,2,3</sup>



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# Contrast enhancement



50g - 4 days  
100g - 5 days  
200g - 8 days  
300g - 11 days

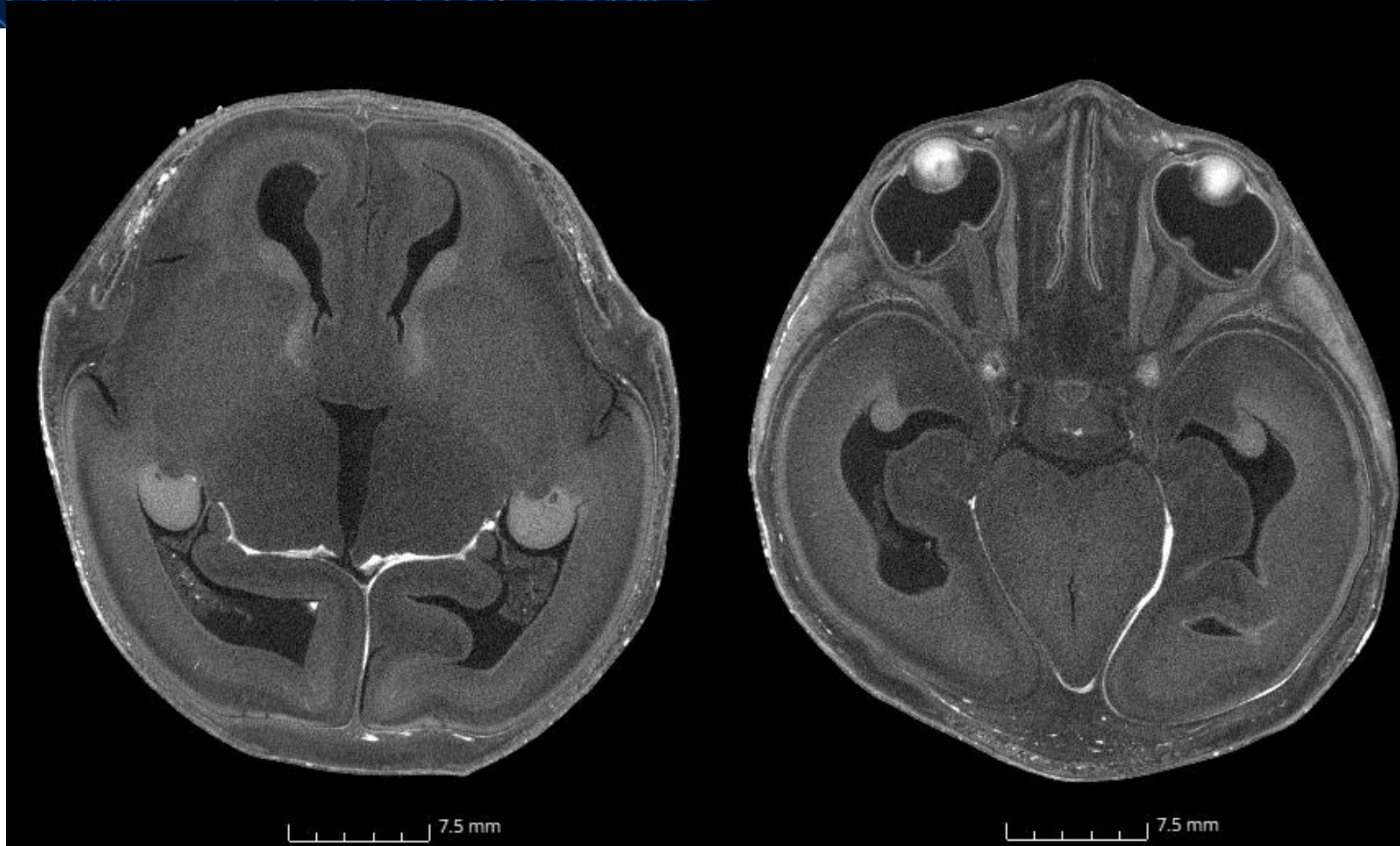
Ref: Simcock IC et al., BJROpen 2024

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# Micro-CT protocol

- Micro-CT high resolution images of neuro and soft tissue structures
- Clinical protocol
- Whole body scan
- Individual head and torso scan
- <90 minute clinical scan time

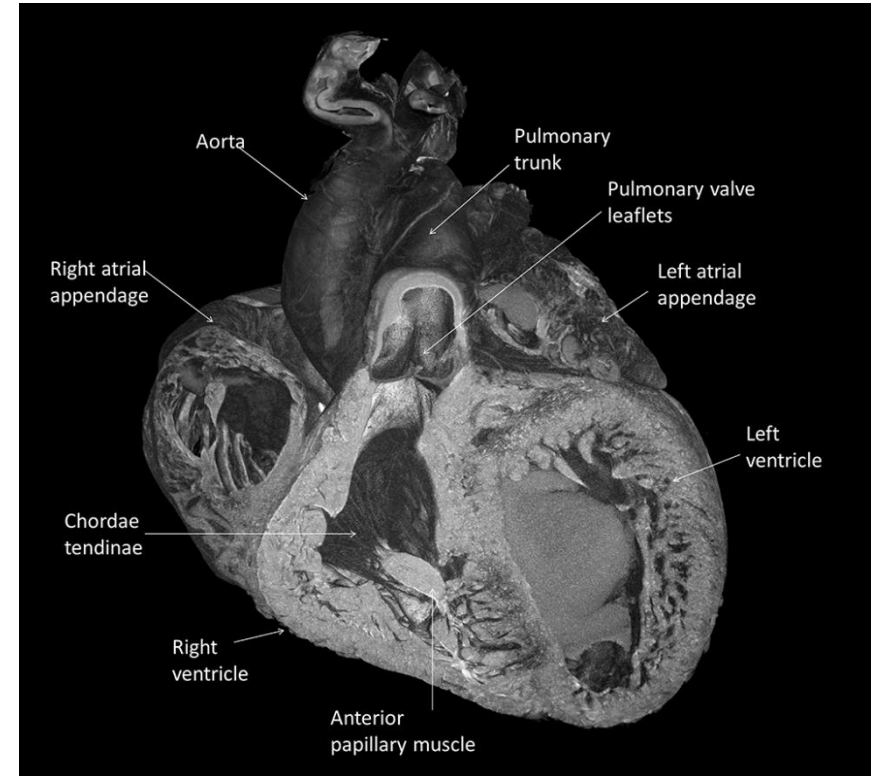
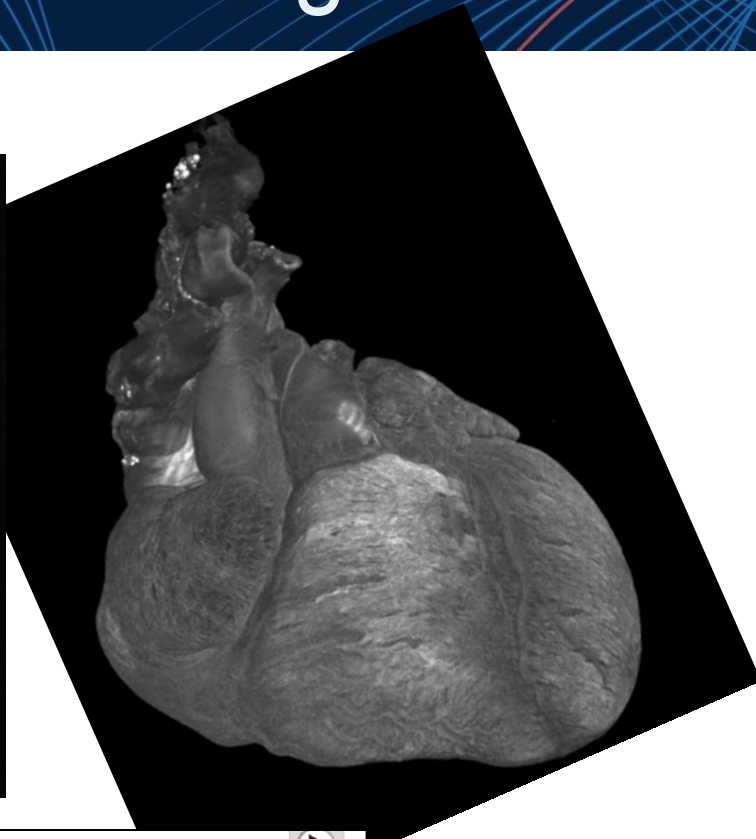


March 19, 2024





# Ex-vivo cardiac scanning



Ultrasound Obstet Gynecol 2016; 47: 58–64  
Published online 2 December 2015 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.15764



## Clinical utility of postmortem microcomputed tomography of the fetal heart: diagnostic imaging *vs* macroscopic dissection

J. C. HUTCHINSON\*†, O. J. ARTHURS\*‡, M. T. ASHWORTH†, A. T. RAMSEY§, W. MIFSUD\*†, C. M. LOMBARDI¶ and N. J. SEBIRE\*†

\*Institute of Child Health, UCL, London, UK; †Department of Histopathology, UCL Institute of Child Health & Great Ormond Street Hospital for Children, London, UK; ‡Paediatric Radiology, Great Ormond Street Hospital for Children NHS Trust, London, UK; §Nikon Metrology, Tring, UK; ¶Department of Radiology, Studio Diagnostico Eco, Vimercate, Milan, Italy

