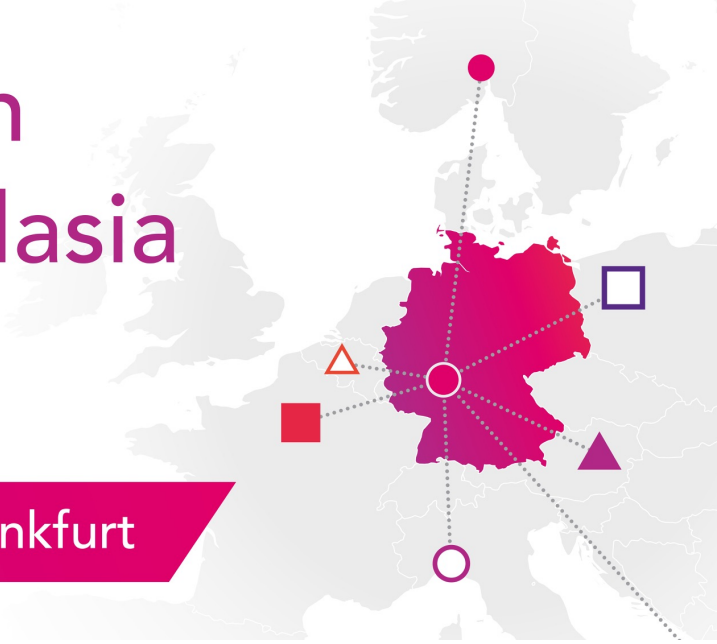
EAF

European Achondroplasia Forum

Advances in Achondroplasia 2023

21–22 April 2023, Frankfurt



DECISION-MAKING AS PART OF THE MDT

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Disclosures

- Melita Irving: Receives honoraria from BioMarin, QED, Ascendis, Novo Nordisk, Sanofi
- Josef Milerad: None to declare
- Simone Riganti: None to declare

CASE 1: INFANT

Case 1: Infant

Background:
- Diagnosed prenatally
- Referred to specialist centre at birth by the local team

Who is part of the core MDT?

Orthopaedics | Clinical genetics | Paediatric endocrinology | Therapy team | Other

What assessments should be undertaken?

MRI | Audiometric assessment | Sleep study

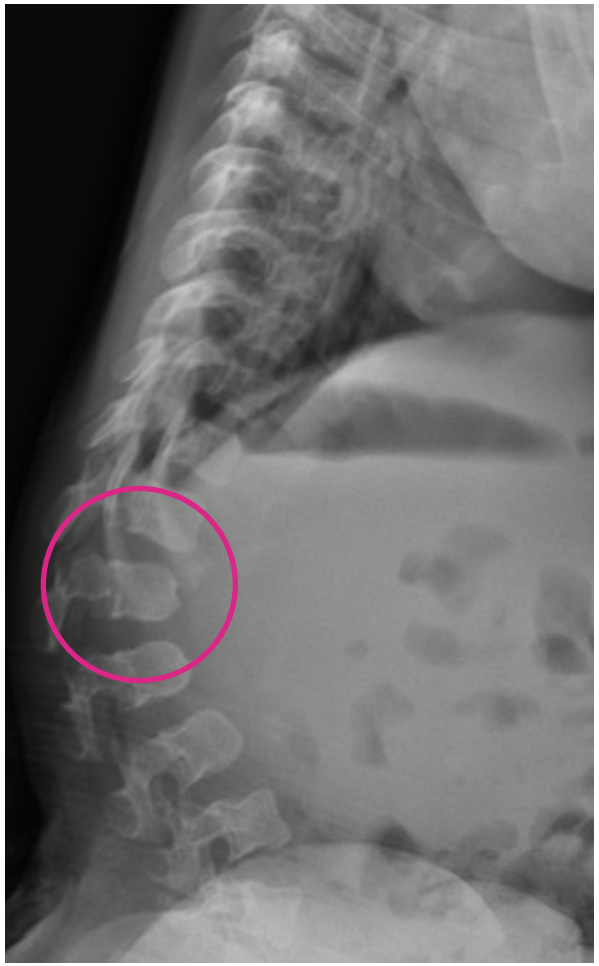
Case 2

MRI, Magnetic resonance imaging; MDT, Multidisciplinary team.

