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EUROPEAN REFERENCE NETWORK
ON RARE BONE DISEASES

Clinical decision-making in Rare Bone Diseases

A survey among EPOS and ERN-BOND members (September 2025)

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This presentation is generated within the European Reference Network on rare BONe Diseases which is partly co-funded by the European Union (Specific Grant Agreement no. 101157116). The contents of this presentation represents the views of the presenter(s) and is their sole responsibility; and do not necessarily reflect the opinion of the European Union.

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Introduction

Rare bone diseases

- Rare disease prevalence (</=1/2000)
- 771 Rare bone disorders
- Key challenges:
  - Clinical outcomes: highly variable
  - Delayed diagnosis
  - Limited research on pathophysiology
  - Limited standardized frameworks
  - Extensive multidisciplinary needs

Image: www.jnj.com

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Aims

Survey on Clinical Decision Making in Rare Bone Diseases

- European Pediatric Orthopaedic Soceity (EPOS)
- European Reference Network for Rare Bone Disorders (ERN BOND)

Objectives of the survey

- Describe current clinical practices in RBDs
- Identify unmet needs
- Outline minimum standards for patient assessment

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Methods

Structure

-Online survey (Google Forms)

-35-items:
(Multiple-choice, Likert scale, Open text)

Participants

Distributed to

EPOS & ERN-BOND members

(Sept 2025)

Requirements

Experience with RBD

(clinic, laboratory, research, regulatory aspects)

Ethical considerations

-Anonymous survey

-Completion and submission implied consent for participation

Data analysis

-Descriptive statistics

-Thematic analysis

Dissemination

-Report through EPOS/ERN-Bond channels

-Peer-reviewed article

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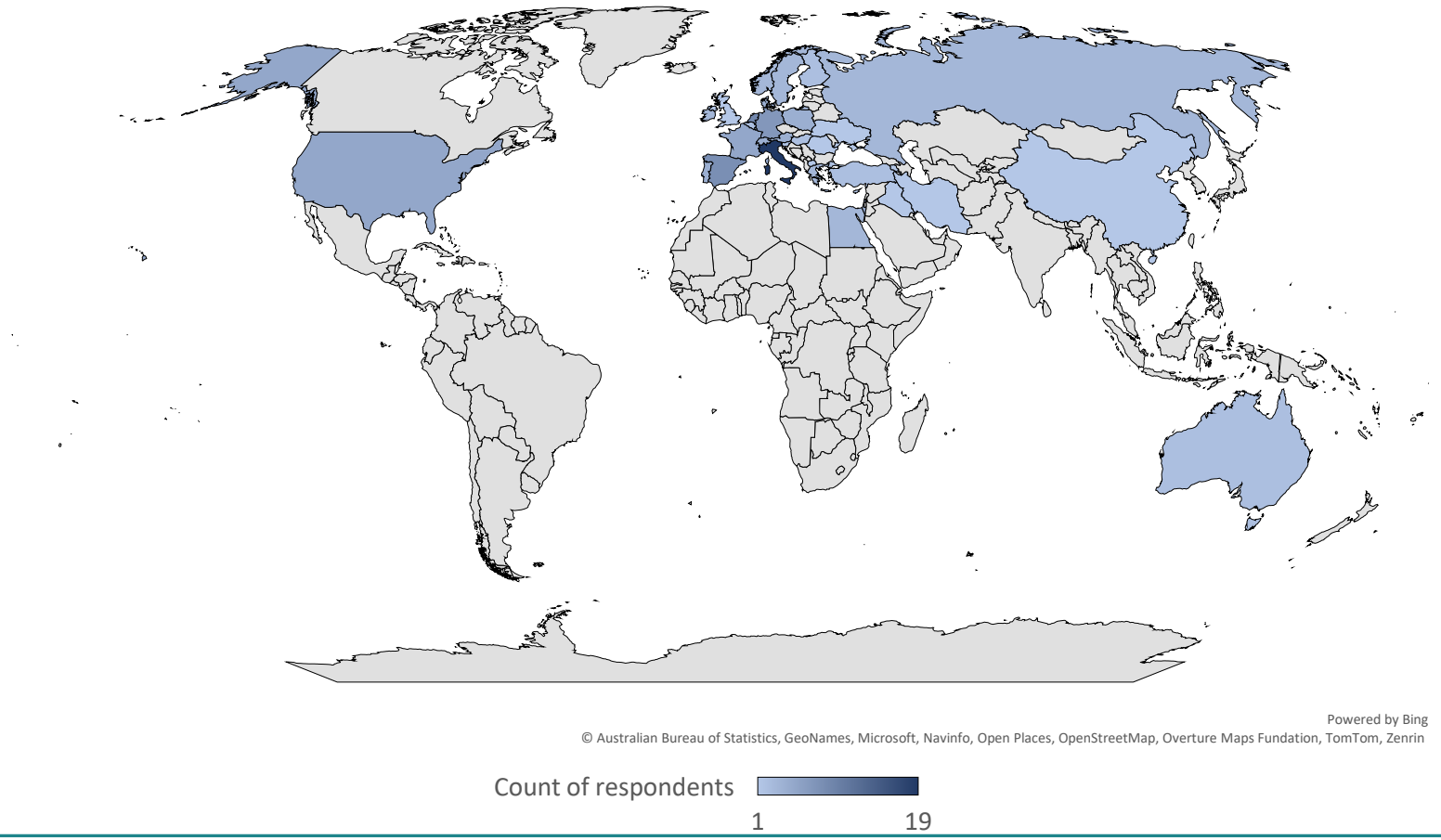
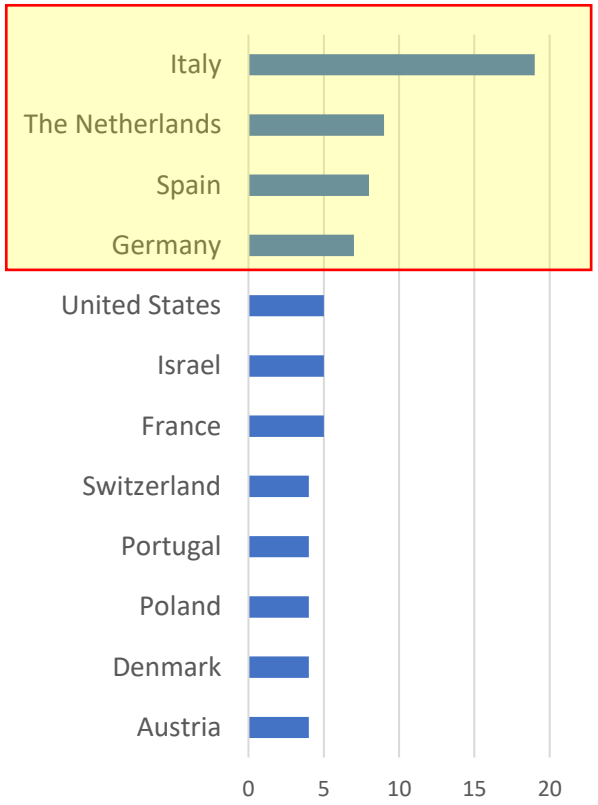
Respondent Profile