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# Skeletal dysplasias

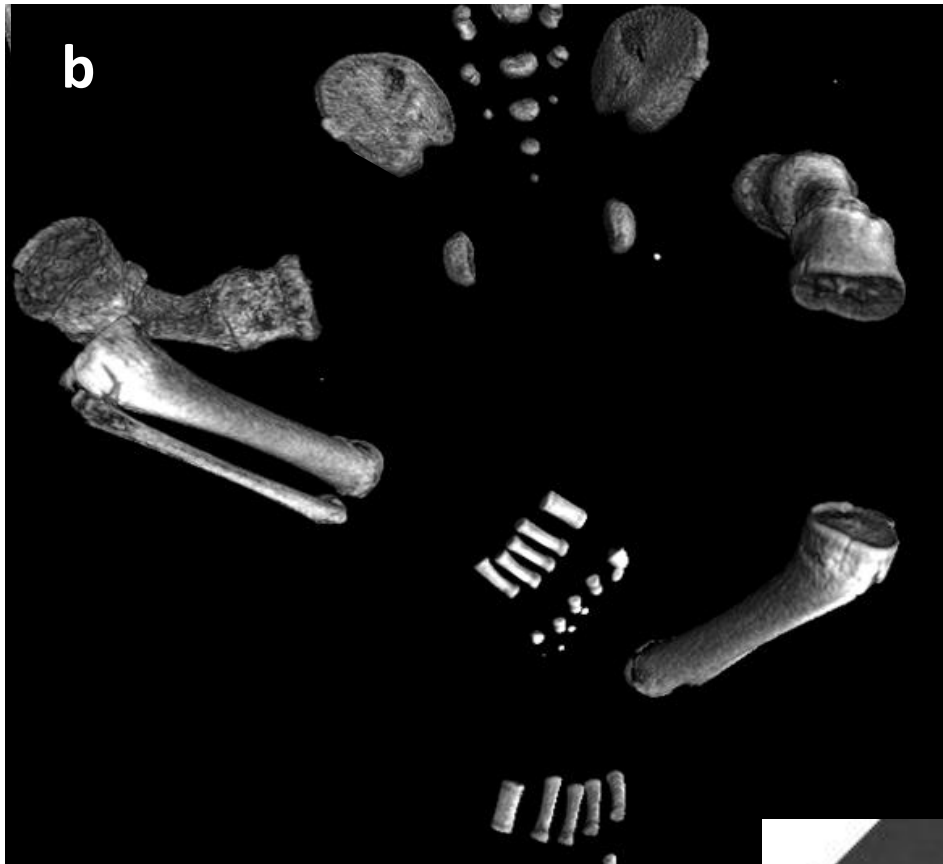
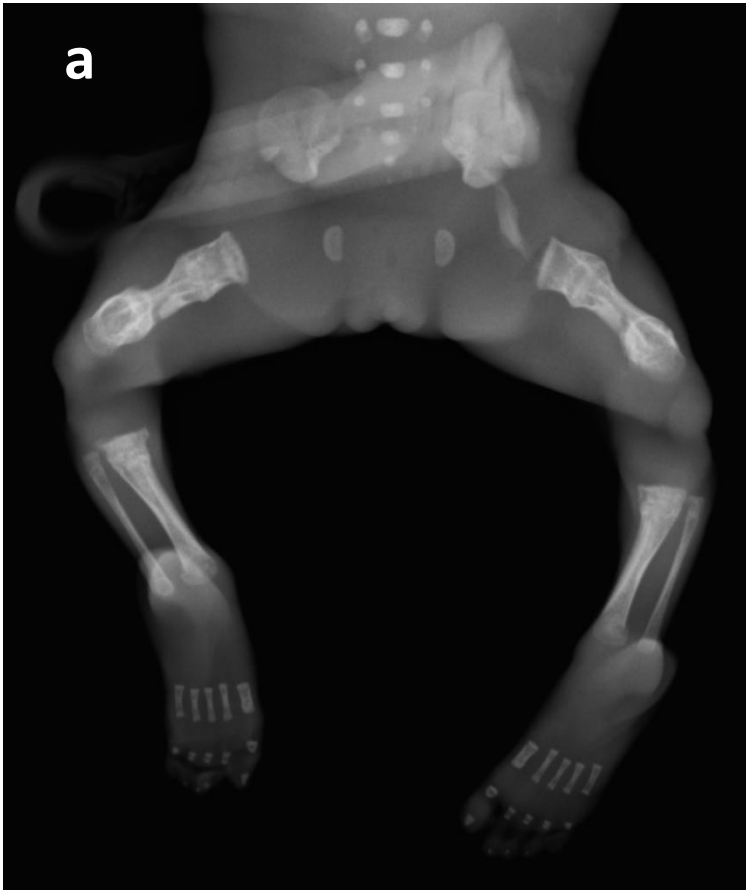
- OI is characterised by bone fragility, susceptibility to multiple bone fractures and low bone mass.
- There are multiple different distinct types of OI, with type 2 being lethal in the perinatal period
- The prevalence of OI varies between 1/10,000 to 1/20,000 although specifically for type 2 the prevalence is unknown.



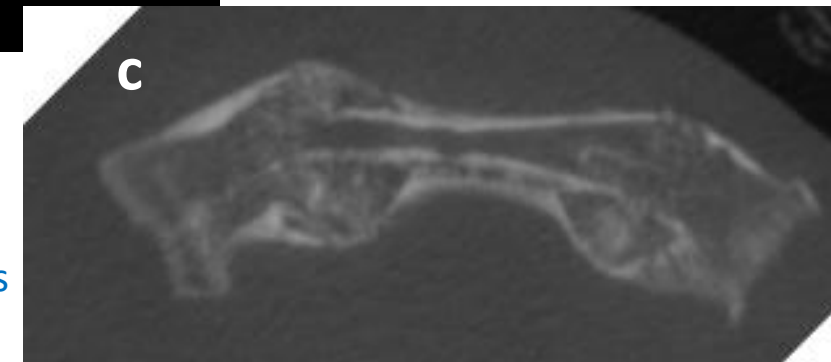
OI Type 2b a, radiograph b, micro-CT displays better the fracture of the distal humeral metaphysis and periosteal reaction at the proximal humeral metadiaphysis.



# Osteogenesis imperfecta



Ref: Shelmerdine et al., 2017  
RSNA



19-week gestation fetus with OI type 2b.

(a) radiograph of the lower limbs with multiple fractures

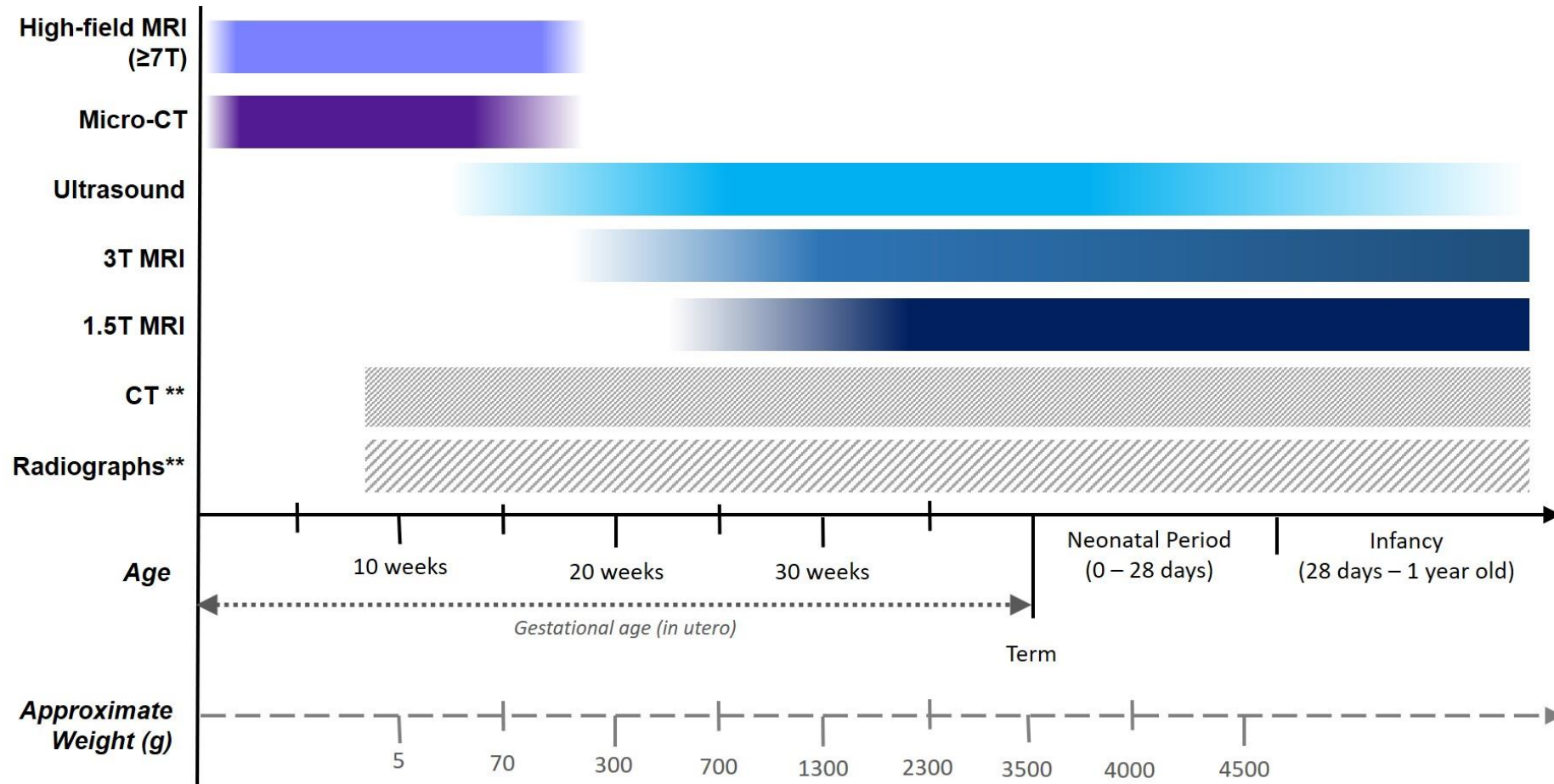
(b) volume rendered micro-CT image showing bent and deformed limbs from multiple fractures

(c) micro-CT to resolution of 80um of the left femur showing detailed bony architecture



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# PM Imaging by gestation



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# Topic 3

## Parental and wider views on less invasive autopsy

Dr Celine Lewis; UCL Great Ormond Street Institute of Child Health, London, UK

*March 19, 2024*



# Parental and Wider Views on Less-Invasive Autopsy

Dr Celine Lewis

UCL GOS Institute of Child Health



March 19, 2024



# Historically low rates of perinatal PM uptake



## UK perinatal & paediatric consent rates

	Stillbirth	Neonatal death
Offer of PM	98%	81%
Uptake of PM	49%	29%

2018: Draper et al. *MBRRACE-UK Perinatal Mortality Surveillance Report: UK Perinatal Deaths for Births from January to December 2016*

