Welcome!

November 9, 2022

The webinar will begin at 6 p.m. CT

Agenda for the evening

6 p.m. | **Welcome** by Erika Peterson, MD

6:05 p.m. | **Skeletal Dysplasias in Pregnancy** by Donald Basel, MD and Jessica Smith, MD

6:50 p.m. | **Supporting Families Through an Uncertain Pregnancy: Evolution and Current State of Perinatal palliative Care** by Steven Leuthner, MD, Erin Rholl, MD and Carrie Hecox, APNP

7:35 p.m. | **Update on the Heart Disease and Pregnancy Clinic** by Scott Cohen, MD

7:45 p.m. | **Wrap up** by Erika Peterson, MD

Q&A sessions to follow after each presentation.

Physician Liaisons are here for you

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Southeast Wisconsin

- **Serve as a link between Children's Wisconsin and referring providers**
- **Provide information about Children's Wisconsin services and programs, including continuing education opportunities**
- **Facilitate solutions for referral and communication issues**

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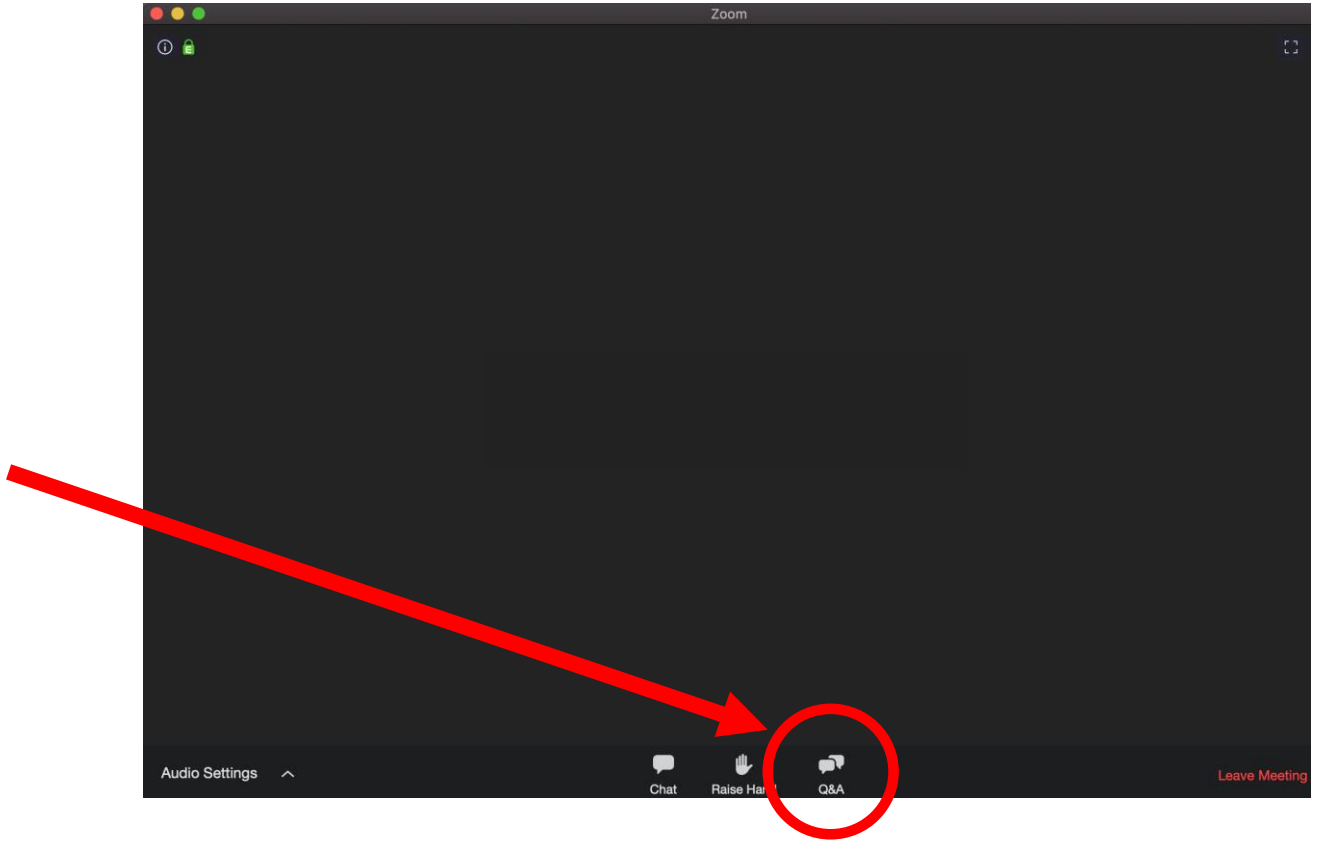
Asking the presenter a question

At any time during the lecture, you can submit your question by **clicking the Q&A icon** at the bottom of your screen



Donald Basel, MD

- Medical Director of the Genetics Center and Co-program director for neurofibromatosis and RASopathy; since 2010
- Professor and Chief of the Division of Genetics in the Department of Pediatrics at the Medical College of Wisconsin
- Board certified in Medical Genetics
- Earned his medical degree from the University of the Witwatersrand, South Africa
  - Fellowship in Genetics at the University of Cape Town
  - Postdoctoral research fellowship at University of Connecticut
  - Pediatric and Genetics training in Portland Oregon
- Dr. Basel sees patients at the Milwaukee Hospital campus

Jessica Smith, MD

- Clinical geneticist and genomist at Children's Wisconsin since 2019
- Professor and Chief of the Division of Genetics in the Department of Pediatrics at the Medical College of Wisconsin
- Board certified in clinical genetics and genomics, maternal-fetal medicine and obstetrics gynecology
- Earned her medical degree from the University of Cincinnati Medical Center
- Completed her fellowships in maternal fetal medicine at the University of Michigan Medical Center
- Dr. Smith sees patients at the Milwaukee Hospital campus

Skeletal Dysplasias in Pregnancy

Jessica Smith, MD & Donald Basel, MD

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Disclosures

We have no relevant financial interests/relationships to disclose

Objectives

- Define skeletal development and skeletal dysplasias
- Antenatal diagnosis of skeletal dysplasia
- Nomenclature involved with skeletal dysplasias
- A few “tips” for antenatal diagnosis of severe disorders

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Defining Skeletal Development & Dysplasias

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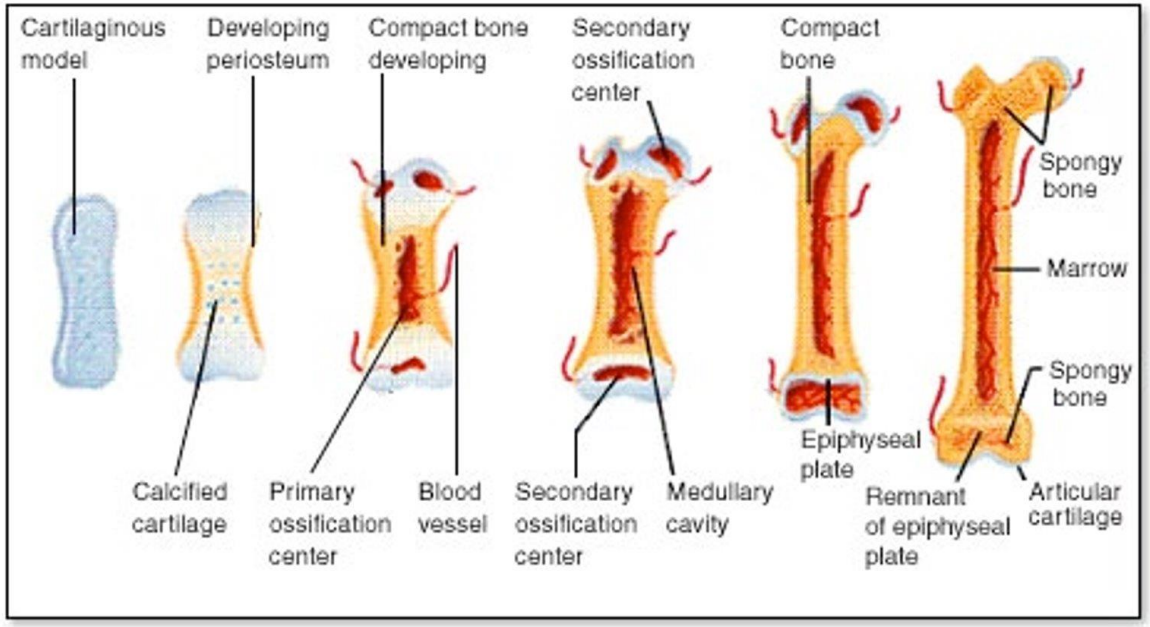
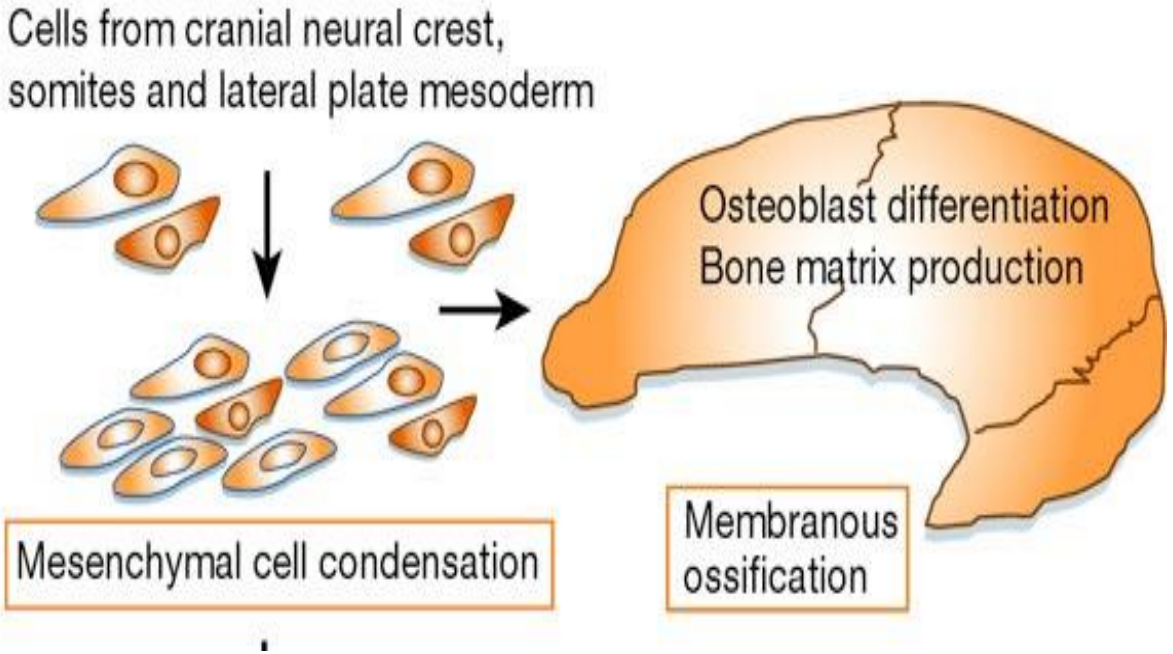
Skeletal Ossification

Endochondral Ossification



Axial/appendicular skeleton, cartilage model, osteoblasts replace with bone at 9-14 weeks gestation

Membranous Ossification



Craniofacial, clavicles, direct differentiation from mesenchyme to osteoblasts

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WHAT ARE SKELETAL DYSPLASIAS

- DISORDERS OF BONE DUE TO ABNORMALITY OF:
  - CARTILAGE MATRIX IN THE PHYSIS
    - •COMPOSITION
    - •REGULATION
  - MATRIX MINERALIZATION AT THE PHYSIS &/OR IN THE BONE ITSELF
  - QUALITY OF BONE MATRIX

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Rare but not Uncommon

- Skeletal dysplasias are rare individually

- Common as a whole
  - Incidence estimated at 2.4/10,000 births
    - 0.95 to 1.5 per 10,000 live births are lethal skeletal dysplasias
  - Approximately 460 well defined disorders
  - 100 with prenatal onset

    ~1% perinatal deaths (9.1/1000)

Norton, *Callen's Ultrasonography in Obstetrics and Gynecology*

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Antenatal Detection

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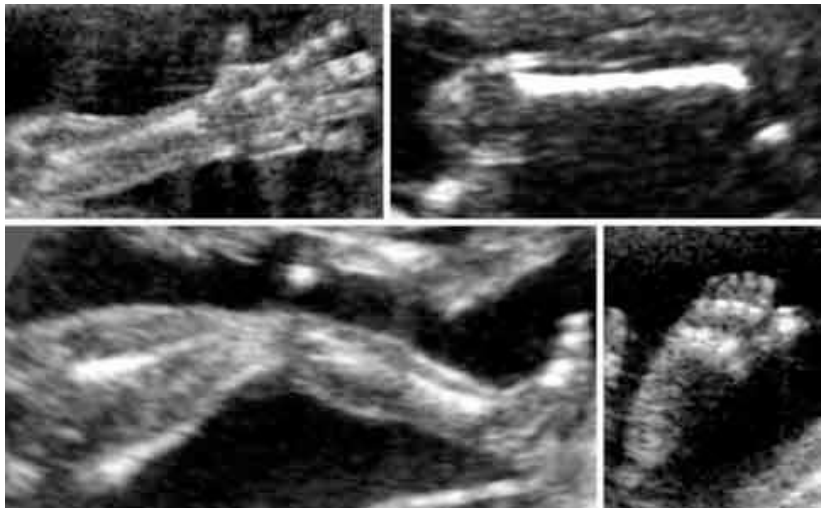
Imaging Fetal Skeleton

- Limb buds - 8 weeks
- Femur, humerus - 9 weeks
- Tib/fib, rad/ulna- 10 weeks
- Digits - 11 weeks



ALL LONG BONES ARE CONSISTENTLY SEEN FROM 11 WEEKS

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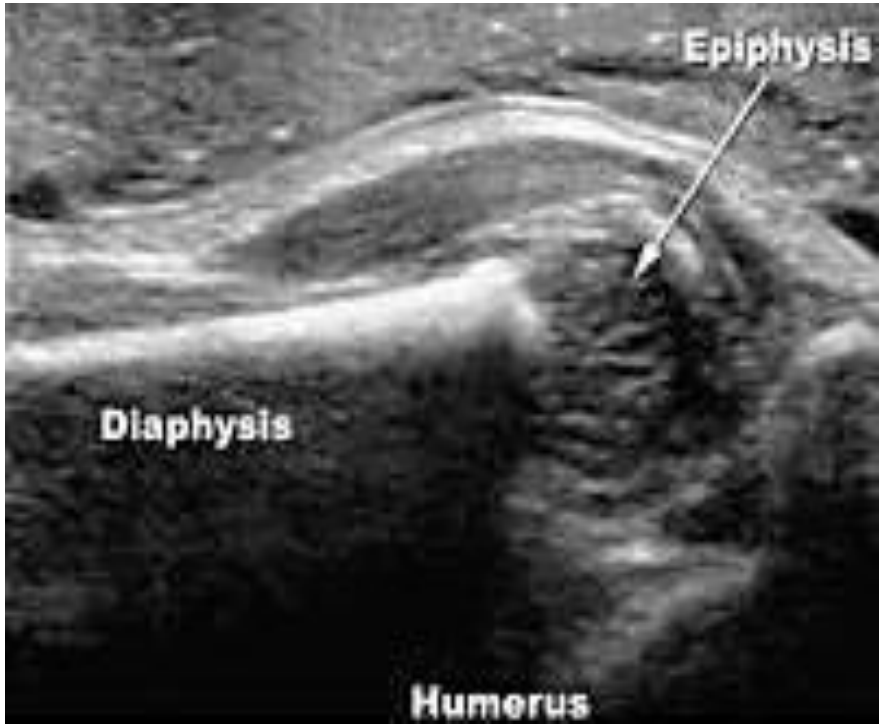
Imaging Fetal Skeleton

When are epiphyses seen:

- Distal femoral 32-33 weeks
- Proximal tibial 34-35 weeks
- Proximal humeral 37-38 weeks



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An Approach to Antenatal Detection

- Measure all long bones
- Appearance long bones
- Evaluate thorax appearance
- Hands, feet
- Cranium
- Facial features
- Spine

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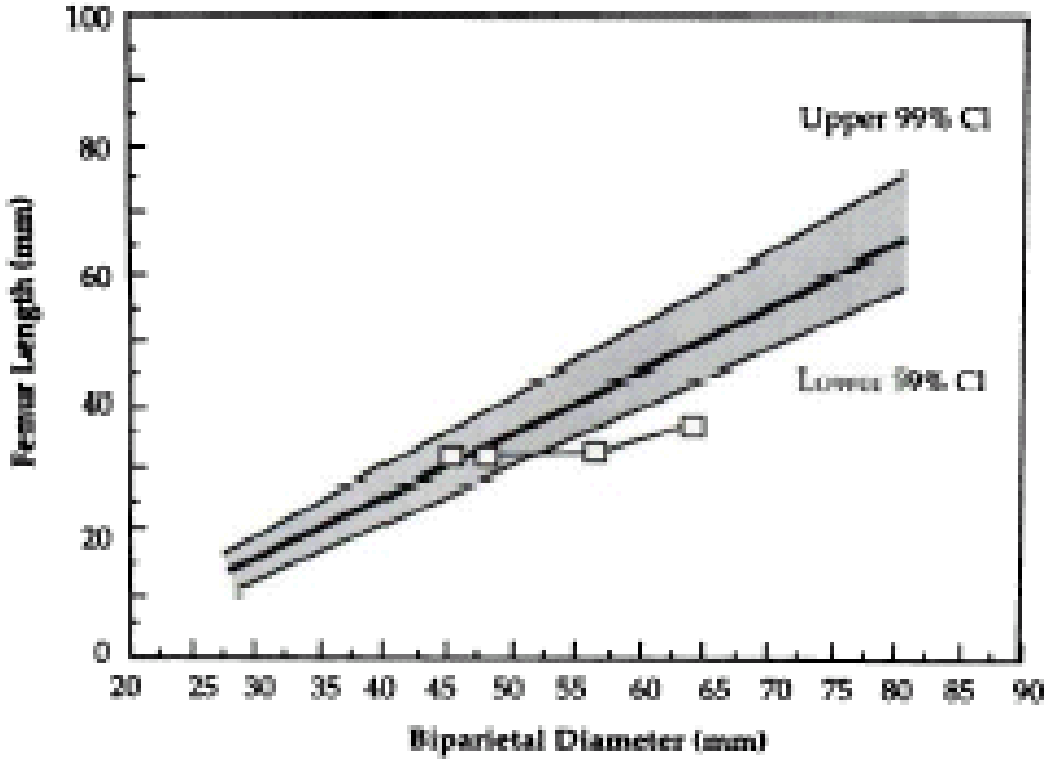
Antenatal Detection

- Bone measurement nomograms
- < 3rd percentile
- Will identify normals as well - currently available nomograms don't have enough patients to differentiate 3-1%ile



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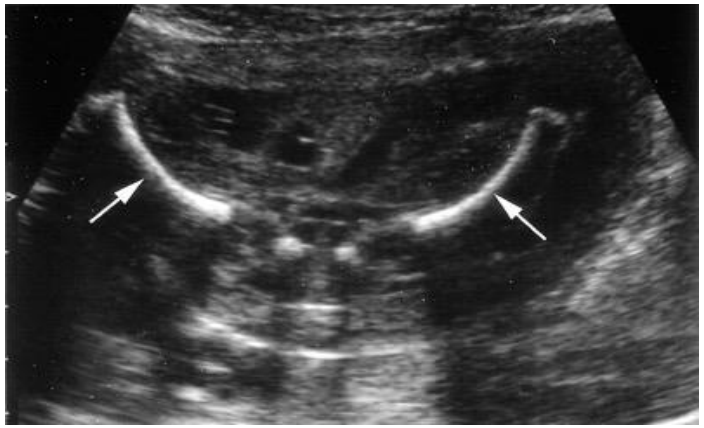
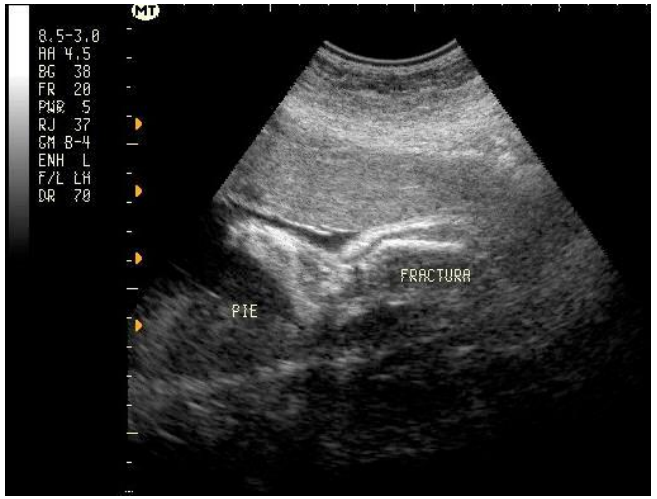
Long bones

- Measure all extremities
- Determine type of shortening (rhizo, meso, micro...)
- Exclude absence/hypoplasia of individual bones
- Fibula, tibia, ulna, radius, clavicles, scapulae
- Characterize mineralization
- Acoustic shadowing
- Absent/decreased echogenicity of spine, cranium
- Long bone bowing, fractures



Limb bowing



Fractures

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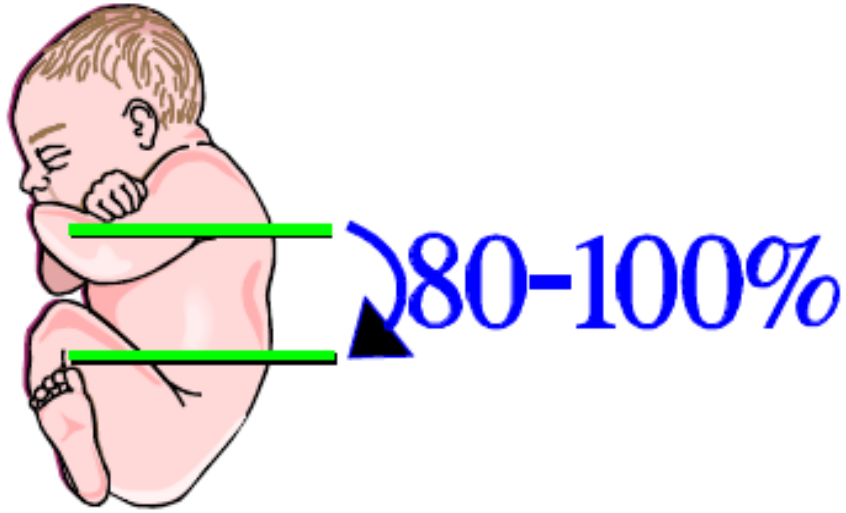
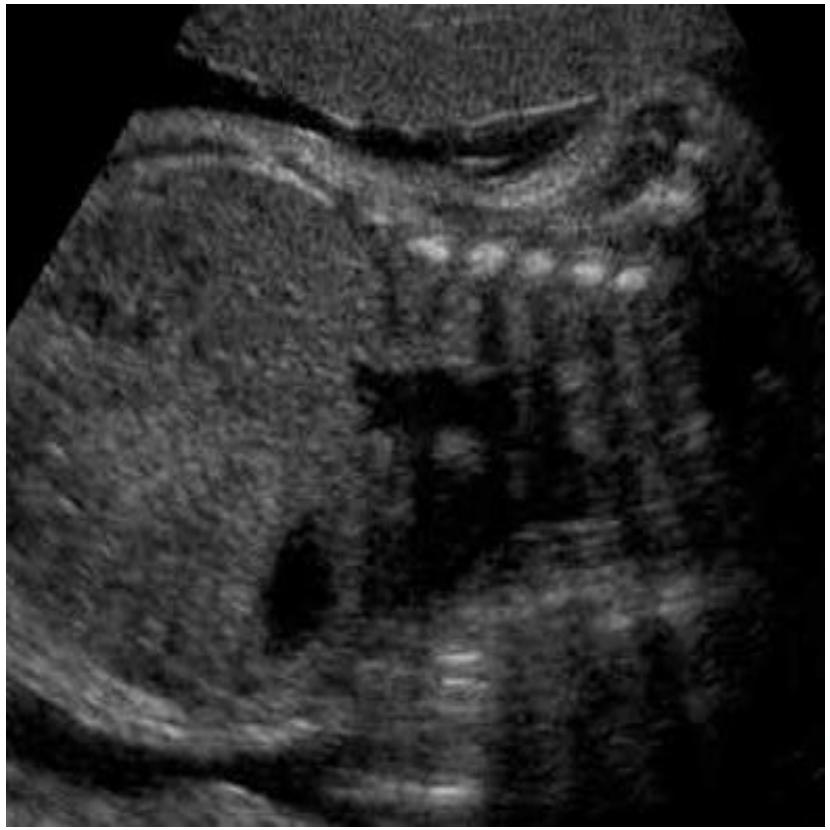
Thoracic Evaluation

