Review of Infant Deaths due to Congenital Anomalies

Wednesday, February 19, 2020
2:00 PM – 3:00 PM ET

NATIONAL CFRP
Saving Lives Together
The National Center for Fatality Review and Prevention

Housekeeping Notes

- **Webinar is being recorded and will be available within 2 weeks on our website: www.ncfrp.org**
- All attendees will be muted and in listen only mode
- Questions can be typed into the "Questions and Answer" (Q & A) pane
  - Due to the large number of attendees, we may not be able to get to all questions in the time allotted
  - All unanswered questions will be posted with answers on the NCFRP website

Introduction

Sonsy Fermín, MSW, LCSW, CDR, USPHS,
Healthy Resources and Service Administration (HRSA)
Acting Chief, East Branch, Healthy Start and Perinatal Services
Federal project officer, National Center for Fatality Review and Prevention

About the National Center

- The National Center for Fatality Review and Prevention (NCFRP) is a resource and data center that supports child death review (CDR) and fetal and infant mortality review (FIMR) programs around the country.
- Supported with funding from the Maternal and Child Health Bureau at the Health Resources and Services Administration, the Center aligns with several MCHB priorities and performance and outcome measures such as:
  - Healthy pregnancy
  - Child and infant mortality
  - Injury prevention
  - Safe sleep

HRSA's Overall Vision for NCFRP

- Through delivery of data, training, and technical support, NCFRP will assist state and community programs in:
  - Understanding how CDR and FIMR reviews can be used to address issues related to adverse maternal, infant, child, and adolescent outcomes
  - Improving the quality and effectiveness of CDR/FIMR processes
  - Increasing the availability and use of data to inform prevention efforts and for national dissemination
- Ultimate Goal:
  - Improving systems of care and outcomes for mothers, infants, children, and families

Acknowledgement

This webinar was made possible in part by Cooperative Agreement Numbers UG7MC28482 and UG7MC31831 from the US Department of Health and Human Services (HHS), Health Resources and Services Administration (HRSA), Maternal and Child Health Bureau (MCHB) as part of an award totaling $1,099,997 annually with 0 percent financed with non-governmental sources. Its contents are solely the responsibility of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

Presentation goals

- Understand the most common congenital anomalies and their prevalence
- Discuss steps for evaluation and prevention of possible anomalies
- Provide tips for effective review of congenital anomalies
- Give local examples of fatality review findings that lead to successful interventions that address prevention of congenital anomalies

Key Note Speaker

Kelly S. Gibson, MD, Maternal Fetal Medicine
Dept of Obstetrics and Gynecology
Cleveland MetroHealth System

National Center for Fatality Review and Prevention

Fetal and Infant Mortality Reviews: How do we review congenital anomaly cases?

Wednesday, February 19, 2020

Kelly S Gibson, MD FACOG
Director, Maternal Fetal Medicine,
The MetroHealth System

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Disclosures

The following speaker(s) have no relevant financial relationships to disclose:

- Kelly S Gibson, MD

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Objectives

- To review the epidemiology of the relationship between the infant mortality rate and congenital anomalies
- To describe the most common congenital anomalies and infections and their association with fetal demise
- To discuss the steps for an effective evaluation of possible anomalies and tips for an effective review

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Outline

- **Background**
- Common congenital anomalies
- Common congenital infections
- Steps for evaluation of possible anomalies
- Tips for an effective review
- Case examples

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Congenital Anomalies: Background

- Infant mortality rate (IMR) refers to deaths that occur during infancy—the first year of life, or from a live birth to age one
  - Deaths per 1,000 live births
  - In USA IMR was 6.6 in 2008 -> 5.79 in 2017
    - Non-Hispanic Black: 10.97 vs Non-Hispanic White 4.67
    - < 28 weeks 384.39 vs 37-41 weeks 2.10

- Worldwide ~7% of neonatal deaths vs ~20-25% in developed countries due to congenital anomalies

Ely et al National Vital Statistics Reports, vol 68 no 10. 2019.

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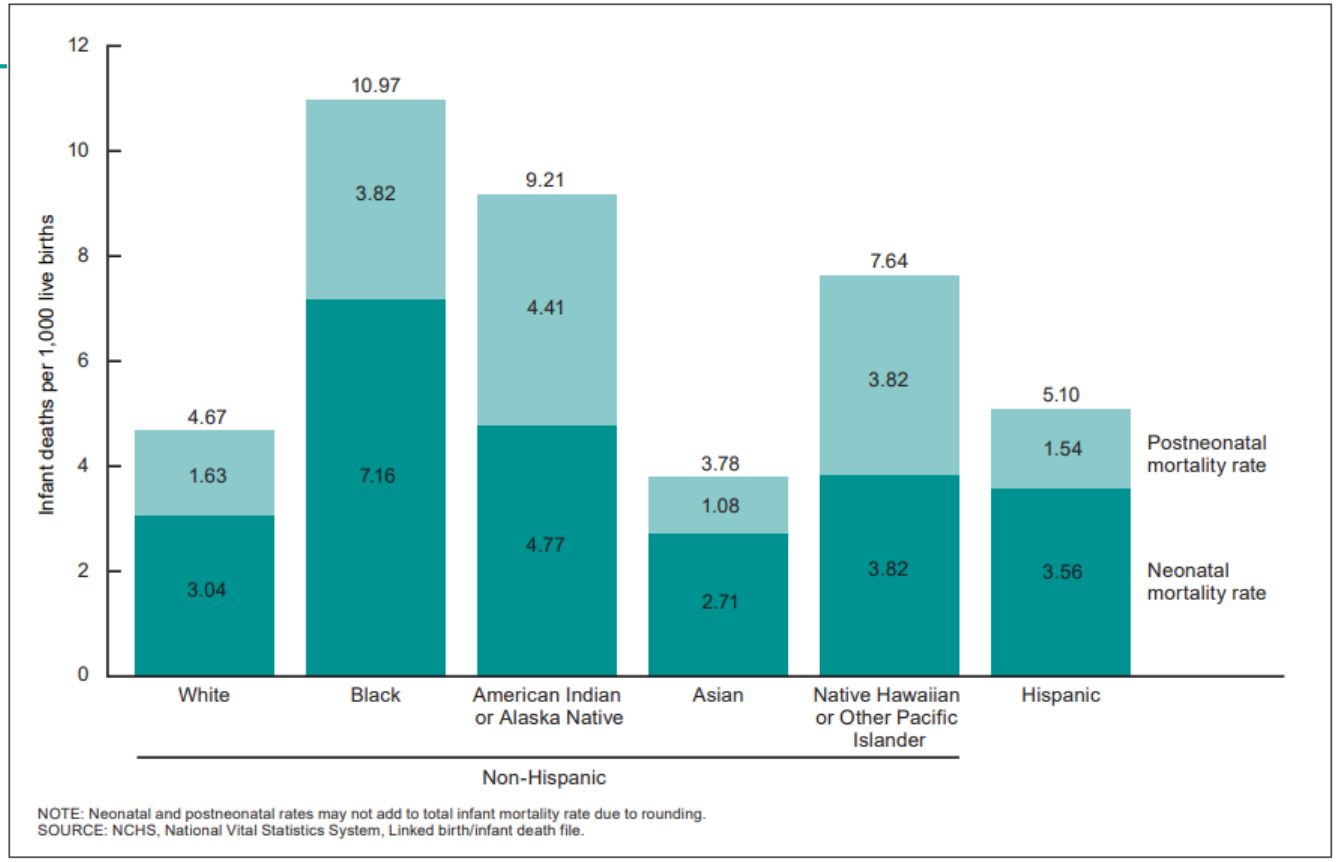

Figure 2. Infant, neonatal, and postneonatal mortality rates, by race and Hispanic origin: United States, 2017

Ely et al National Vital Statistics Reports, vol 68 no 10. 2019.

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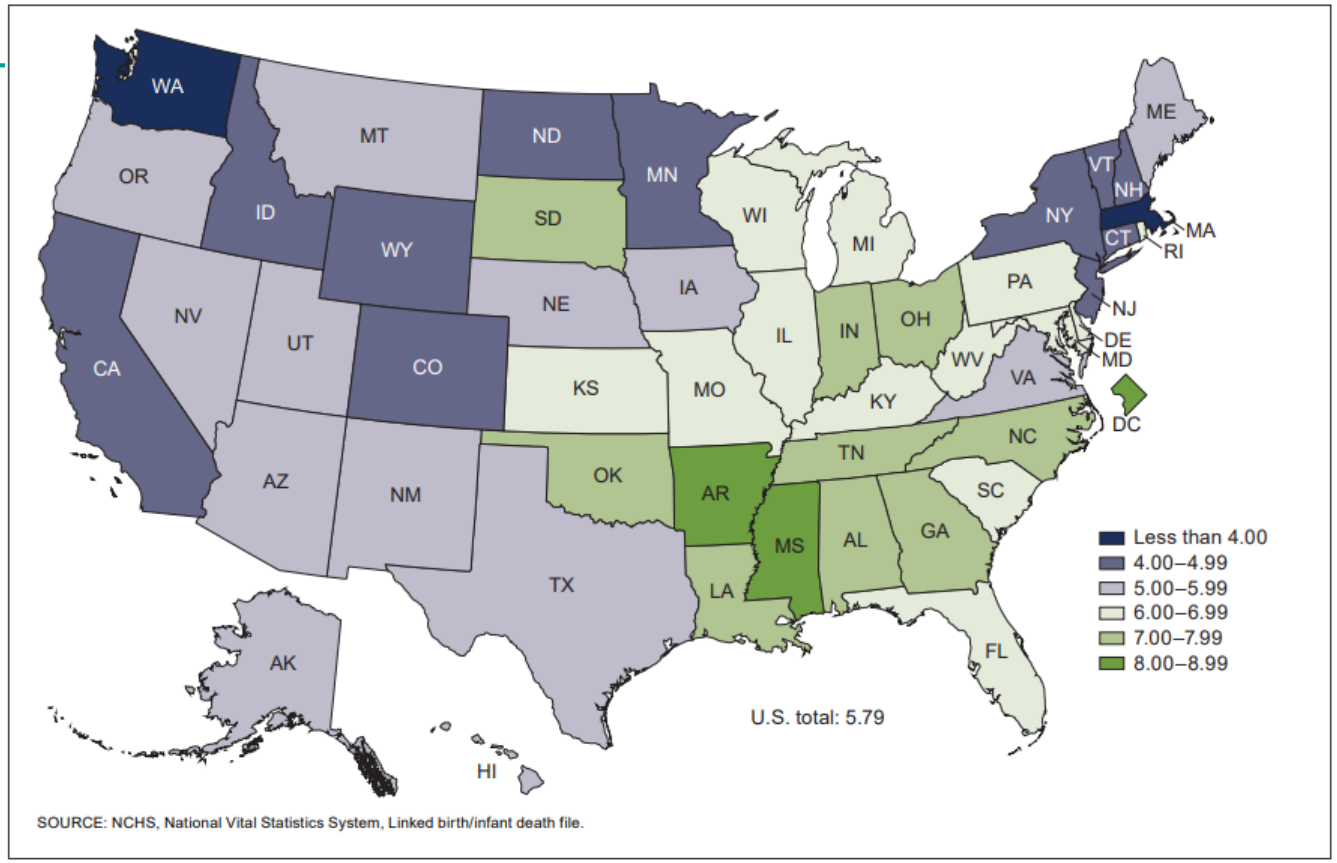

Figure 3. Infant mortality rates, by state: United States, 2017

Ely et al National Vital Statistics Reports, vol 68 no 10. 2019.