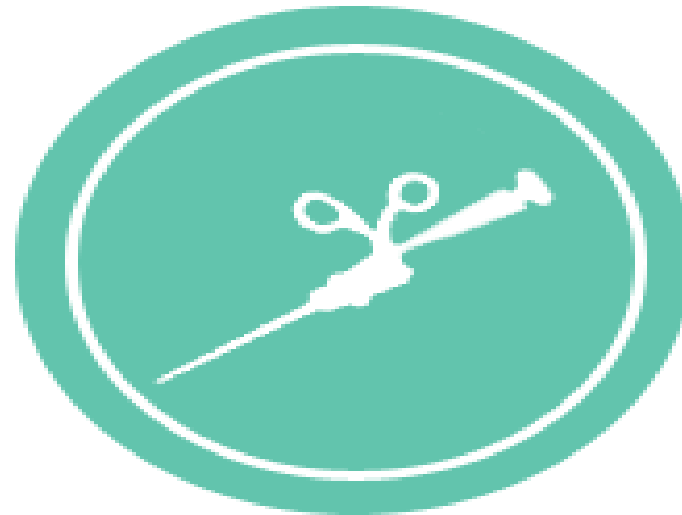


# Study aims

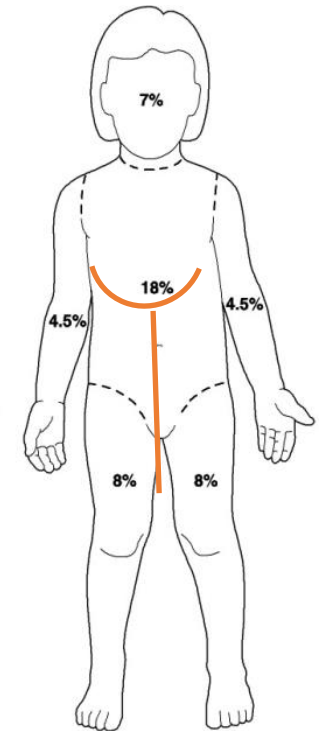
1. Acceptability of different types of PM for bereaved parents
2. Whether less invasive PM is likely to improve uptake rates



**Non-invasive autopsy or NIA**  
imaging only



**Minimally-invasive autopsy or MIA**  
Imaging plus targeted biopsy



**Standard autopsy**  
Organs removed & examined

# Study design

## Survey



**Recruitment:** (June 2016 – Dec 2017)

**Retrospective recruitment:** Online through website/FB page of 4 UK support groups

**Prospective recruitment:** Through 6 antenatal/neonatal units

**Inclusion criteria:** Open to anyone who had experienced pregnancy loss (ether miscarriage, ToP for fetal anomaly, stillbirth) or neonatal or infant death irrespective of whether they were offered PM or accepted PM

# Study design

## Survey



### **Description of standard autopsy**

Would you agree to it? *Yes, No, Not sure*

How acceptable is it? *Very acceptable – not at all acceptable*

*Free-text responses*

### **Description of non-invasive autopsy**

Would you agree to it? *Yes, No, Not sure*

How acceptable is it? *Very acceptable – not at all acceptable*

*Free-text responses*

### **Description of minimally invasive autopsy**

Would you agree to it? *Yes, No, Not sure*

How acceptable is it? *Very acceptable – not at all acceptable*

*Free-text responses*

### **Preference:**

standard, MIA, NIA, none, no strong preference

*Free-text responses*



# Response rate

## Survey



**Total: n=857**  
Retrospective: n=789  
Prospective: 68  
51% free-text comments

# Study design

## Interviews



## Recruitment

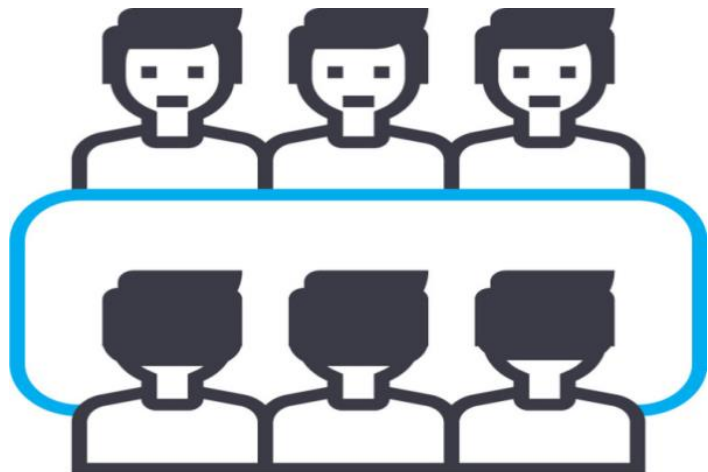
**Sample:** Survey responders who indicated they were willing to take part in a telephone interview

**Variation:** Demographic characteristics, experience of loss, views regarding acceptability of (less invasive) PM

**Uptake:** 20 bereaved parents (56% recruitment rate)

# Study design

## Focus groups with Muslim and Jewish participants



## Recruitment (Sept 2016 – May 2017)

### Sites:

Muslim Community Centres in London and Midlands  
Synagogue in N London  
Rabbi from Ultra-Orthodox community in N London

### Inclusion criteria:

Parents

Males and females

Childbearing age (18-40)

English, Bengali or Urdu speaking (translator present)