- 119 respondents from 35 countries completed the survey





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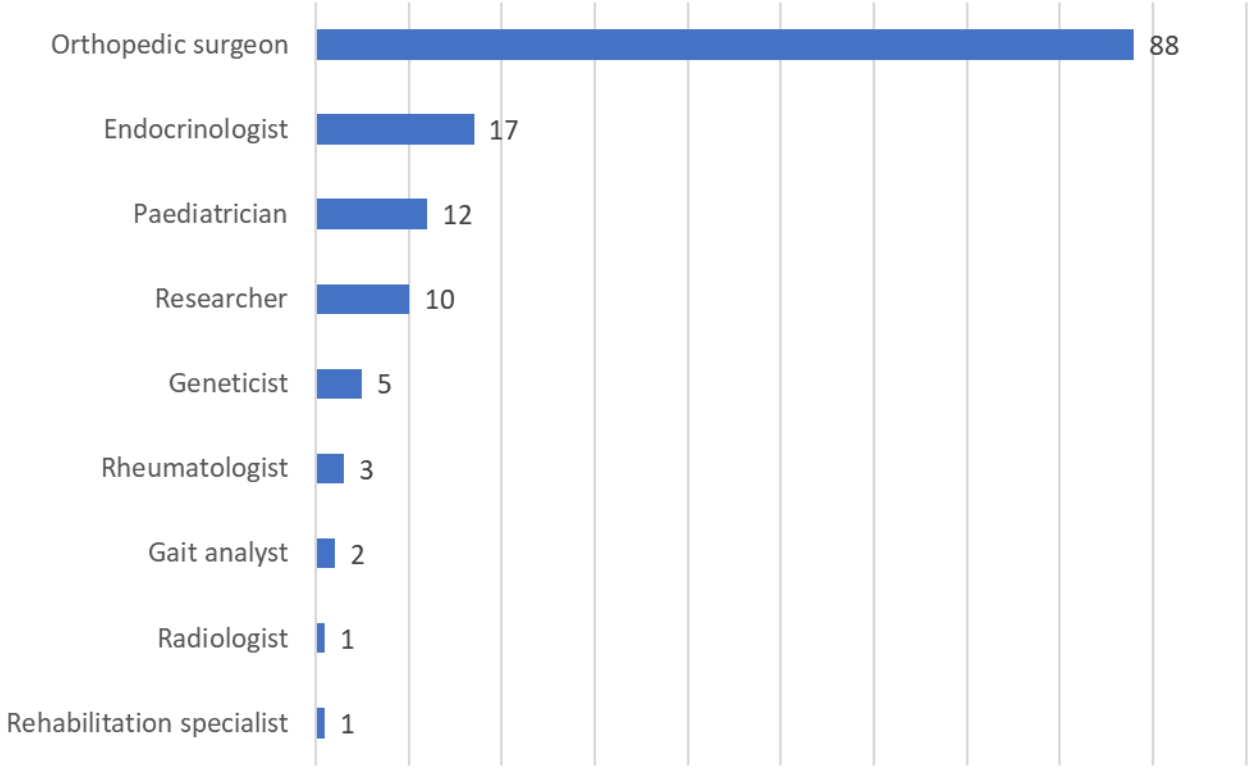
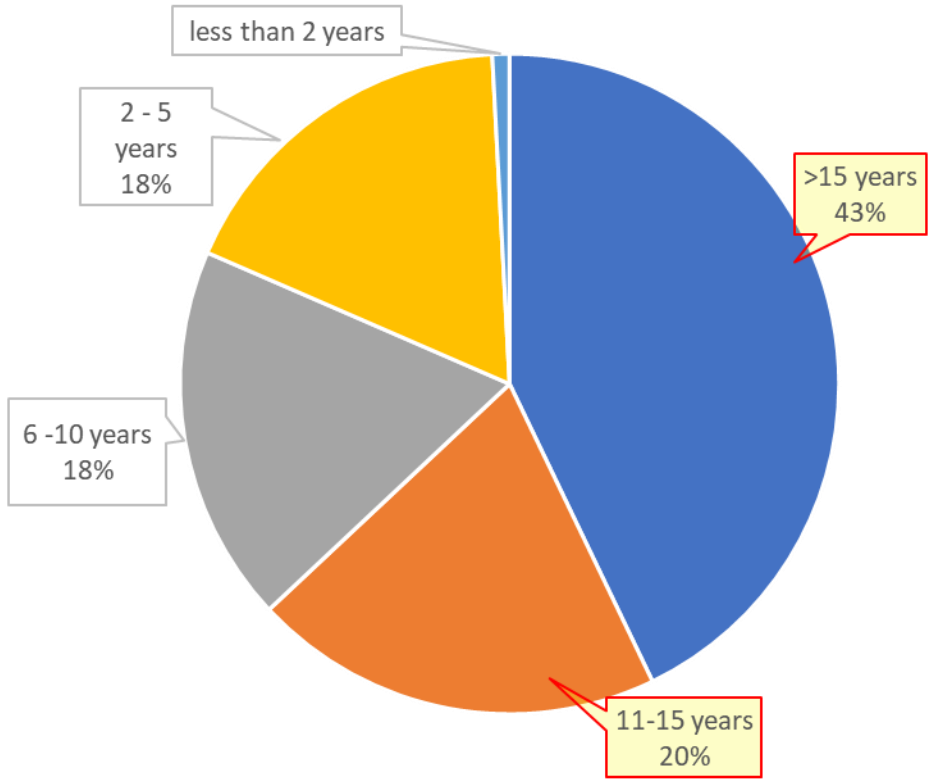
Respondent Profile

Profession



Years of RBD experience



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RBD types

- Most manage common RBDs (osteogenesis imperfecta, fibrous dysplasia, achondroplasia, and multiple osteochondromas)
- Few have experience with ultra-rare conditions (FOP)