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Background: Causes by Race/Ethnicity

Table 4. Infant deaths, percentage of

Year	Number	Percent	Rate (d per 10 live bi
2017	4,596	20.6	119
2016	4,823	20.8	122
2015	4,847	20.7	121
2014	4,754	20.5	119
2013	4,778	20.4	121
2012	4,967	21.0	125
2011	5,016	20.9	126
2010	5,115	20.8	127

NOTE: The five leading causes of death were the same c

SOURCE: NCHS, National Vital Statistics System, Linke

Congenital malformations (Q00–Q99)

Year	Number	Percent	Rate (deaths per 100,000 live births)
2017	4,596	20.6	119.2
2016	4,823	20.8	122.2
2015	4,847	20.7	121.8
2014	4,754	20.5	119.2
2013	4,778	20.4	121.5
2012	4,967	21.0	125.7
2011	5,016	20.9	126.9
2010	5,115	20.8	127.9

2017

	Accidents (unintentional injuries) (V01–X59)		
deaths 0,000 irths)	Number	Percent	Rate (deaths per 100,000 live births)
.3	1,313	5.9	34.1
.0	1,217	5.3	30.8
.4	1,289	5.5	32.4
.6	1,163	5.0	29.2
.7	1,150	4.9	29.2
.4	1,163	4.9	29.4
.2	1,167	4.9	29.5
.5	1,107	4.5	27.7

Ely et al National Vital Statistics Reports, vol 68 no 10. 2019.

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Table 5. Infant deaths and mortality rates for the five leading causes of infant death, by race and Hispanic origin of mother: United States, 2017

[Rates are per 100,000 live births in specified group]

Cause of death (based on the *International Statistical Classification of Diseases, 10th Revision*, 1992)	All races			White			Black			American Indian or Alaska Native			Asian[1]		
	Rank	Number	Rate	Rank	Number	Rate	Rank	Number	Rate	Rank	Number	Rate	Rank	Number	Rate
All causes	…	22,341	579.5	…	9,306	467.1	…	6,152	1,097.2	…	276	921.3	…	943	378.3
Congenital malformations, deformations and chromosomal abnormalities (Q00–Q99)	1	4,596	119.2	1	2,138	107.3	2	822	146.6	1	50	166.9	1	211	84.7
Disorders related to short gestation and low birth weight, not elsewhere classified (P07)	2	3,757	97.4	2	1,260	63.2	1	1,354	241.5	2	31	103.5	2	162	65.0
Newborn affected by maternal complications of pregnancy (P01)	3	1,436	37.2	5	470	23.6	3	467	83.3	5	10	*	3	83	33.3
Sudden infant death syndrome (R95)	4	1,360	35.3	3	688	34.5	5	391	69.7	4	23	76.8	8	19	*
Accidents (unintentional injuries) (V01–X59)	5	1,313	34.1	4	619	31.1	4	397	70.8	3	25	83.5	7	25	10.0

Cause of death (based on the *International Statistical Classification of Diseases, 10th Revision*, 1992)	Total Hispanic			Mexican			Puerto Rican			Central and South American[2]		
	Rank	Number	Rate	Rank	Number	Rate	Rank	Number	Rate	Rank	Number	Rate
All causes	…	4,584	509.9	…	2,588	505.3	…	459	648.2	…	653	448.4
Congenital malformations, deformations and chromosomal abnormalities (Q00–Q99)	1	1,199	133.4	1	727	142.0	2	66	93.2	1	190	130.5
Disorders related to short gestation and low birth weight, not elsewhere classified (P07)	2	764	85.0	2	419	81.8	1	87	122.9	2	114	78.3
Newborn affected by maternal complications of pregnancy (P01)	3	310	34.5	3	162	31.6	3	33	46.6	3	50	34.3
Sudden infant death syndrome (R95)	5	174	19.4	5	94	18.4	5	23	32.5	7	19	*
Accidents (unintentional injuries) (V01–X59)	4	186	20.7	4	104	20.3	4	25	35.3	4	24	16.5

… Category not applicable.

* Rate does not meet NCHS standards of reliability; based on fewer than 20 deaths in the numerator.

[1]For non-Hispanic Asian persons, Bacterial sepsis of newborn (P36) was the fourth leading cause of death, with 40 deaths and a rate of 16.0. Diseases of the circulatory system (I00–I99) was the fifth leading cause of death, with 27 deaths and a rate of 10.8.

[2]For Central and South American persons, Newborn affected by complications of placenta, cord and membranes (P02) was the fifth leading cause of death, with 24 deaths and a rate of 16.5.

NOTES: Reliable cause-specific infant mortality rates cannot be computed for Cuban or non-Hispanic Native Hawaiian or Other Pacific Islander persons because of the small number of infant deaths. Race and Hispanic origin are reported separately on birth certificates. Race categories are consistent with the 1997 Office of Management and Budget standards. Persons of Hispanic origin may be of any race.

SOURCE: NCHS, National Vital Statistics System, Linked birth/infant death file.

Ely et al National Vital Statistics Reports, vol 68 no 10. 2019.

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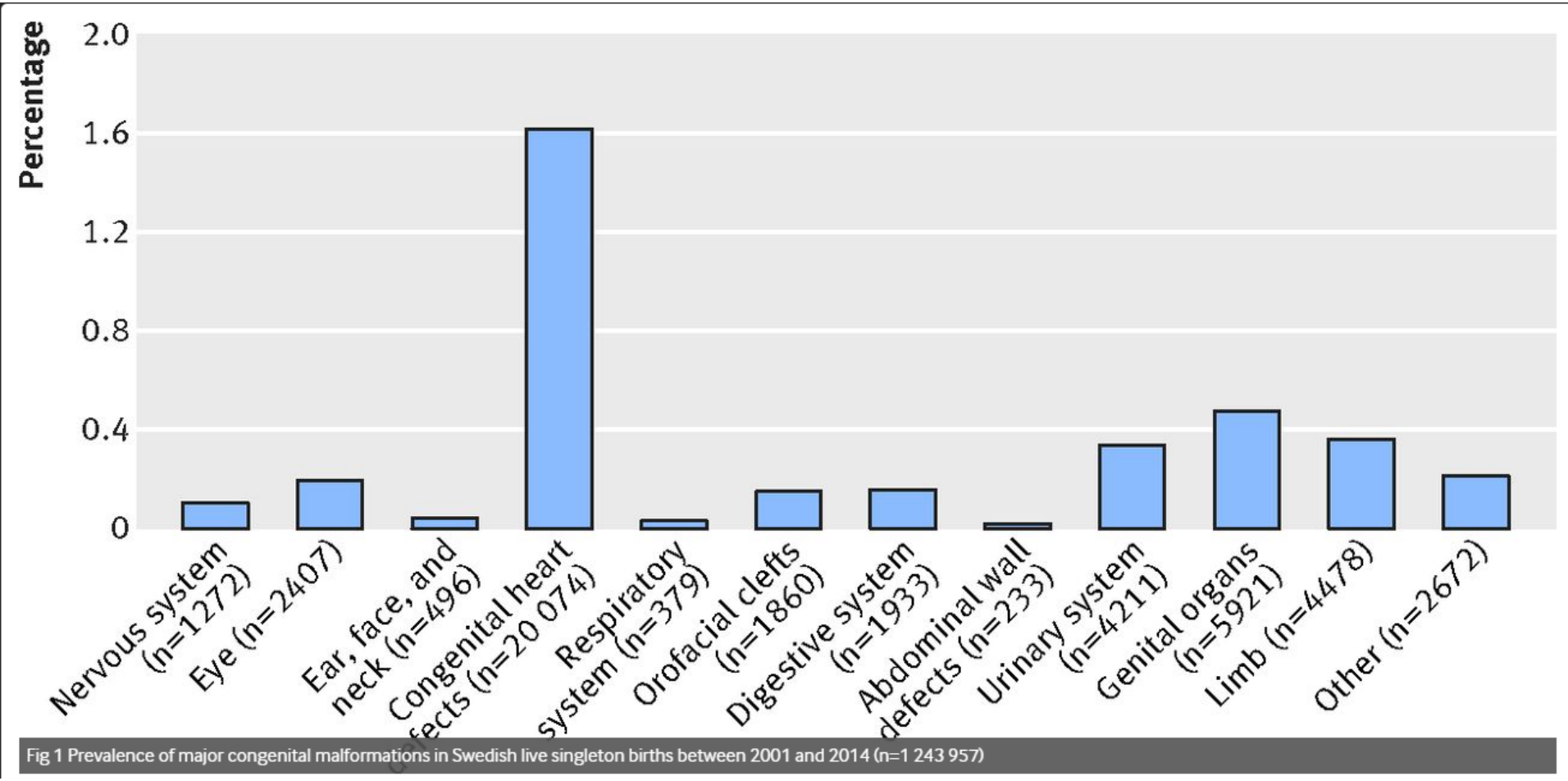

Fig 1 Prevalence of major congenital malformations in Swedish live singleton births between 2001 and 2014 (n=1 243 957)

Perrson, BMJ 2017

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Definition

- Congenital anomalies or birth defects
  - Structural or functional abnormalities, including metabolic disorders, which are present from birth.
  - Also include inborn errors of metabolism and blood disorders
- Can cause spontaneous abortion, stillbirth, and neonatal death
- A significant but underrecognized cause of mortality and disability among infants and children under five years of age
- Can be life-threatening, result in long-term disability, and negatively affect individuals, families, health-care systems and societies

World Health Organization. Birth Defects. 2010

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Background

- Exact number and cause difficult to tract
  - May be due to genetic or environmental causes
  - Many countries lack standard definitions or tracking systems

- The most common serious congenital disorders are:
  - Congenital heart defects
  - Neural tube defects
  - Down syndrome
  - Hemoglobinophathies (thalassemia and sickle-cell), G6PD deficiency

World Health Organization. Birth Defects. 2010

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Diverse Causes

- Genetics
  - Genetics Single gene defects
  - Chromosomal disorders/aneuploidy
- Multifactorial inheritance
- Environmental teratogens
  - Chemicals and high doses of radiation
- Micronutrient deficiencies
  - Iodine and folic acid deficiency
- Maternal infectious
  - Syphilis and rubella
- Maternal illnesses
  - Diabetes mellitus
  - Exposure to medicines and recreational drugs including alcohol and tobacco

World Health Organization. Birth Defects. 2010

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Prevention and Treatment

- Family planning
- Preconception screening and counselling
- Optimizing women's diet before and throughout pregnancy
- Preventing and treating teratogen-induced infections before and throughout pregnancy
- Optimizing preconception maternal health and treatment
- Antenatal screening and Prenatal diagnosis
- Fetal treatment

World Health Organization. Birth Defects. 2010

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Prevention and Treatment

- Newborn infant examination
- Newborn infant screening
- Medical treatment
- Surgery
- Rehabilitation and palliative care

World Health Organization. Birth Defects. 2010

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Outline

- Background
- **Common congenital anomalies**
- Common congenital infections
- Steps for evaluation of possible anomalies
- Tips for an effective review
- Case examples

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Neurologic: Movement Disorders

- 1/3,000 live births
- Hypertonia, hypotonia, seizures
- Associations with demise depend on underlying cause
  - Typically progressive, but some can be treated
  - Abnormal movement can lead to arthrogryoposis
    - Lack of extremity motion despite fetal stimulation
      - Persistent unusual or abnormal posturing of limbs
      - Early finding often clubfeet and clenched hands
  - Seizures may be due to other underlying condition
- Evaluation: genetics, Fetal MR of spine and brain
  - Spinal Muscular Atrophy (SMA)