### 10 focus groups

60 Muslim participants

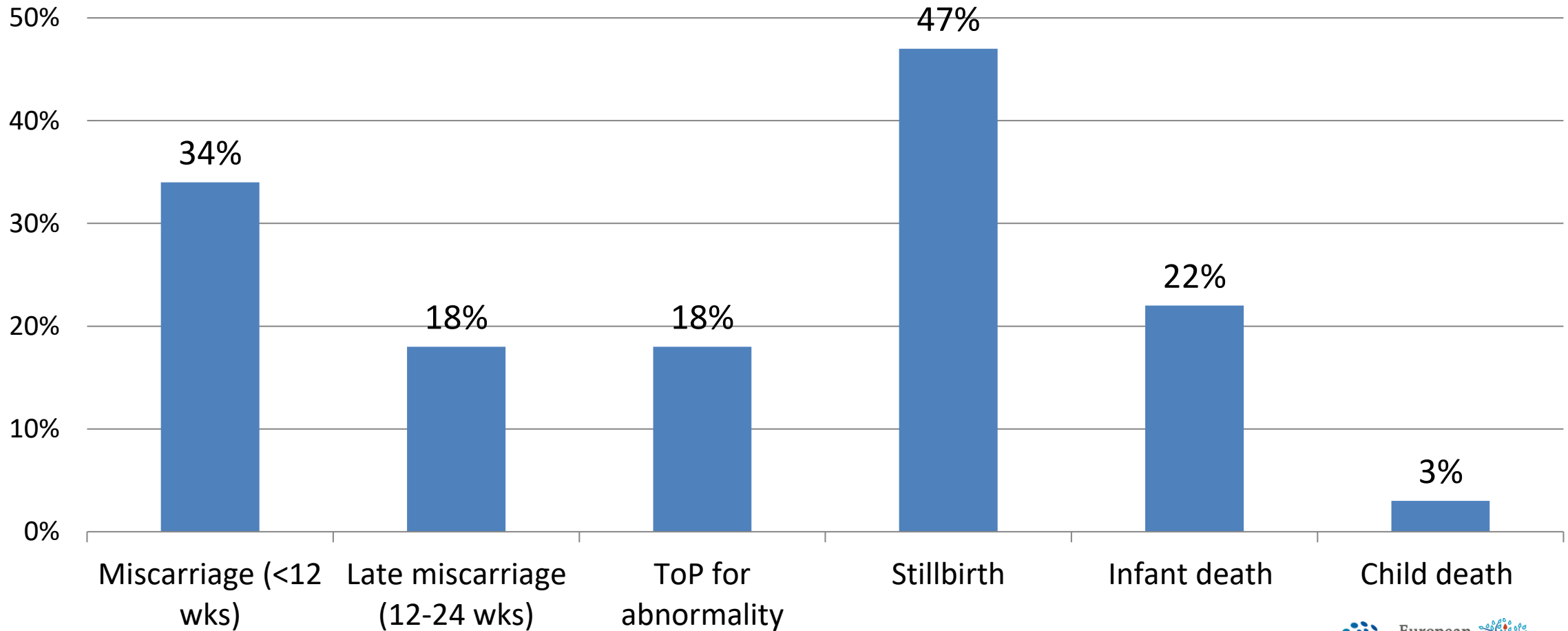
16 Jewish participants



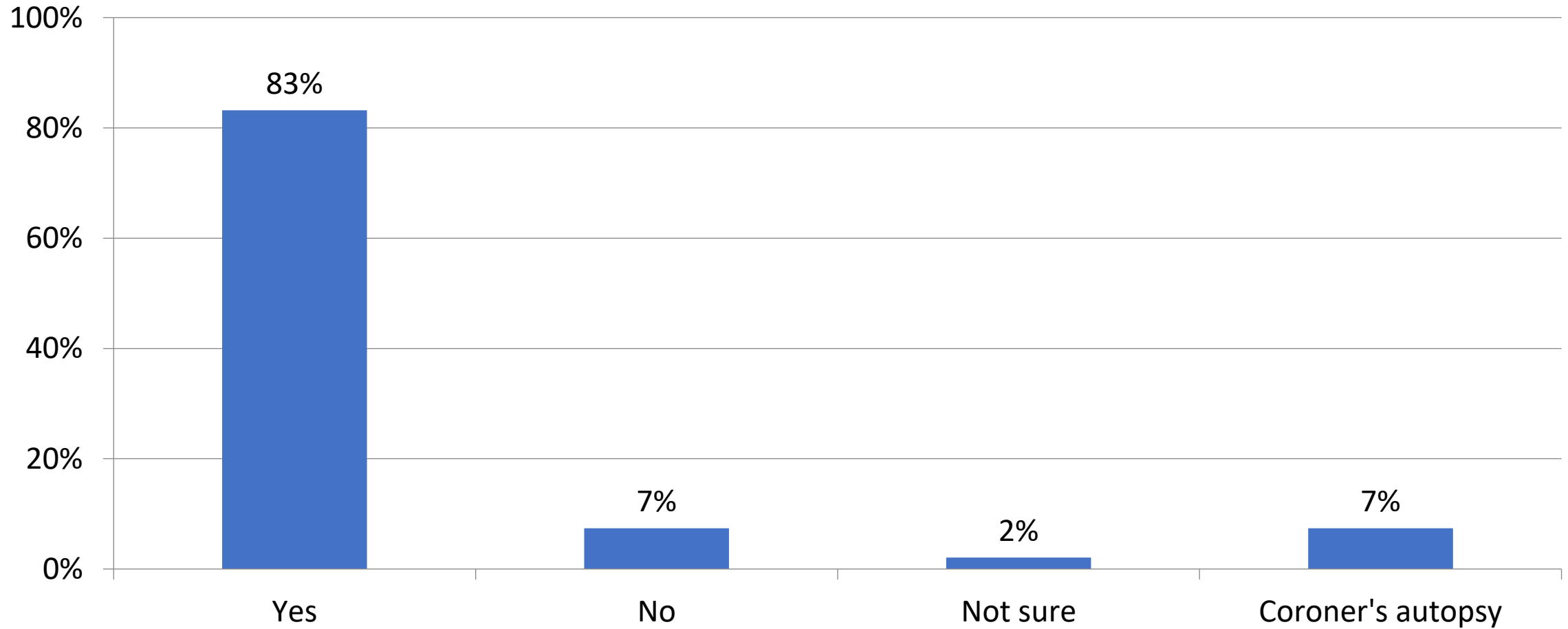
# Study Results

# Survey responders experience of loss

Mirrors proportions from recent national data



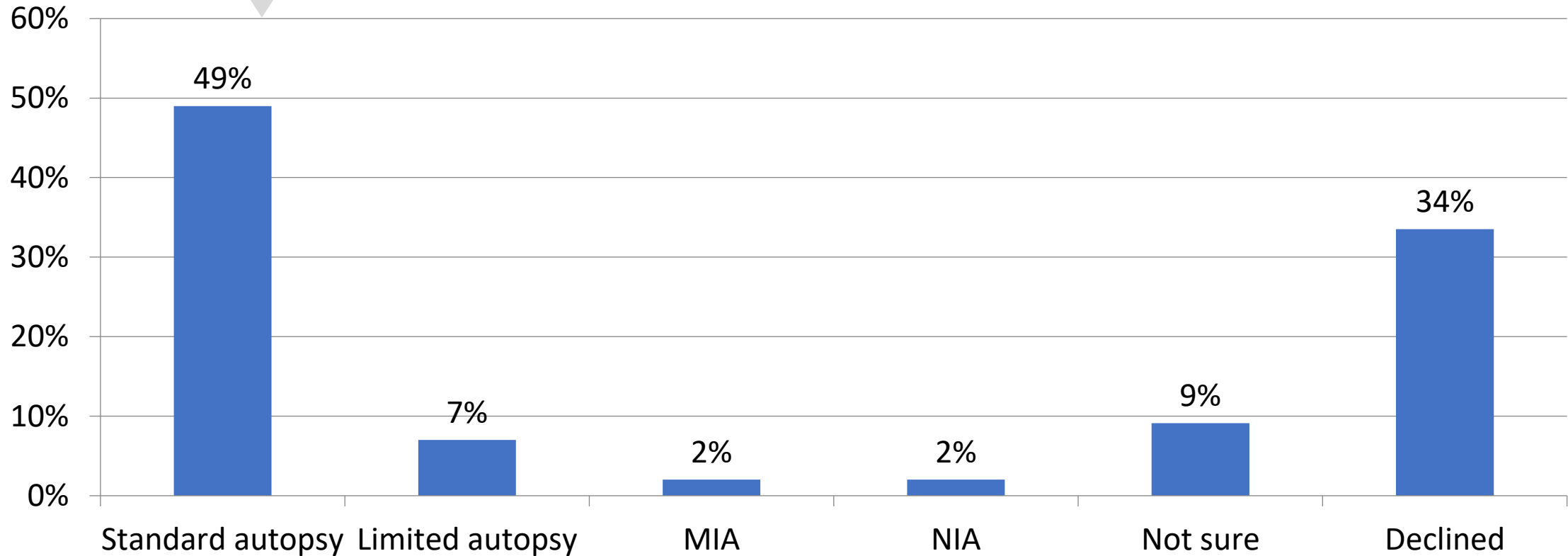
# Were you offered A PM?



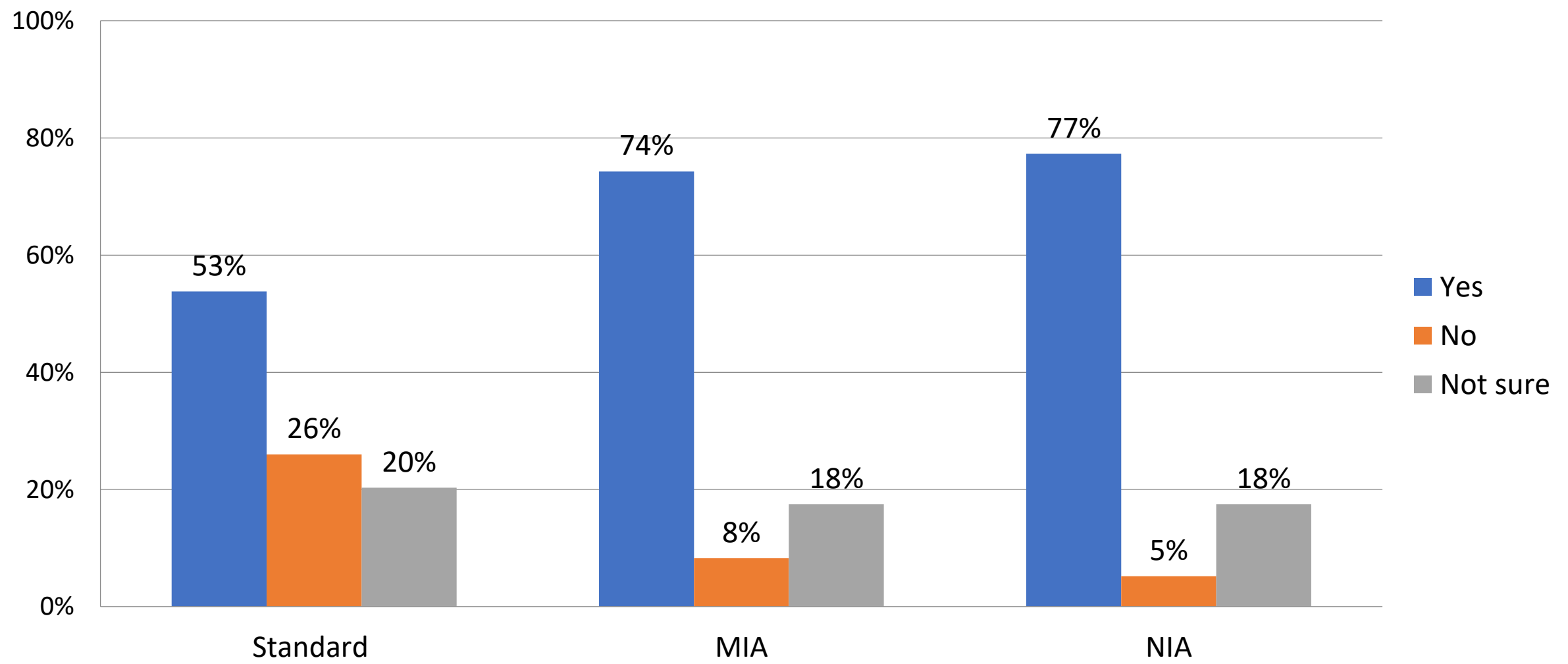


# Did you consent to a PM?

Mirrors proportions from recent national data



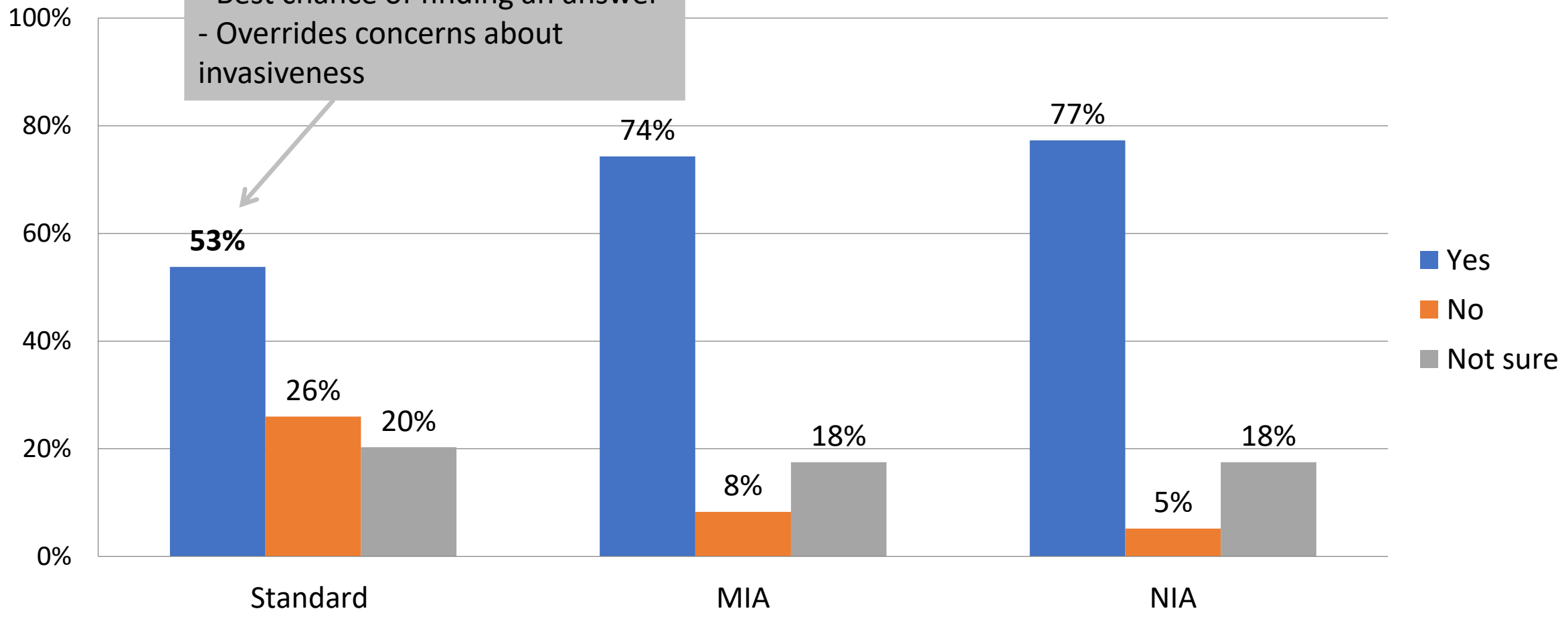
# Would you consent to the following types of PM?