- Chest restriction --> pulmonary hypoplasia
- Subjective comparison to abdominal size
- Thoracic circumference, level of the four chamber heart

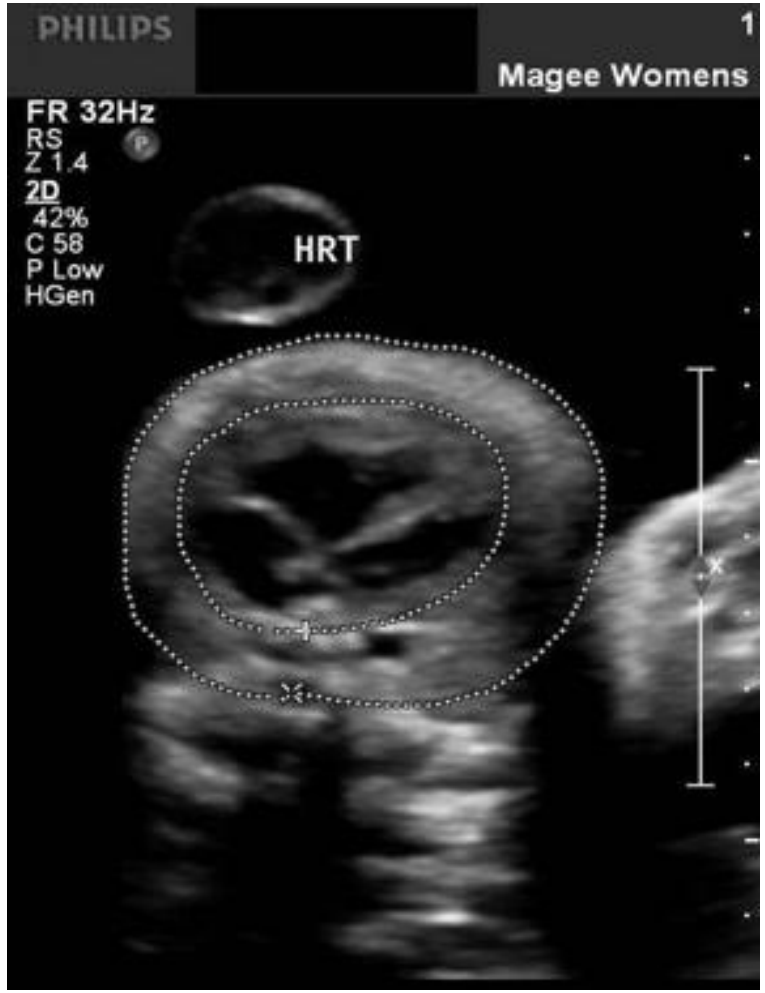


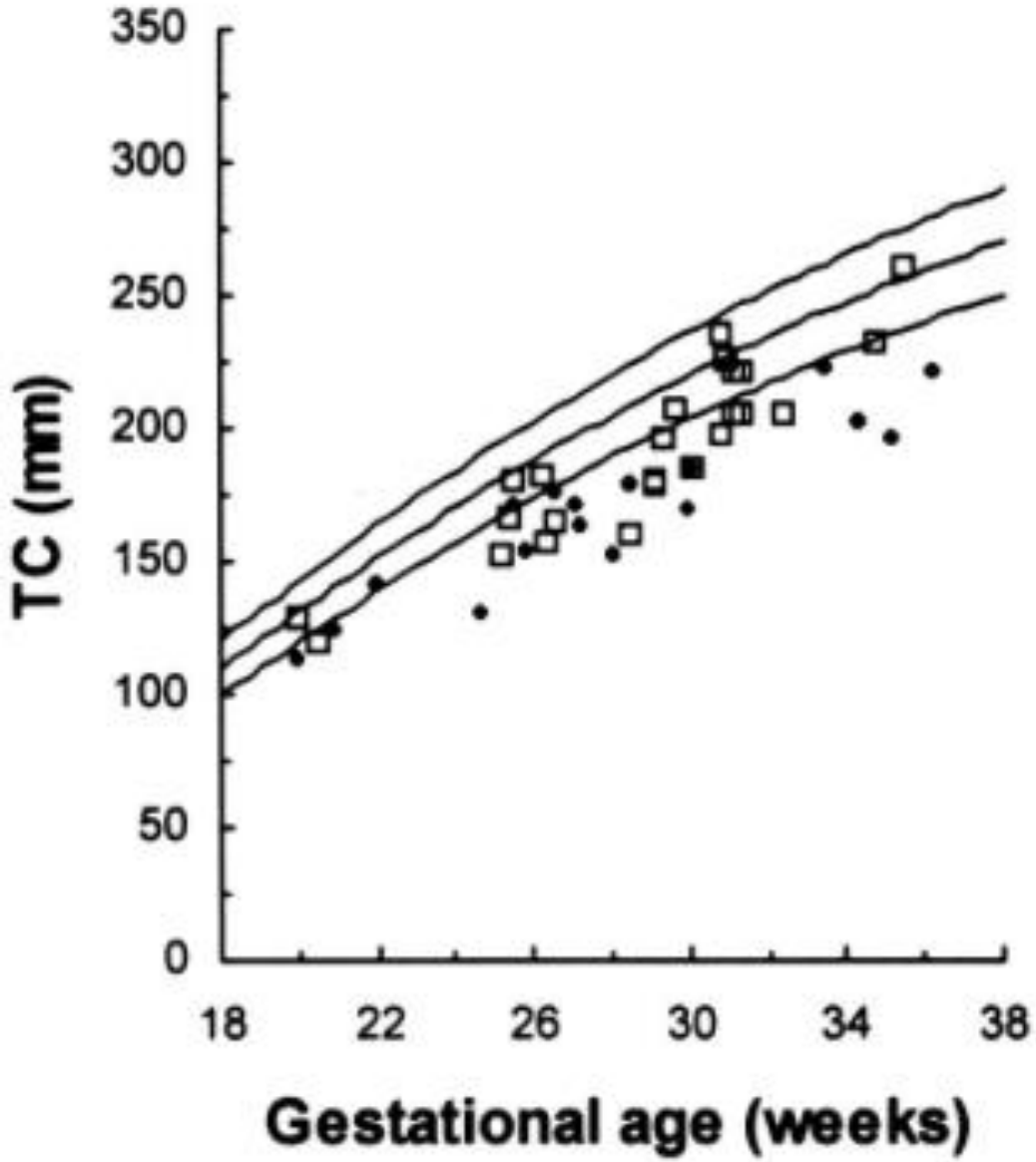
chest diameter =80-100% abdominal diameter (AC)

hypoplastic



Cardiothoracic (CT) Ratio

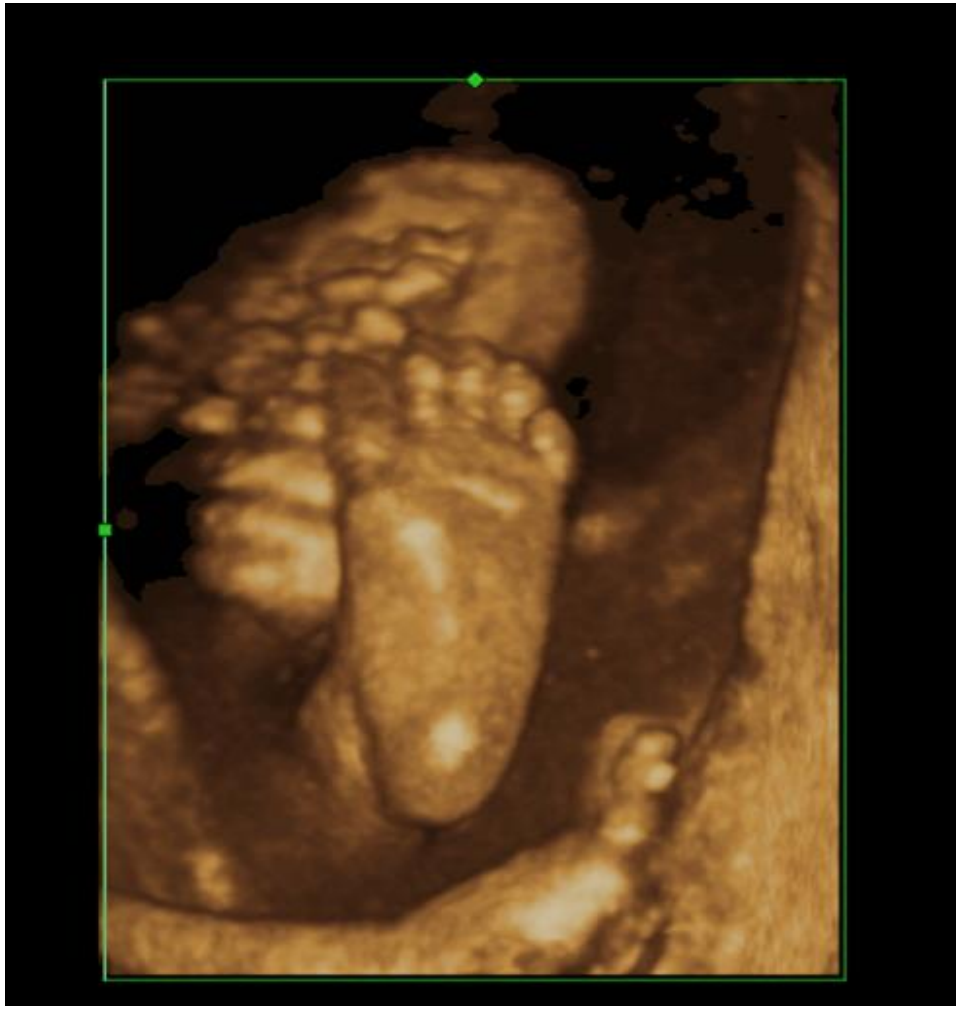




Hands & Feet

- Polydactyly
- Brachydactyly
- Postural deformities
- Fetal foot size
- Femur:Foot length

  (Jeanty, 2005)
  - Nearly constant 14-40 weeks
  - 0.99 +/- 0.06
  - < 0.87 abnormal



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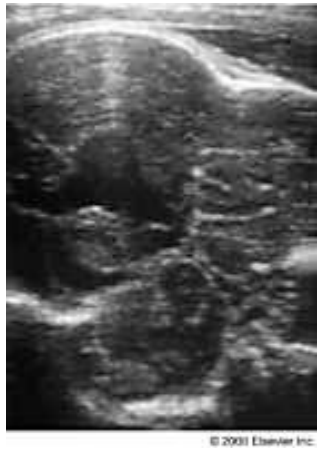
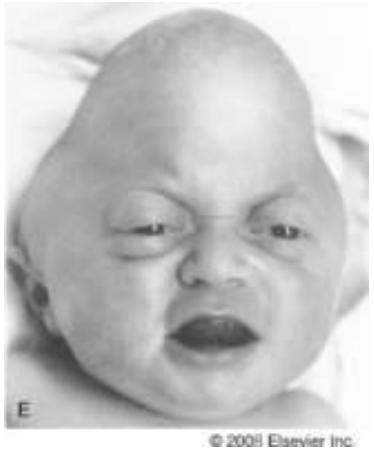
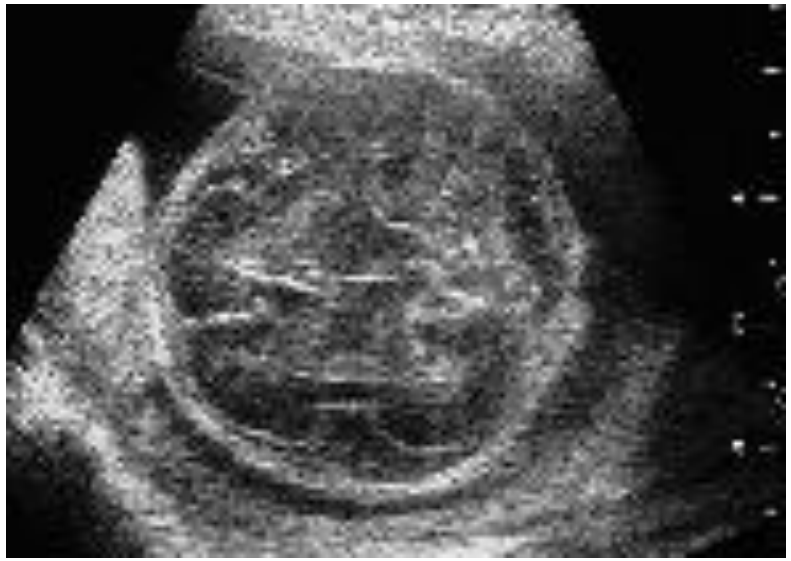
Cranium

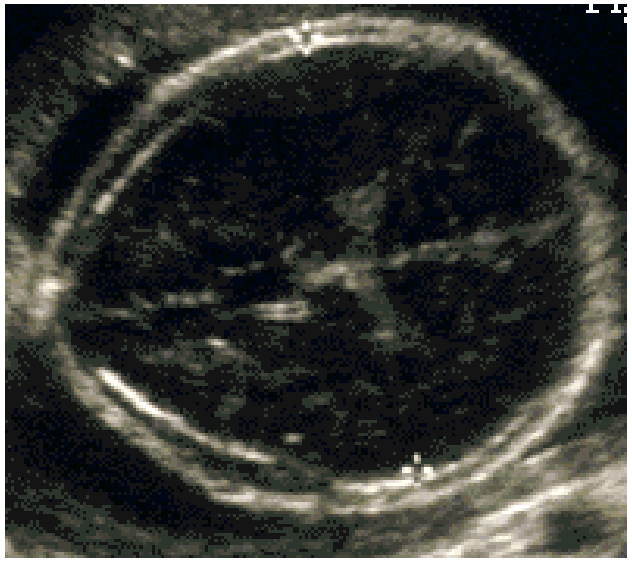
- Poor ossification
- Frontal bossing
- Cloverleaf deformity
- Shape and internal structures
- Face : clefting, micrognathia, hyper/hypotelorism



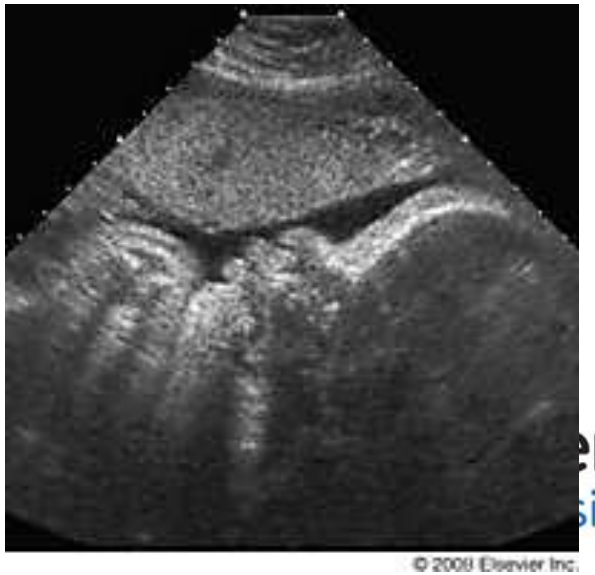




Jeanty 2005

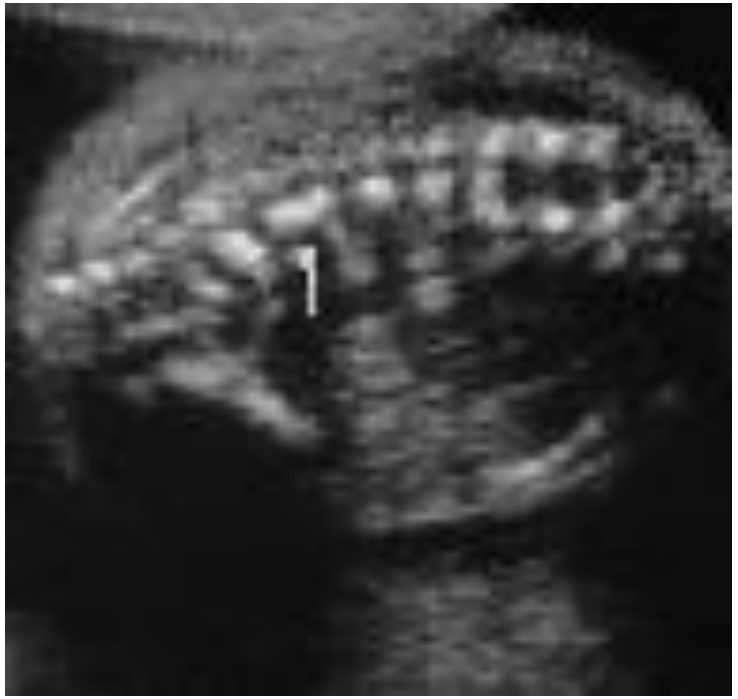


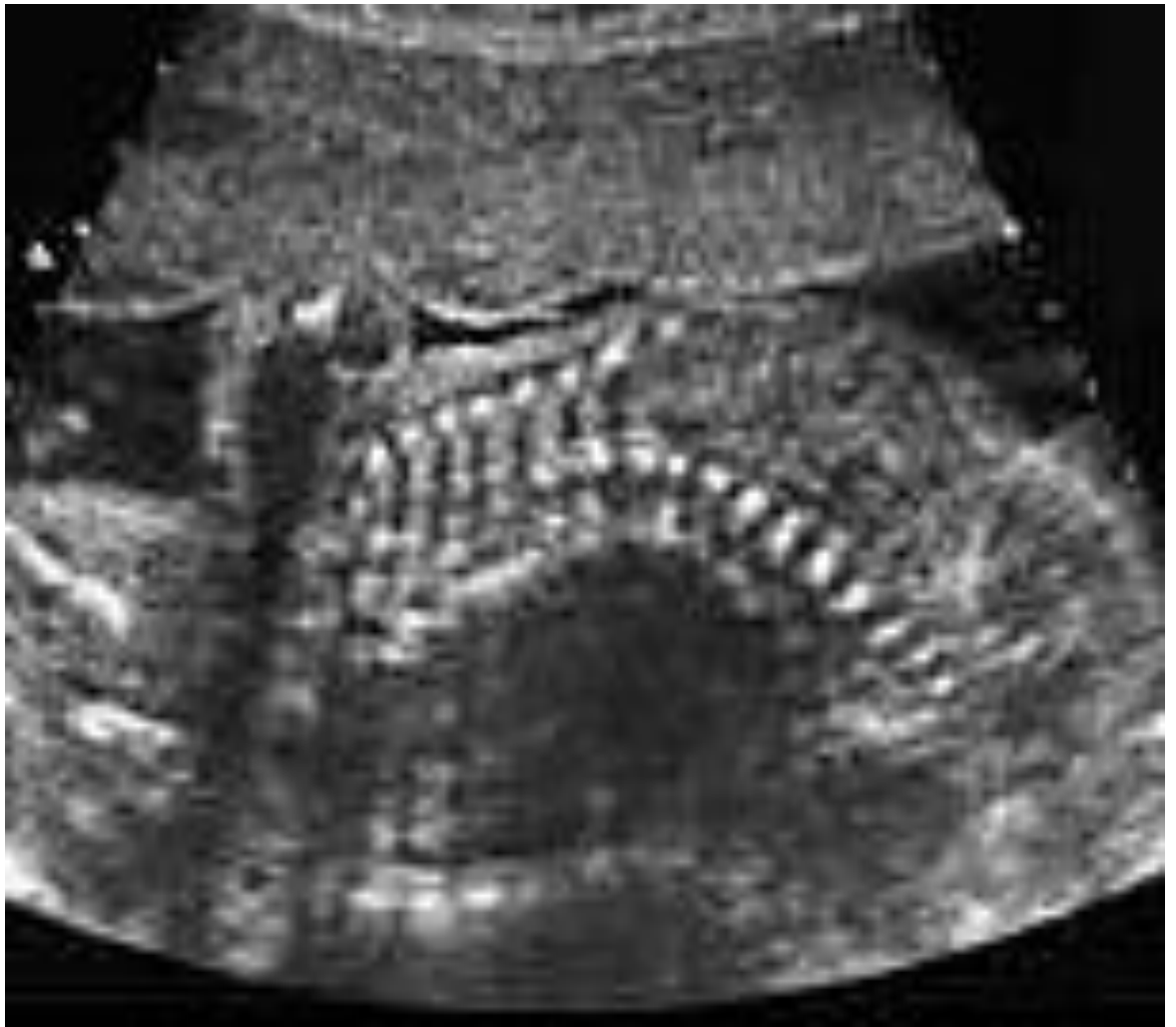
sonoworld.com, 2009



Spine

- Hemivertebrae
- Butterfly vertebrae
- Block vertebrae
- Congenital scoliosis
- Platyspondyly





Additional Evaluation

- Systemic involvement? - kidneys, heart, etc
- Radiographs - consider fetal CT
- Newborn evaluation
- Geneticist