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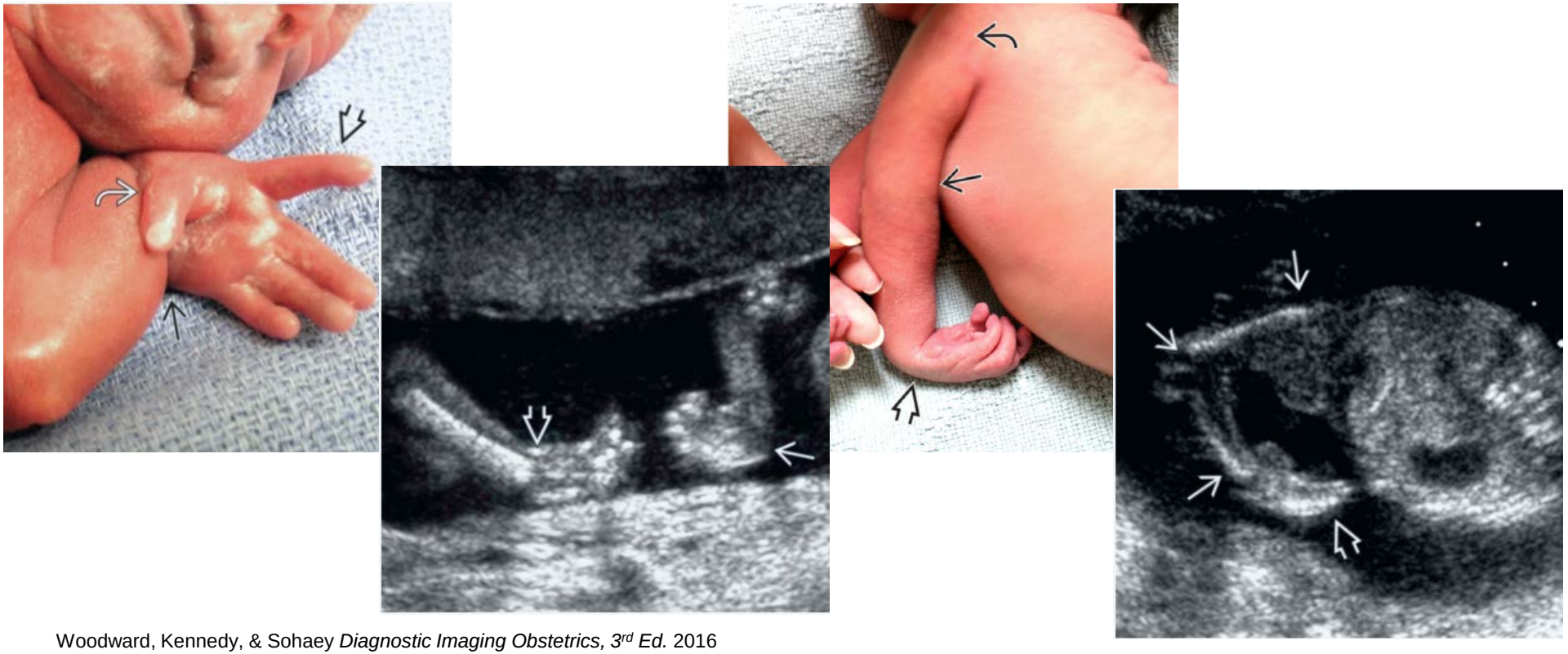
Neurologic: Movement Disorders



Woodward, Kennedy, & Sohaey *Diagnostic Imaging Obstetrics, 3rd Ed.* 2016

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Neurologic: Cranial Defects

- 3/10,000 births
- Cephalocele: defect in the skull/dura with protrusion and exposure of intracranial contents
  - Whole skull (leads to anencephaly) or partial
- Typically fatal postnatally
  - Associated with diabetes, obesity, hyperthermia, and low folic acid
  - Exposure to amniotic fluid leads to neurologic injury
- Evaluation: genetics, screen for risk factors

Woodward, Kennedy, & Sohaey *Diagnostic Imaging Obstetrics, 3rd Ed.* 2016
Bianchi, Crombleholme, D'Alton, Malone *Fetology, 2nd Ed*. 2000

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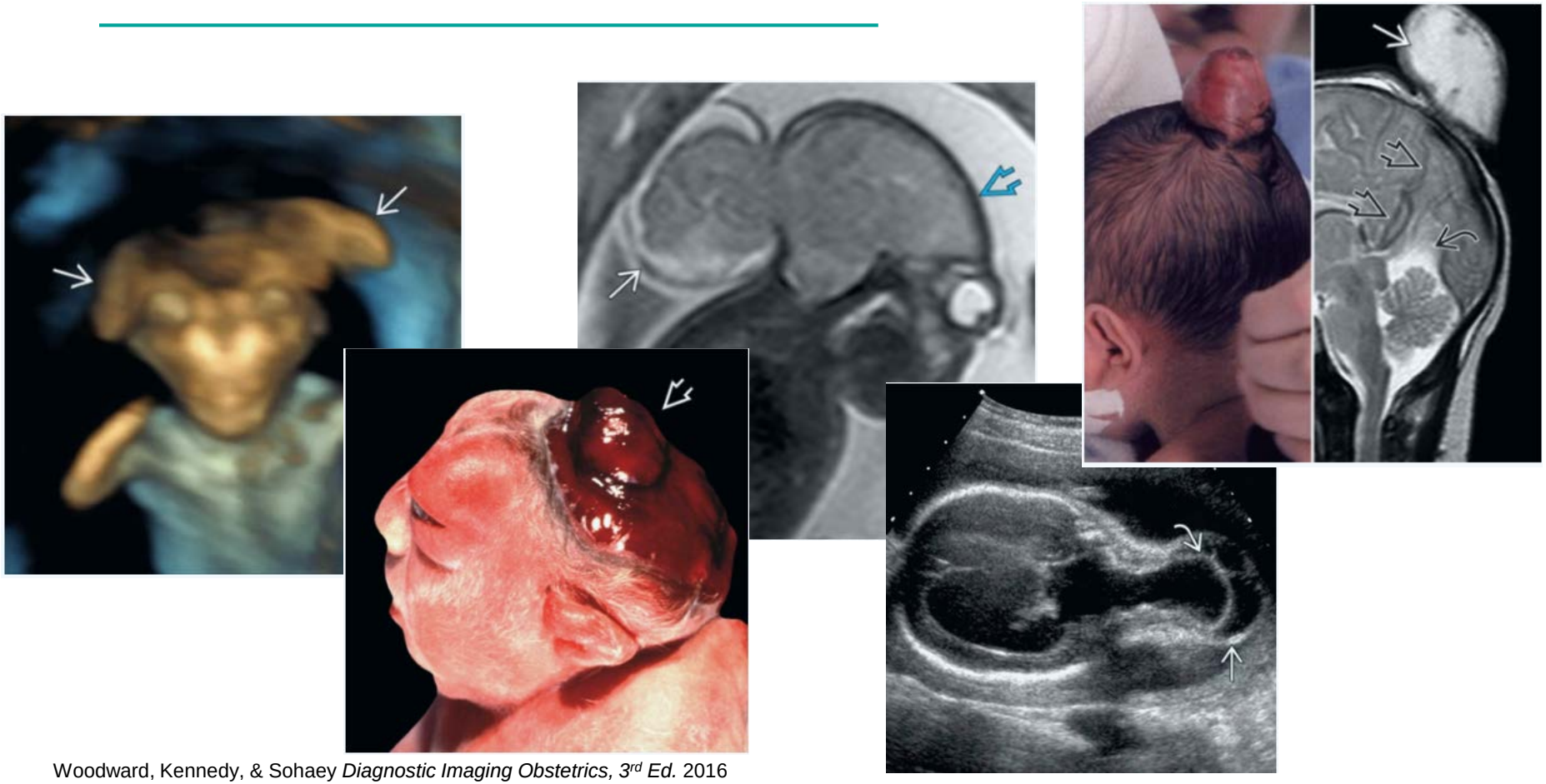
Neurologic: Cranial Defects



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Neurologic: Midline Anomalies

- Approximately 1-2% of population vs 1/8,000 births
- Corpus callosum disruption (pathway connecting hemispheres) vs Holoprosencephaly (hemispheres not separated) vs Cysts
- Outcome varied based on extent of involvement
  - Multiple genetics associations
  - Lower associated with diabetes
- Evaluation: genetics (common), screen for risk factors

Woodward, Kennedy, & Sohaey *Diagnostic Imaging Obstetrics, 3rd Ed.* 2016
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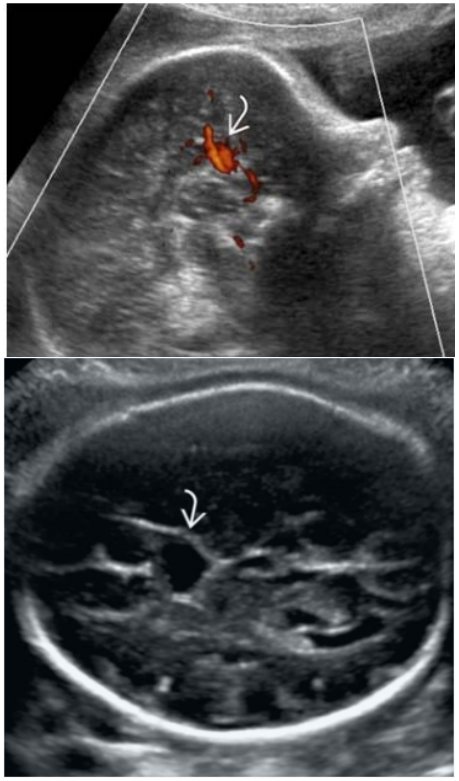
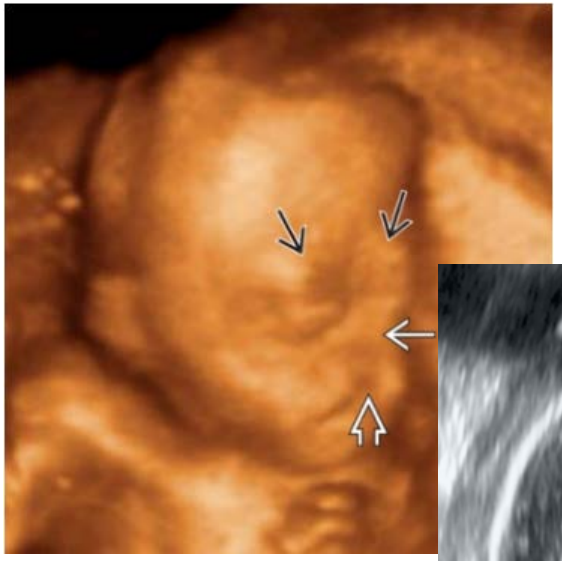
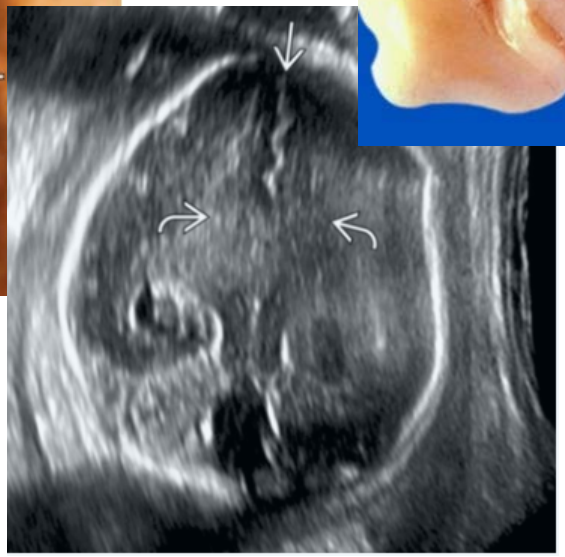
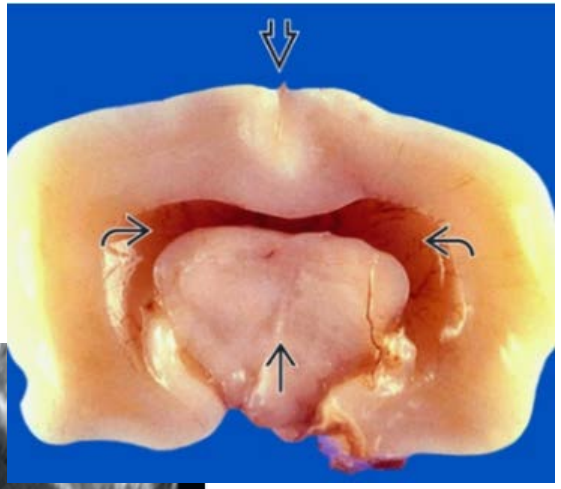
Neurologic: Midline Anomalies