Most invasive ← → Least invasive

# Would you consent to the following types of PM?

**Thoroughness**  
- Best chance of finding an answer  
- Overrides concerns about invasiveness



Most invasive ←————→ Least invasive



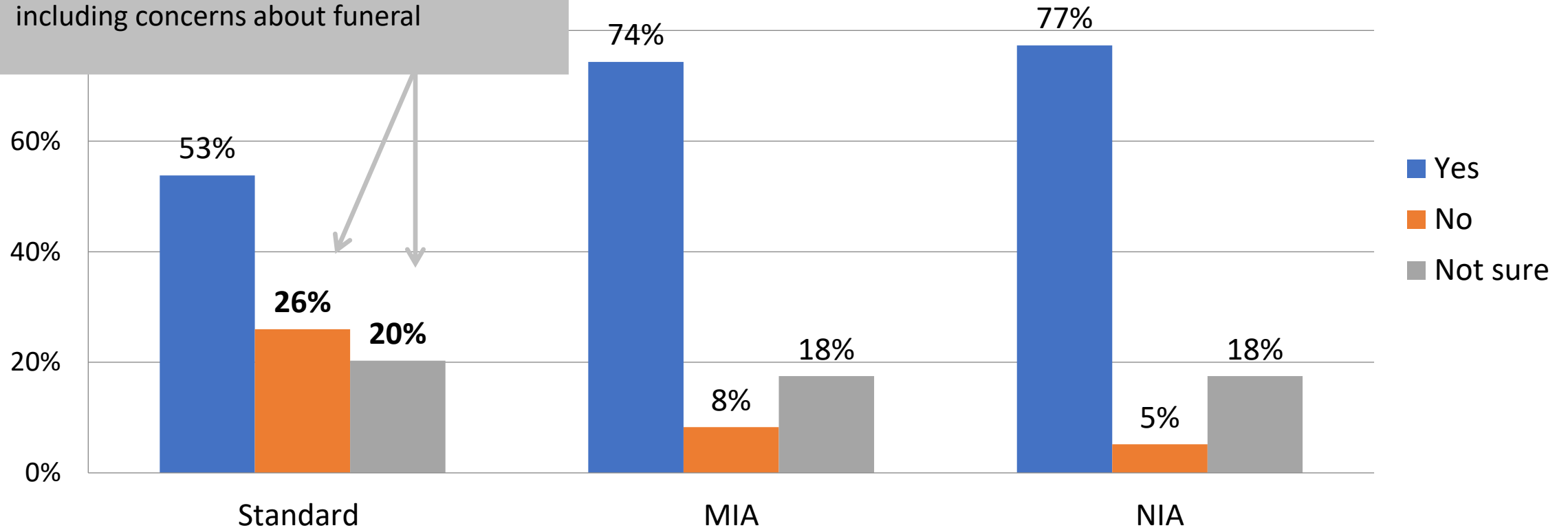
*“I understood that the procedure was invasive on my daughter's body however **it was extremely important for us to understand why she died** after a seemingly healthy pregnancy. It was difficult to think of what would happen to her body however **it was worth this distress to get an answer.**”*

Sands, experienced stillbirth, consented to standard autopsy

# Would you consent to the following types of PM?

## Invasive as problematic

- Prohibited for parents from Muslim/Jewish faith
- 'Cutting' an additional trauma
- Disturbing the baby's appearance including concerns about funeral



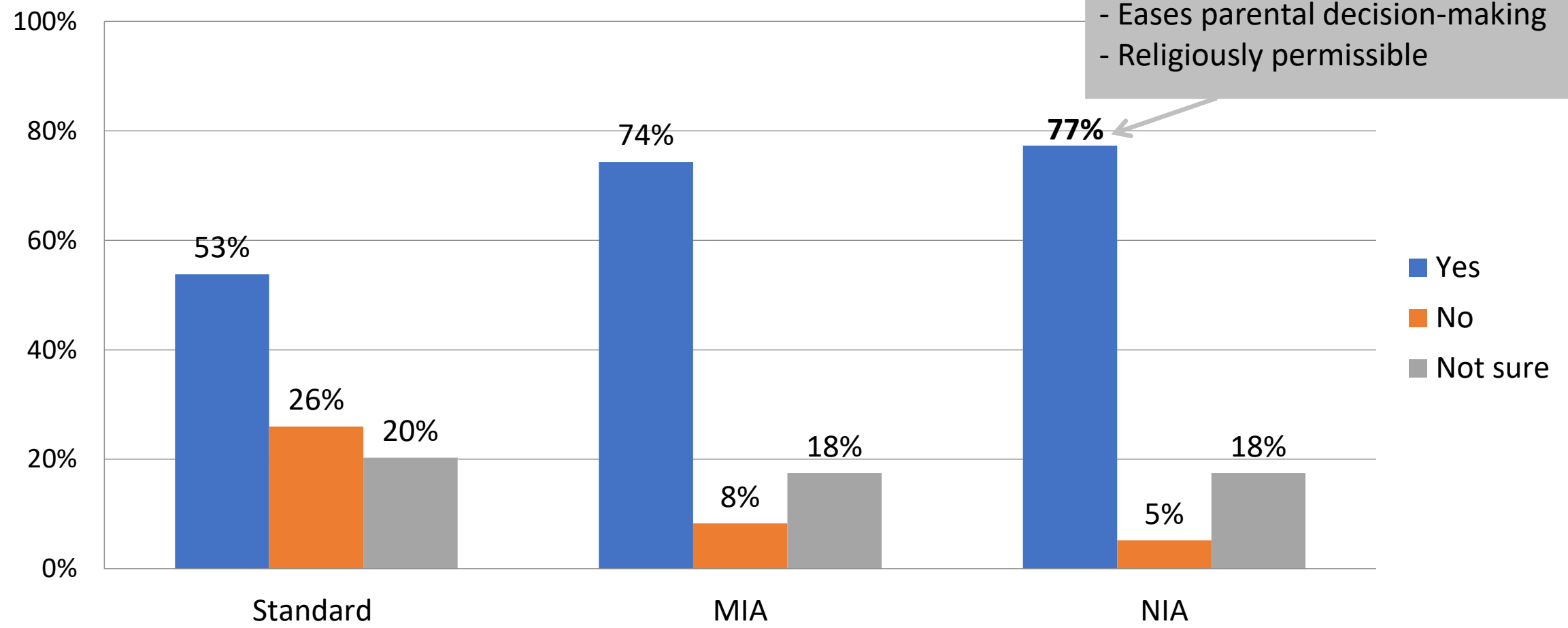
Most invasive

Least invasive

# Would you consent to the following types of PM?

**Removes the 'burden' of cutting**

- Lets child rest in peace
- Eases parental decision-making
- Religiously permissible



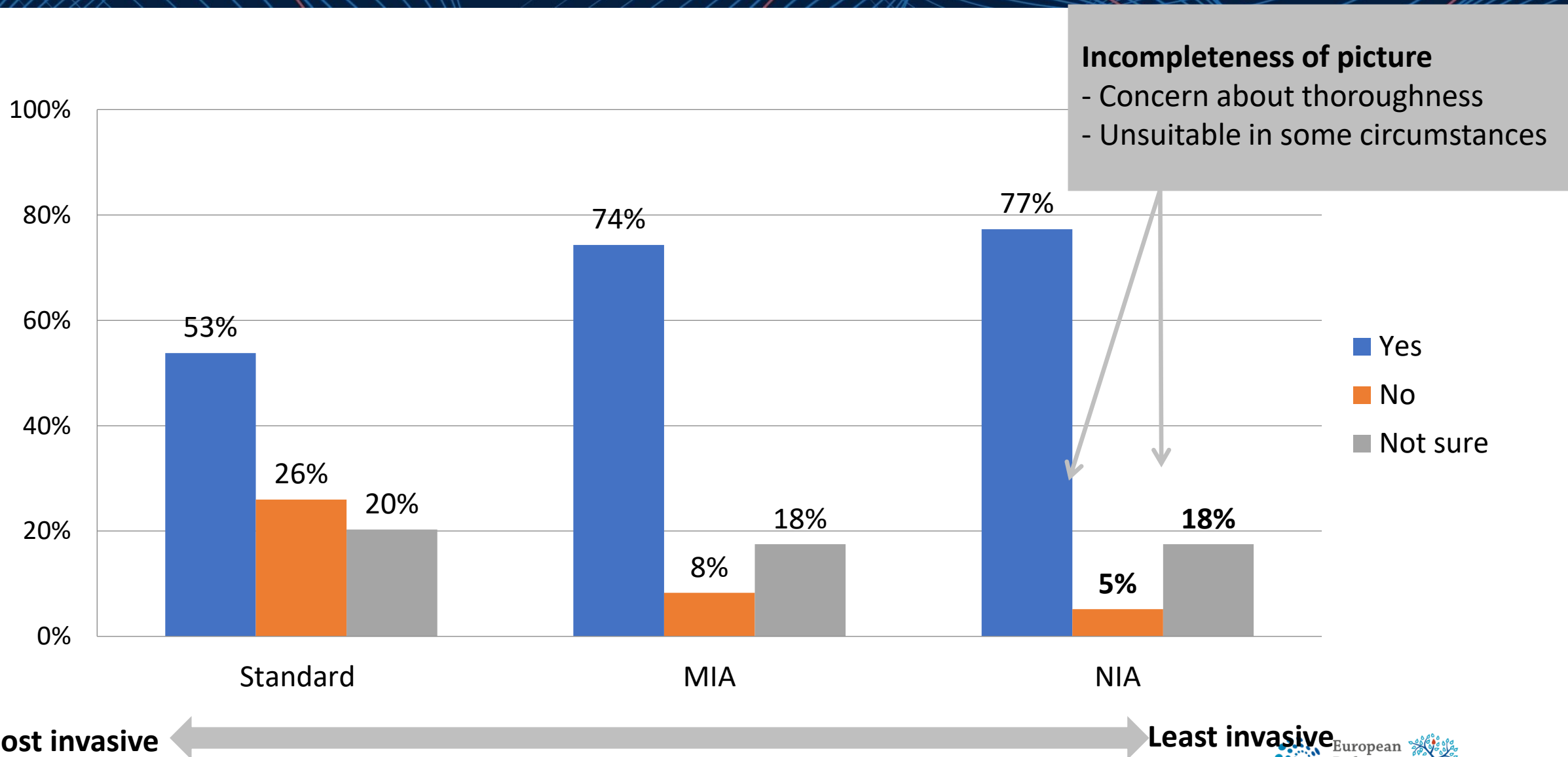
Most invasive ←—————→ Least invasive



*““My initial thought would be, **absolutely 100% I would go for a non-invasive [autopsy]**, without any hesitation... you’re keeping the body intact.”*