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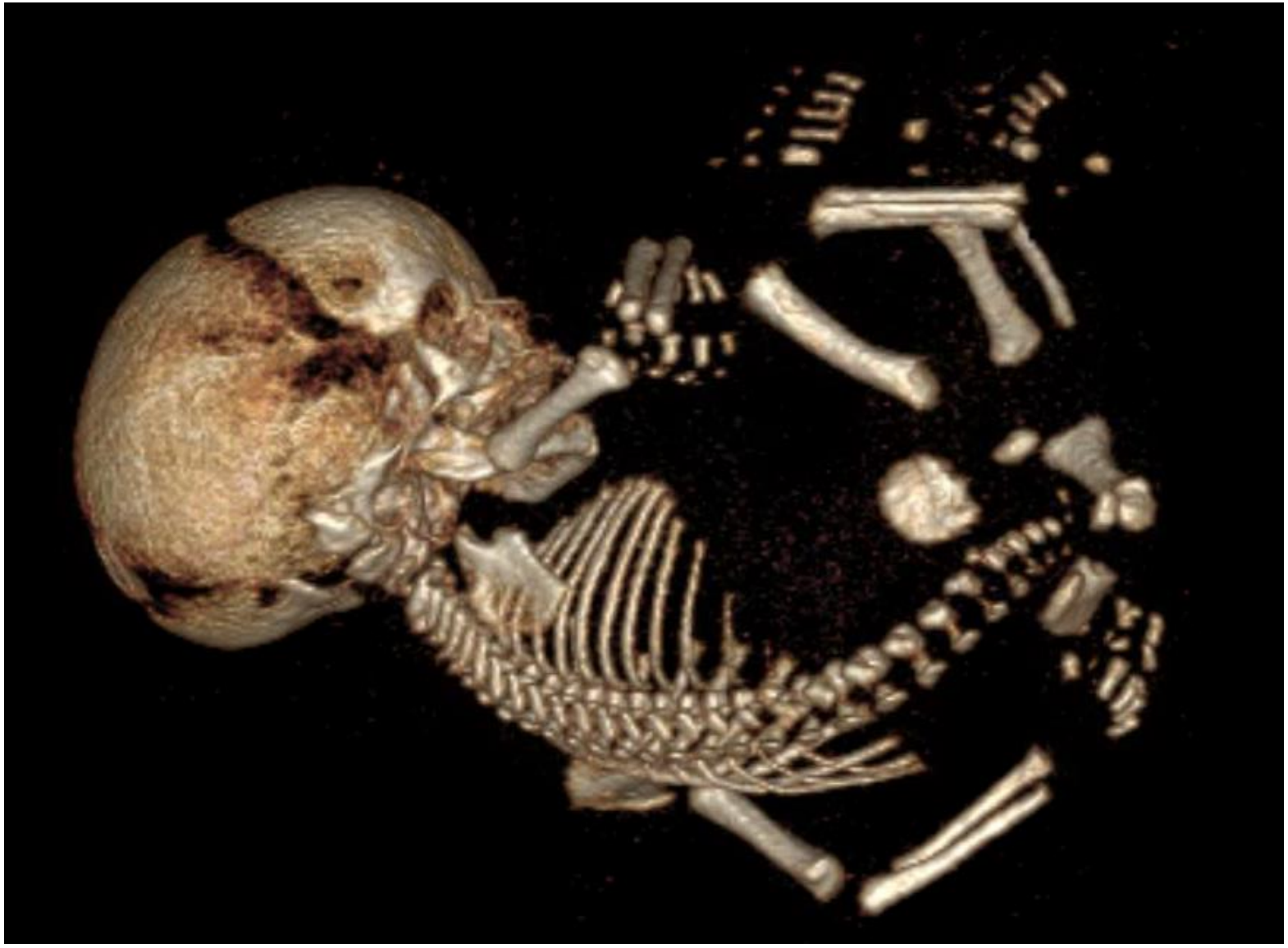
Emerging Techniques



- Fetal CT

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Nomenclature involved with Skeletal Dysplasias

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The Name Game

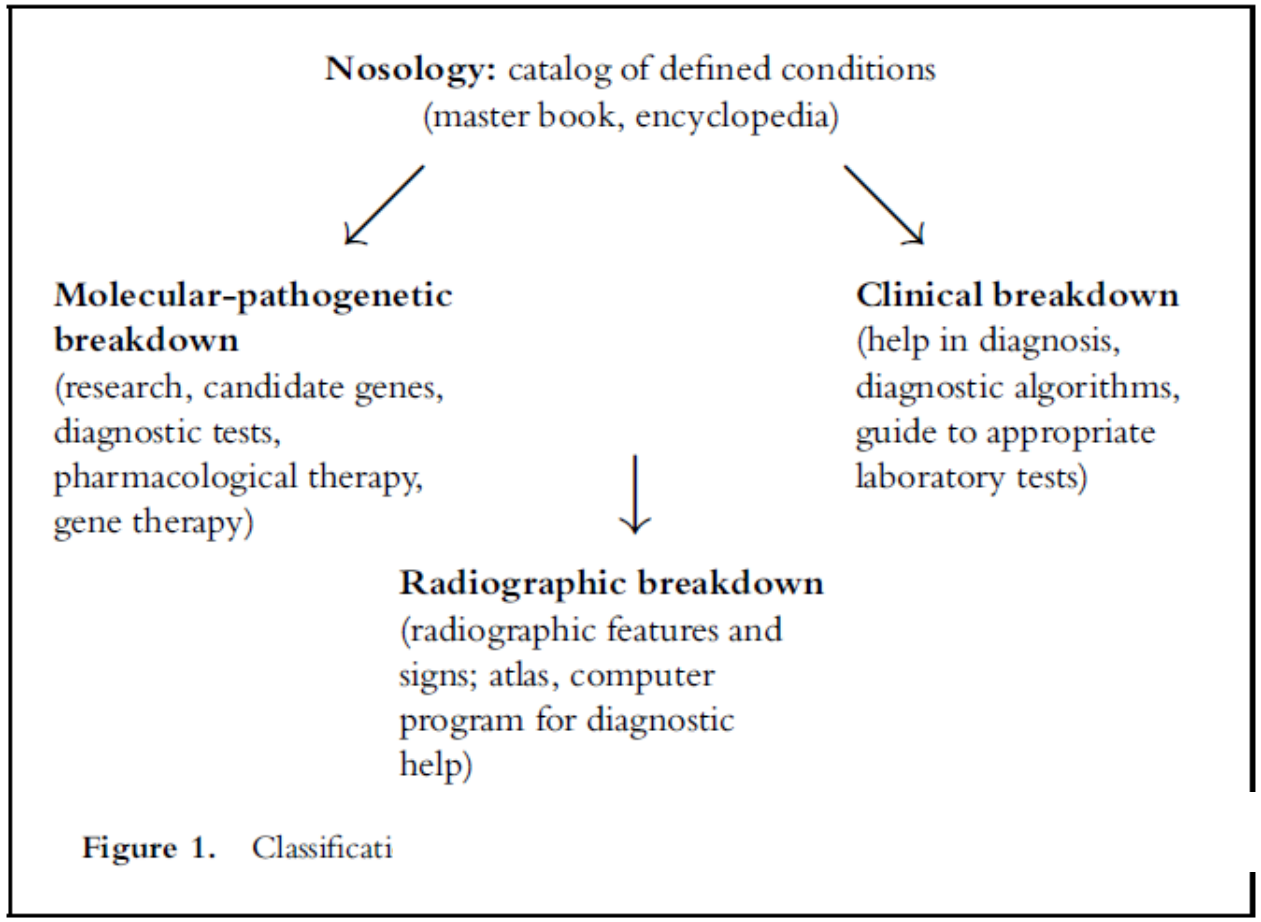

Figure 1. Classificati

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The International Skeletal Dysplasia Society

- International Nomenclature of Constitutional Diseases of Bone
  - Revisions:
    - 1970
    - 1977
    - 1983
    - 1992
    - 2001
    - 2005
    - 2010
    - 2015
    - 2019

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1970 International Nomenclature of Constitutional Diseases of Bone

- **Osteochondrodysplasias**
  - Abnormalities of cartilage and/or bone growth and development.
    - Defects of growth of tubular bones and/or spine
      - A. Identifiable at birth
      - B. Identifiable in later life
  - Disorganized development of cartilage and fibrous components of skeleton
  - Abnormalities of density of cortical diaphyseal structure and/or metaphyseal modeling
- **Dysostoses**
  - Malformation of individual bones singly or in combination.
    - Dysostoses with predominant axial involvement
    - Dysostoses with predominant involvement of extremities
  - Idiopathic Osteolyses
- **Chromosomal Aberrations**
- **Primary Metabolic Abnormalities** : Calcium /phosphorus ; Complex carbohydrates; Lipids ; Nucleic acids; Metals

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2001 Nosology

- Group 1: Defects in extracellular structural proteins
- Group 2: Defects in metabolic pathways -including enzymes, ion channel transporters
- Group 3: Defects in folding, processing and degradation of macromolecules.
- Group 4: Defects in hormone and signal transduction mechansisms
- Group 5: Defects in nuclear proteins and transcription factors
- Group 6: Defects In oncogenes and tumor-supressor genes
- Group 7: Defects in RNA and DNA processing and metabolism

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2006 Nosology

Sub classified into **37** Groups

1. FGFR3 group
2. Type 2 collagen group
3. Type 11 collagen group
4. Sulphation disorders group
5. Perlecan group
6. Filamin group
7. Short-rib dysplasia (SRP) (with or without polydactyly) group
8. Multiple epiphyseal dysplasias and pseudoachondroplasia group
9. Metaphyseal dysplasias
10. Spondylometaphyseal dysplasias (SMD)
11. Spondylo-epi(-meta)physeal dysplasias (SE(M)D)
12. Severe spondylodysplastic dysplasias
13. Moderate spondylodysplastic dysplasias (brachyolmias)
14. Acromelic dysplasias
15. Acromesomelic dysplasias
16. Mesomelic and rhizo-mesomelic dysplasias
17. Bent bones dysplasias
18. Slender bone dysplasias
19. Dysplasias with multiple joint dislocations
20. Chondrodysplasia punctata (CDP) group
21. Neonatal osteosclerotic dysplasias
22. Increased bone density group (without modification of bone shape)
23. Increased bone density group with metaphyseal and/or diaphyseal involvement
24. Decreased bone density group
25. Defective mineralization group
26. Lysosomal Storage Diseases with Skeletal Involvement (Dysostosis Multiplex Group)
27. Osteolysis group
28. Disorganized development of skeletal components group
29. Cleidocranial dysplasia group
30. Craniosynostosis syndromes and other cranial ossification disorders
31. Dysostoses with predominant craniofacial involvement
32. Dysostoses with predominant vertebral and costal involvement
33. Patellar dysostoses
34. Brachydactylies (with or without extraskeletal manifestations)
35. Limb hypoplasia–reduction defects group
36. Polydactyly-Syndactyly-Triphalangism group
37. Defects in joint formation and synostoses

Classification evolved from **purely clinical-pathological** descriptions to a nosology that **reflects the underlying molecular etiology** while **retaining the clinical distinction**

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AJMG AMERICAN JOURNAL OF medical genetics PART A

ORIGINAL ARTICLE

Nosology and classification of genetic skeletal disorders: 2019 revision

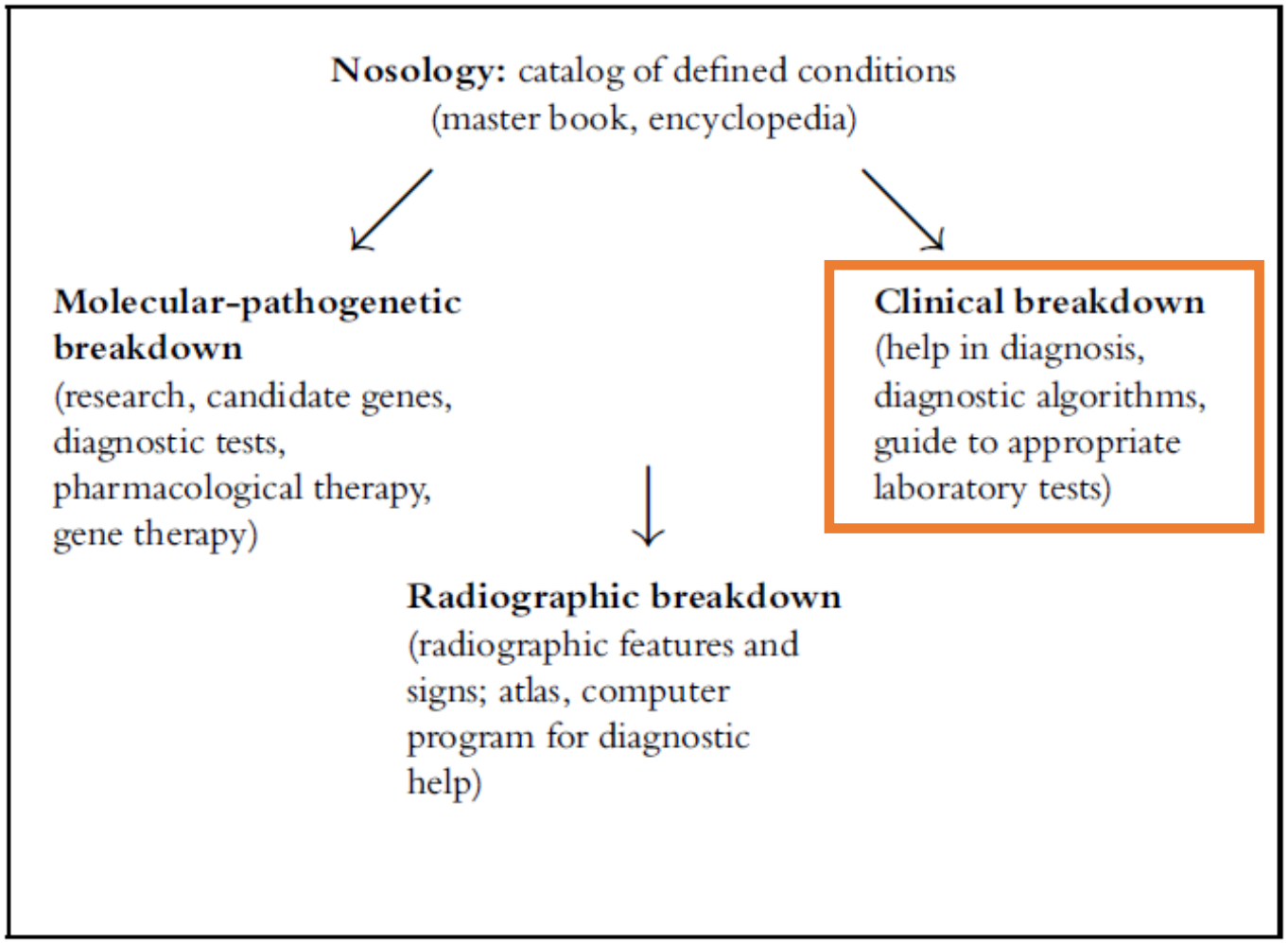
Geert R. Mortier, Daniel H. Cohn, Valerie Cormier-Daire, Christine Hall, Deborah Krakow, Stefan Mundlos, Gen Nishimura, Stephen Robertson, Luca Sangiorgi ... See all authors

First published: 21 October 2019 | https://doi.org/10.1002/ajmg.a.61366 | Citations: 267

Abstract

The application of massively parallel sequencing technology to the field of skeletal disorders has boosted the discovery of the underlying genetic defect for many of these diseases. It has also resulted in the delineation of new clinical entities and the identification of genes and pathways that had not previously been associated with skeletal disorders. These rapid advances have prompted the Nosology Committee of the International Skeletal Dysplasia Society to revise and update the last (2015) version of the Nosology and Classification of Genetic Skeletal Disorders. This newest and tenth version of the Nosology comprises 461 different diseases that are classified into 42 groups based on their clinical, radiographic, and/or molecular phenotypes. Remarkably, pathogenic variants affecting 437 different genes have been found in 425/461 (92%) of these disorders. By providing a reference list of recognized entities and their causal genes, the

Clinical Classification



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Proportionate vs Disproportionate

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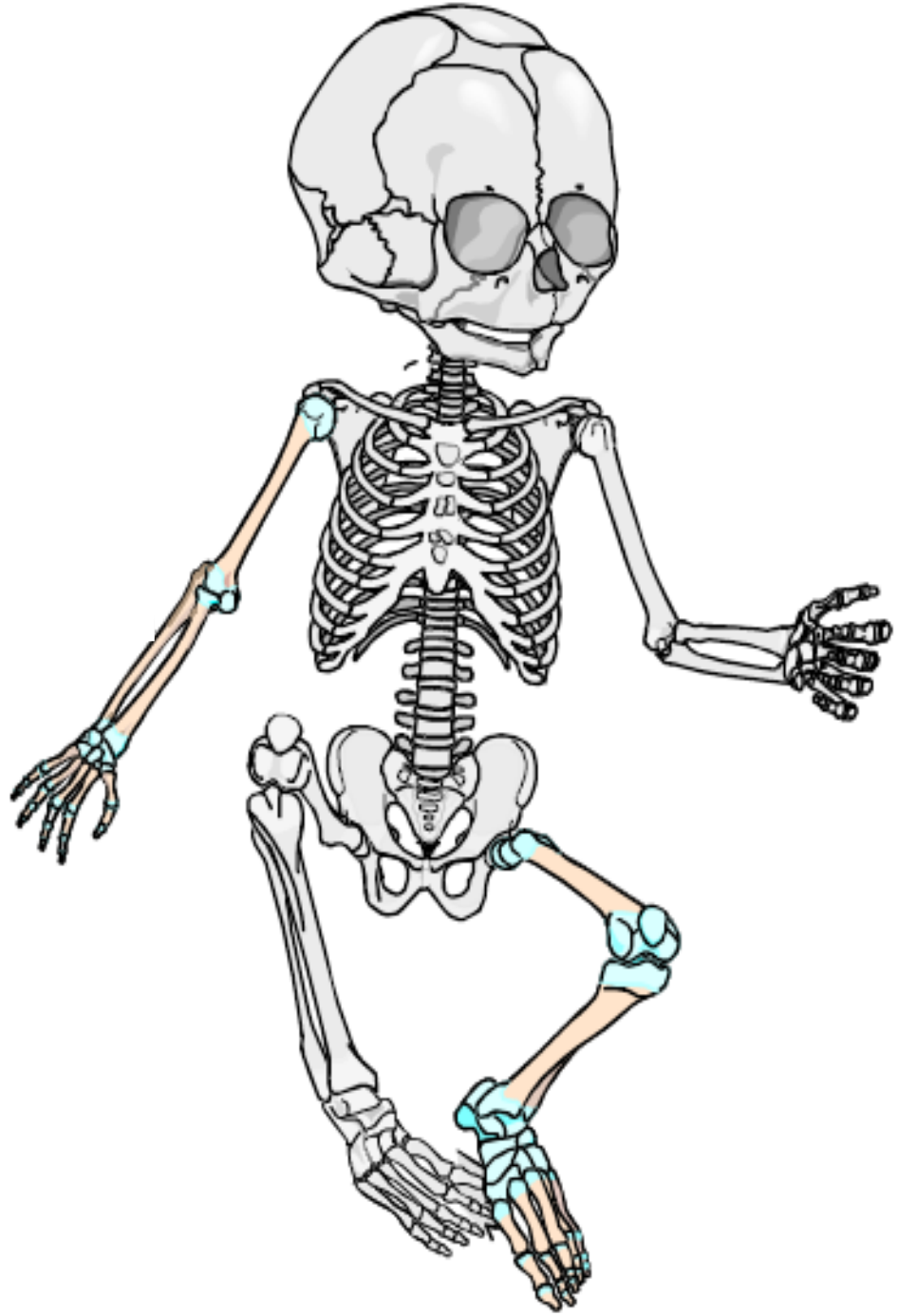

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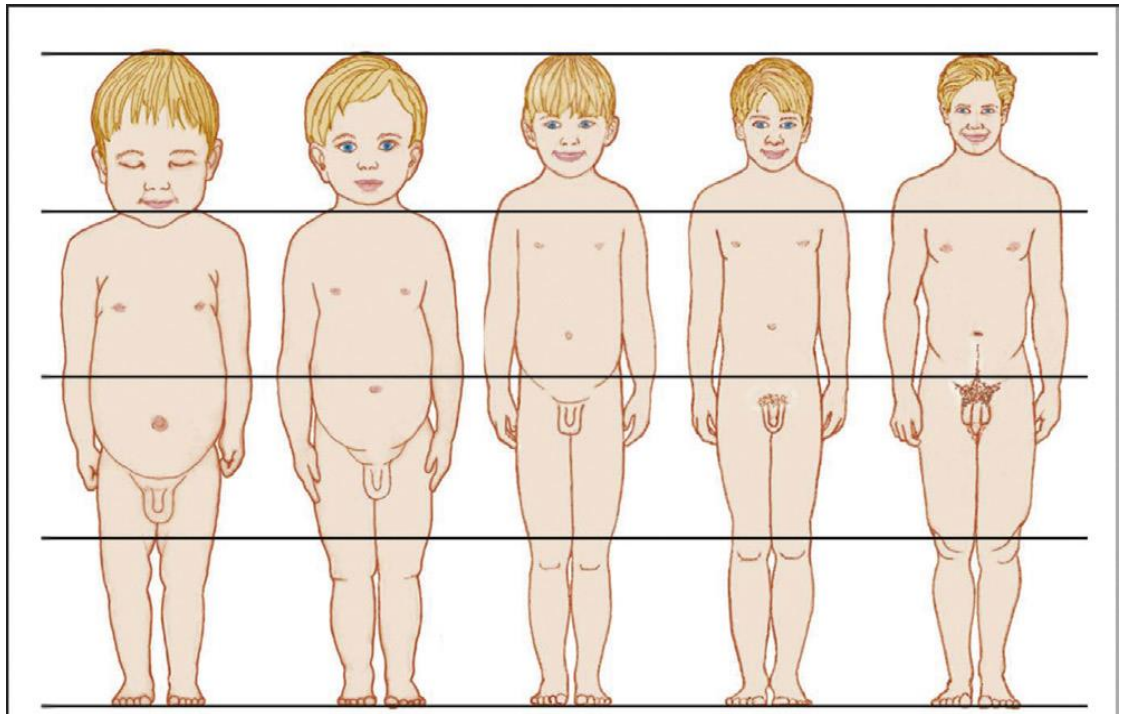
Clinical Assessment

- Key feature
  - **Disproportionate** short stature
  - Measure Upper : Lower segment ratio
    - 1.7 (infant); 1.1 (toddler); 0.95 adult



Leifer G. *Introduction to Maternity & Pediatric Nursing.* Philadelphia: Saunders; 2011:347–385, Fig. 15-2.