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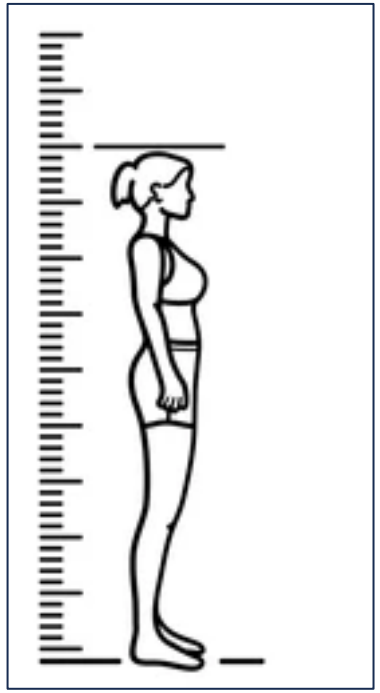
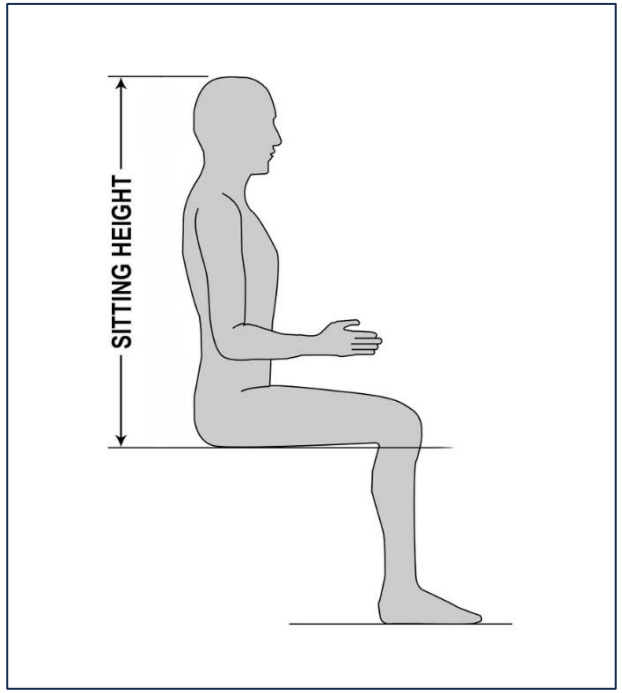
Clinical practices

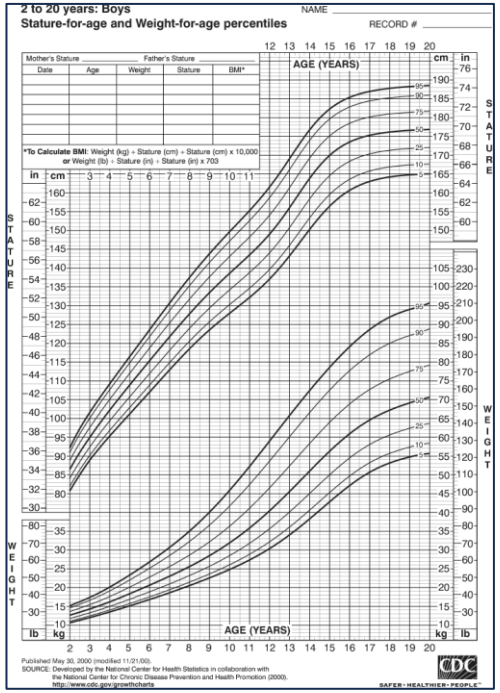
Similar practice in use anthropometric and growth parameters

- Height
- Weight
- Sitting height
- Growth chart
- Body proportions
- BMI

	Height	Sitting height	Growth charts
Used:	100%	79%	89%







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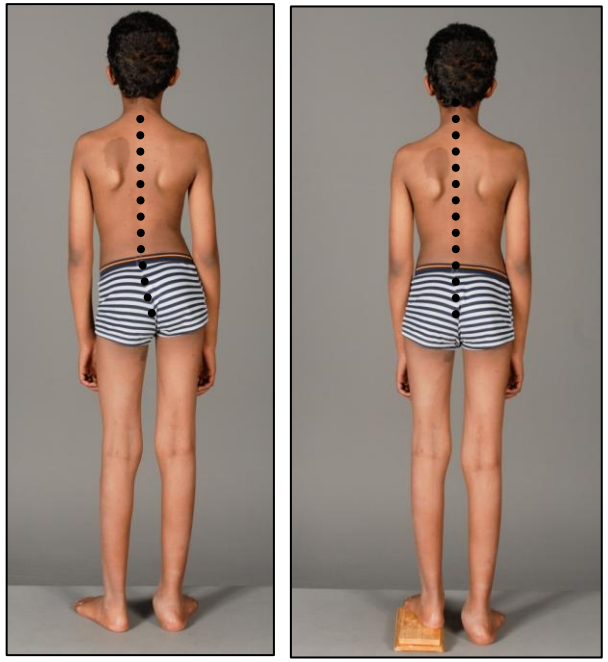
Clinical practices

Similar practice in use posture and alignment in standing position and observation of gait