Jewish Orthodox parent

# Would you consent to the following types of PM?



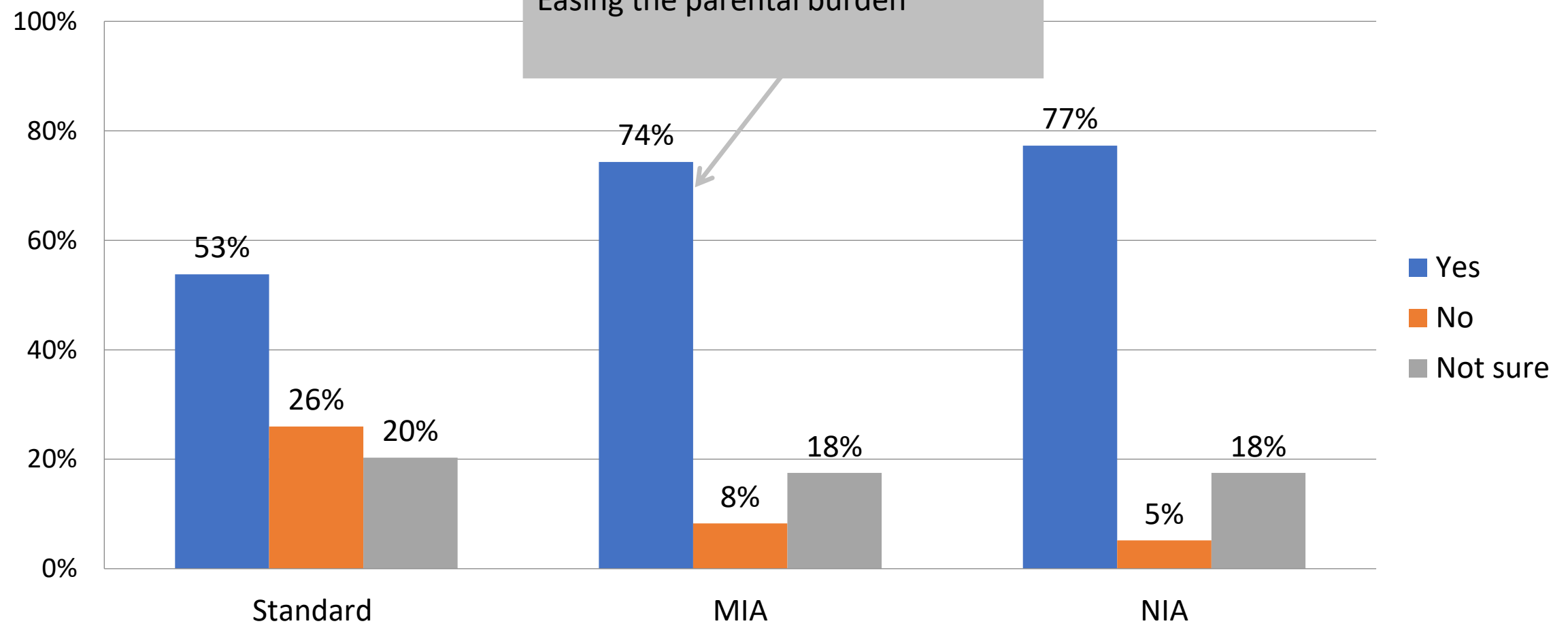
*“A less invasive procedure would have appealed to me at the time. However I felt the need to know everything I possibly could about what was wrong with my baby, so would struggle with the idea that **something might be missed that could have been picked up from a different method.**”*

ARC, ToP and stillbirth, consented to standard autopsy



# Would you consent to the following types of PM?

**Best of both worlds**  
Information with 'desecration'  
Easing the parental burden



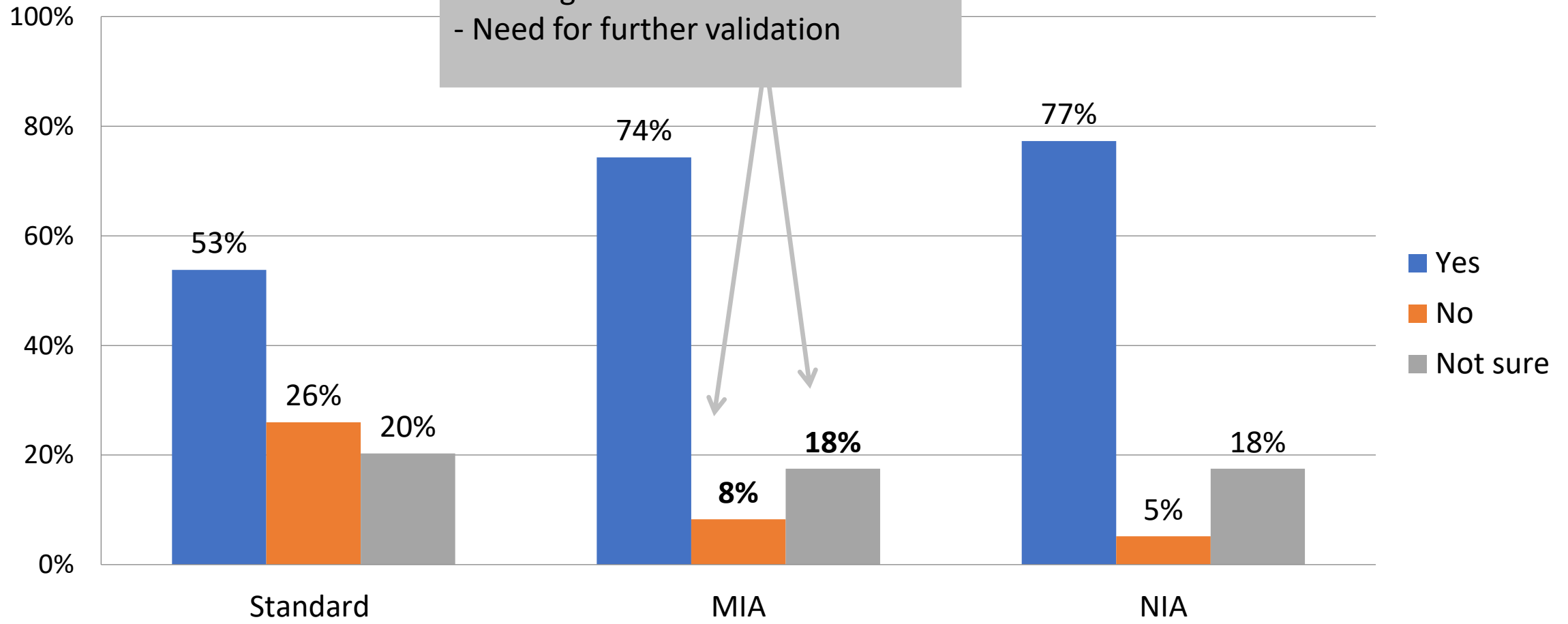
Most invasive ←————→ Least invasive

*“This seems like the **best of both worlds** - you can physically see organs, and take samples, whilst at the same time being minimally invasive and less distressing for parents to agree to”*

ARC, ToP for abnormality, consented to standard autopsy

# Would you consent to the following types of PM?

**1. Comparability to complete PM**  
- Leaving 'no stone unturned'  
- Need for further validation



Most invasive ←————→ Least invasive

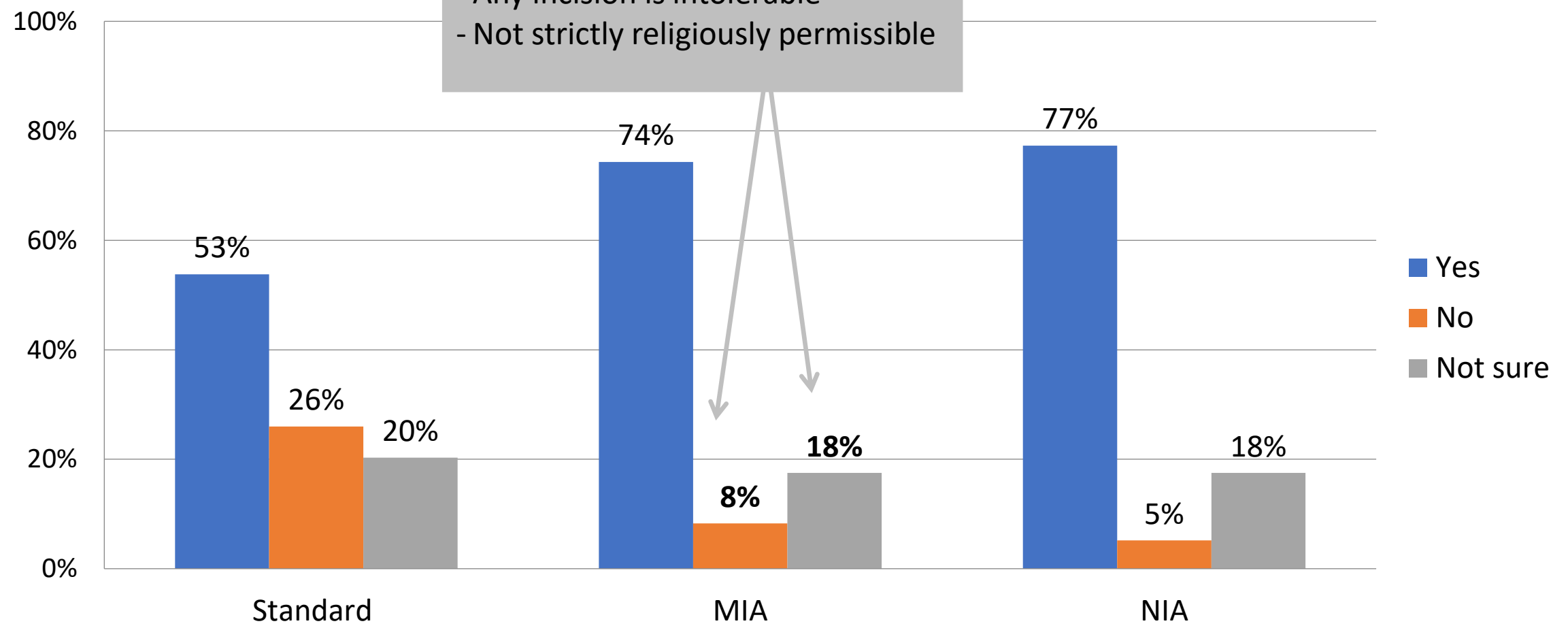
*“As it's new **would it find the answers parents need from an autopsy? If results came back inconclusive would parents then be left wondering if the answer was in a full autopsy?**”*