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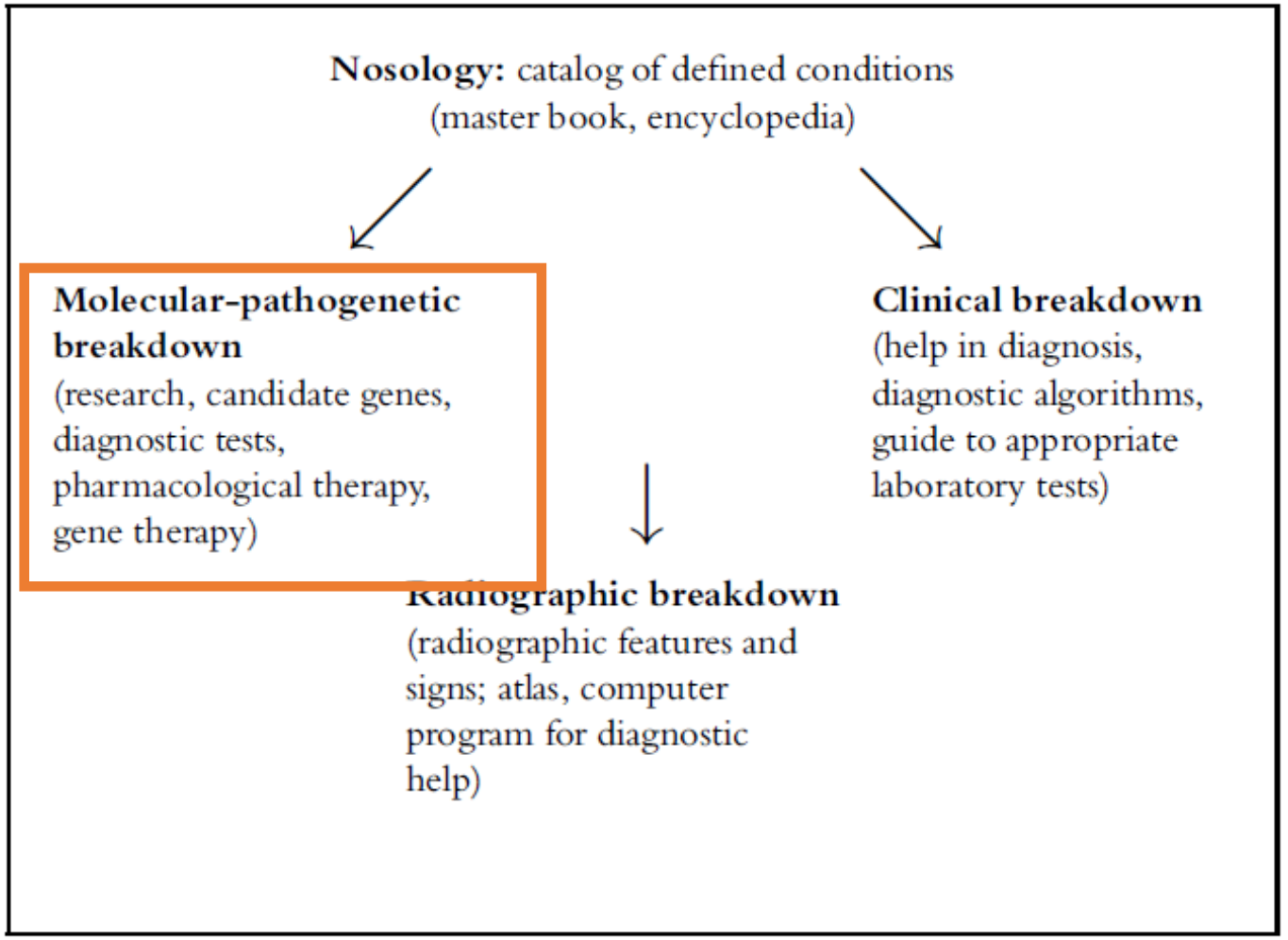
Molecular Classification



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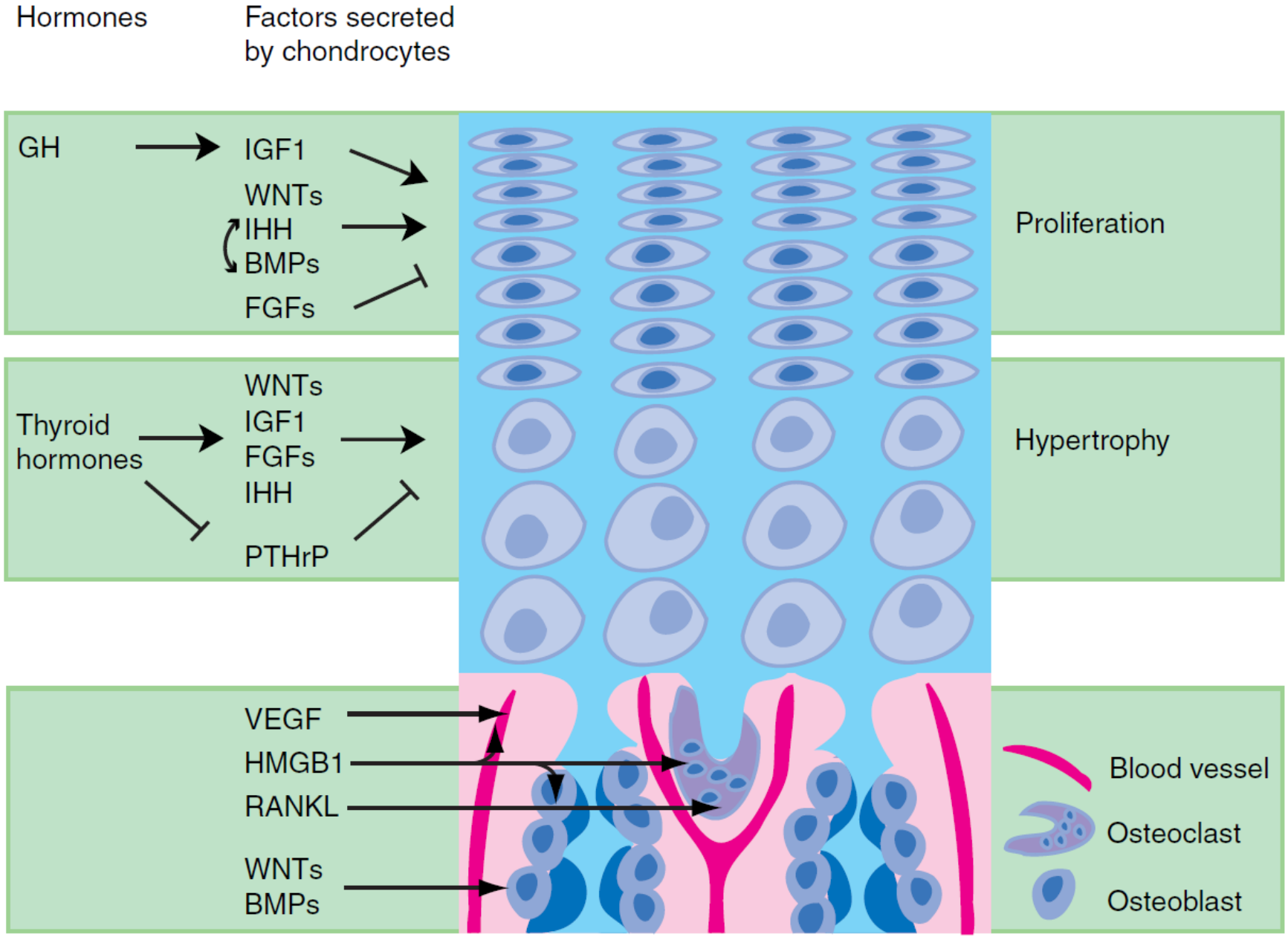

(Mackie et al 2011)

Group/name of disorder	Inheritance	Gene(s)	number	code	Notes
7. Filamin group and related disorders					
Frontometaphyseal dysplasia	XL AD AD	*FLNA* *MAP3K7* *TAB2*	305620 617137	1826	
Cardiospondylocarpofacial syndrome	AD	*MAP3K7*	157800	3238	
Melnick–Needles syndrome	XL	*FLNA*	309350	2484	Includes osteodysplasty
Otopalatodigital syndrome type 1 (OPD1)	XL	*FLNA*	311300	90650	
Otopalatodigital syndrome type 2 (OPD2)	XL	*FLNA*	304120	90650	
Terminal osseous dysplasia (TOD)	XL	*FLNA*	300244	88630	Includes digitocutaneous dysplasia
Atelosteogenesis type 1 (AO1)	AD	*FLNB*	108720 112310	1190 1263	Includes Boomerang dysplasia, Piepkorn dysplasia, and spondylohumerofemoral (giant cell) dysplasia
Atelosteogenesis type 3 (AO3)	AD	*FLNB*	108721	56305	
Larsen syndrome (dominant)	AD	*FLNB*	150250	503	
Spondylocarpotarsal synostosis syndrome	AR AD, AR	*FLNB* *MYH3*	272460	3275	
Frank-ter Haar syndrome	AR	*SH3PXD2B*	249420	137834	Includes Borrone dermatocardioskeletal syndrome
See also group 4 for recessive Larsen syndrome and group 20 for conditions with multiple dislocations					
8. TRPV4 group					
Metatropic dysplasia	AD	*TRPV4*	156530	2635	Includes "hyperplastic," lethal and nonlethal forms. Can also result from somatic mosaicism for a *TRPV4* mutation
Spondyloepimetaphyseal dysplasia, Maroteaux type (pseudo-Morquio syndrome type 2)	AD	*TRPV4*	184095	263482	Includes parastremmatic dwarfism (OMIM 168400)
Spondylometaphyseal dysplasia, Kozlowski type	AD	*TRPV4*	184252	93314	
Brachyolmia, autosomal dominant type	AD	*TRPV4*	113500	93304	
Familial digital arthropathy with brachydactyly	AD	*TRPV4*	606835	85169	
See also groups 4 and 13 for other forms of brachyolmia					
9. Ciliopathies with major skeletal involvement					
Chondroectodermal dysplasia (Ellis-van Creveld)	AR AR AR AR	*EVC1* *EVC2* *WDR35* *DYNC2LI1*	225500	289	See also Weyers acrofacial (acrodental) dysostosis in group 34
Short rib–polydactyly syndrome (SRPS) type 1/3 (Saldino–Noonan/Verma–Naumoff)	AR AR AR AR AR	*DYNC2H1* *IFT80* *WDR34* *WDR60* *DYNC2LI1*	613091	93270 93271	There is significant clinical and radiological overlap between SRP1/3 and ATD. Some forms of both remain unlinked to the known genes.
Asphyxiating thoracic dysplasia (ATD; Jeune)	AR AR AR	*DYNC2H1* *DYNC2LI1* *WDR34*	613091	474	Dynein motor

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ORIGINAL ARTICLE

AMERICAN JOURNAL OF medical genetics PART A WILEY

Nosology and classification of genetic skeletal disorders: 2019 revision

437 Genes

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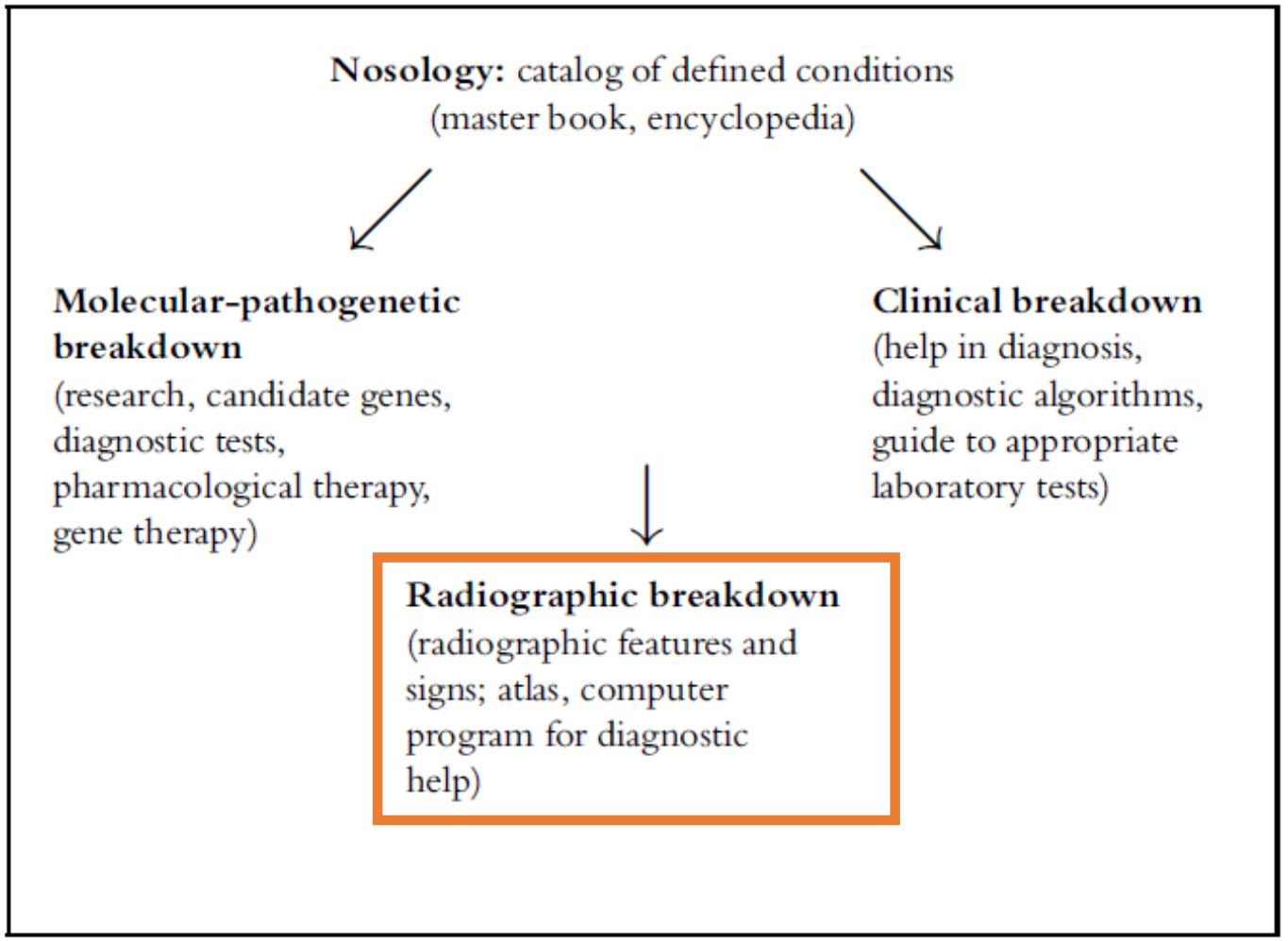
Radiological Classification



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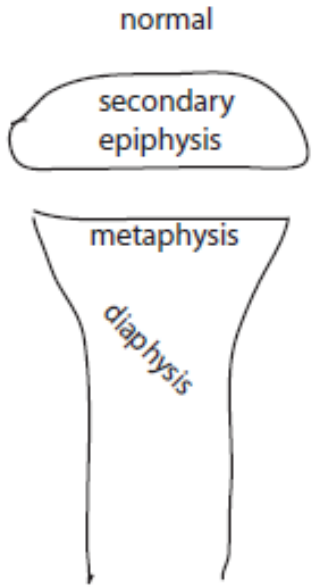
Radiology



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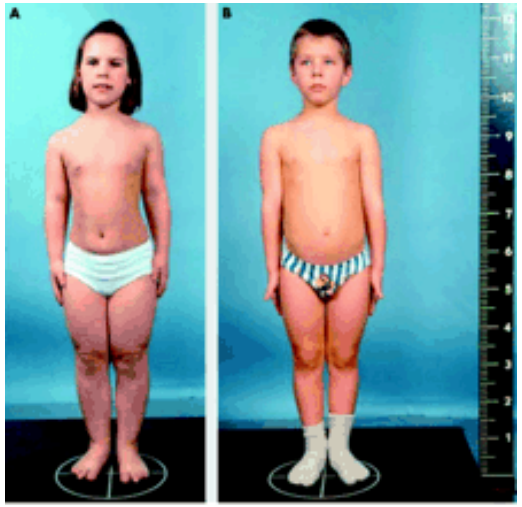
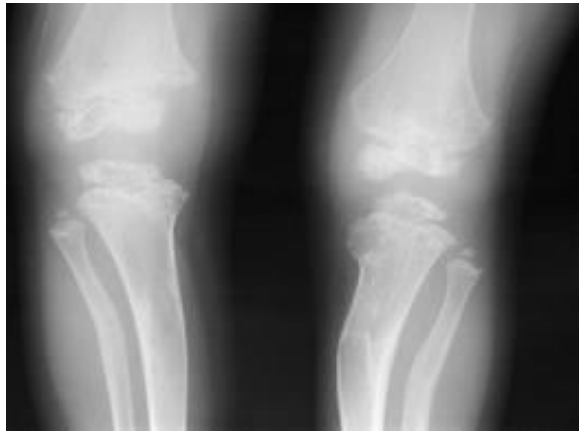
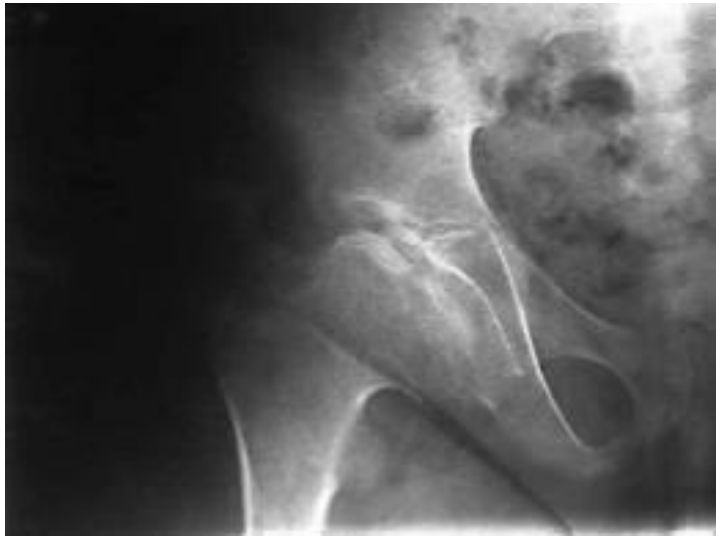
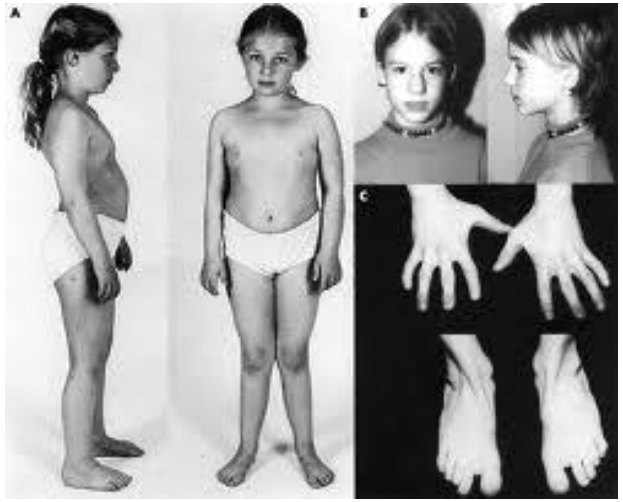
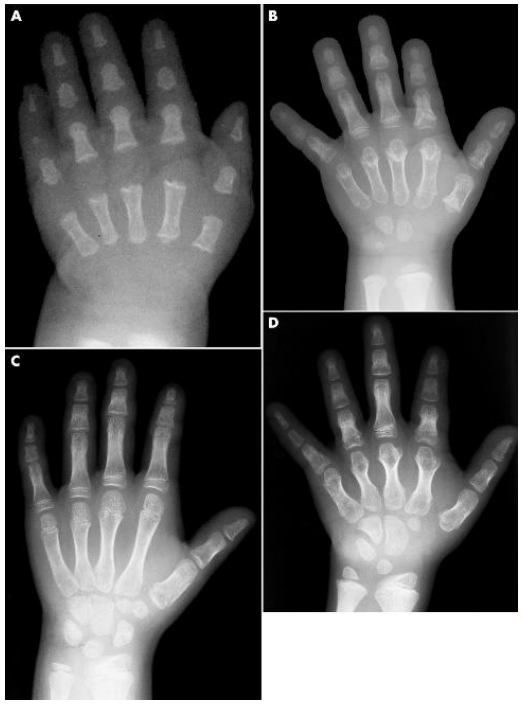
Epiphyseal Dysplasias











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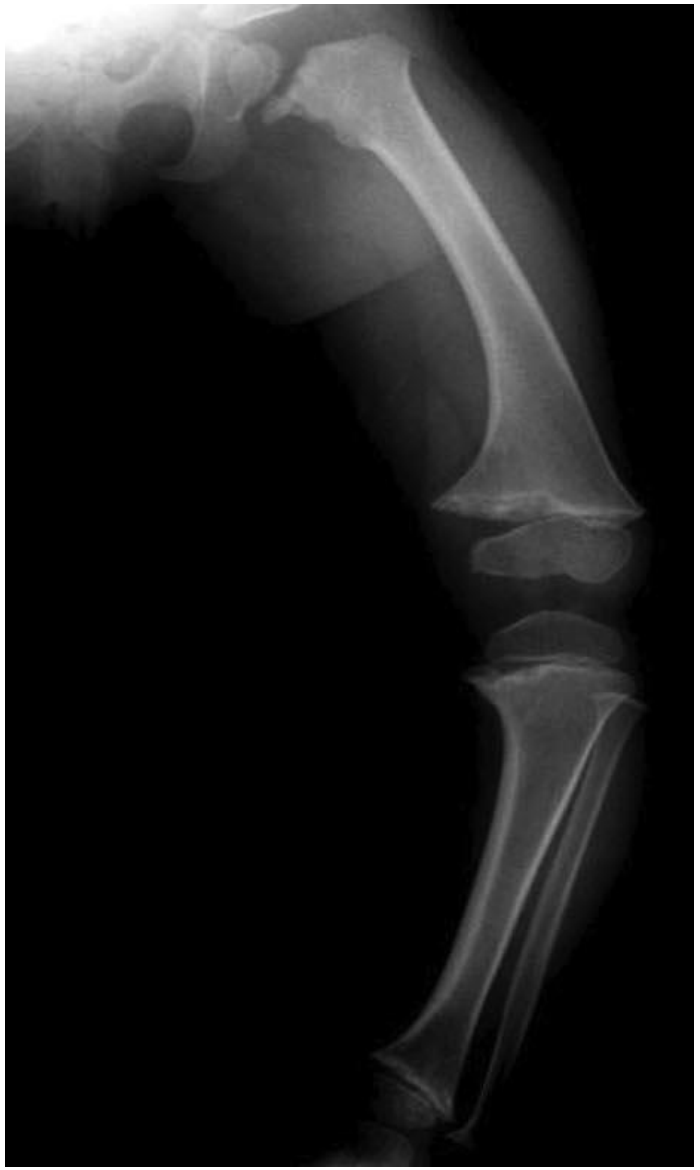
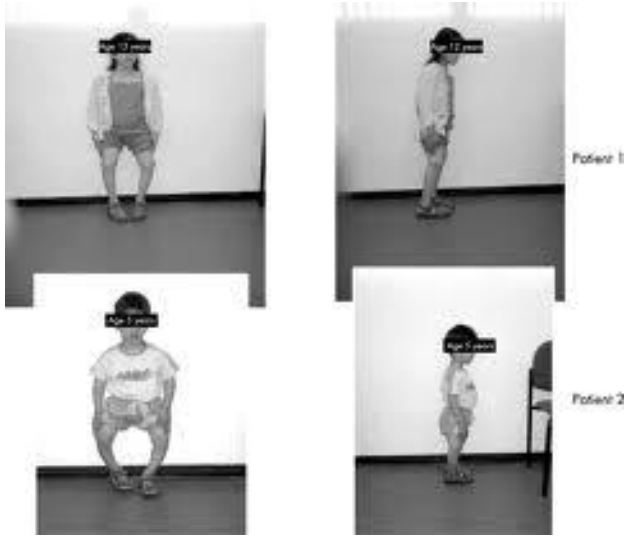
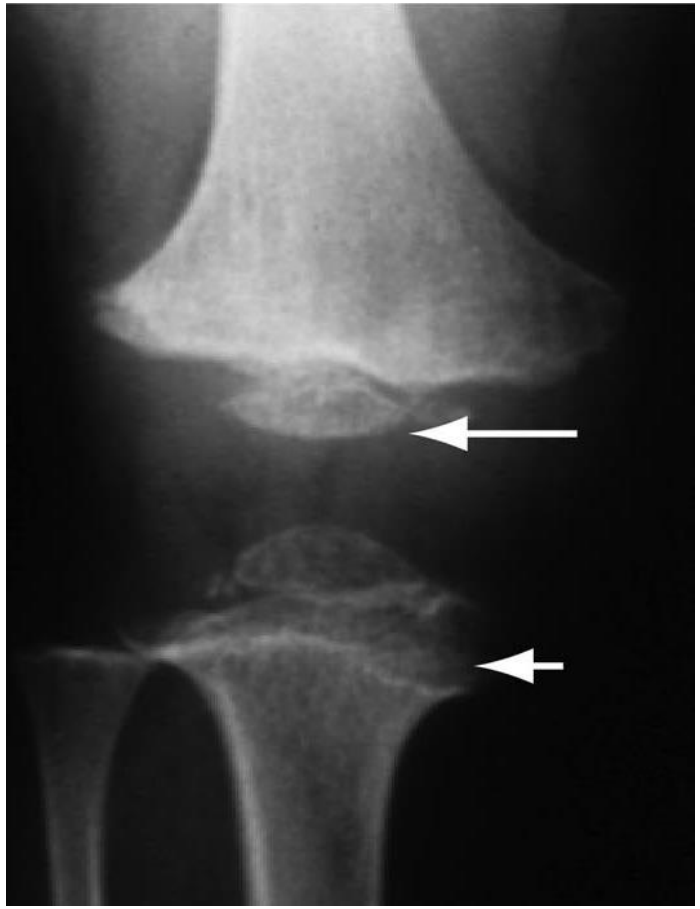
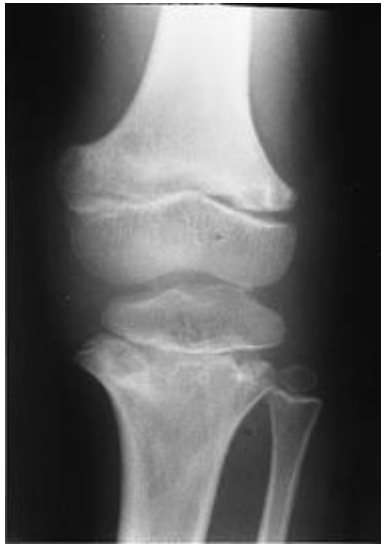
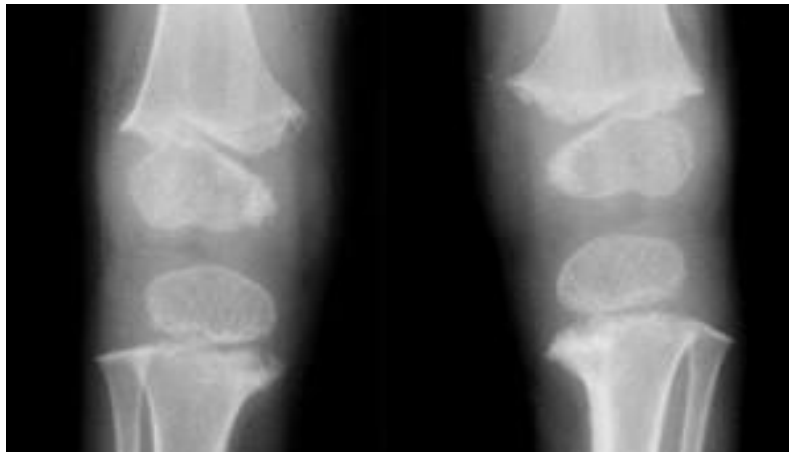
Metaphyseal Dysplasia