- Pelvic tilt, LLD, shoulder height, spine



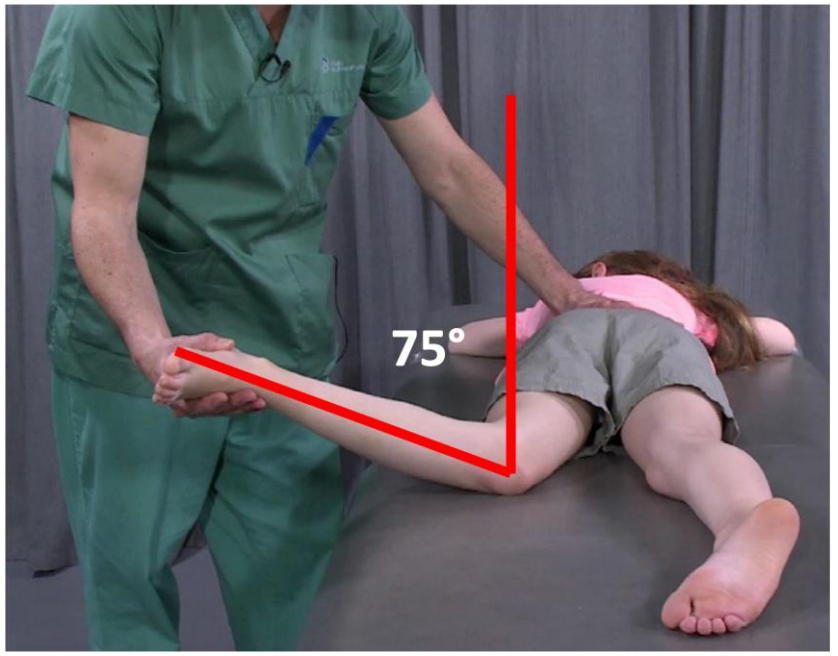
Used: > 80%

- Limb alignment

- Observation of gait

> 90%

- Range of motion testing



95%

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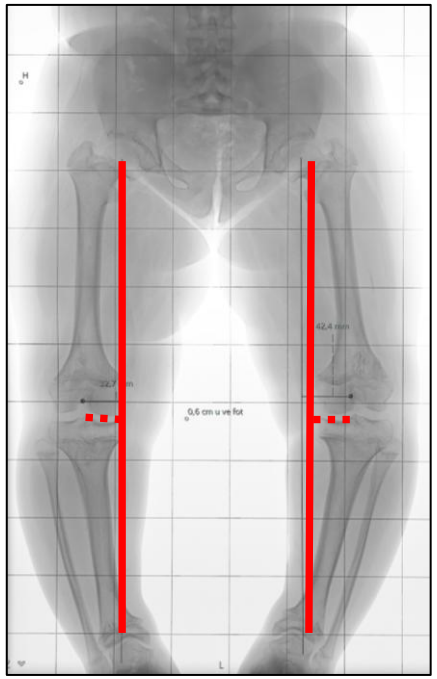
Clinical practices

Variable use of Radiological parameters

- Segmental radiographs

Used: 100%

- Long standing radiographs

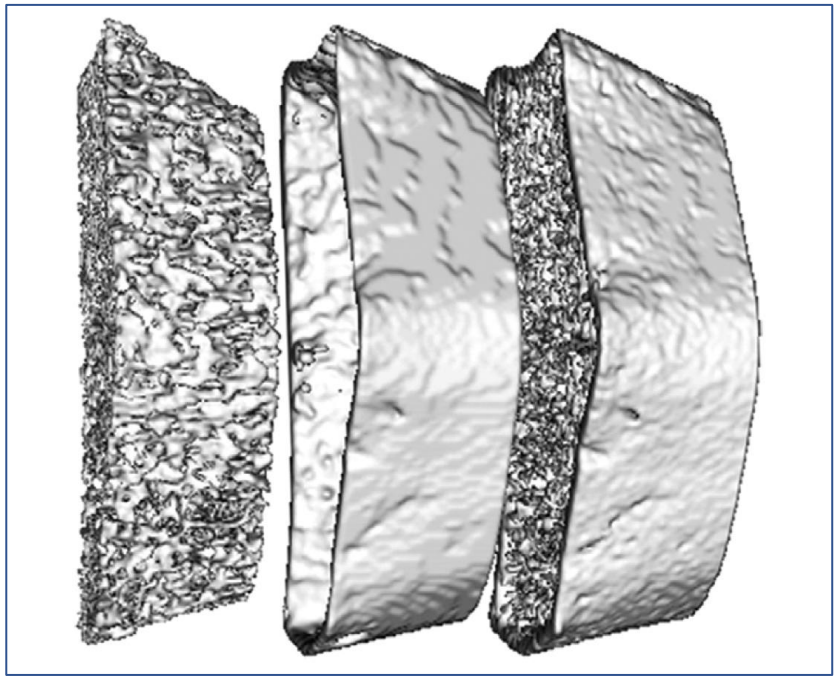


96%

- Weight bearing CT

31%

- High resolution imaging (HR-pQCT)



30%

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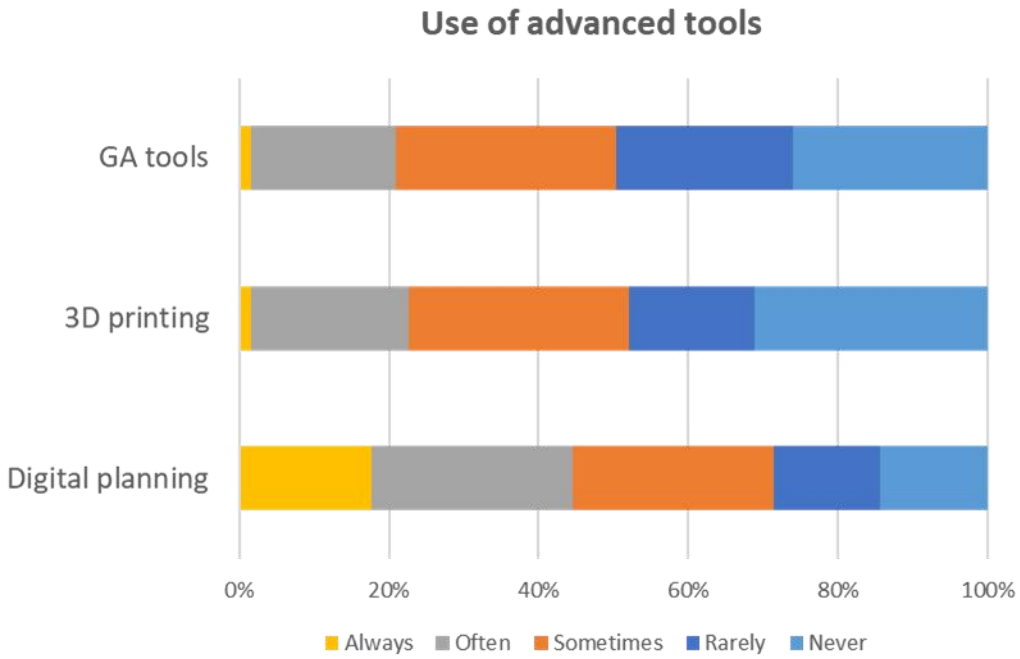
Clinical practices