Sands, stillbirth, declined autopsy



# Would you consent to the following types of PM?

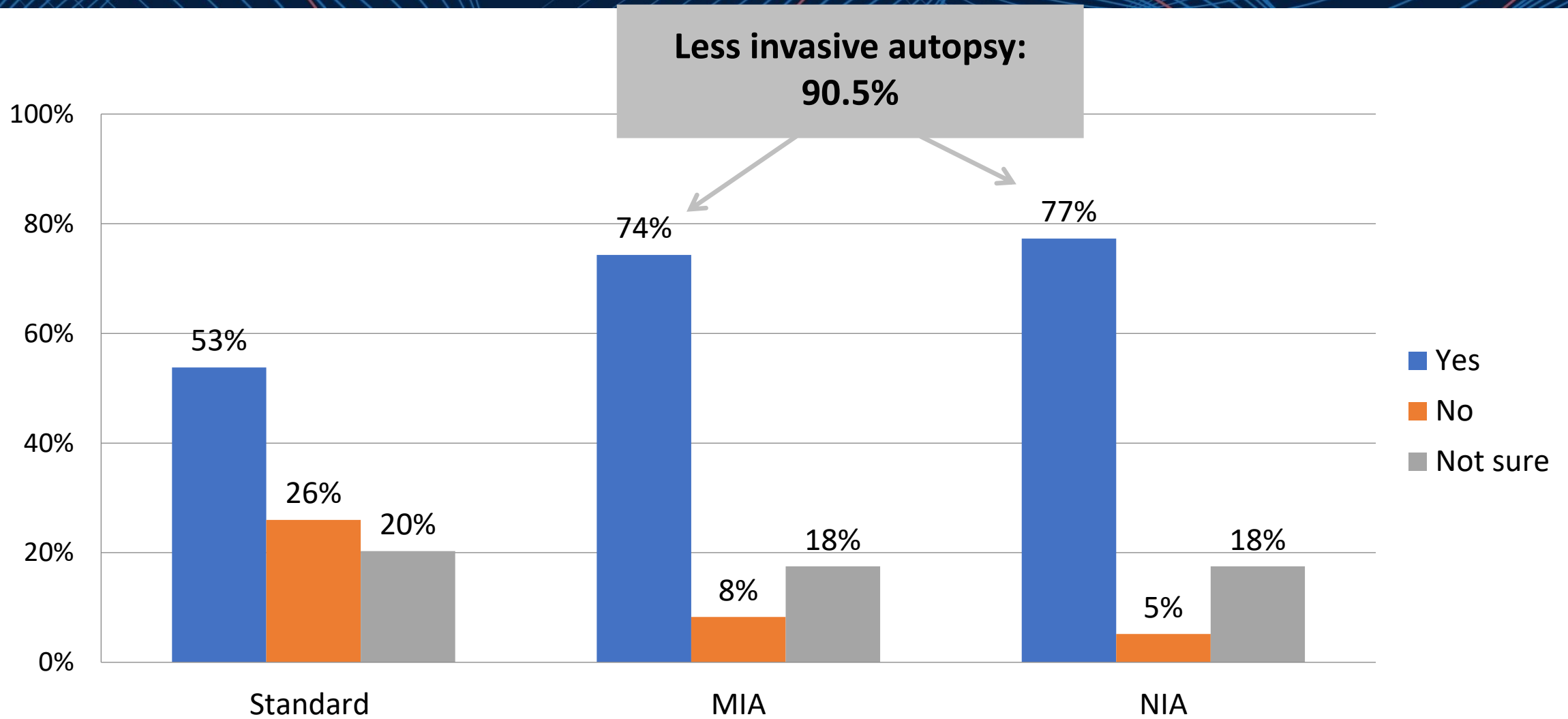
**2. Still invasive**  
- Any incision is intolerable  
- Not strictly religiously permissible



Most invasive ←————→ Least invasive

*“In the Islamic perspective, it’s **still forbidden** because of the cutting, whether it’s a big cut or a small cut.”* (FG7, Mixed Ethnicity, Muslim, Women).

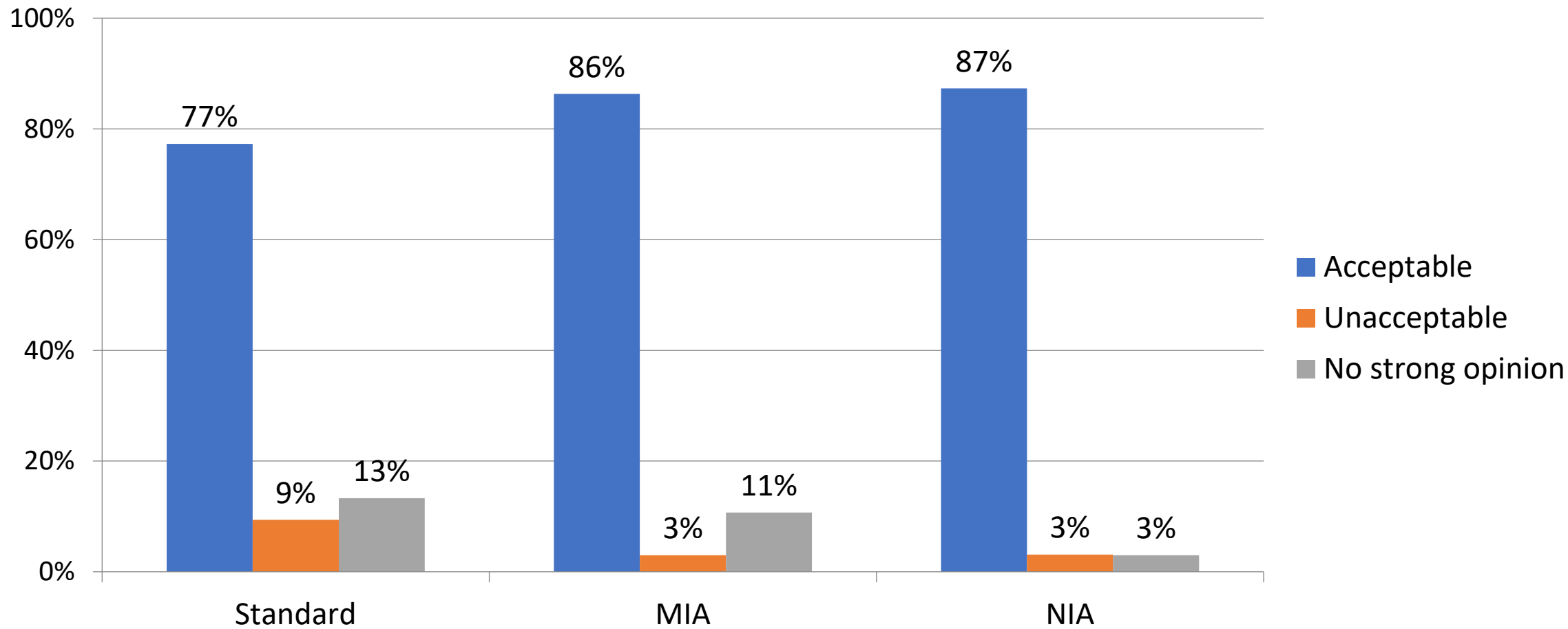
# Would you consent to the following types of PM?