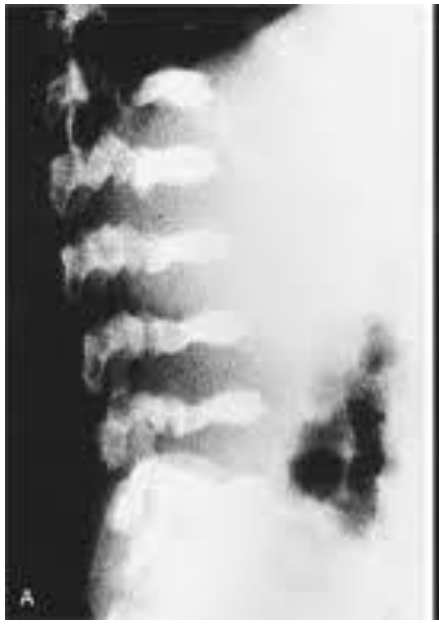


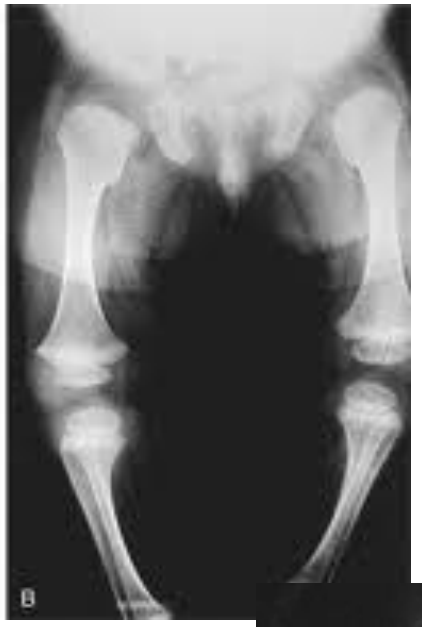


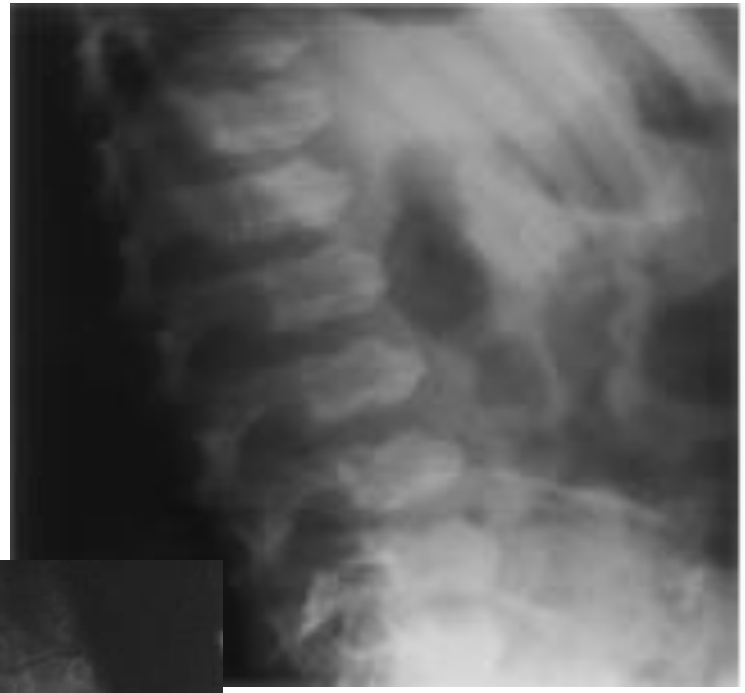


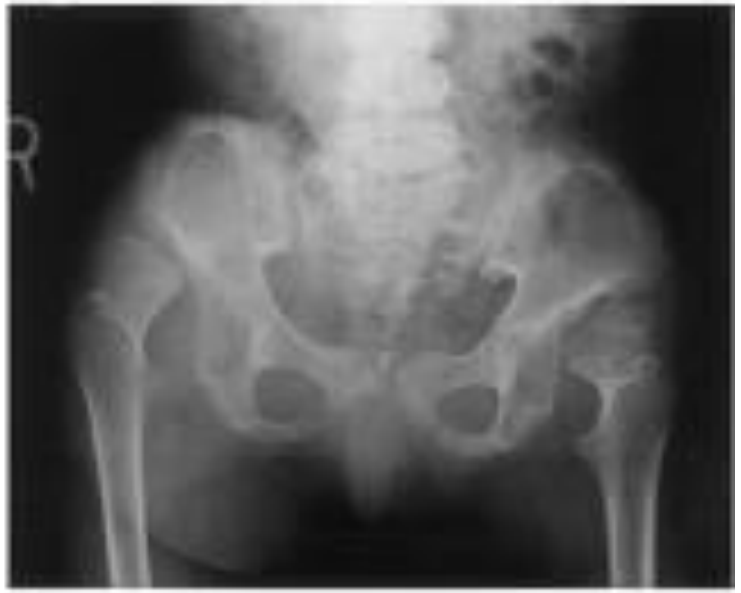


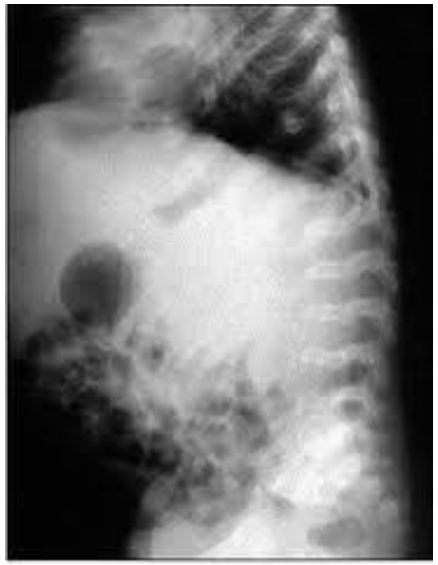
Spondylo + (more often not isolated)

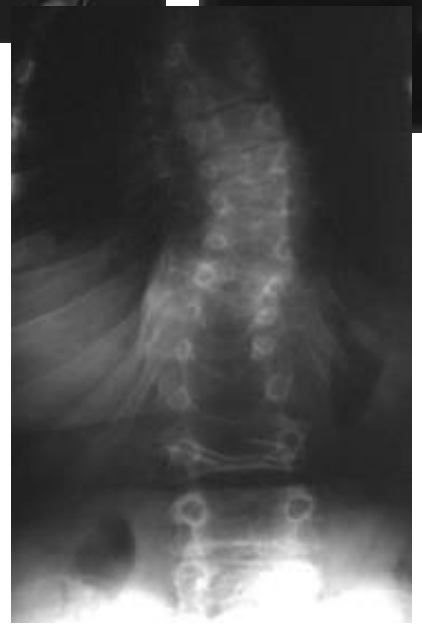












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A Few Tricks for diagnosis of severe ‘common’ (lethal) skeletal dysplasias

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Differential of Curved Femurs

- Thanatophoric Dysplasia
- Osteogenesis Imperfecta (severe forms)
- Hypophosphatasia (congenital subtype)
- Campomelic Dysplasia

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‘Lethal’ Skeletal Dysplasias

- Not viewed as lethal in all cases but generally prognosis is poor
- Most common:
  - Thanatophoric dysplasia
  - Osteogenesis imperfecta (type 2)
  - Achondrogenesis
- Three disorders represent 40-60% of all lethal skeletal dysplasias

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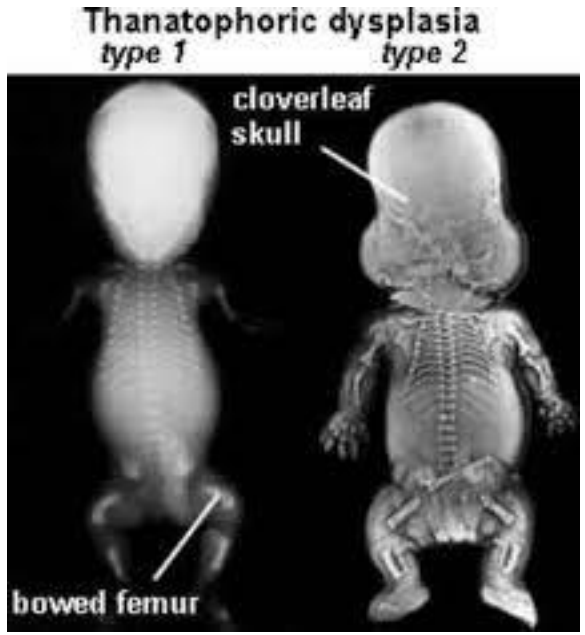
Thanatophoric

- Early severe micromelia
- Femur: Curved - TD1
  : Straight - TD2

(TD2 – clover leaf skull)

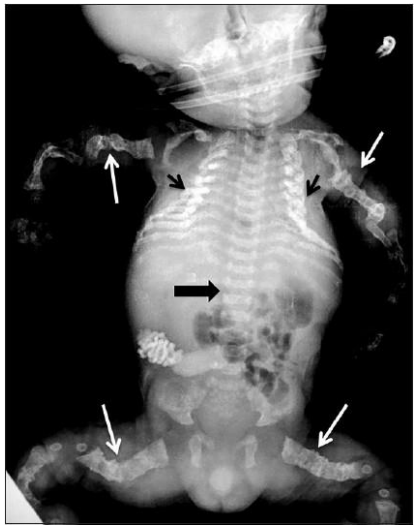
- Narrow chest with relatively large abdomen



Cohen MM. Oxford University Press

Osteogenesis

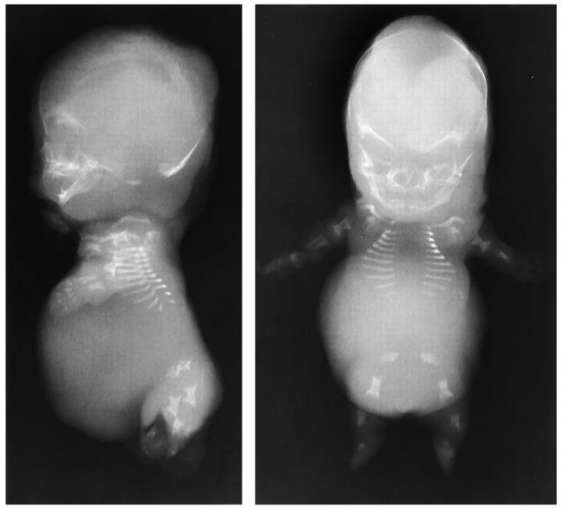
- "Type II"
- Calvarial Ossification spared
- Ribs: Beaded or Crumpled
- Short / Crumpled long bones
- Poor Px = hyperextension of neck



IMAGES IN CLINICAL NEONATOLOGY(2015) 4:1, 60-61

Achondrogenesis

- General lack of mineralization
- Non-ossification of sacrum, pubis, talus, vertebrae
- Extreme shortening of limbs
- Frequent Hydrops



https://doi.org/10.1074/jbc.270.4.1747

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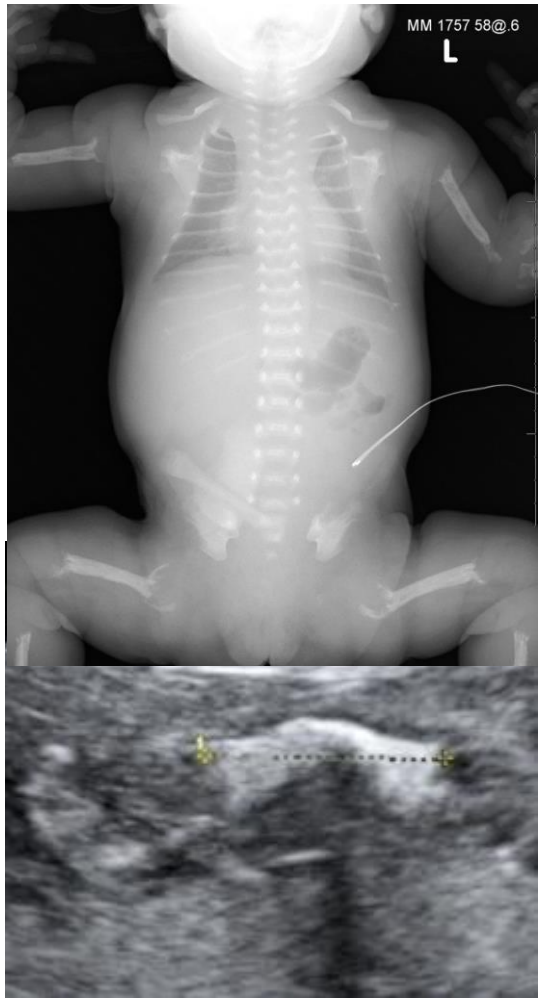
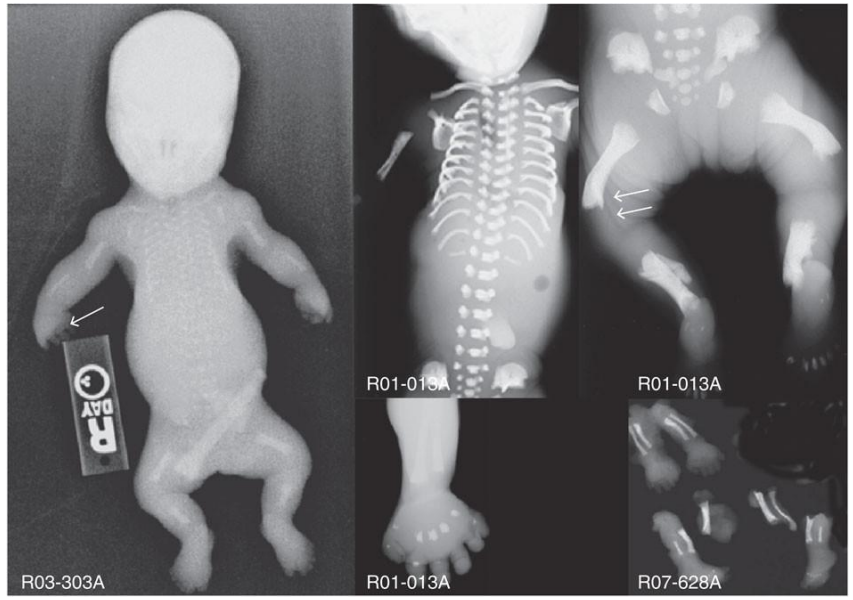
Hypophosphatasia

- Poor mineralization
- Metaphyses = “V” shaped



Short Rib Polydactyly

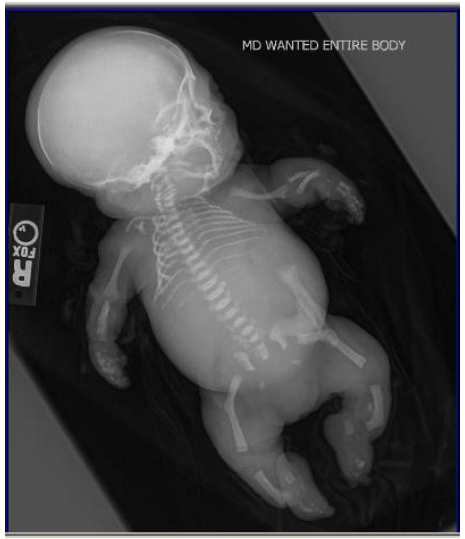
- Extremely short ribs
- Polydactyly (not always)
- “Ciliopathy” :
  - Renal cysts
  - ‘Heterotaxy’ – heart / brain
  - Bowel malrotation
- Tibia often appear oval



https://doi.org/10.1038/ncomms8092

Campomelic

- Hypoplastic scapulae
- Non-ossification of thoracic vertebral pedicles
- Bowed femora
- Cardiac / Renal malformation



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https://doi.org/10.1038/ncomms8092

Thank You

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- Pediatric neonatologist and perinatologist at Children's Wisconsin since 2015
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- Board certified in neonatal-perinatal medicine
- Earned his medical degree at University of Chicago
- Completed his neonatology McGaw Medical Center of Northwestern University and Northwestern University
- Dr. Luethner sees patients at the Milwaukee Hospital Campus

Erin Rholl, MD, MA

- Pediatric neonatologist and pediatric palliative care physician at Children’s Wisconsin since 2022
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- Board certified in pediatrics and neonatal perinatal medicine
- Earned her medical degree at XXXX
- Completed her Neonatal Perinatal Medicine fellowship at the Medical College of Wisconsin and a Hospice & Palliative Medicine Fellowship, Children's National Hospital, Washington, DC
- Dr. Rholl sees patients at the Milwaukee Hospital Campus

Carrie Hecox, APNP

- Pediatric nurse practitioner at Children's Wisconsin since 2017
- Certified in pediatric nurse practitioner
- Completed her graduate degree at Marquette University
- Carrie sees patients at the Milwaukee Hospital Campus

Supporting Families Through an Uncertain Pregnancy: Evolution and Current State of Perinatal Palliative Care

Erin Rholl MD MA, Steven Leuthner MD MA, Carrie Hecox APNP

Fetal Innovations: Conversation with the Experts

November 9, 2022

Disclosures

We have no relevant financial interests/relationships to disclose.

Objectives

- Identify a shared understanding of palliative care
- Describe some history of perinatal palliative care
- Summarize the experience at CW Fetal Concerns Center
- Prescribe the current and future state of perinatal palliative care

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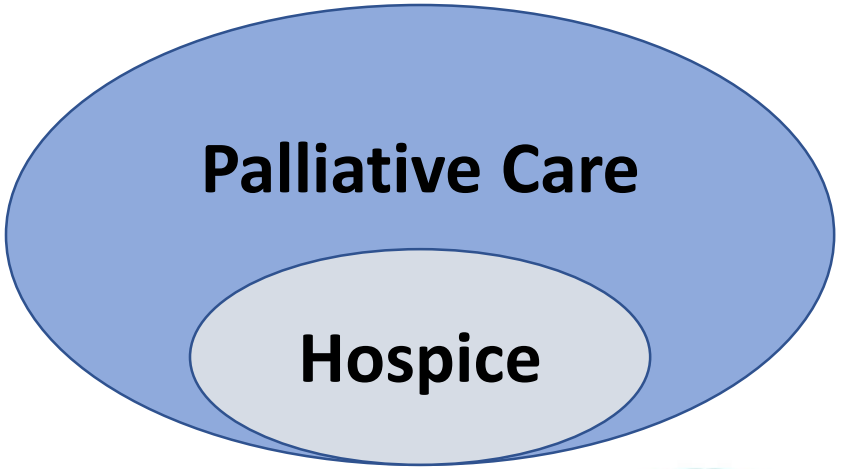
What is Palliative Care?