High variability in use of advanced tools in routine RBD care

- ~50% used any type of gait-analysis tools/3D printing



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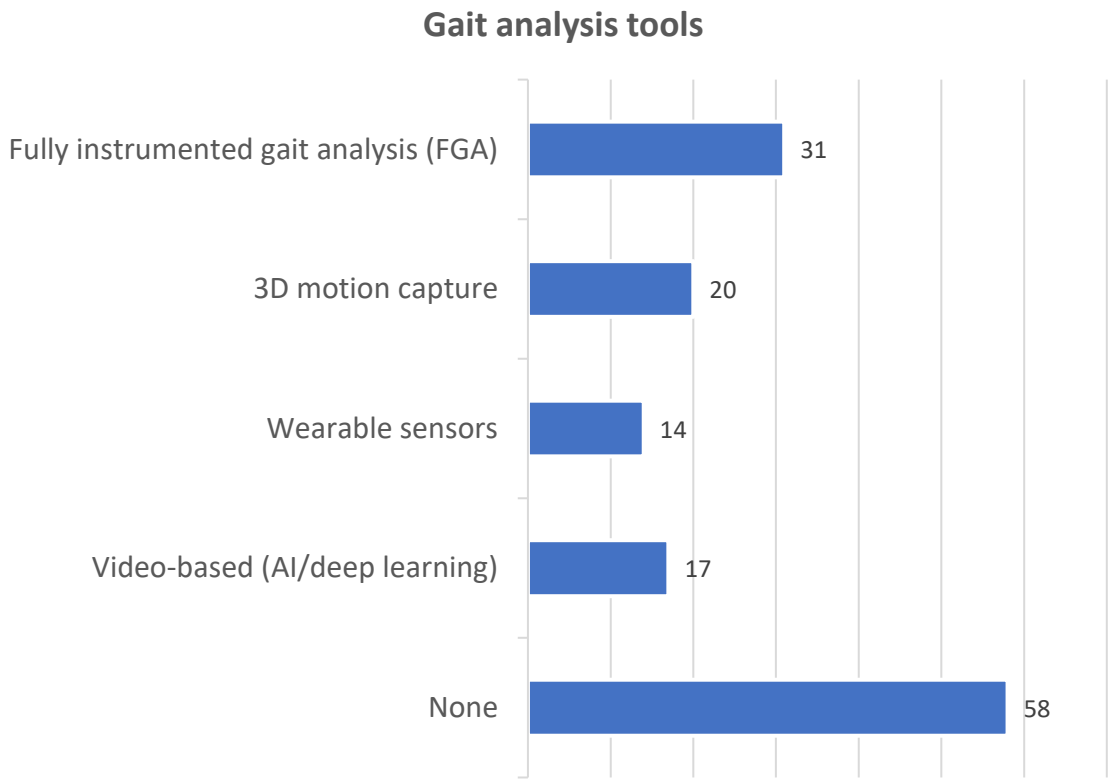
Clinical practices

Most commonly used gait analysis tool-FGA



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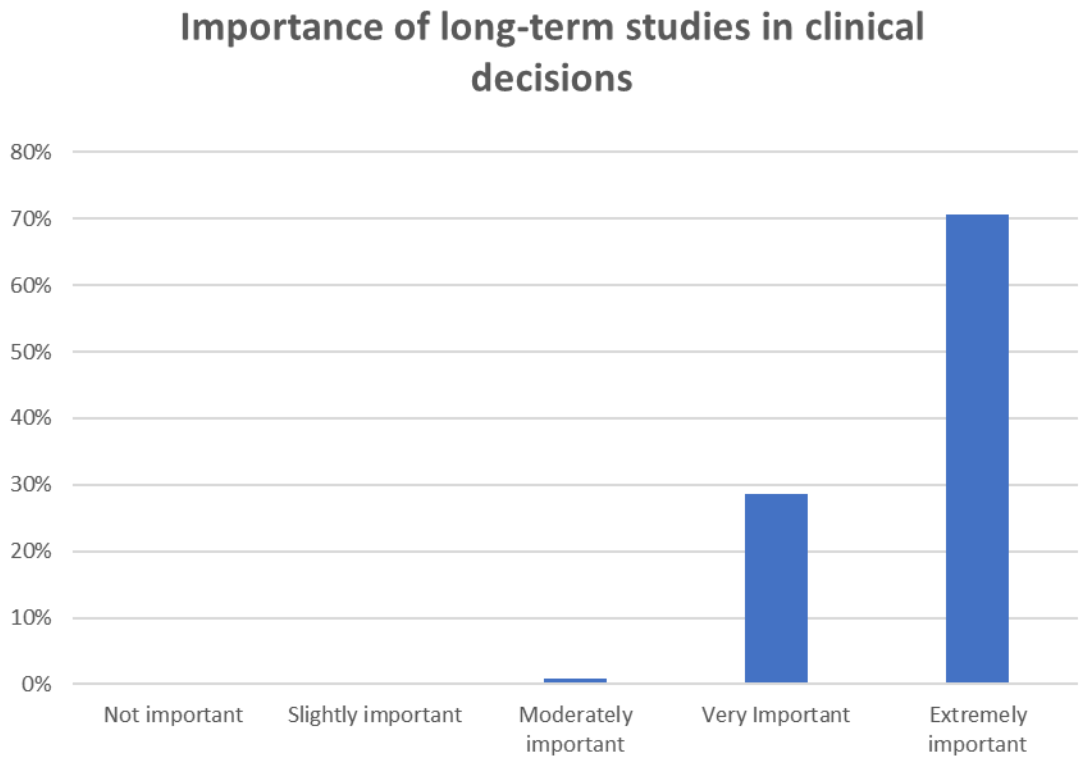
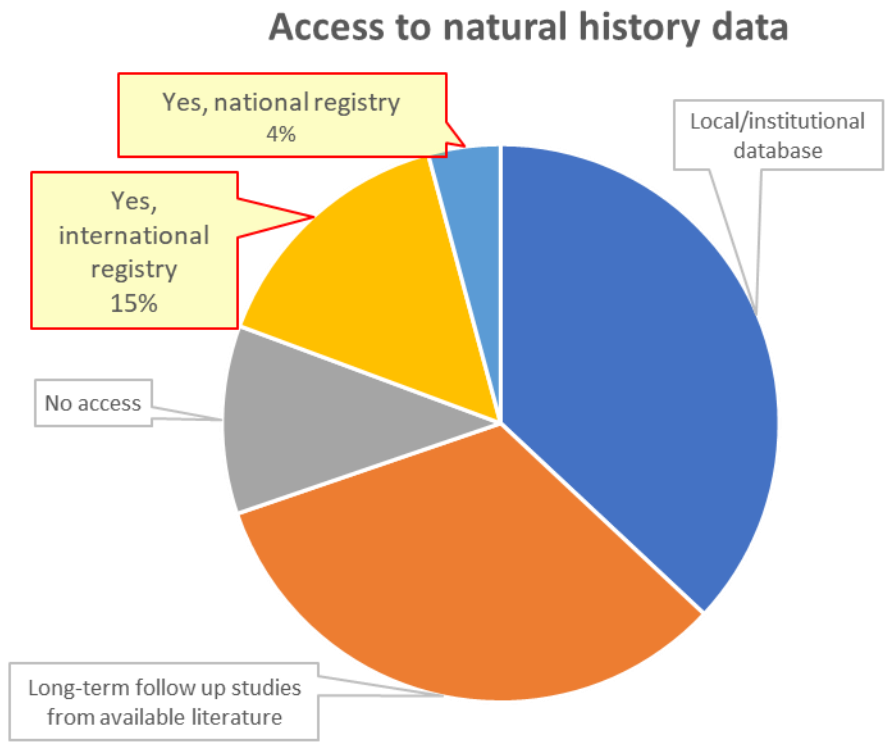
Clinical practices

- All acknowledged the importance of registry data
- But only 19% had access to national or international registry data to guide decisions.