Most invasive ←

→ Least invasive

# How acceptable are the different types of PM?

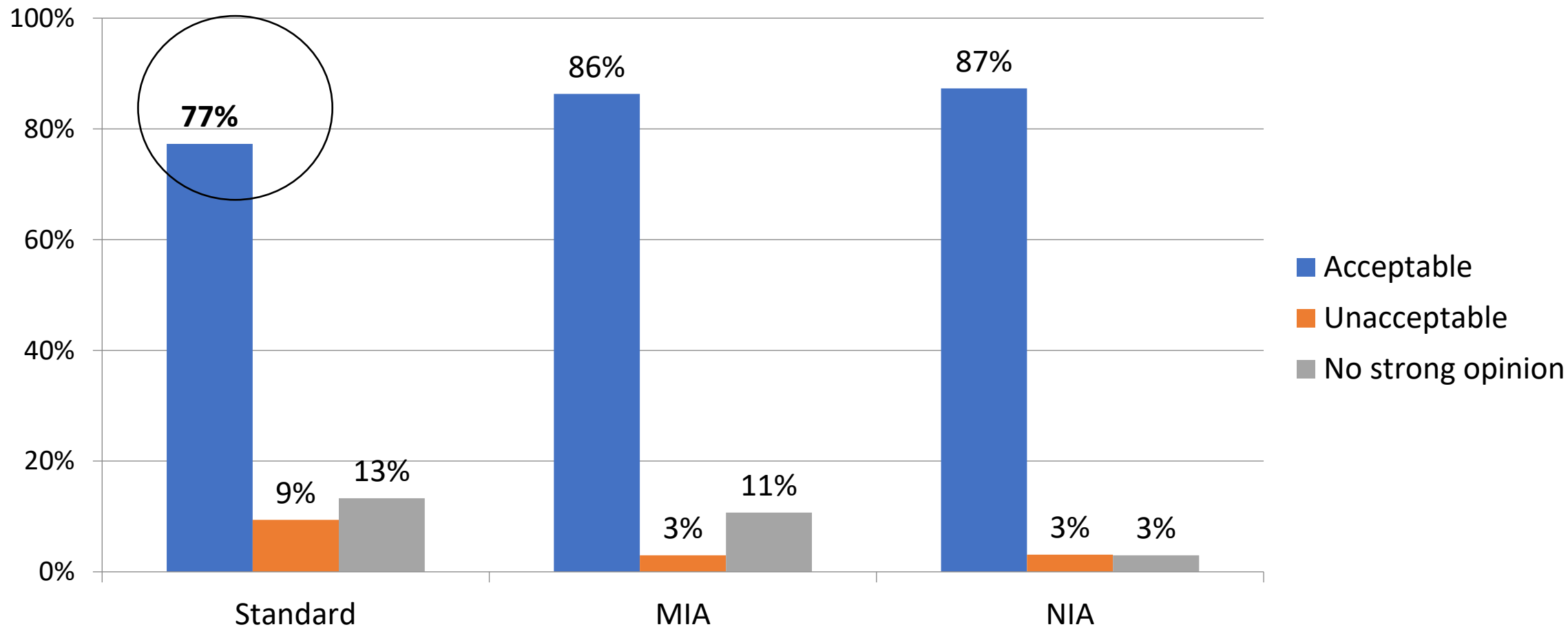


Most invasive ←

→ Least invasive

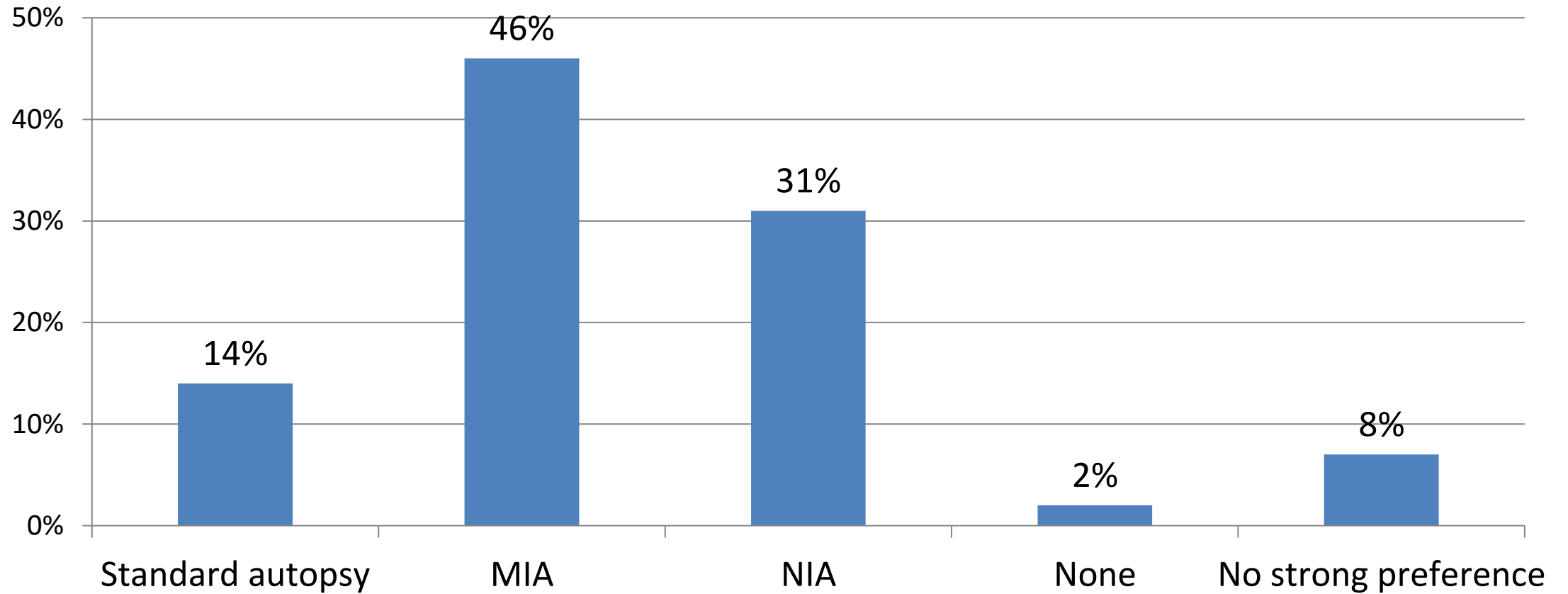


# How acceptable are the different types of PM?

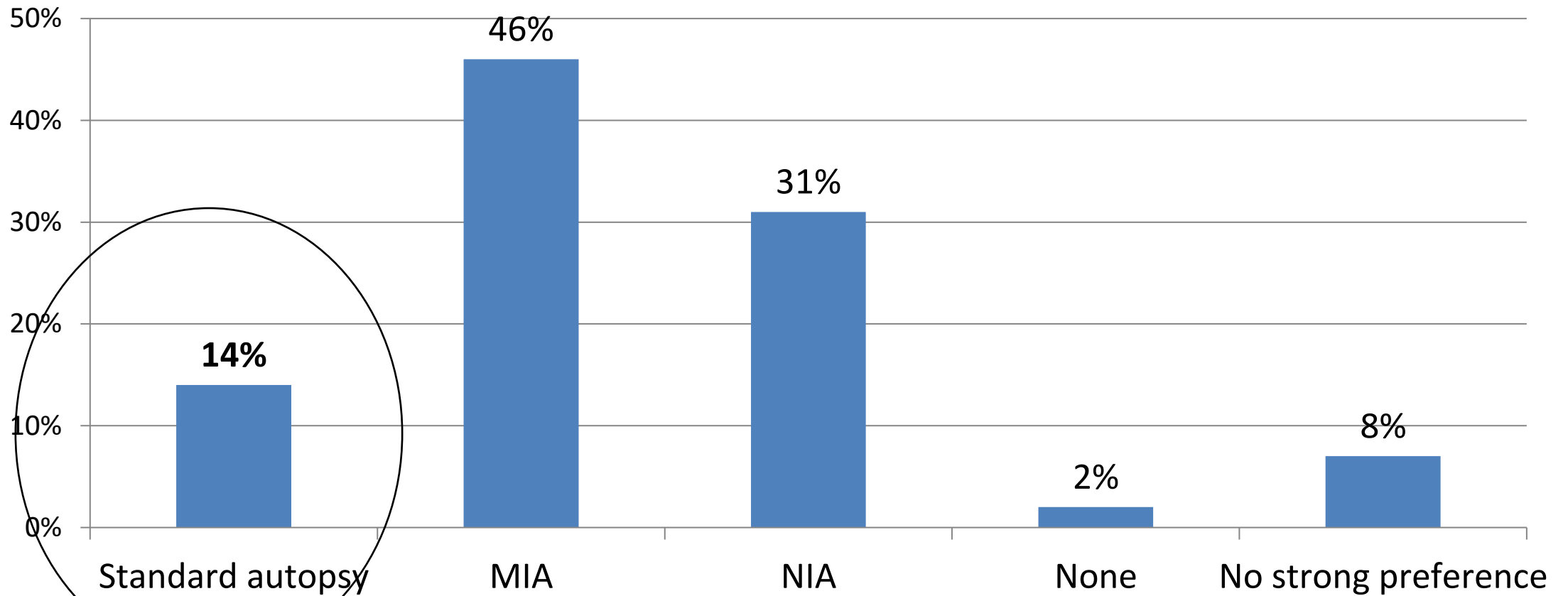


Most invasive ←—————→ Least invasive

# If you had the option, which would you choose?



# If you had the option, which would you choose?



# Summary

- Less invasive methods would open up **opportunities for people who would otherwise decline including religious groups**
- **Likely to be a significant increase in uptake** if personalised and more acceptable approaches were routinely available
- Some people will **still prefer standard autopsy** – important to offer choice
- Further work to **assess effectiveness of LIA in specific circumstances** to guide counselling
- Need for **economic/implementation evaluations**
- **REQUIRES SIGNIFICANT BUY-IN FROM HOSPITAL MANAGERS, PATHOLOGISTS & RADIOLOGISTS IN TERMS OF TRAINING, EQUIPMENT & WILL TO CHANGE**



# Publications

## Health professionals' and coroners' views on less invasive perinatal and paediatric autopsy: a qualitative study

Celine Lewis,<sup>1,2</sup> Melissa Hill,<sup>1,2</sup> Owen J Arthurs,<sup>2,3</sup> John C Hutchinson,<sup>2,4</sup> Lyn S Chitty,<sup>1,2</sup> Neil Sebire<sup>2,4</sup>

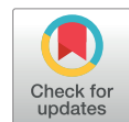


RESEARCH ARTICLE

“We might get a lot more families who will agree”: Muslim and Jewish perspectives on less invasive perinatal and paediatric autopsy

Celine Lewis<sup>1,2\*</sup>, Zahira Latif<sup>3,4</sup>, Melissa Hill<sup>1,2</sup>, Megan Riddington<sup>5</sup>, Monica Lakhanpaul<sup>6,7</sup>, Owen J. Arthurs<sup>6,8</sup>, John C. Hutchinson<sup>9</sup>, Lyn S. Chitty<sup>1,2</sup>, Neil J. Sebire<sup>9</sup>

1 North East Thames Regional Genetics Service, Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom, 2 Genetics and Genomic Medicine, The UCL Great Ormond Street Institute of Child Health, London, United Kingdom, 3 College of Medical and Dental Sciences, University of Birmingham, Birmingham, United Kingdom, 4 School of Medicine, Faculty of Medicine and Health Sciences, University of Nottingham, Nottingham, United Kingdom, 5 Department of Psychological Services, Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom, 6 Faculty of Population Health Sciences, UCL Great Ormond Street Institute of Child Health, London, United Kingdom, 7 Community Paediatrics, Whittington Health NHS Trust, London, United Kingdom, 8 Department of Radiology, Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom, 9 Department of Histopathology, Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom



Received: 5 June 2019 | Revised: 20 August 2019 | Accepted: 29 September 2019

DOI: 10.1002/pd.5575

ORIGINAL ARTICLE

WILEY PRENATAL DIAGNOSIS

“The communication and support from the health professional is incredibly important”: A qualitative study exploring the processes and practices that support parental decision-making about postmortem examination

Celine Lewis<sup>1,2</sup> | Megan Riddington<sup>3</sup> | Melissa Hill<sup>1,2</sup> | Charlotte Bevan<sup>4</sup> | Jane Fisher<sup>5</sup> | Lucy Lyas<sup>6</sup> | Ann Chalmers<sup>7</sup> | Owen J. Arthurs<sup>8</sup> | John C. Hutchinson<sup>9</sup> | Lyn S. Chitty<sup>1,2</sup> | Neil Sebire<sup>9,10</sup>



General obstetrics | Open Access |

Availability of less invasive prenatal, perinatal and paediatric autopsy will improve uptake rates: a mixed-methods study with bereaved parents

C Lewis, M Riddington, M Hill, OJ Arthurs, JC Hutchinson, LS Chitty, C Bevan, J Fisher, J Ward, NJ Sebire

First published: 21 December 2018 | <https://doi.org/10.1111/1471-0528.15591> | Citations: 22

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Lyn Chitty

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## **Participating Sites**

**Basildon & Thurrock NHS  
Foundation Trust**

**Southend Hospital NHS Foundation  
Trust**

**Homerton University Hospital**

**The University Hospitals of  
Leicester NHS Trust**

**Newham University Hospital**

**University College London Hospital  
NHS Foundation Trust**

## **Participating Support Groups**

**Antenatal Results and Choices**

**Sands – Stillbirth and neonatal death  
charity**

**The Lullaby Trust**

**Child Bereavement UK**

*March 19, 2024*

# ESHG-Young



ESHG-Y Committee mission is to represent and support the young European geneticists by developing strategies and programs that aim for a better education.

<https://www.eshg.org/eshgy>



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[eshgy@eshg.org](mailto:eshgy@eshg.org)



ESHG-Y: European Society of Human Genetics - Young



Young (Human) Geneticists Network

March 19, 2024



# Thank you for your attention !

- Register to the Web site to get NewsLetter and calls for collab !

## Satisfaction Survey

<https://forms.office.com/e/SZre6cSaDG>

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European  
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complex diseases

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Malformations (ERN ITHACA)



March 19, 2024