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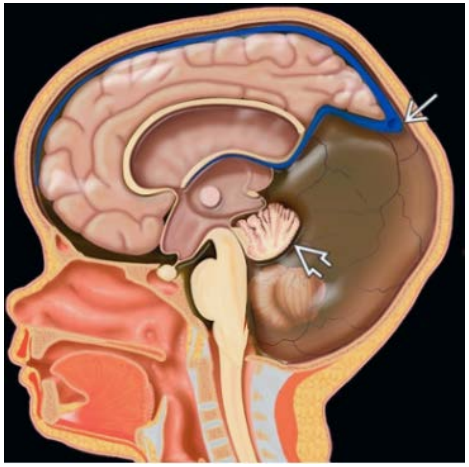
Neurologic: Posterior Fossa Anomalies

- 1/3-5,000 live births
- Dandy Walker or Arnold-Chiari Malformations, cerebellar hypoplasia, rhomboencephalosynapsis, aqueductal stenosis
- Outcome varied, usually not fatal
  - Multiple genetics associations
- Evaluation: genetics



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Neurologic: Spine and Neural Tube

- 1-2/2,000 live births
- Open or closed spina bifida (without or with skin coverage)
- Outcome depends on site of lesions, available of post-natal care and surgery
  - Associated with low folic acid, diabetes
- Evaluation: genetics, screening for diabetes
- May be candidate for fetal surgery

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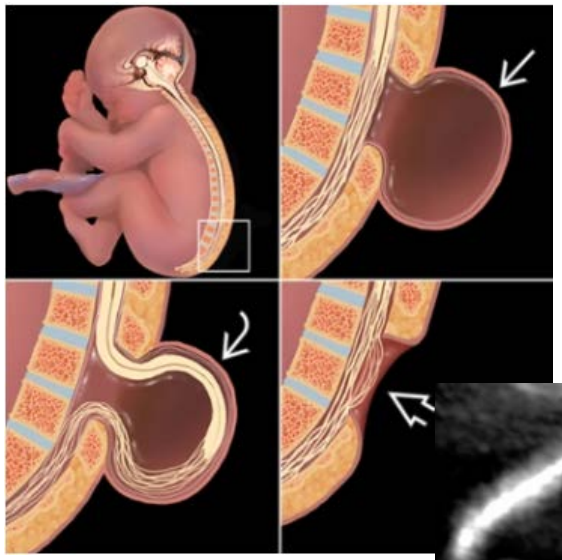
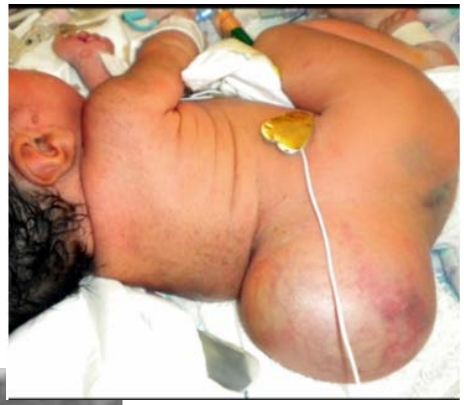
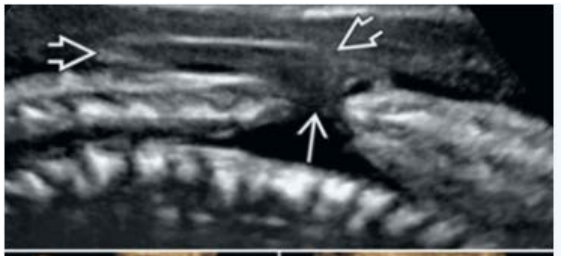
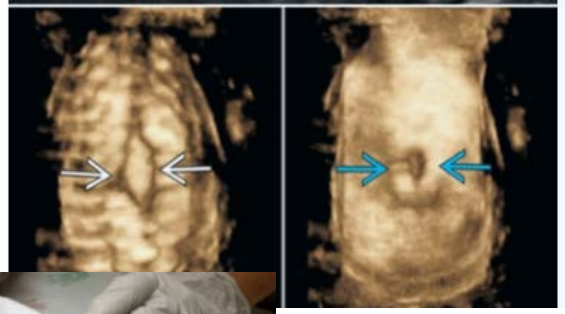
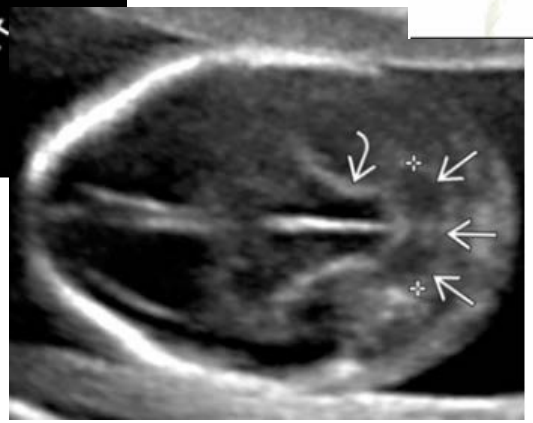
Neurologic: Spine and Neural Tube











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~~Pulmonary: Hernias, Masses~~, and Agenesis

- 1-5/10,000 live births
- Mass of tissue preventing normal lung development
  - Herniation of abdominal contents into thoracic cavity
  - Congenital Pulmonary Airway Malformation
  - Broncho-Pulmonary Sequestration or cyst
- Outcome depends on size and development of lung tissue
- Evaluation: genetics, fetal echo
- May need echo and surgery after delivery

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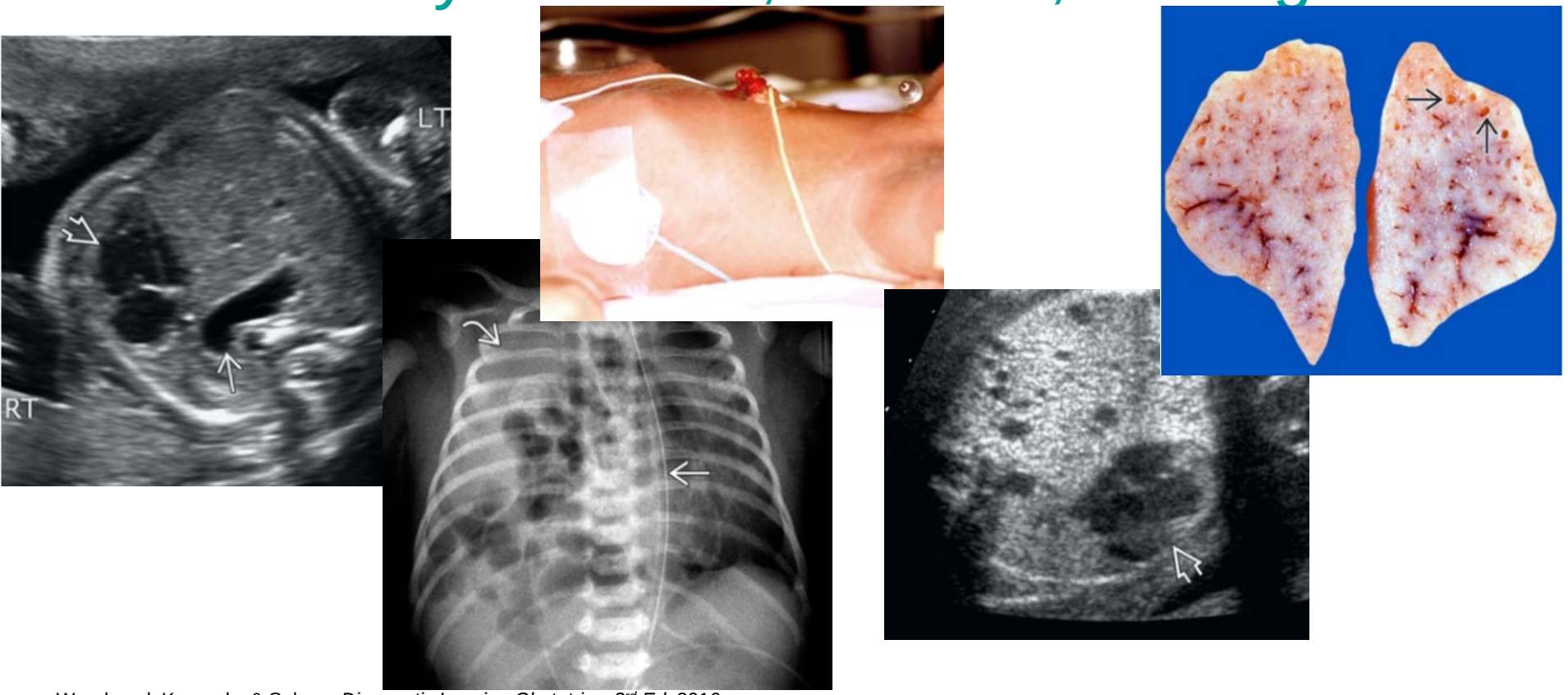
~~Pulmonary: Hernias, Masses~~, and Agenesis



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~~Cardiac: Abnormal Axis~~

- 6/1,000 live births severe CHD (up to 75/1,000 mild CHD)
- Normal axis 35-45º to left of mildline
  - Compare with stomach for heterotaxy or situs inversus
  - May be cono-truncal malformation
  - May be pulmonary mass
- Outcome depends on underlying defect and severity
- Evaluation: genetics, fetal echo
- May need echo and surgery after delivery

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Bianchi, Crombleholme, D'Alton, Malone *Fetology, 2nd Ed*. 2000