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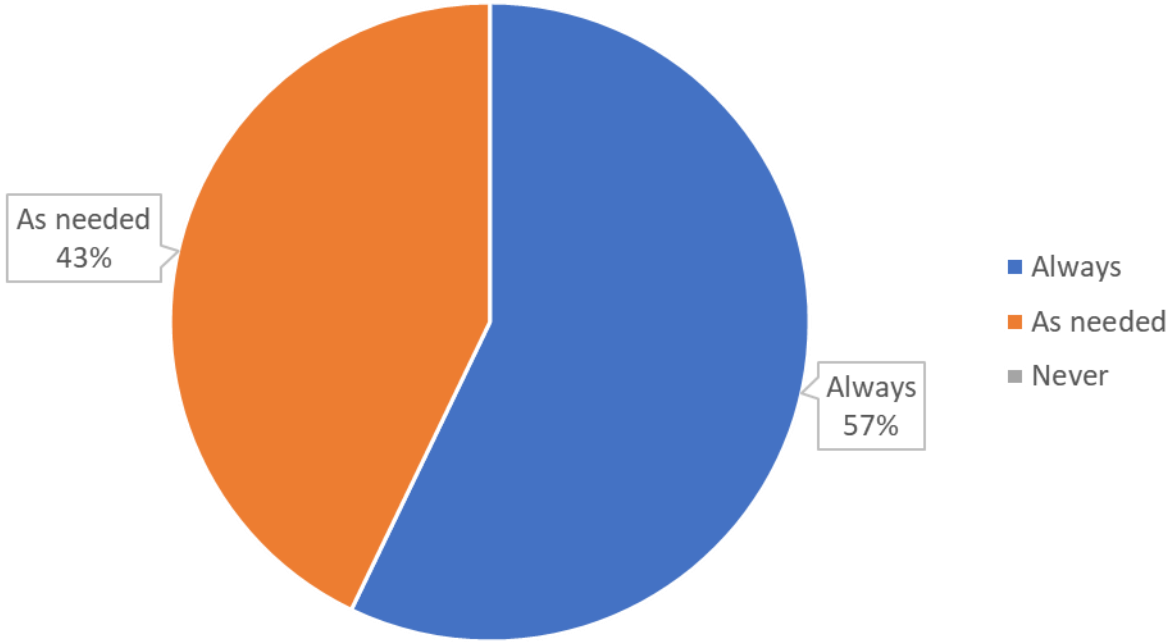
Interdisciplinary & Transition care

Interdisciplinary approach



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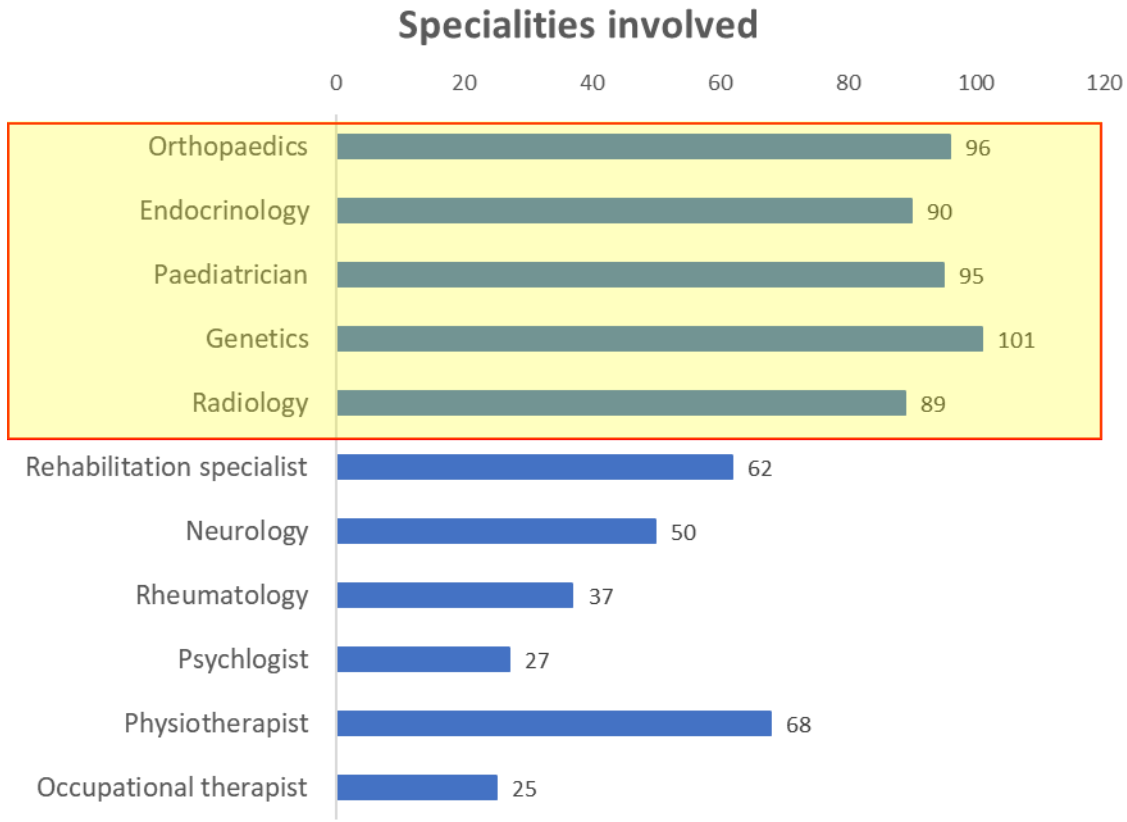
Interdisciplinary & Transition care