- Focused on improving the QOL of patients and families dealing with a life-limiting illness via an interdisciplinary care model:
    - Preventing and relieving suffering
    - Celebrating life while recognizing death as a natural process
    - Neither speeding up nor prolonging death
    - Additional layer of support
- Appropriate at any stage of illness



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Pediatric Palliative Care

- Children with a life-limiting, life-threatening, or serious illness.
- Begins at diagnosis and continues regardless of whether a child receives disease directed treatment
- Services can be provided with curative or intensive therapies
  - Affordable Care Act: Concurrent Care for Children

WHO: Palliative Care
Carter, 2022

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Pediatric Death is Difficult

- Conflict between the expected life sequence
- Miracles of NICU & PICU
  - Advances in medicine & surgery
  - Innovation & marketing raise expectations

- More children die in the perinatal period than any other time in childhood…

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Perinatal Death

- Congenital anomalies #1 cause of infant mortality
- Many anomalies diagnosed before birth
- Leads to grief crisis and numerous, critical decisions
- Acute grief reaction comparable if terminate, stillborn or neonatal death
- Clearly, they need help and support

Lloyd J Br Med J 1985;290(6472):907-9
Sherry LM, et al. NCHS Data Brief 2021; 427

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A Brief History of Perinatal Palliative Care

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Development of Perinatal Palliative Care

- *The Application of Hospice Concepts to Neonatal Care* Whitfield, (1982)
  - Decision-making process
  - Family Room - physical environment
  - Involvement of family
  - Hospice training of NICU nurses

- *Home Death and Hospital Follow-up of the Dying Infant* Mangurten, MD. (1990)
  - 6 patients (Potter's sequence, MM and Hydrocephalus, T18, Triploidy and Werdnig-Hoffman)
  - Home care by intermittent pediatric house officer
  - Death pronounced in ER

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Development of Perinatal Palliative Care

- Appreciation of grief experienced after pregnancy termination (Zeanah, 1993)
  - Just as intense after termination as still birth and neonatal death

- Perinatal Hospice (Hoeldke, Calhoun 2001)
  - As an alternative to pregnancy termination

- Fetal Concerns Program (Leuthner S, Jones EL 2007)
  - Model for perinatal palliative care

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Fetal Concerns Center of Wisconsin

- Began as a cooperative effort: Children's Wisconsin, Froedtert Hospital, & The Medical College of Wisconsin Departments of Pediatrics & OB/GYN
- Goal: support all pregnancy related diagnosis in which the baby should be at CHW
- Has evolved administratively and now located in CW

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Froedtert & Medical College

Children's Hospital of Wisconsin

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Supporting Prenatal Diagnoses; A Team Approach

- A team approach to cover *all aspects* of prenatal care, delivery, and postnatal management

- "The Team . . .composition should extend beyond midwives and obstetricians to include pediatricians, surgeons, radiologists, sonographers, general practitioners, bereavement counselors, as well as representatives of parent groups with experiences of fetal abnormality."
    - Wilcox. Pediatric Clinics of North America 1993
    - Madsen. *Neurosurgery Clinics of North America* 1998
    - Nichols. *Pediatrics* 1996
    - Marteau. Prenatal Diagnosis 1995

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Neonatology

Perinatology

Referring MD

Nurse Coordinator

Poison Control Center

Fetal Echo CV

Genetics

Radiology

General Surgery

Urology/ Nephrology

ENT

Orthopedics

Palliative Care Program

Neurology/ Neurosurgery

Infectious Disease HIV

Support Services

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Support Services

- Child-life specialist
- Tours
- Social work
- Financial counselor
- Spiritual care
- Lactation consultant
- Research coordinators
- RTS and bereavement counselors
- Palliative care team

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FEATURE ARTICLE

Fetal Concerns Program

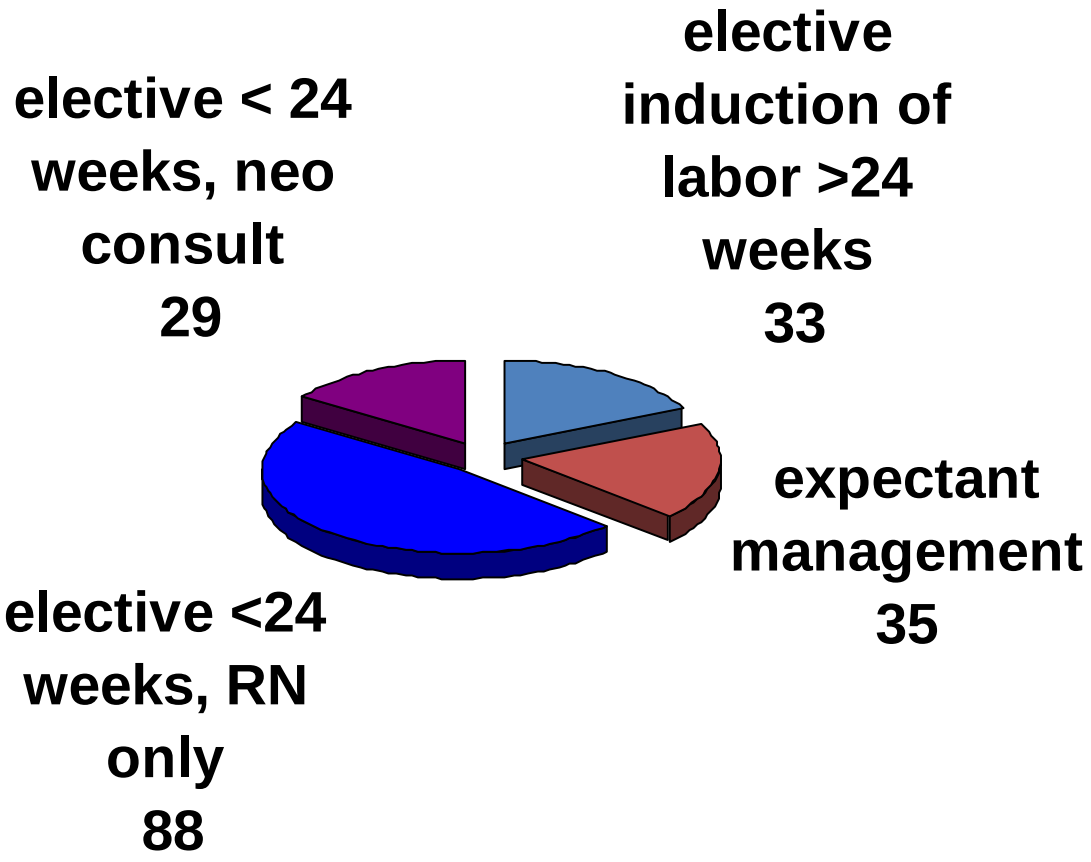
A Model for Perinatal Palliative Care

Leuthner, Steven MD, MA; Jones, Emilie Lamberg BSW, RN

Author Information

MCN, The American Journal of Maternal/Child Nursing: September 2007 - Volume 32 - Issue 5 - p 272-278

FCC Palliative Consultations & Pregnancy Decision-Making



Total of 185 (13.7%) of 1354 FCP referrals, 09/2000-2007

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CW Palliative Consultation Outcomes

- Advance care planning requests
  - 67% of families chose comfort care
  - 25% requested initial resuscitation & evaluation after birth
  - All patients had personalized care plans documented
- 23% of the time Palliative care team present at birth
- 51% stillborn and 49% born alive
- Of those born alive, 76% died - from minutes to 4.5 months
- 2 infants died in the NICU
- 3 died at home with hospice care
- 4 infants are still living?

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Current State of Perinatal Palliative Care

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What Differentiates Perinatal Palliative Care?

- Uniquely consider two patients: mother and baby
- Care plans weigh the needs of both
- Starts during pregnancy and continues after the birth

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Perinatal Palliative Care

- Interdisciplinary care for families that end or continue a pregnancy

- Support parents during disrupted pregnancy & parent narratives
  - What does it mean to be a parent now?

- Assist in complex medical decision making
  - Decisions surrounding pregnancy, delivery & postnatal care
  - Explore parent value systems → goal concordant care

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Biggest Decisions at the Worst Time

- What we expect the families to do:
  - Become fully educated in a rare medical condition
  - Explore and potentially challenge their (& their partner's) spiritual & ethical beliefs
- When?
  - When their brain is physiologically in shock
  - When they are grieving
  - Exhausted, ill, in pain
  - And often, very quickly
- How?
  - Relationship-based care

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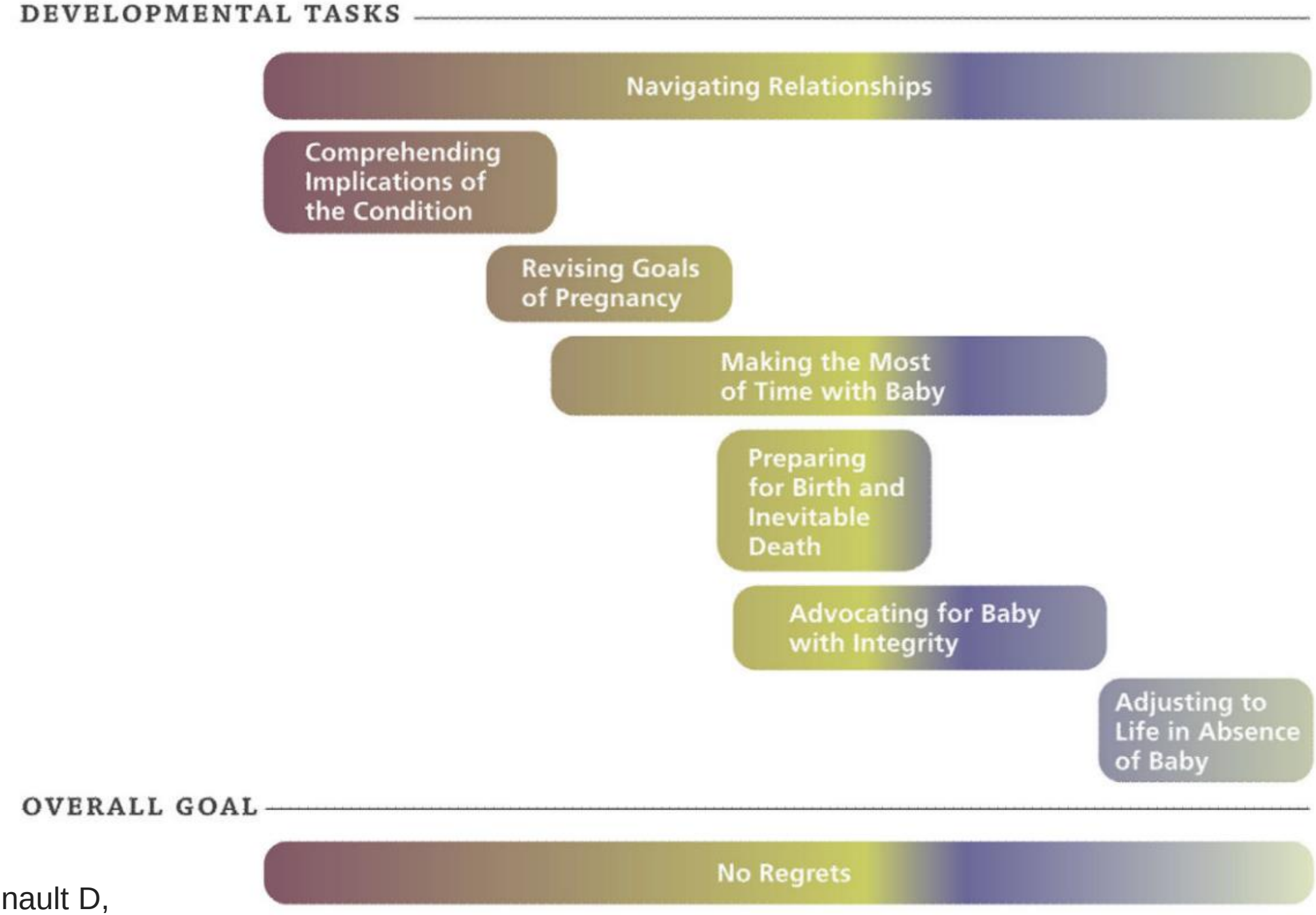

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Côté-Arsenault D,

Creating a Birth Plan

- Tool assisting with communication and advocacy
- Give parents sense of control in an uncertain situation
- Exploring parent values, hopes, worries → goal concordant plan

Whitford, 2014

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Components of a Birth Plan

Advanced Care Planning:

- Environment and site of care
- Care of newborn
- Family-centered care
- Spiritual/psychosocial care
- After death issues

Leuthner SR. Fetal Palliative Care. *Clin Perinatol* 2004: 31; 649-665

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Environment and Site of Care

- Location not as important as
  - “mind set”
  - People present
- Attitude of staff & desire to be present
  - Avoid judgment, abandonment, or “giving up”
  - Balance with privacy
- Prenatal discussion should include possibility of home with hospice

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Consistency in Obstetrical & Neonatal Plan

- Goals for family help determine plan
  - Lowest risk for mother and future pregnancy
  - Avoid stillbirth
  - Aggressive obstetrical monitoring & neonatal resuscitation for further evaluation

- Delivery options may include
  - Termination
  - Early induction for maternal health with palliative care
  - Expectant management and palliative care
  - Vaginal delivery vs. C/S for liveborn infant

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Newborn Care

- Address parameters of treatment initiation, evaluation and potential withdrawal

- In delivery room
  - Comfort care from the start
  - Transitional support to offer time
  - Aggressive resuscitation and trial of Rx

- In the NICU
  - Intervention limits (i.e., no ECMO, no dialysis)

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Newborn Care

- Prenatal anticipatory guidance – how will the baby die and what does it look like?
- Avoid predicting time to death
  - Share a reasonable time frame
  - Celebrate whatever time available
- Pain and symptom management
  - Medications, skin care, feedings
- Elements of normal newborn care
  - Eye ointment, Vit K, bilirubin levels

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Family Centered Care

- Assess who constitutes family
- Sensitive to cultural, ethnic, and religious beliefs
- Empower family choices & participation, including siblings
- Provide privacy & be empathetic
- Call child by name, use touch when appropriate
- Continue support until family leaves

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Memory Making & Bereavement

- Bereavement Photography
- Plaster Molds
- Memory Books & Boxes
- Bereavement Coordinator
  - Follow up phone calls & cards
  - Resolve Through Sharing
  - Community support services

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Spiritual/Psychological Care

- Address religious preferences and needs
- Baptism or blessings?
- Help personal pastoral support to be present
- Planning memorial service, cremation or burial before birth is supportive
- Be present at these events

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After Death Issues

- Further diagnostic information to collect?
  - Cord blood
  - Skin biopsy
  - Autopsy – 10-40% with added or new info
  - Valuable in:
    - Confirmation
    - Future planning
    - Grief support/alleviate guilt
    - Altruism

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Addressing Hope

- Parents have many hopes
  - Hope that we are wrong
  - Hope their child will be ok
- They can hold these hopes while appreciating the diagnosis

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REMAP

Reframe medical understanding and prognostic awareness

- Help family explore "What does all of this mean?"
- Acknowledge uncertainly and balance of "hope and reality"

Expect emotion

- Ask open ended questions (i.e., what are you most worried about?)
- Validate and respond to emotion. Be okay with silence

Map out value system family uses to makes decisions

- Inquire about faith, spirituality, religion
- Help to put those values in context of child's care (i.e., what is most important to you when thinking about how you want the team to care for your child?)

Align with values and hopes

- Reflect and summarize what we are hearing
- Seek clarification and verification

Propose a plan

- Suggest how identified goals may be achievable
- "I am hearing you ask that we create a plan that focuses on _____ for Baby's care after delivery" (time, comfort, better understanding diagnosis, etc.)

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Cortezzo, 2020

Delivery Location

Primary perinatal palliative care

- Primary pediatrician of family practice provider
- Nursing staff and/or care coordinator
- Access to bereavement care (may be community based)

Secondary perinatal palliative care

- Primary subspecialists
- Care coordinator
- Access to social & spiritual care providers with peds experience
- Access to consultant in peds palliative care

Tertiary perinatal palliative care

- On-site social services, spiritual care, child life, child psych & other interdisciplinary palliative care specialists
- On-site specialists in peds palliative care
- Research support staff
- Education support staff

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Leuthner & Jones, 2018

Experiences of Other Programs

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J Perinatol. 2021 Sep;41(9):2196-2200. doi: 10.1038/s41372-021-00966-2. Epub 2021 Feb 17.

Outcomes following perinatal palliative care consultation: a retrospective review

Megan H Tucker [1], Kelstan Ellis [2], Jennifer Linebarger [2]

Children's Mercy - Fetal Health Center

- PaCT met 430 mothers
- 390 live-born infants
- 218 survived > 1 year
- 109 children still alive
- 96 children discharged from the program

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Prenatal diagnostic category	Number of perinatal consults, $n = 436$ (%)	Example conditions
Cardiac	172 (39.4%)	Hypoplastic left heart syndrome
Congenital anomalies	99 (22.7%)	Congenital diaphragmatic hernia
Neurological	60 (13.8%)	Encephalocele
Genetic	49 (11.2 %)	Trisomy 18, trisomy 13
Renal	47 (10.7%)	Bilateral renal dysplasia
Pulmonary	6 (1.3%)	Congenital pulmonary airway malformation
Other	3 (0.6%)	Severe intrauterine growth restriction, sacrococcygeal teratoma

- Neonatologists reported positive changes
  - Longitudinal partnership extending beyond the NICU
  - Involvement across a spectrum of diagnoses
  - Partnership in delivering bad news

Tucker, 2021

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Randomized Controlled Trial > Cardiol Young. 2018 Apr;28(4):561-570.
doi: 10.1017/S1047951117002761. Epub 2018 Jan 10.

A randomised trial of early palliative care for maternal stress in infants prenatally diagnosed with single-ventricle heart disease

Hayley S Hancock [1], Ken Pituch [2], Karen Uzark [3], Priya Bhat [4], Carly Fifer [3], Maria Silveira [5], Sunkyung Yu [3], Suzanne Welch [3], Janet Donohue [3], Ray Lowery [3], Ranjit Aiyagari [3]

- Both groups with high anxiety
- Early palliative care group →
  - Decreased anxiety
  - Higher positive reframing
  - Improved communication and family relationships

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Take Home Points

- Families receiving perinatal palliative care benefit from a team approach
- Perinatal palliative care is appropriate for parents desiring a range of interventions
- Earlier palliative care involvement can help parents and staff

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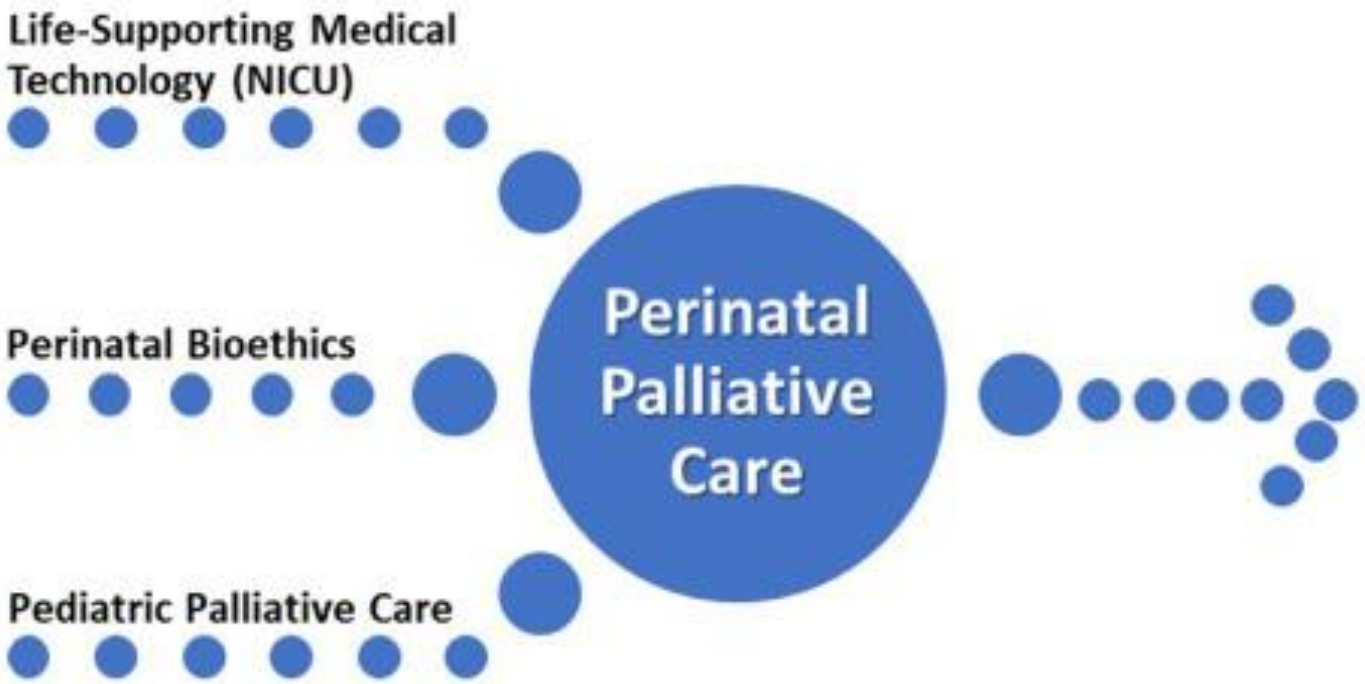
The Evolution of Perinatal Palliative Care



Prenatal Diagnoses & Counsel
- The pregnancy narrative
- Goals & values
- Birth plan

Delivery Room Presence & Support
- Psycho-social-spiritual
- Being present

Post-Delivery Care
- NICU
- Nursery
- Home

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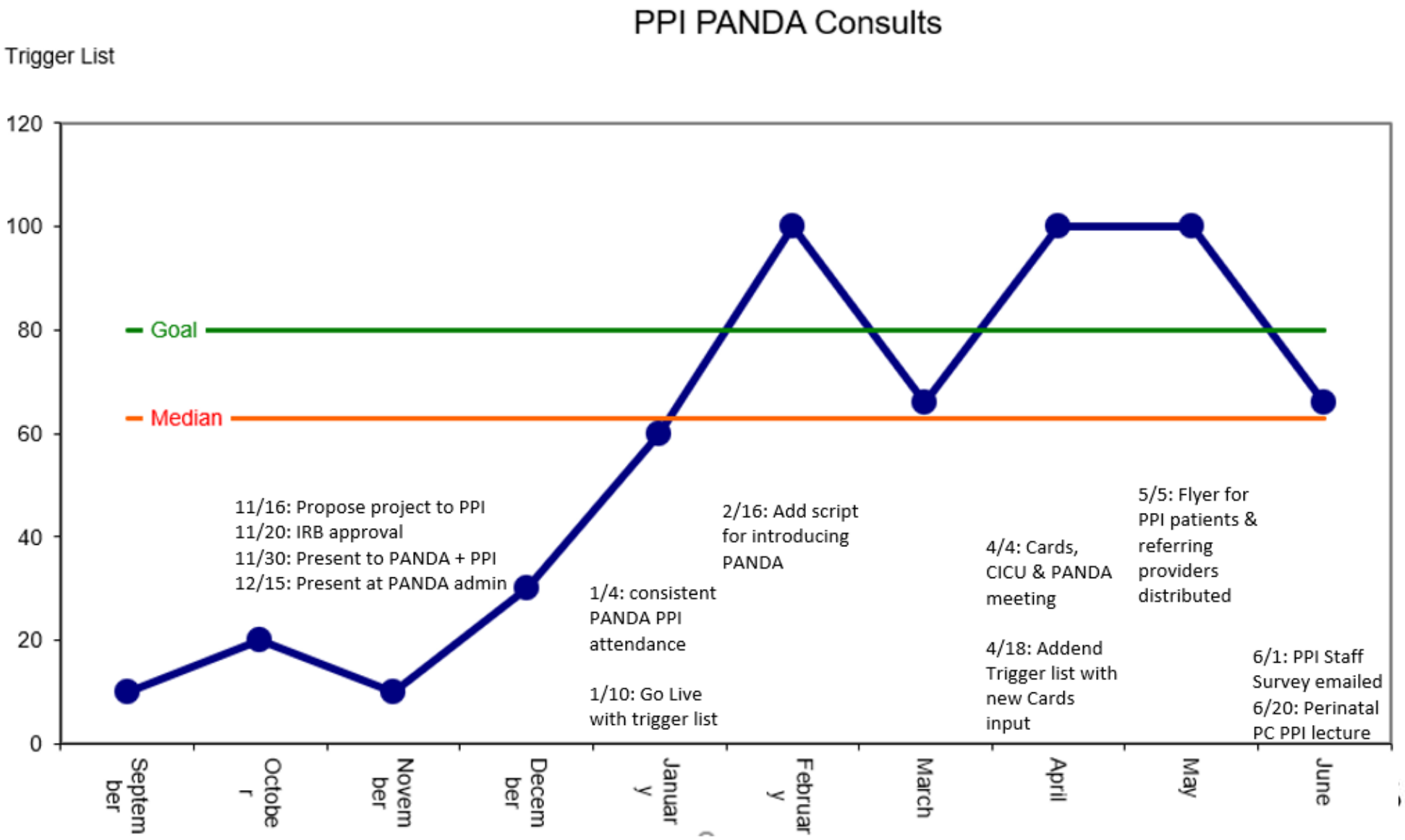
Carter BS, 2022