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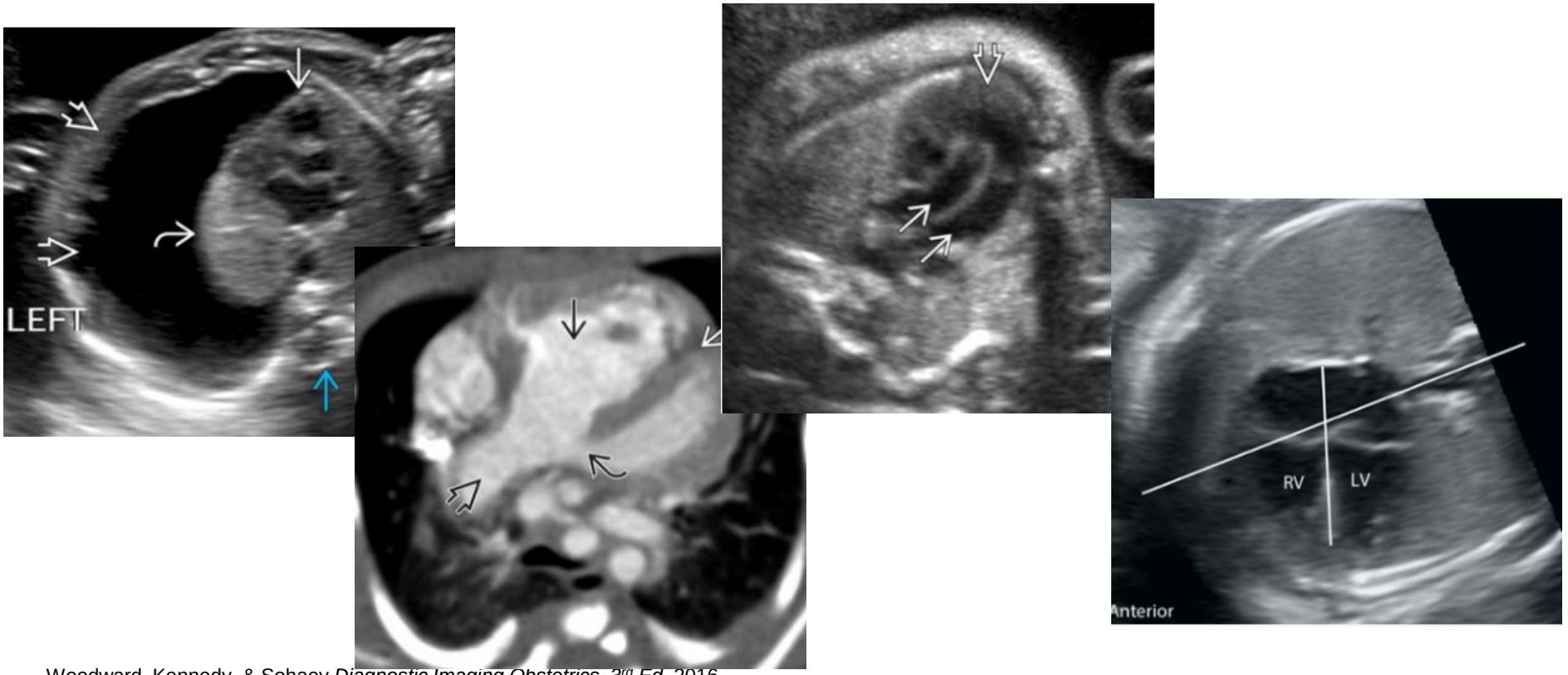
~~Cardiac: Abnormal Axis~~



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~~Cardiac: Chamber Asymmetry~~

- Chambers, Septums, or Valves can affected
- Univentricle: Hypoplastic left or right heart
  - Typically due to valve atresia or stenosis, without septal defect
- Asymmetric, but still two ventricles
  - Septal defects, AV canal, aortic coarctation
- Outcome depends on size and extent of lesion
- Evaluation: genetics, fetal echo
- Typically needs surgery, if compatible with life

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Bianchi, Crombleholme, D'Alton, Malone *Fetology, 2nd Ed*. 2000

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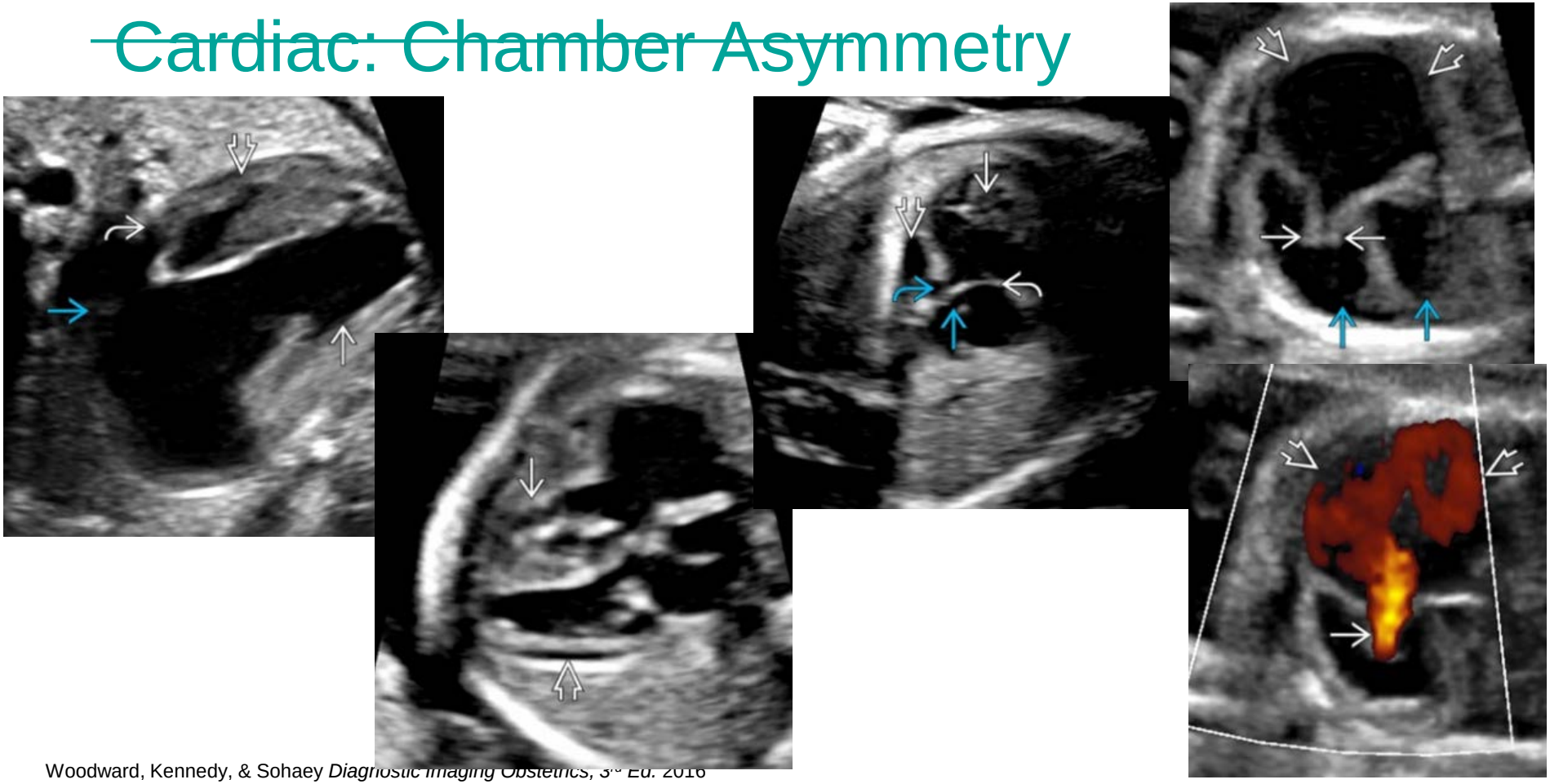
Cardiac: Chamber Asymmetry



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GI: Ventral Wall Defects

- 1/2-5,000 live births
- Bowel herniation through abdominal wall defect
  - Right paramedian without membrane: gastroschisis (simple or complex)
  - Midline membrane-covered into base of cord: omphalocele
- Evaluation: genetics, fetal echo, maternal smoking
- Serial scans to evaluate bowel wall edema and growth
- Hernia can be corrected with surgery, but risk infection

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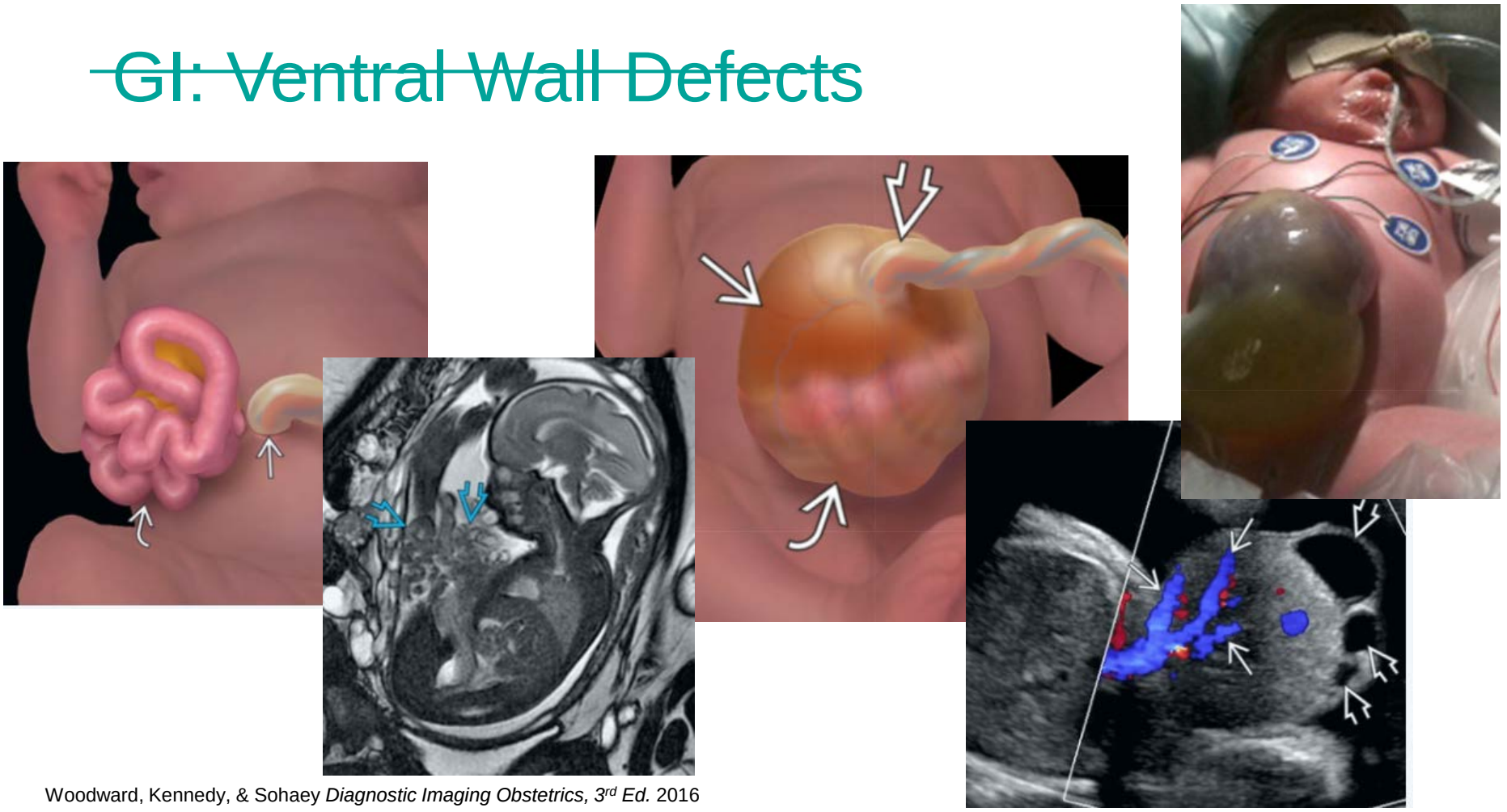
GI: Ventral Wall Defects



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GI: Atresia

- 1-3/10,000 live births
- Complete blockage of the lumen
  - Esophageal, Duodenal, Ileal, Jejunal, Colonic, or Anal
  - Proximal bowel dilated, distal bowel difficult to visualize
  - Polyhydramnios often present
- Outcome depends on size and development of remaining bowel
- Evaluation: genetics
- Should be NPO after delivery until surgery