Interdisciplinary approach



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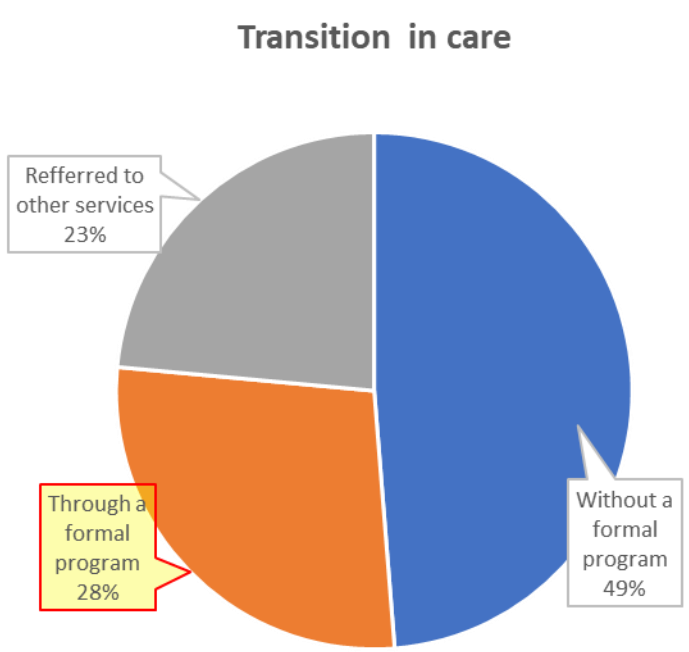
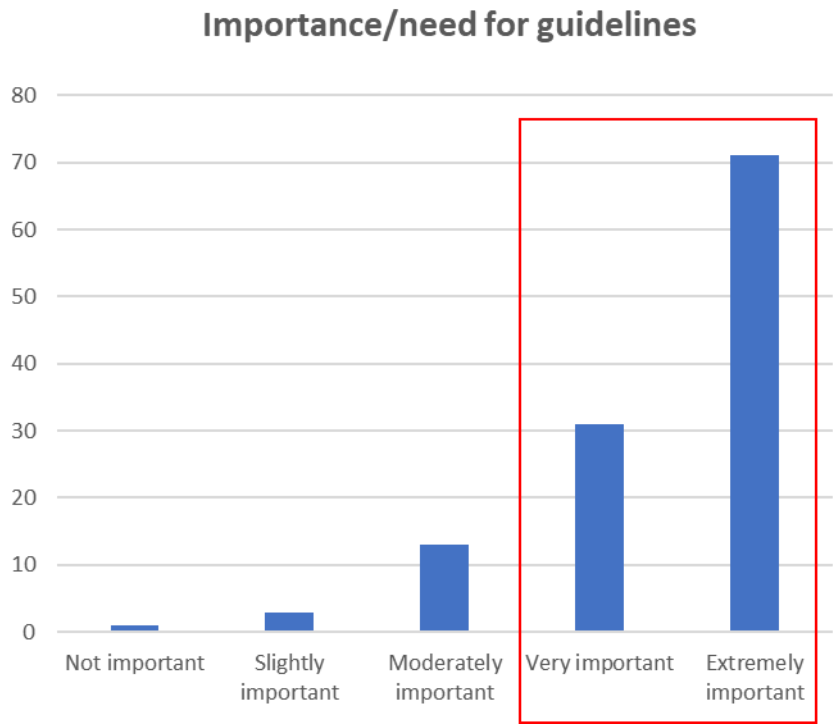
Interdisciplinary & Transition care

Transition in care from paediatric to adult setting in RBD care

- Only 28% have a formal transition protocol
- >85% acknowledged the need for a formal protocol





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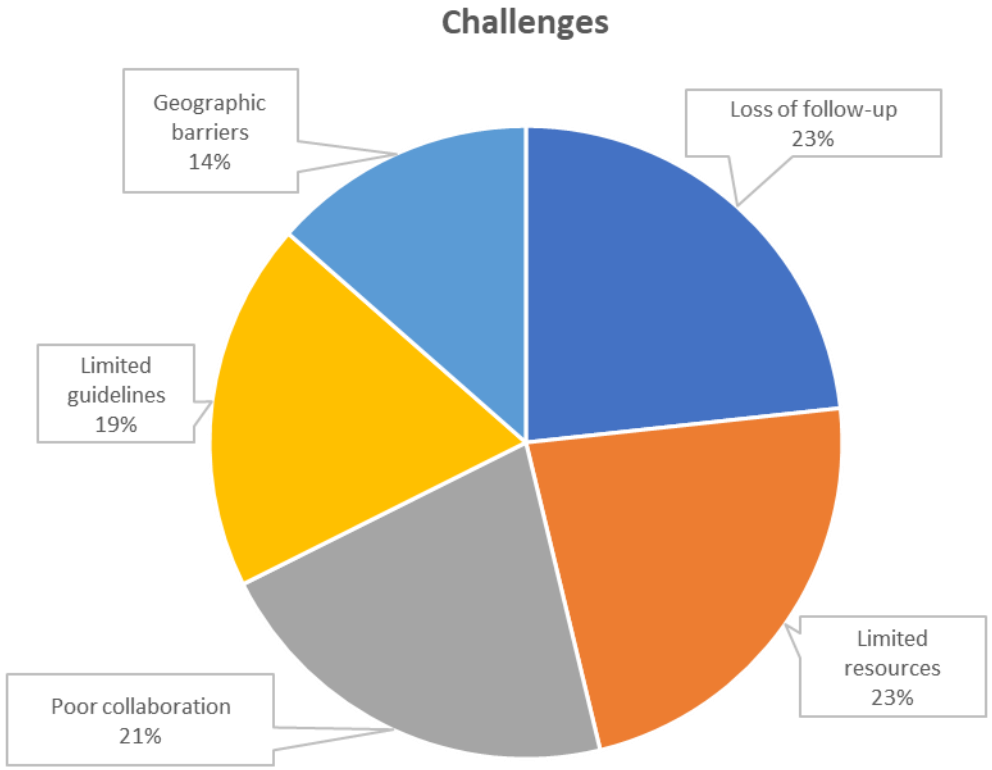
Interdisciplinary & Transition care

Transition in care from paediatric to adult setting in RBD care

- >50% named 3 or more challenges
  - Loss of follow-up or patient disengagement
  - Limited resources in adult services
  - Poor paediatric-adult care collaboration
  - Limited guidelines/protocols
  - Geographic barriers