Orthopaedics

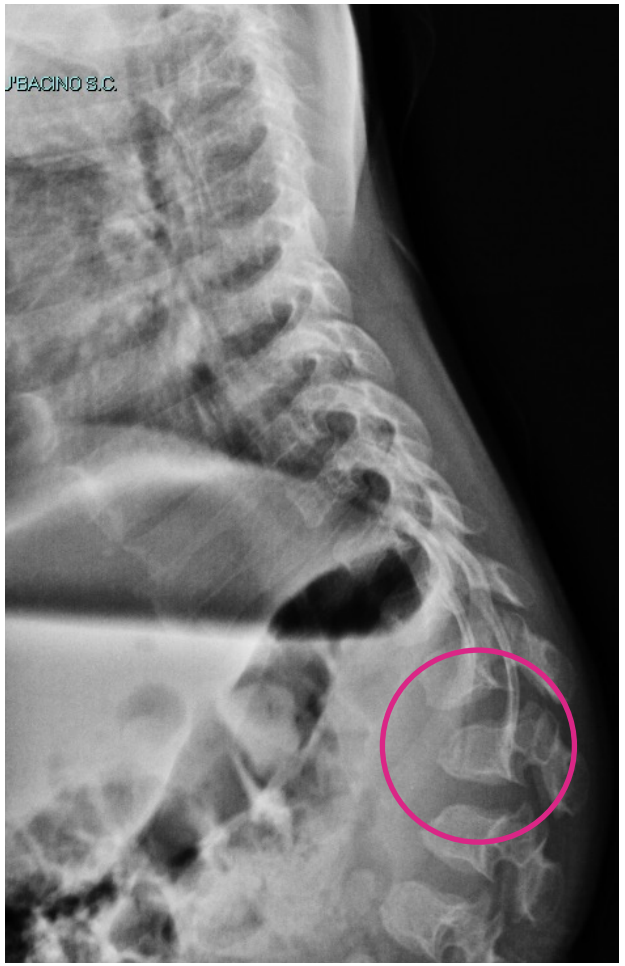
- What is the role of the orthopaedic specialist for an infant?
- What interventions do you consider?
- What are the next steps?

Spinal evaluation: possible spinal deformity



Postural hyperkyphosis at the dorsolumbar passage

Physiotherapy



Anterior wedge deformity of the vertebral body

Brace

Back

Clinical genetics

- What is the role of the clinical geneticist for an infant?
- What interventions do you consider?
- What are the next steps?

Back

Paediatric endocrinology

- What is the role of the paediatric endocrinologist for an infant?
- What interventions do you consider?
- What are the next steps?

Back

Therapy team

- What is the role of the therapy team for an infant?
- What interventions do you consider?
- What are the next steps?

Back

Other

- Patient Advocacy Groups
- Any other specialities that should form part of the core MDT?
  - Neurosurgeon?
  - Neurologist?
  - Psychologist?

Back

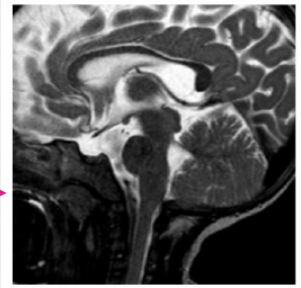
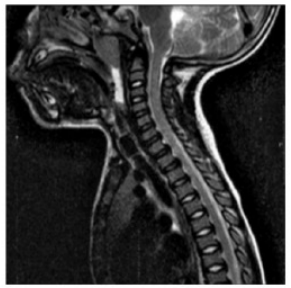
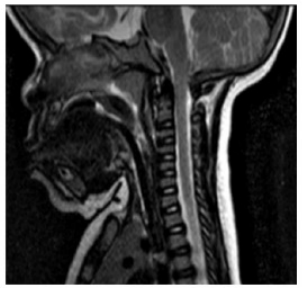
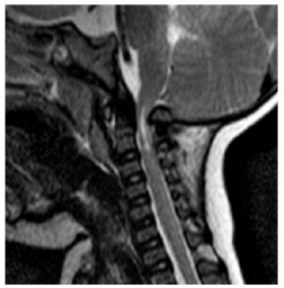
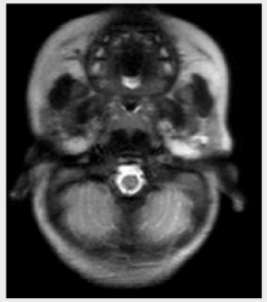
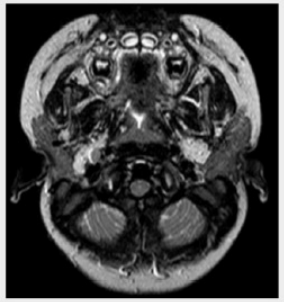
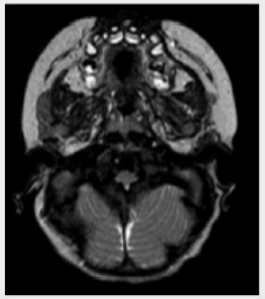
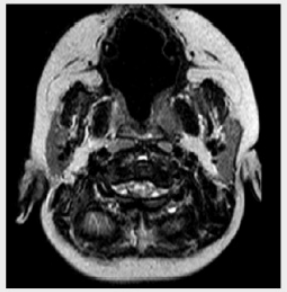
MDT, Multidisciplinary team.