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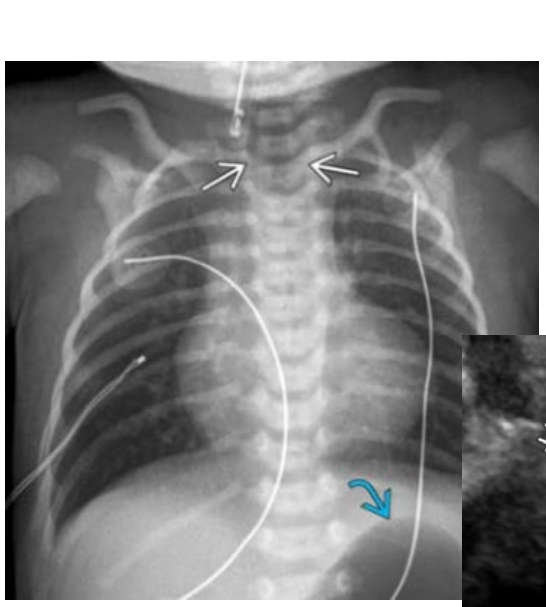
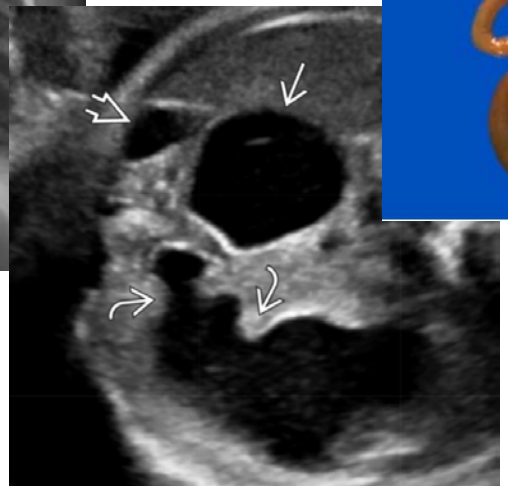
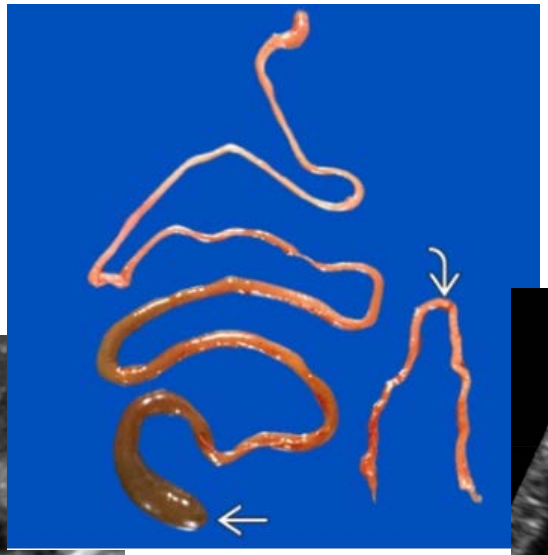
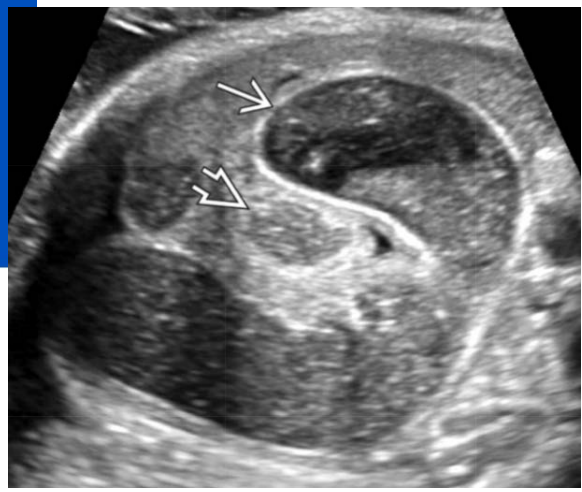
GI: Atresias









Woodward, Kennedy, & Sohaey *Diagnostic Imaging Obstetrics, 3rd Ed.* 2016

MetroHealth

GU: Obstruction, Dilation, and Cysts

- 5/1000 live births
- Abnormal number, location, echogenicity, or size of kidneys
  - Dilation of urinary tract
  - Cystic renal disease
- Outcome depends on residual renal function and laterality
- Evaluation: genetics, cystocentesis, serial ultrasounds
- If bilateral, may develop Potter's Sequence (pulmonary hypoplasia) or need neonatal renal transplant

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Bianchi, Crombleholme, D'Alton, Malone *Fetology, 2nd Ed*. 2000

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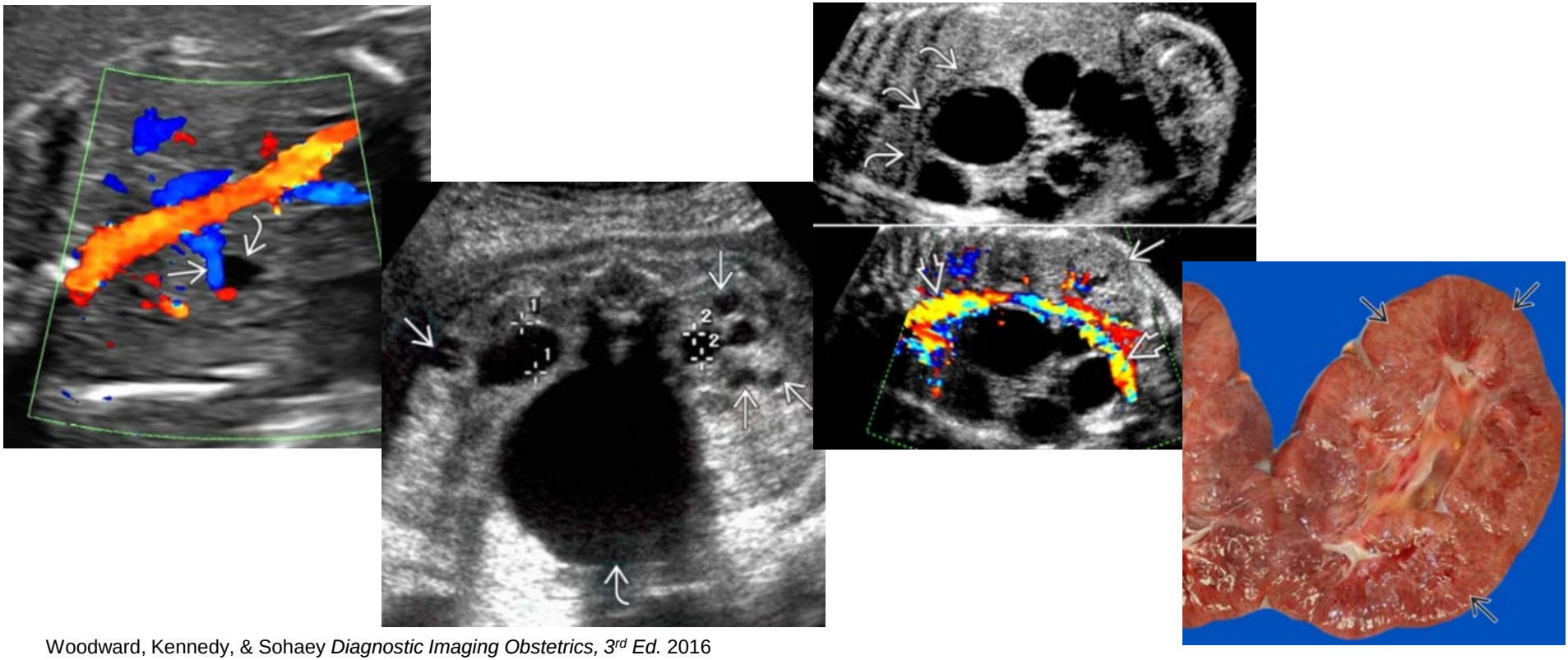
GU: Obstruction, Dilation, and Cysts



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MetroHealth

Skeletal: Dysplasias

- 1/4-5,000 live births (lethal 1/10,000)
- Heterogenous group involving abnormal bone development and growth
  - Several hundred have been described
  - Earlier detection more likely to be lethal
- Outcome depends on etiology and thoracic size
- Evaluation: genetics, serial ultrasounds

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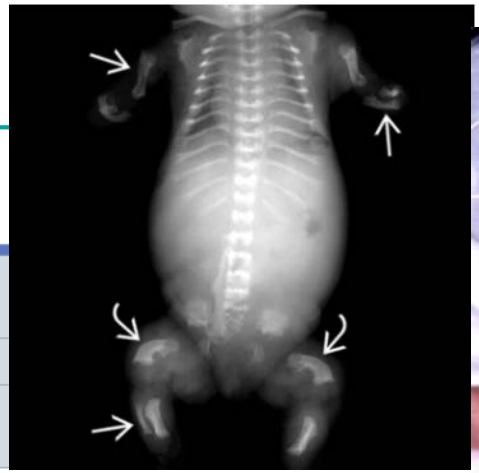
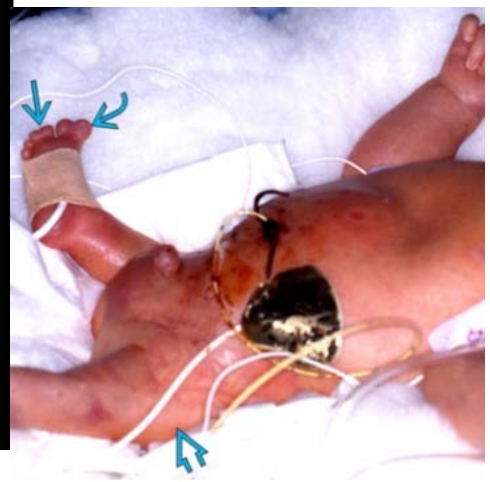
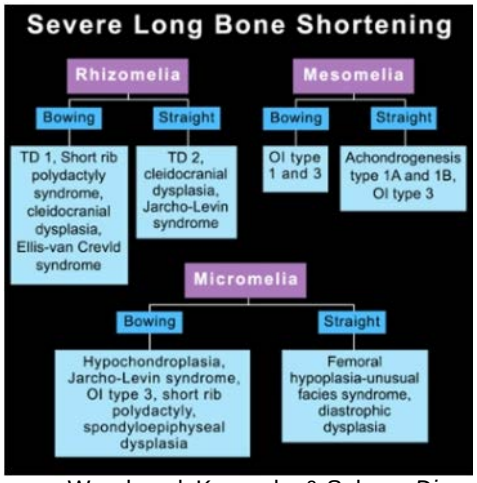
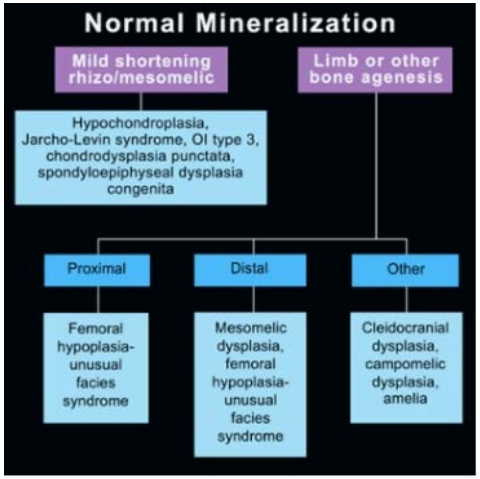
Skeletal: Dysplasias

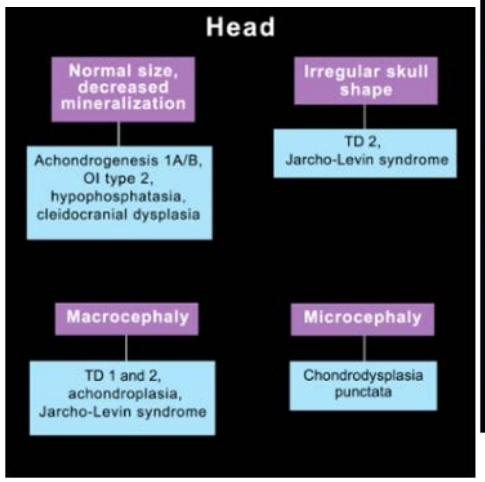
Femur length:foot length ratio	< 1 suggests skeletal dysplasia
Femur length:abdominal circumference ratio	< 0.16 suggests lethality
Chest circumference:abdominal circumference ratio	< 0.8 suggests lethality