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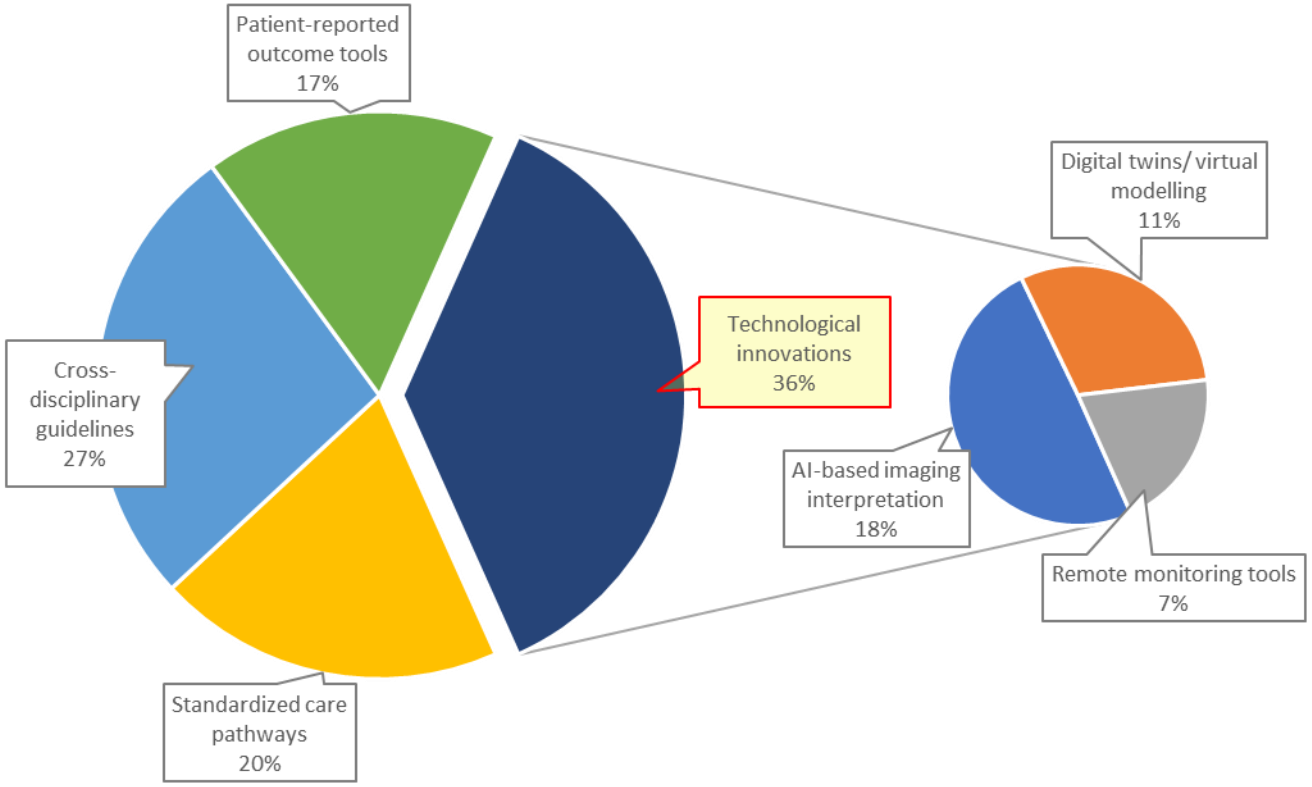
Future Perspectives

Innovations shaping the future of RBDs

- Clinical and patient-centred care frameworks (64%)
- Technological innovations (36%)



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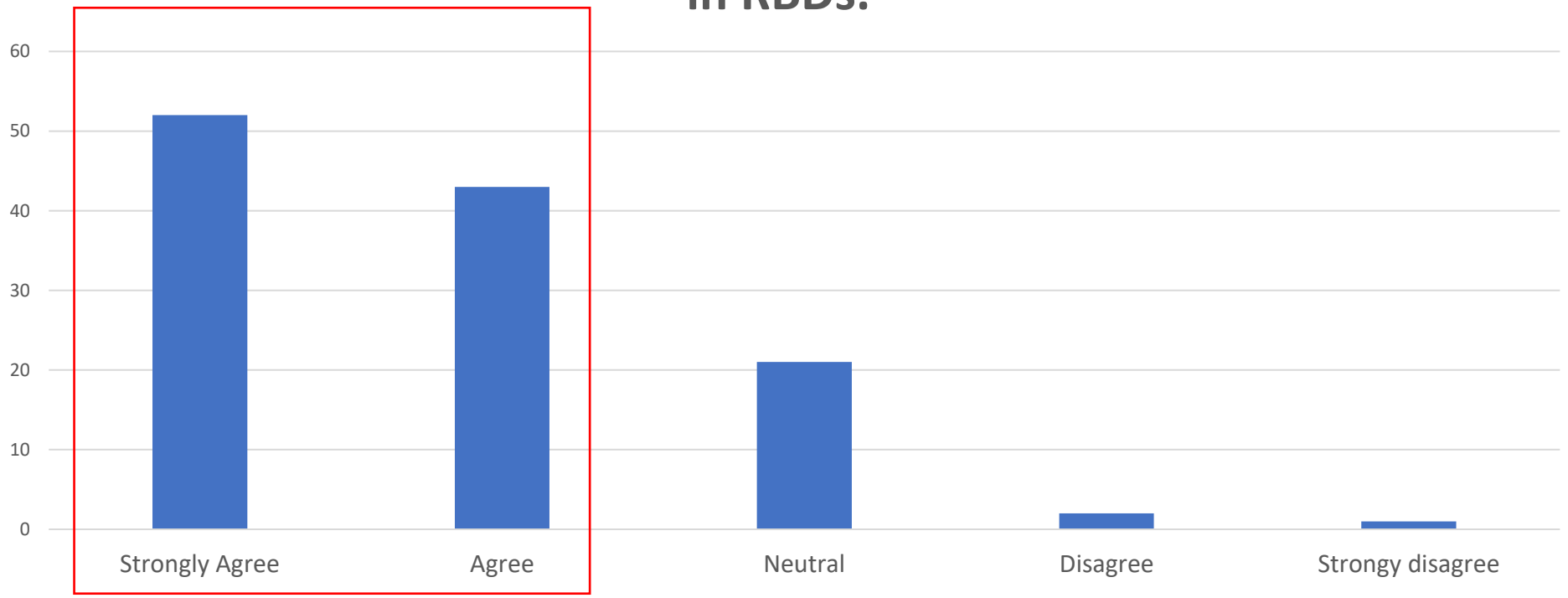
Future Perspectives

"Better integration of clinical, imaging, and functional data would improve clinical decision-making in RBDs."



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Conclusions

- First pan-European survey within ERN-BOND and EPOS to systematically capture clinical decision-making in RBDs
- Similar practice in use of basic clinical parameters
- Considerable variability in use of more advanced techniques
- The findings underscore the importance and need of
    - standardized interdisciplinary care
    - national and international registry data
    - structured transition protocols

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