MRI

Clinical neurological examination
- Hypotonia
- Motor delay
- Myelopathy

Polysomnography
- Abnormalities indicative of FMS

MRI

AFMS0	AFMS1	AFMS2	AFMS3	AFMS4
Normal foramen magnum	Constitutional narrowing of the foramen magnum with preserved CSF (no cord distortion)	Narrowing of the foramen magnum with loss of CSF space surrounding the cord	Loss of the CSF space with cord compression	Cord compression and signal changes (Myelomalacia)
				
				

What is the decision-making process to proceed to decompression surgery?

Who is involved?

Back

FMS, Foramen magnum stenosis; MRI, Magnetic resonance imaging.

Sleep study

- Infants with achondroplasia can have the same complications as those without
- Even severe breathing problems may not show up in wakefulness
- Respiratory control in wakefulness
  - Sensory inputs
  - Speech
  - Exercise
- Respiratory control during sleep
  - Oxygen and CO2 sensors
  - Sensory inputs from the airways

- **Central apnea** – *no breathing movements* >10 sec ***and*** drop in SaO2 >3%
- **Obstructive apnea** – *breathing movements no airflow* >10 sec *and* drop in SaO2 >3%
- ***Hypopnea*** > 30% decrease in respiratory movements or airflow >10 sec and drop in SaO2 >3%
- ***Airflow restriction/UARS*** increased respiratory efforts to compensate for restricted airflow. (RIP) thoraco-abdominal asynchrony. SaO2↓ CO2 ↑

Sleep Study – What should you do?

Increased airway resistance is present in most children with ACH

Decreased oxygenation and elevated CO_2 levels are common

Airway obstruction may arise from brain stem compression leading to asynchronous breathing.

We need a composite index of respiratory impairment – single variables unreliable

Which type of sleep study is best? When? Where?

Back

Audiometric assessment

- Newborn hearing assessment
- Monitoring for otitis media
- How frequently?
- When do you refer to an ENT specialist?

Back

ENT, Ear, nose and throat.

CASE 2: YOUNG CHILD

Case 2: Young child

Background:

- Child aged 2–5
- Core MDT remains unchanged, with discussion of issues arising with extended MDT

What are the main considerations for the MDT in this age group?

Medical

Adaptations

Other

Case 3

MDT, Multidisciplinary team.