Children's National: Prenatal Pediatrics Institute



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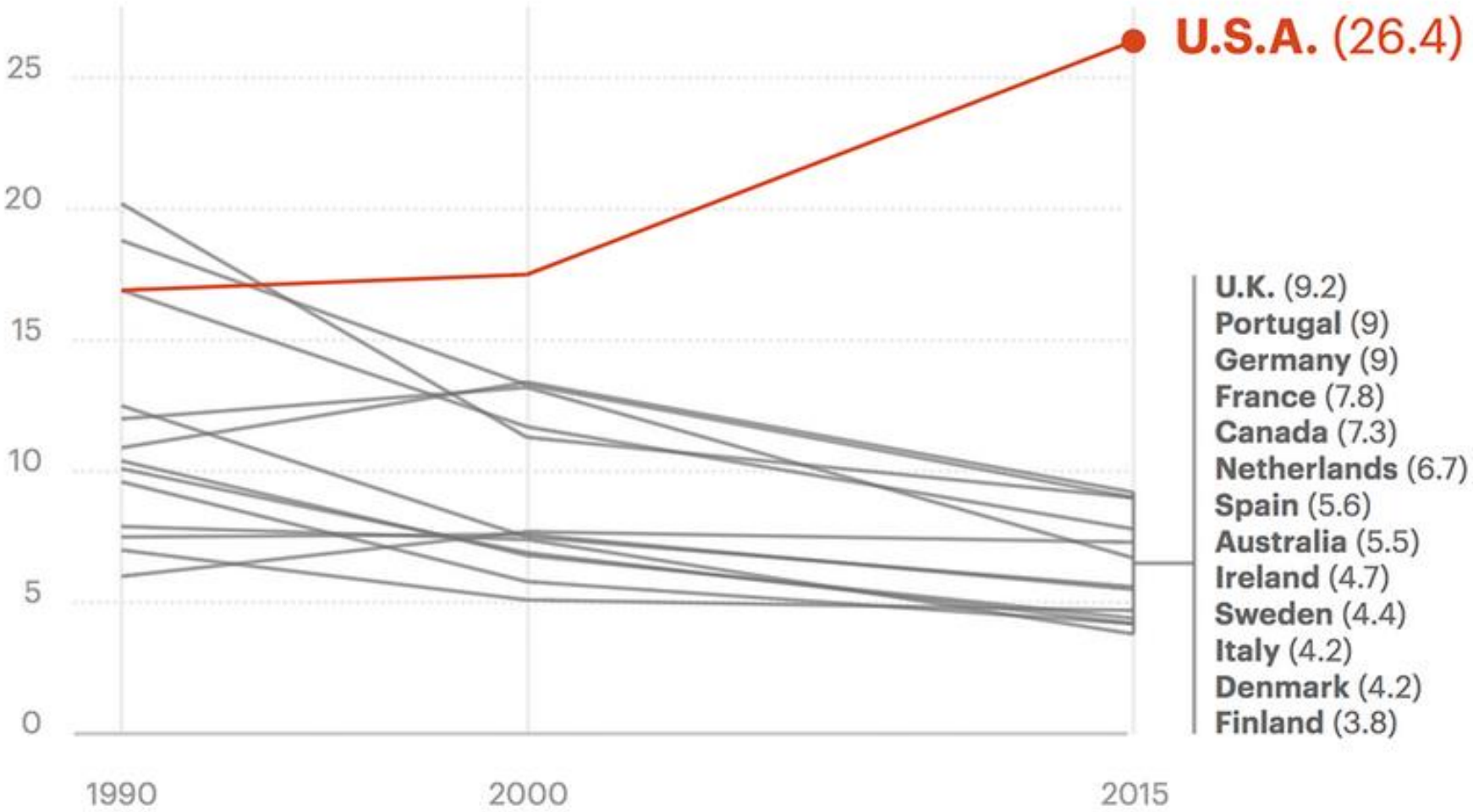
Scott Cohen, MD, MPH

- Adult cardiologist at Children's since 2011
- Program director of the Adult Congenital Heart Disease Program at the Herma Heart Institute
- Board certified in adult congenital heart disease, cardiovascular disease and internal medicine
- Earned his medical degree from Saint Louis University School of Medicine
- Completed his fellowships in pediatric cardiology from the Medical College of Wisconsin, preventative cardiology from Providence VA Medical Center and cardiovascular disease fellowship at Rhode Island Hospital
- Dr. Cohen sees patients at the Milwaukee Hospital campus

Update on the Heart Disease in Pregnancy Clinic

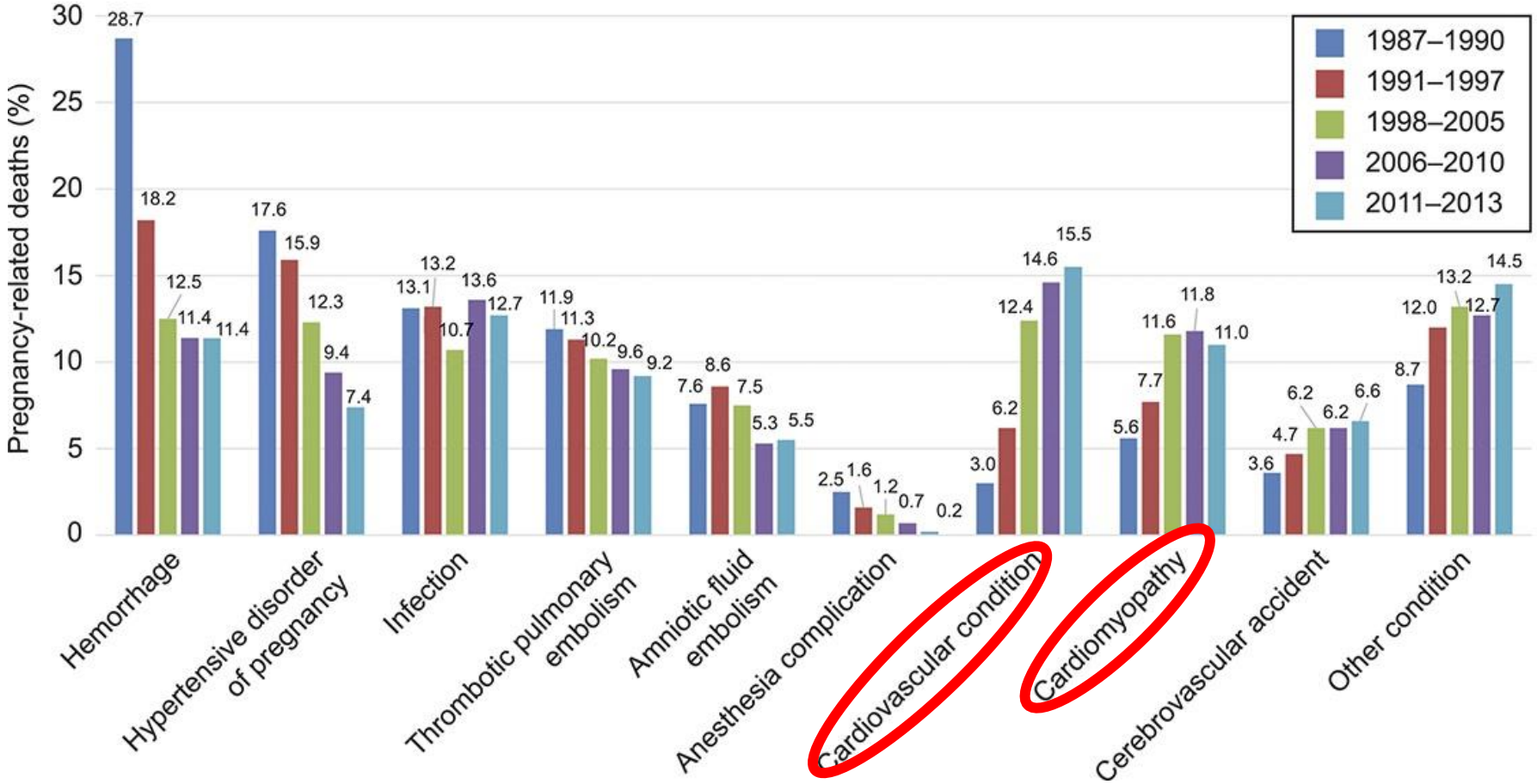
Sarah Thordsen MD, Meredith Cruz MD, MPH, MBA, Ayse Kula MD, Erika Peterson MD, Maria Muravyeva MD, PhD, Scott Cohen MD, MPH

Maternal Mortality Is Rising in the U.S. As It Declines Elsewhere



GBD 2015 Maternal Mortality Collaborators. Lancet, 2016

Pregnancy Mortality in the US



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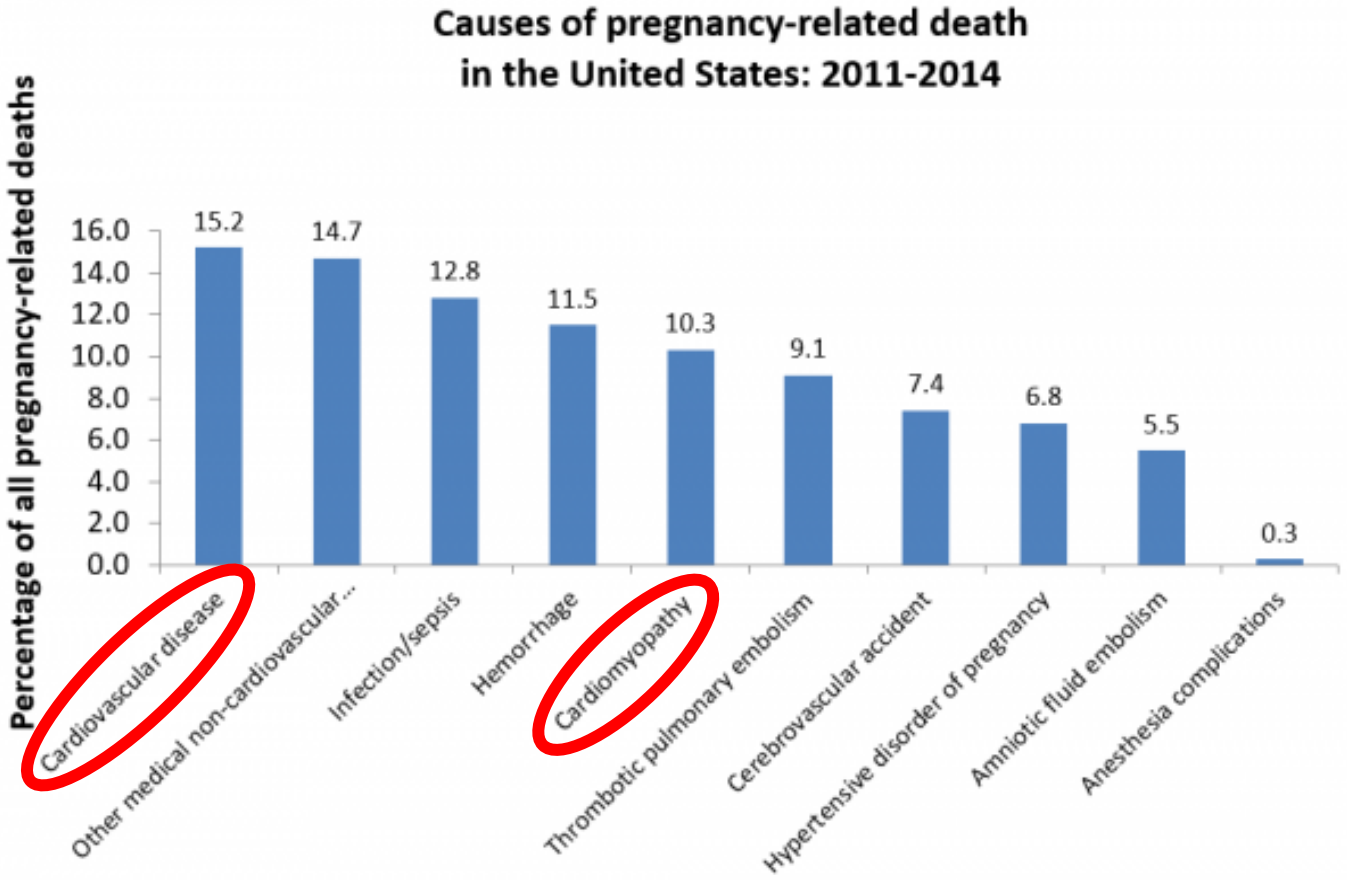
Creanga AA, Syverson C, Seed K, Callaghan WM. Pregnancy-Related Mortality in the United States, 2011-2013. Obstet Gynecol. 2017;130(2),366–373.

Pregnancy Mortality in the US



Note: The cause of death is unknown for 6.5% of all pregnancy-related deaths.

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CDC. Pregnancy Mortality Surveillance System. Website updated 8/2018. Retrieved 11/1/18

2016–17 Wisconsin Maternal Mortality Report

Released April 2022

Cause and Manner of Death

Cause and manner of death are both determined by a coroner, certified medical examiner, or another medical professional who completes the death certificate. The three most common causes of pregnancy-related deaths in 2016–17 were:

- **52% Mental Health Conditions**: includes substance use disorders and overdoses
- **12% Hemorrhage** (excludes aneurysms and cerebrovascular accidents or strokes): blood loss
- **9% Cardiomyopathy:** a disease of the heart muscle that makes it harder for the heart to pump blood to the rest of the body, which can lead to heart failure

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Wisconsin Department of Health Services, 2016-2017 Maternal Mortality report

Cardiac Morbidity During Pregnancy

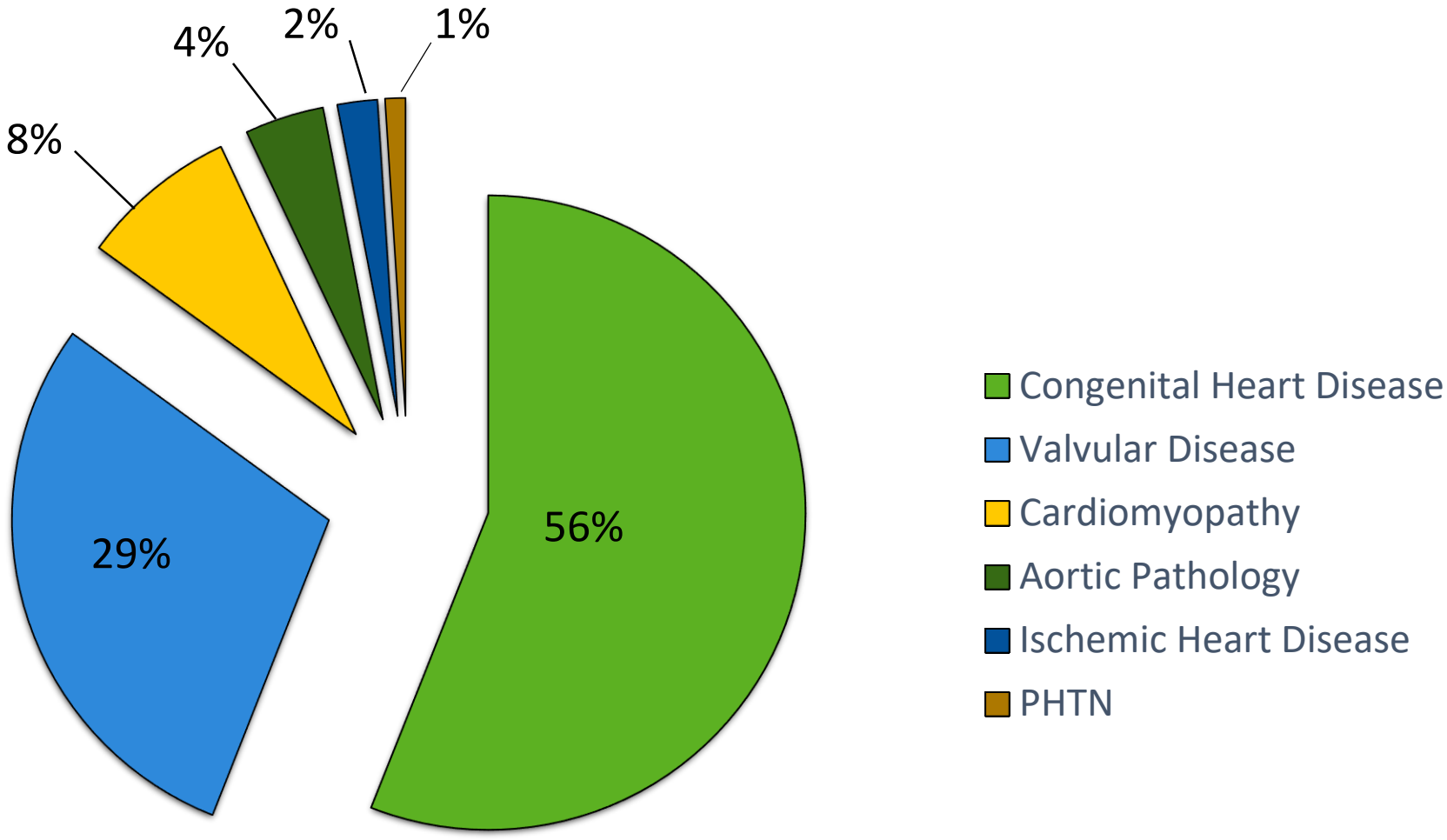
TABLE 2 Incidence of Adverse Cardiac Event Rates During Pregnancy (N = 1,938)

Any maternal cardiac events	307 (15.8)
Maternal cardiac death	6 (0.3)
Maternal cardiac arrest	8 (0.4)
Arrhythmias	181 (9.3)
Any left- or right-sided HF	120 (6.2)
Left-sided HF	106 (5.5)
Right-sided HF	19 (1.0)
Stroke	13 (0.7)
Myocardial infarction	8 (0.4)
Dissection	7 (0.4)
Cardiac thromboembolism	6 (0.3)

et al., JACC 2018Silversides

et al., JACC 2018Silversides

Make up of CV Disease in Pregnancy



ESC ROPAC Study

Clinic Overview

The Heart Disease in Pregnancy Program offers multidisciplinary care located in the same clinic with services provided on the same day

- Held in the MFM clinic space at Froedtert Hospital
- General cardiology
- Adult congenital cardiology
- Maternal fetal medicine
- Obstetric-anesthesia
- Cardiac echocardiography
- Fetal ultrasounds.

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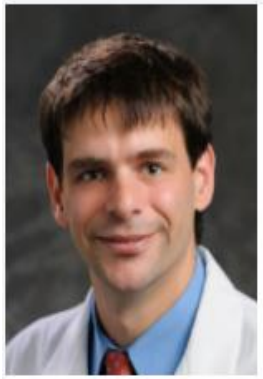
Meet Our Team



Scott Cohen, MD, MPH

Associate Professor; Director, Adult Congenital Heart Disease Program

Meredith Cruz, MD, MPH, MBA

Associate Professor

Ayse Oge Kula, MD

Assistant Professor

Maria Muravyeva, MD, PhD

Assistant Professor

Erika Peterson, MD

Associate Professor

Sarah E. Thordsen, MD, FACC

Assistant Professor

Andrew Pistner, MD

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Services Provided by Team

- Pre-conception cardiovascular risk and counseling
- Consultative cardiac care throughout pregnancy and peripartum period
- Cardiac medication safety recommendations throughout pregnancy and lactation
- Cardiac recommendations for labor and delivery planning
- Coordination between multispecialty teams

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Heart Disease and Pregnancy - Inpatient

- MFM primary team
- Location / Treatment Team
- Cardiovascular Disease Consultation – Cohen/Thordsen
- ACHD Consultation – Cohen/ ACHD Faculty
- Cards consult team serves as conduit to other subspecialties
- If needed advanced heart Failure, EP, Cardiac anesthesia, Pulm HTN team , Interventional cardiology, critical care

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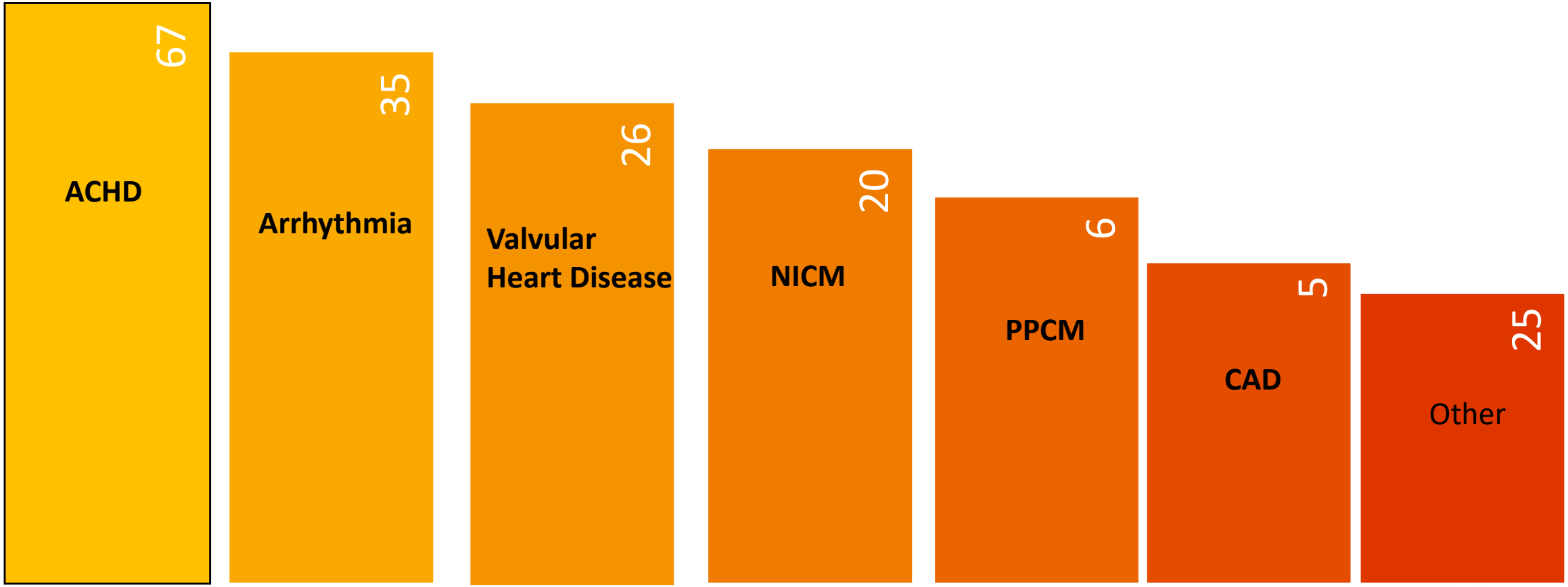
Breakdown of Diagnosis:



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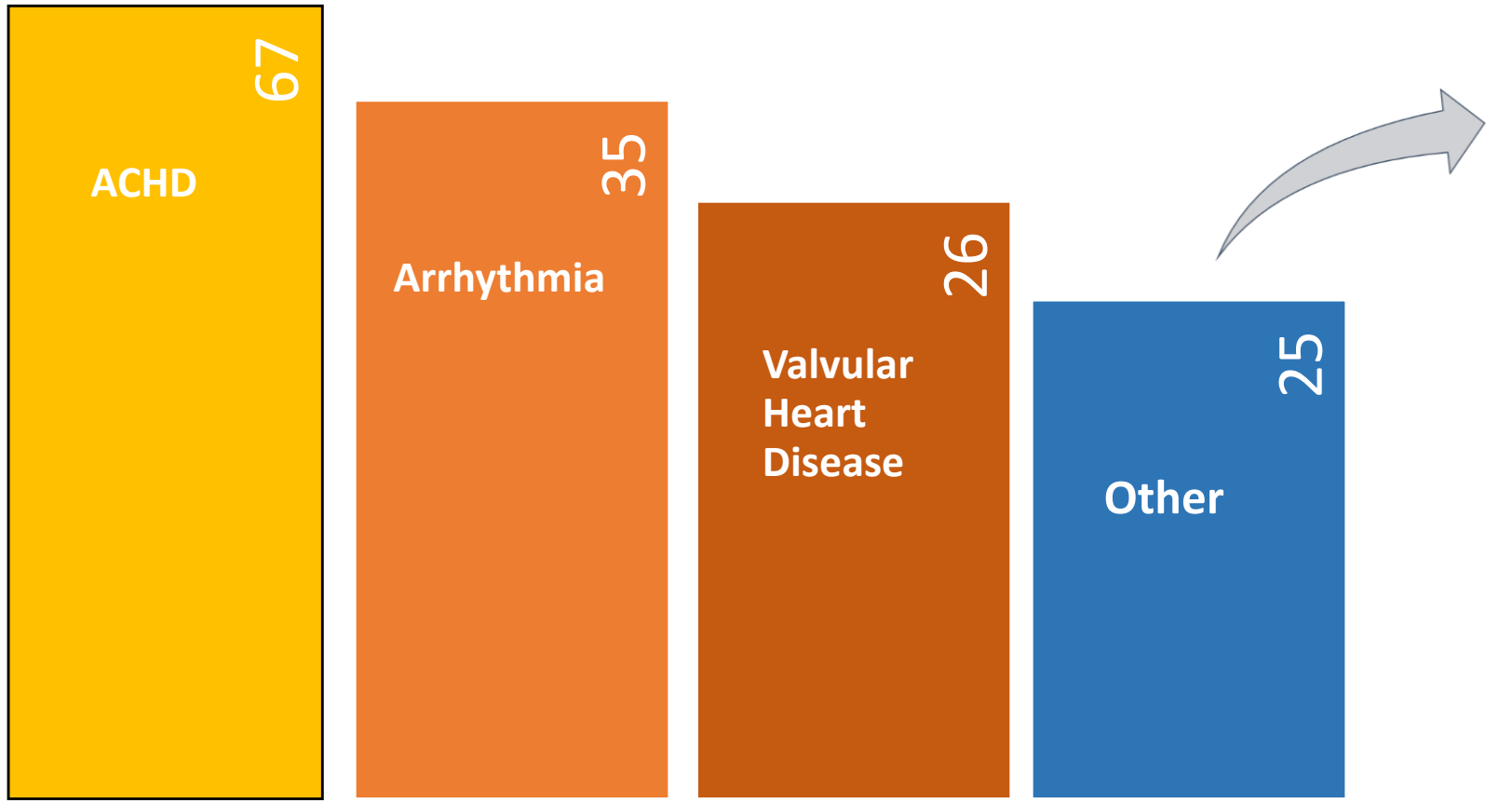
Breakdown of Diagnosis:



- Genetic cardiomyopathy
- Chemotherapy exposure
- Hx of HTN emergency
- Mosiac Turner Syndrome
- PH/CETPH
- Fabry
- Chest pain

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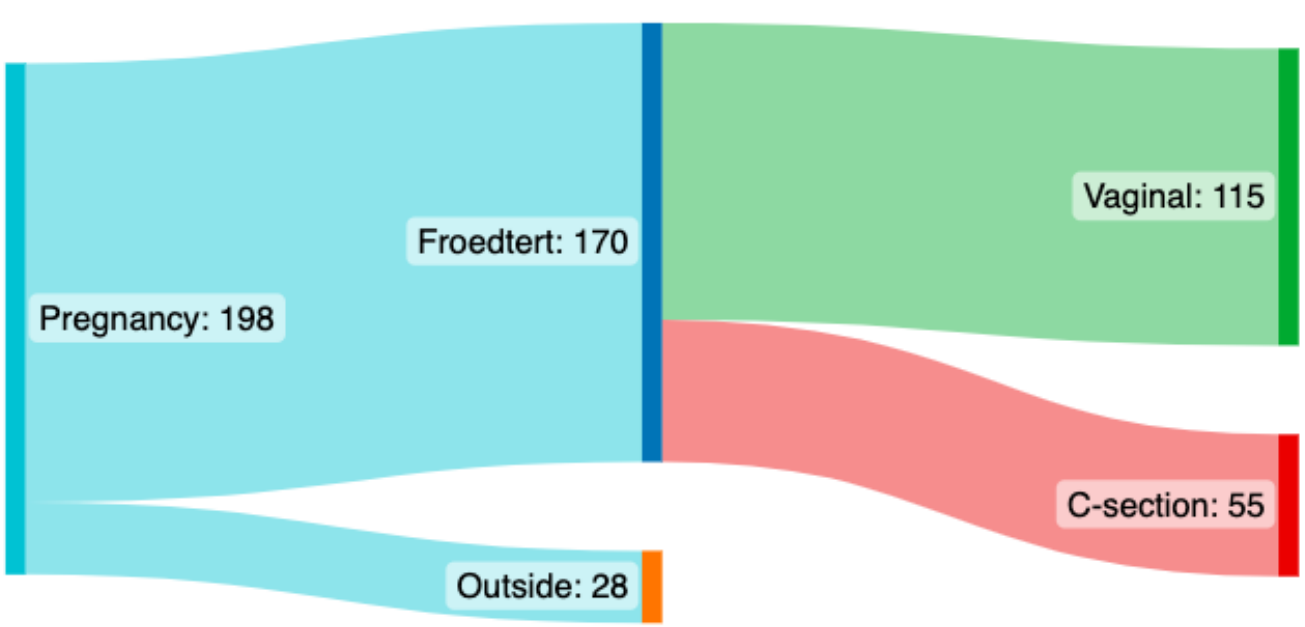
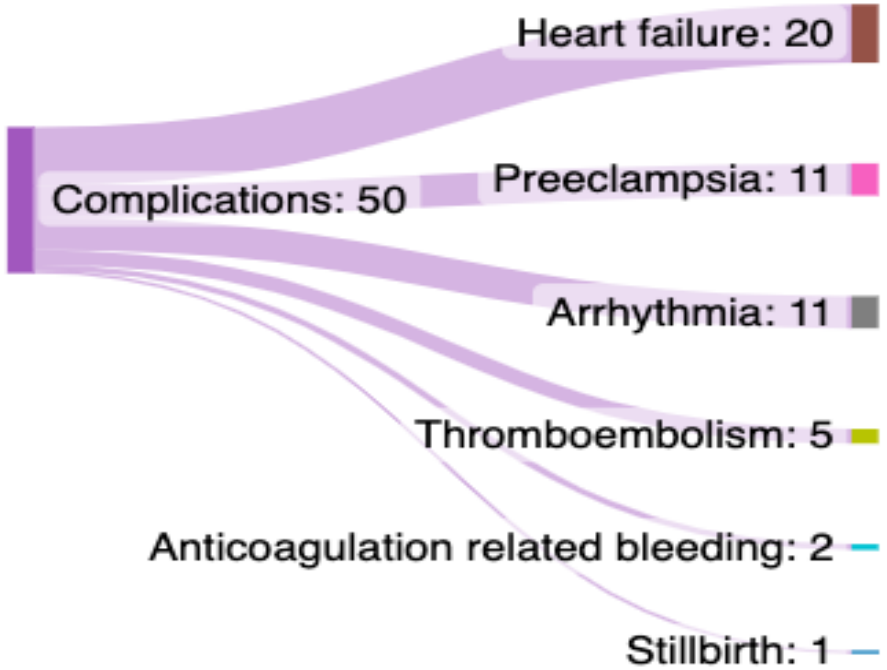
Summary statistics





- Clinic started February 2019
- Data thru June 2022
- No maternal deaths
- 184 patients

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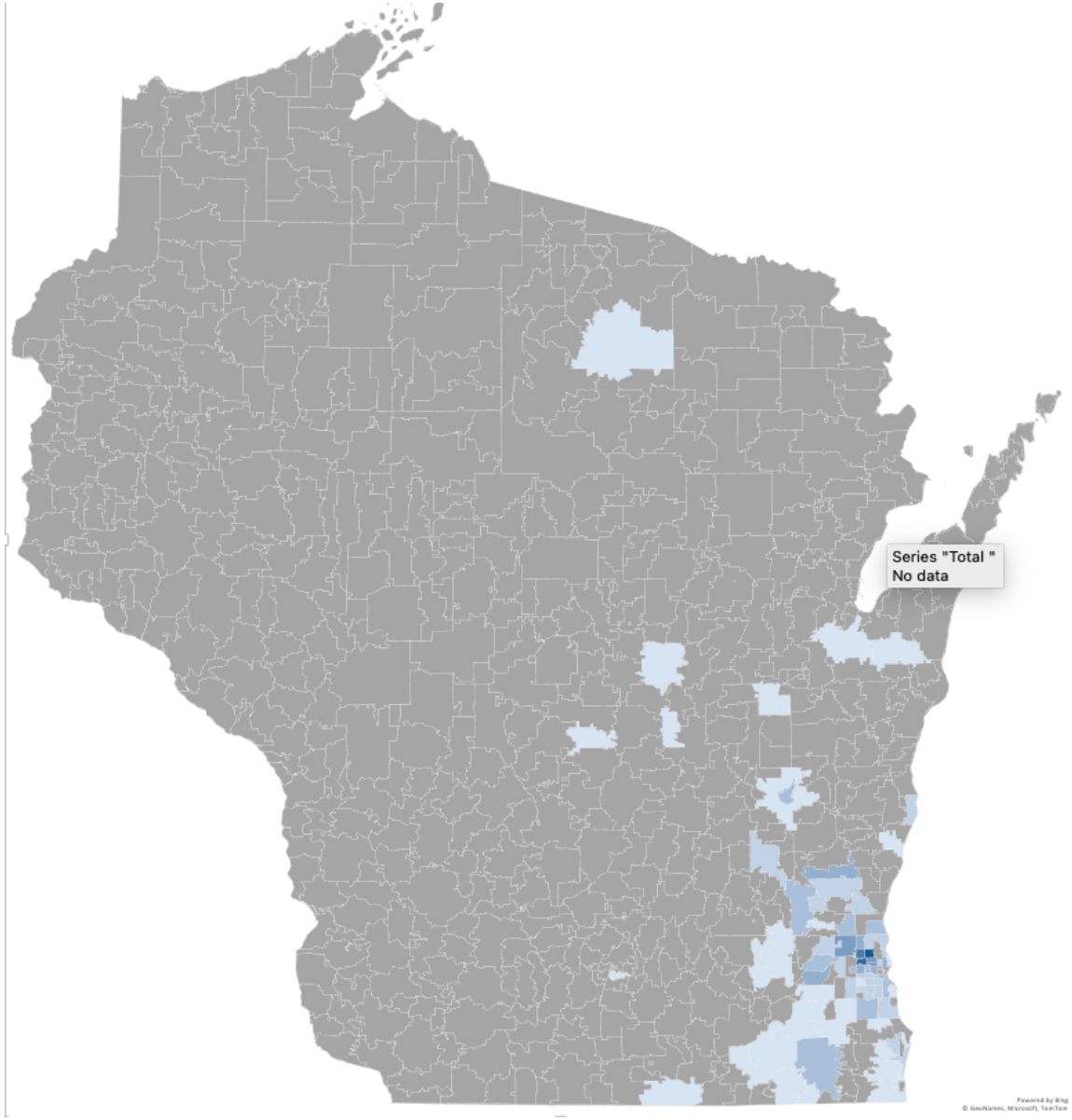
Location of patients seen by zip code

- SE Wisconsin
- 2 patients in Michigan upper peninsula
- 1 patient MN



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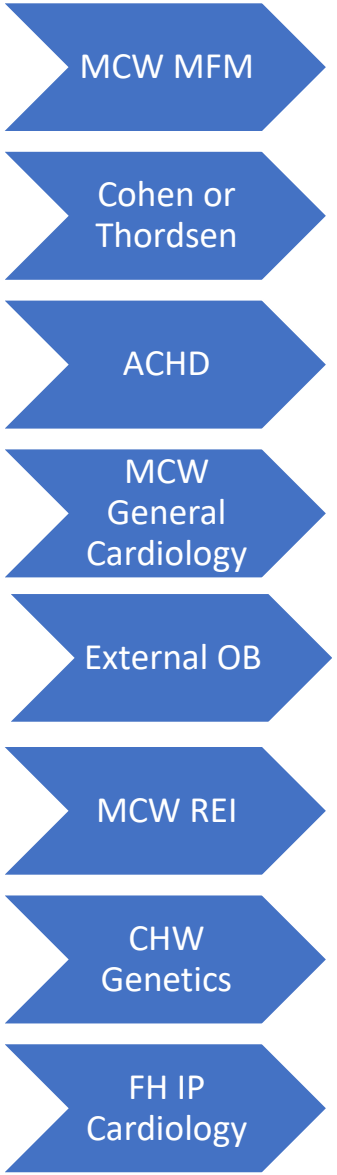
Referral source:

- List of referrals in order of most common to least common (top to bottom)



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Research

- Peripartum Cardiomyopathy Network
  - REBIRTH – Bromocriptine PPCM
- PPCM and Mechanical Circulatory Support
  - Registry – AbioMED
- Mode of delivery study:
  - Vaginal birth vs. cesarean section rate
- PROFAT (PRegnancy Outcomes in Fontans with Anticoagulation Therapy)
  - Multicenter institutional retrospective study
- Pregnancy outcomes in women with coarctation and risk factors for hypertension in women with history of coarctation
  - Abstract and poster presentation at the International Congress on Cardiac problems in Pregnancy conference

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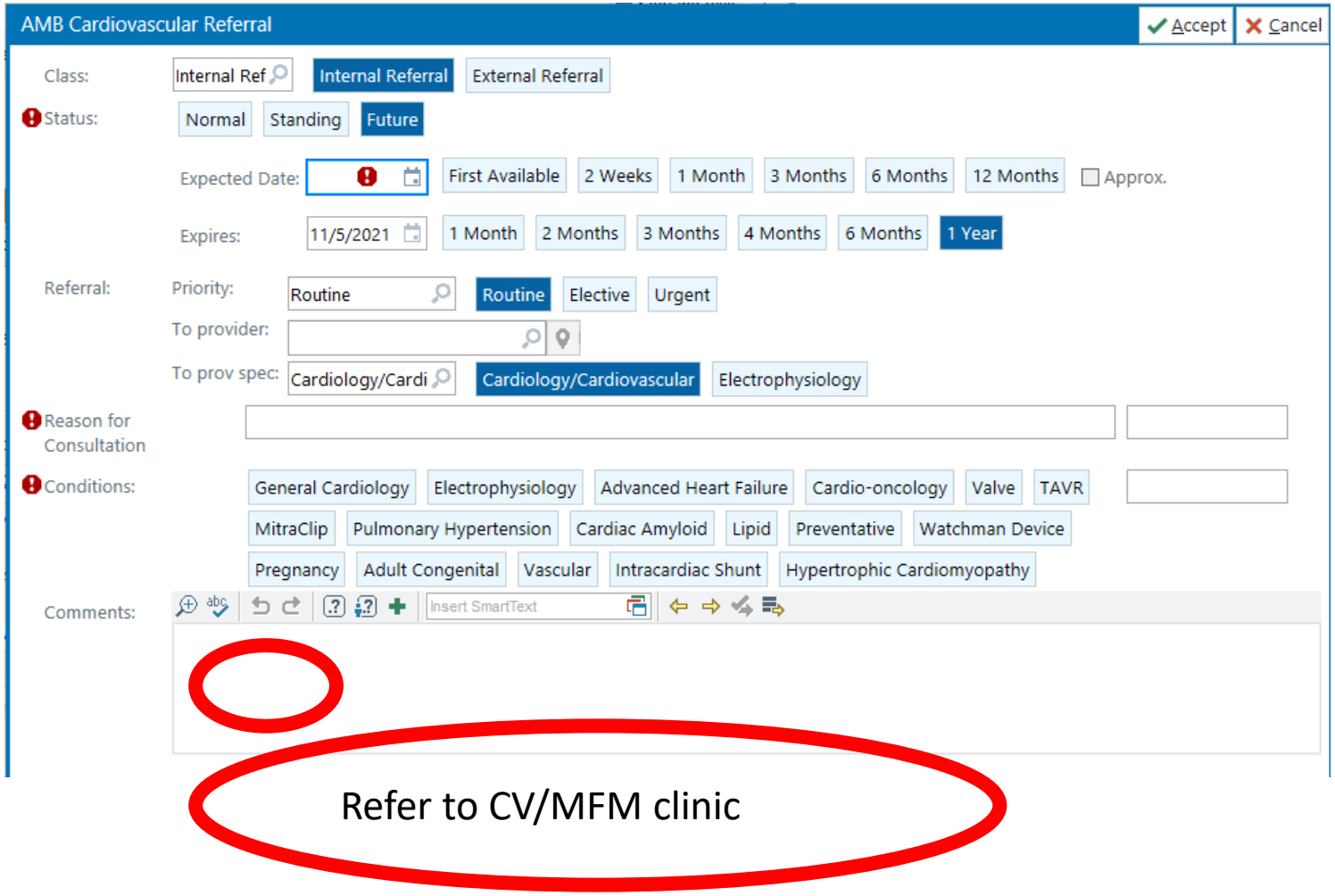
How to refer an outpatient:



- Scheduling coordinator

  Ann Peschek

  414-805-0011

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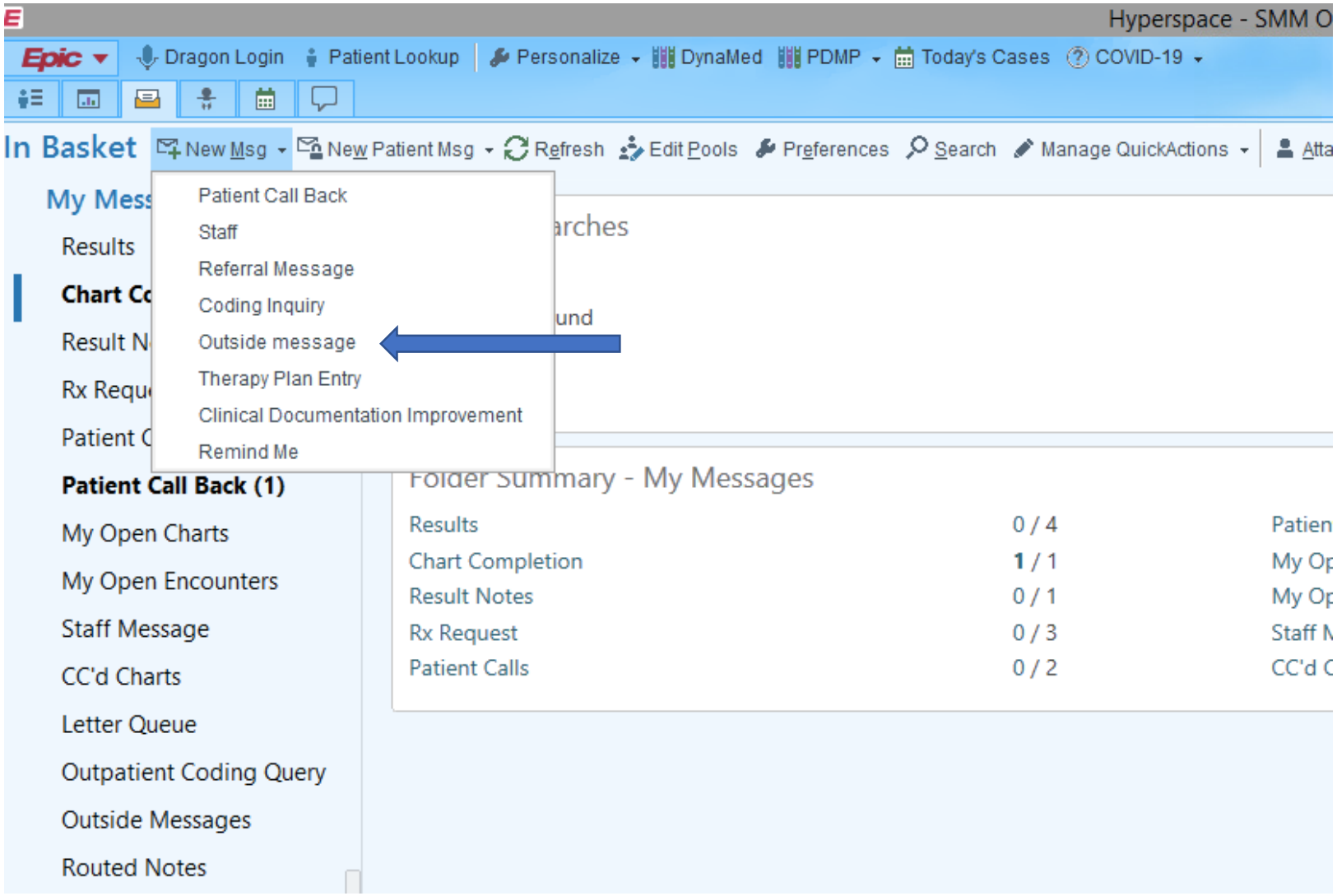
How to refer an outpatient:



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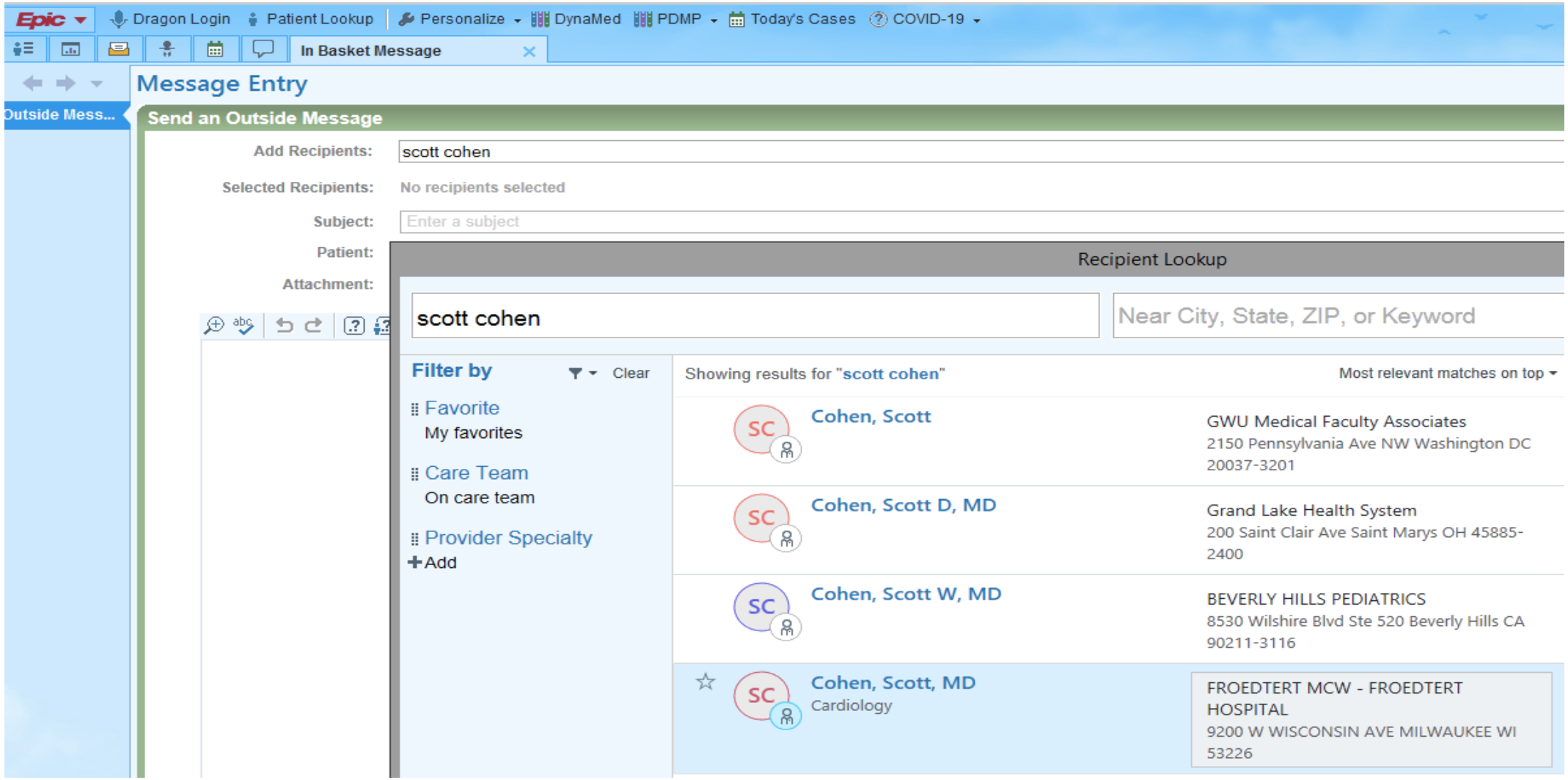
How to refer an outpatient:



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FETAL INNOVATIONS
Conversation with the Experts
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Thank you!