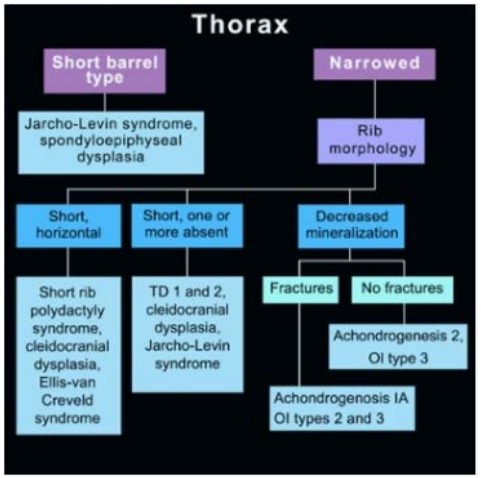












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Aneuploidy: Trisomy 13 (Patau Syndrome)

- 5/1000 births, 3rd most common trisomy
- Multiple anomalies
  - Holoprosencephaly, midline defects, CHD
  - Polydactyly, echogenic kidneys, early IUGR
- Median survival 7-10days, but 5-10% live 12mo+
  - 50% IUFD, 80% of live born pass away on DOL#1
- Evaluation: genetics, serial ultrasounds

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Bianchi, Crombleholme, D'Alton, Malone *Fetology, 2nd Ed*. 2000

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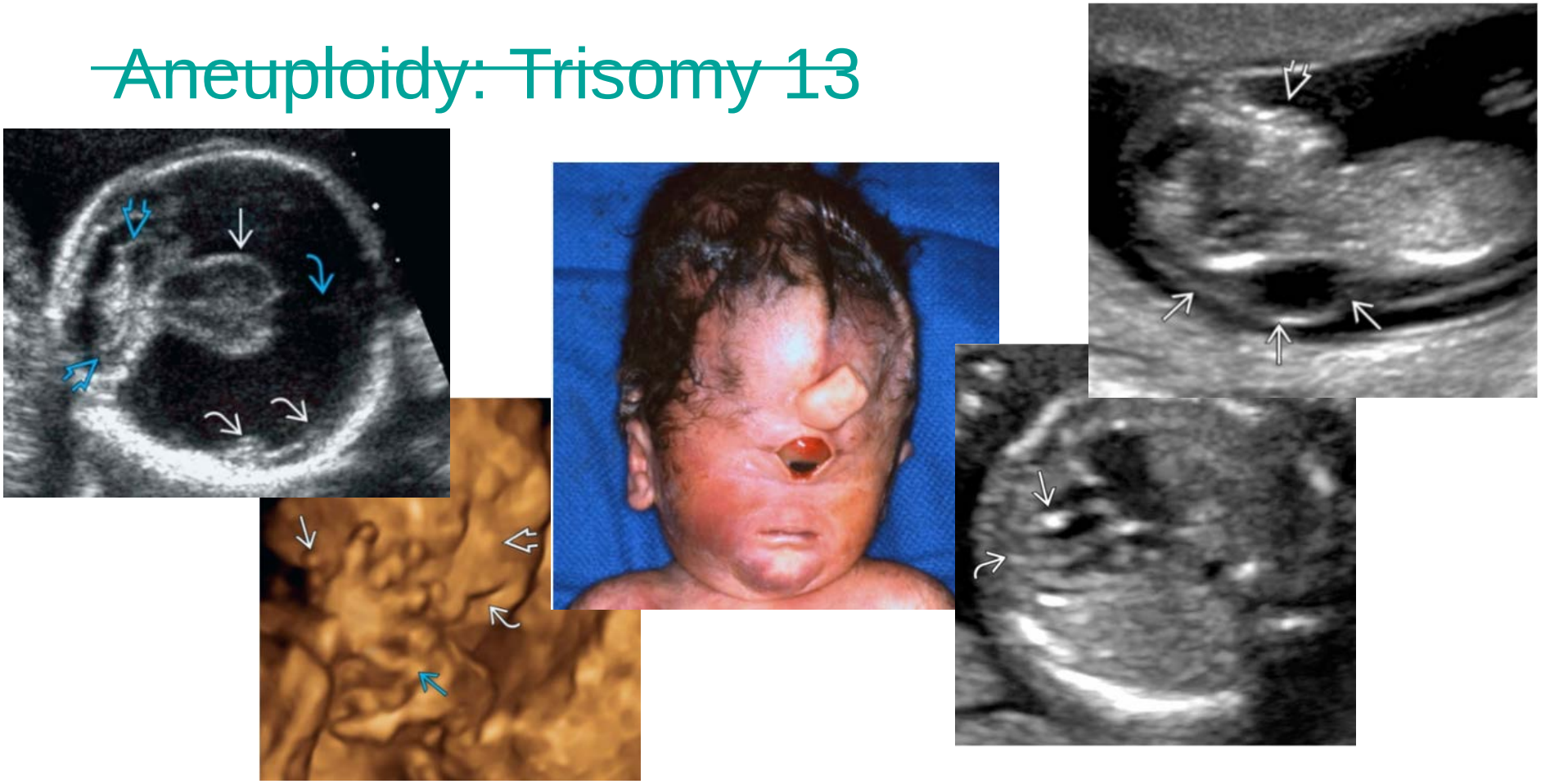
~~Aneuploidy: Trisomy 13~~



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Aneuploidy: Trisomy 18 (Edward's Syndrome)

- 5/3000 births, 2nd most common trisomy
- Multiple anomalies
  - IUGR, CHD, CPCs, CNS anomalies, omphalocele
  - Overlapping fingers/clenched hands
- Median survival 3-13days, but 5-10% live 12mo+
  - 50% IUFD, females tend to live longer than males
- Evaluation: genetics, serial ultrasounds

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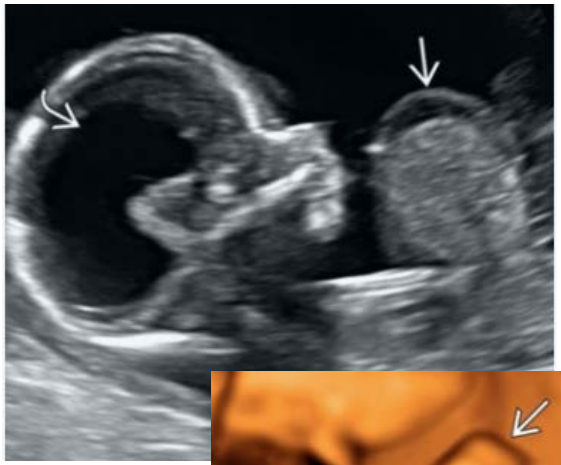
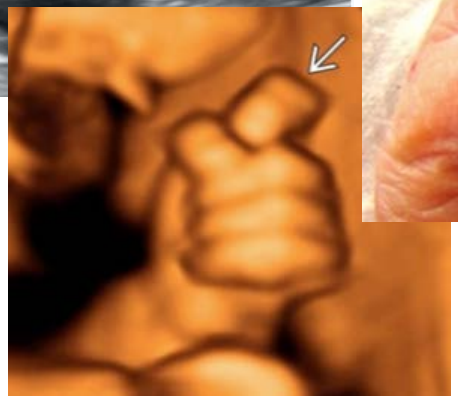
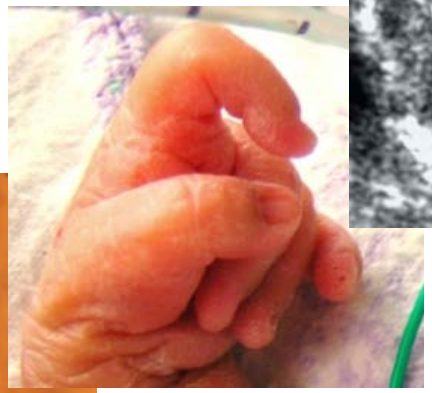
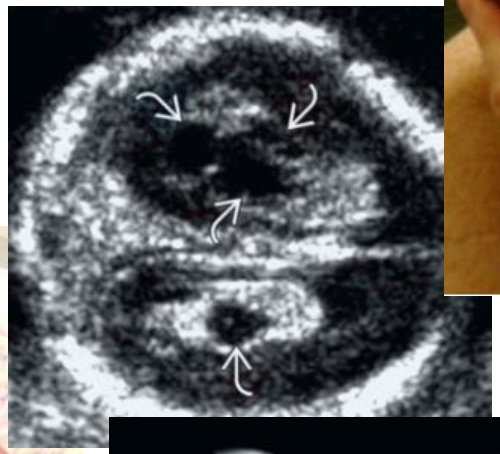
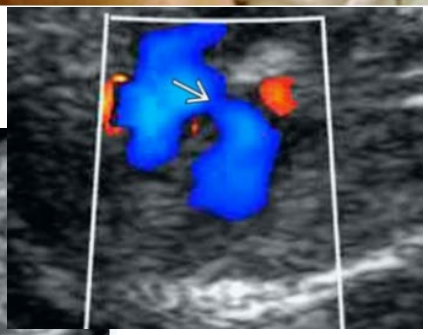
~~Aneuploidy: Trisomy 18~~

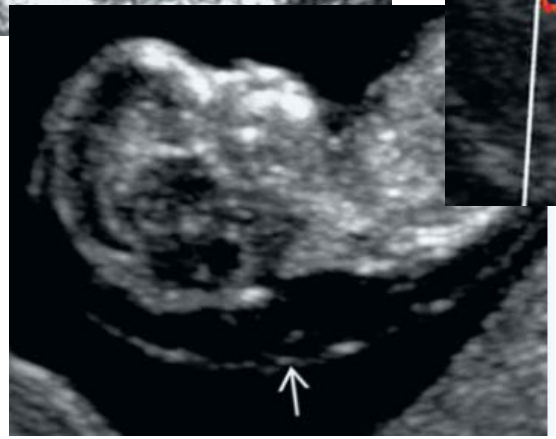












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~~Aneuploidy: Trisomy 21~~ (Down's Syndrome)

- 1/700 live births, Most common trisomy
- Multiple markers
  - Enlarged NT, absent nasal bone, short long bones, pylectasis
  - AV canal defects, duodenal atresia
- 80% live to at least 60yo
  - Prognosis related to underlying anomalies, esp cardiac
- Evaluation: genetics, serial ultrasounds