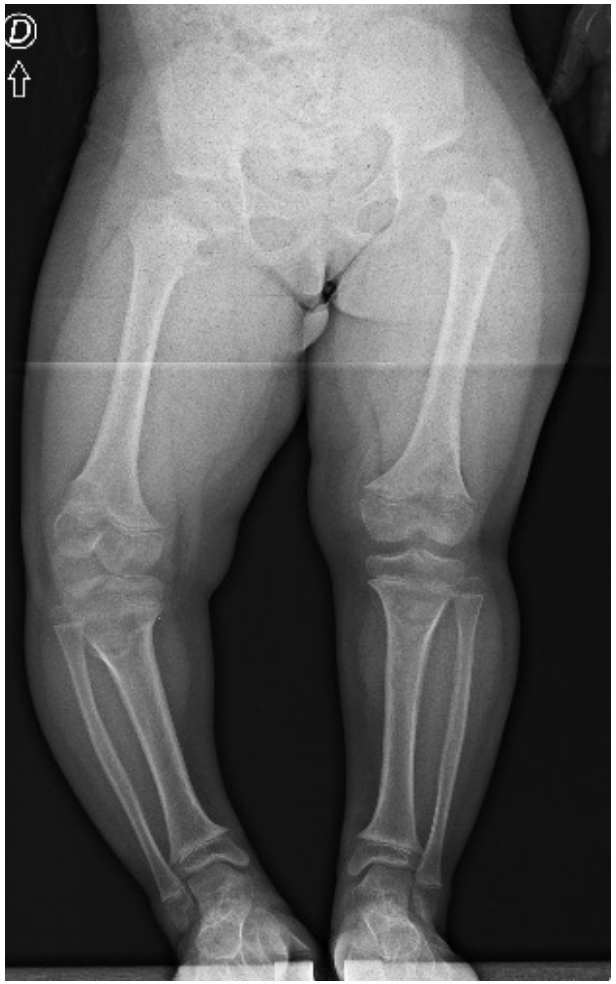
Medical considerations

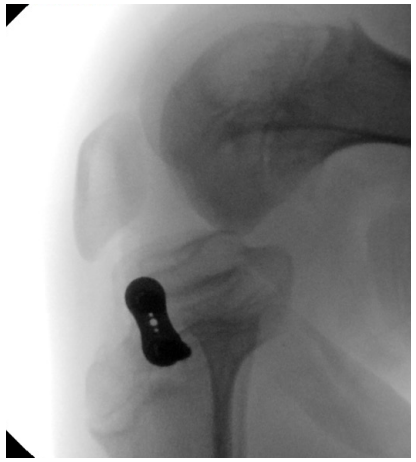
- Children with achondroplasia can have the same complications as those without
- Clinical evaluation – what should you be looking for?
  - Apnoea? Can change as children age and body size changes
- What medical interventions should be carried out in this age group?
- What surgical interventions should be discussed/carried out in this age group?
- Which members of the extended MDT may need to be involved?

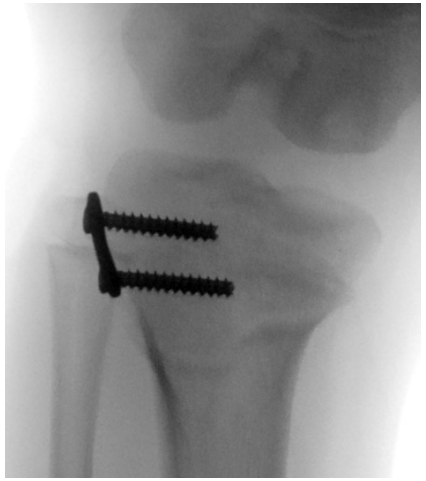
Back

MDT, Multidisciplinary team.

Varus knee deformity







Correction with hemiepiphysiodesis with 8 plates

Adaptations

- At home?
- In preparation for school?

Back

Other

- Accessing support through Patient Advocacy Groups
- Support from pain therapists and physiotherapists during stretching phase of surgical lengthening
- Any other considerations in this age group?

Back

CASE 3: OLDER CHILD

Case 3: Older child

Background:

- Child aged 5–13
- Core MDT remains unchanged, with discussion of issues arising with extended MDT

What are the main considerations for the MDT in this age group?

Medical Adaptations Psychosocial Other

End

MDT, Multidisciplinary team.

Medical considerations

- Older children with achondroplasia can have the same complications as those without
- Clinical evaluation – what should you be looking for?
  - Apnoea? Can change as children age and body size changes
- What medical interventions should be carried out in this age group?
- What surgical interventions should be discussed/carried out in this age group?
- Which members of the extended MDT may need to be involved?

Back

MDT, Multidisciplinary team.

Achondroplasia – Limb Lengthening

- Improvement in limb-trunk proportion
- Functional improvement
- Improve social integration

Achondroplasia – Limb Lengthening

Contraindications

- Severe joints instability → luxation
- Lumbar and/or cervical canal stenosis
- Occipital/Cervical instability
- Poor patient motivation

Back

Adaptations

- At home?
- In preparation for secondary school?
- To enable independence?

Back

Psychosocial

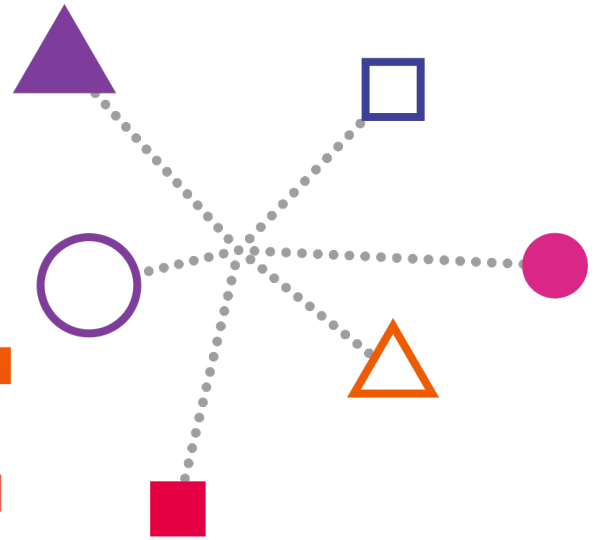
- Preparedness for secondary school
- Independence, confidence, socialisation

Back

Other

- Accessing support through Patient Advocacy Groups
- Considerations for transition from paediatric to adult care
- Any other considerations in this age group?

Back



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