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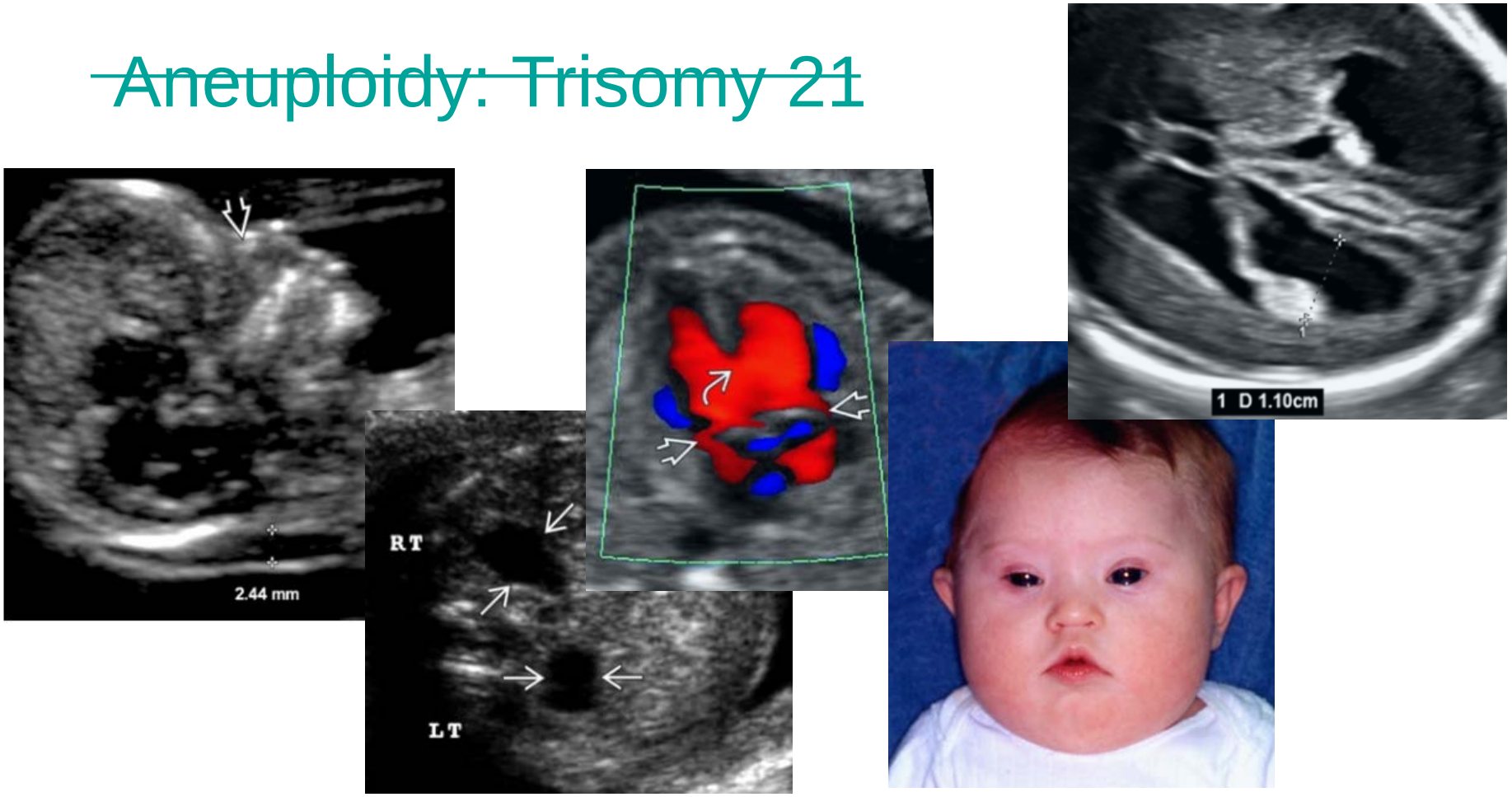
~~Aneuploidy: Trisomy 21~~



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~~Aneuploidy: Monosomy X~~ (Turner's Syndrome)

- 1/2,000 female births, 15% of miscarriages
- Variability in phenotype
  - Mosaics and incomplete X inactivation
- Classic phenotype
  - Webbed neck, broad chest, short limbs, aortic coarctation
- If survive, relatively normal lifespan
- Evaluation: genetics, serial ultrasounds

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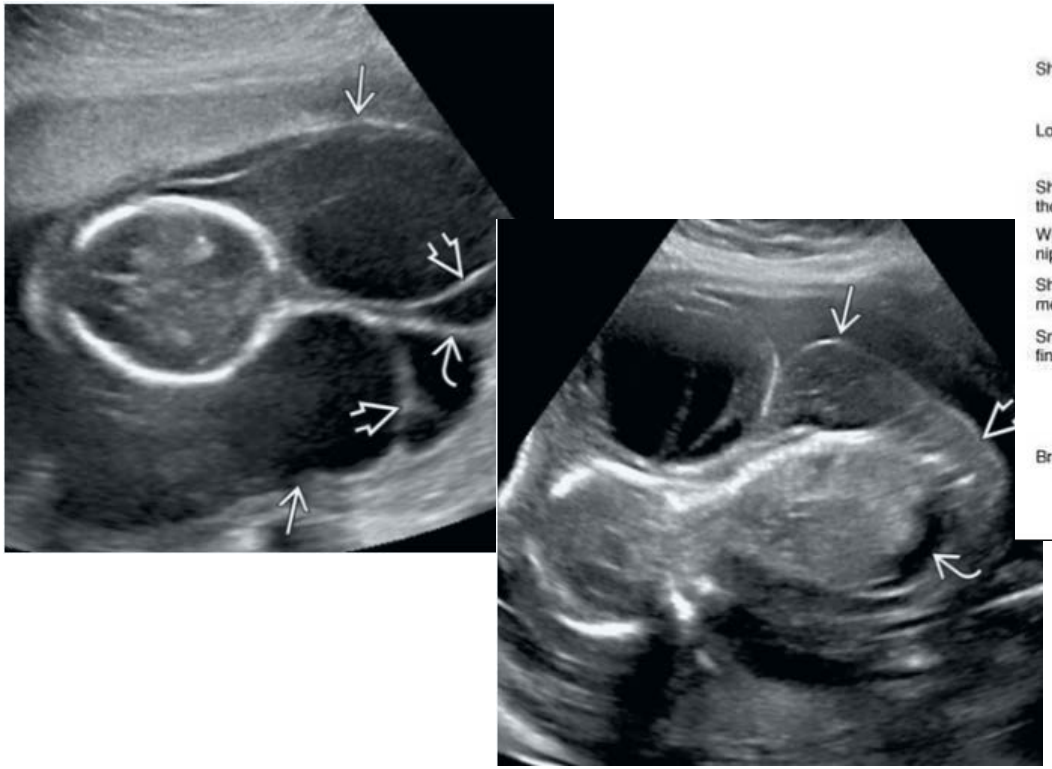
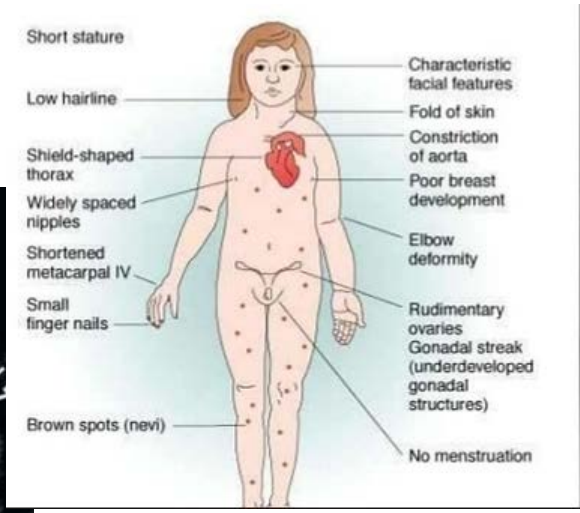
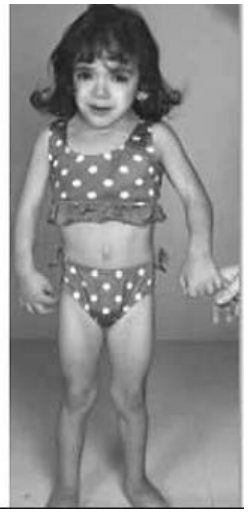
~~Aneuploidy: Monosomy~~ X







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Outline

- Background
- Common congenital anomalies
- **Common congenital infections**
- Steps for evaluation of possible anomalies
- Tips for an effective review
- Case examples

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Congenital Infections: Toxoplasmosis

- 400-4,000 cases per year in USA with 750 deaths
- Transplacental infection with protozoan *Toxoplasma gondii*
- Intracranial and intrahepatic calcifications with IUGR
  - Classic triad of hydrocephalus, intracranial calcifications, chorioretinitis
  - Visual, hearing, motor, cognitive, and other problems in a child
    - May present late
- Severity based on timing of infection
- Evaluation: maternal titers, amniocentesis

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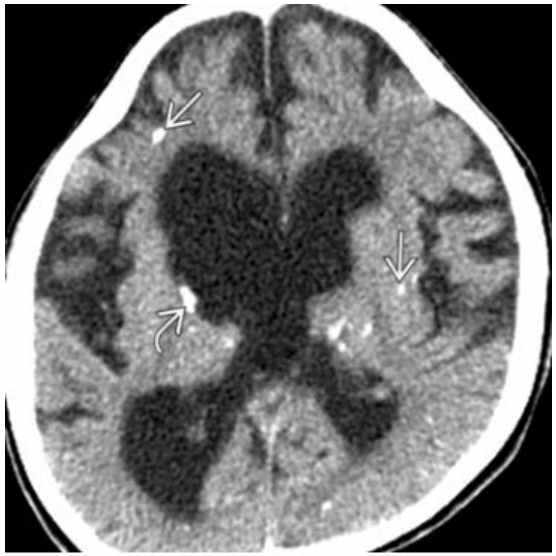
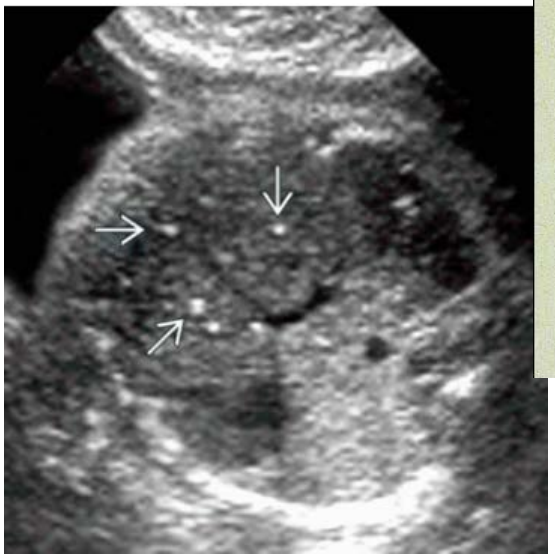
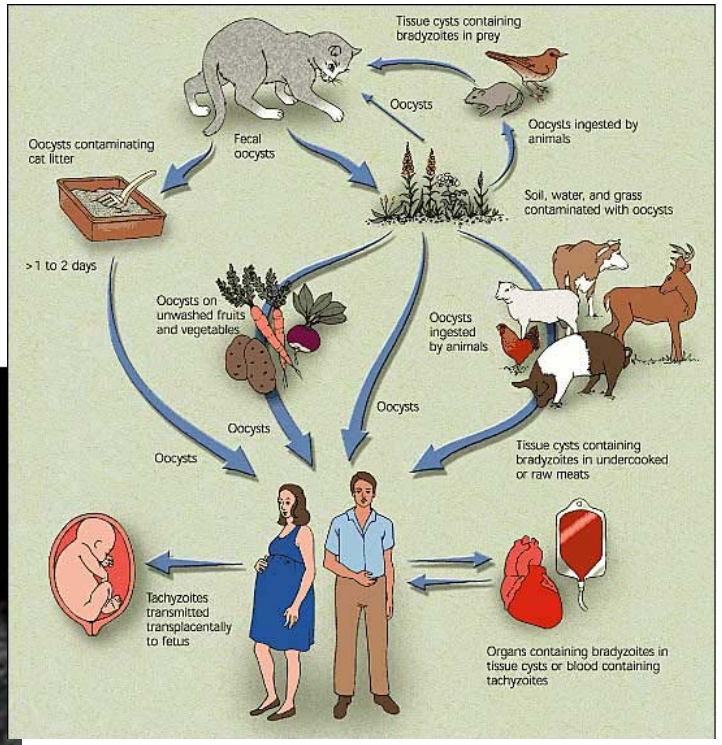
~~Infections: Toxoplasmosis~~







Woodward, Kennedy, & Sohaey *Diagnostic Imaging Obstetrics, 3rd Ed.* 2016;
www.aafp.org

MetroHealth

Congenital Infections: Other: Parvovirus

- Can affect 1-5% of pregnancies
- Transplacental infection with Parvovirus B19
- Classic triad of anemia, heart failure, hydrops
  - 8-27% of hydrops cases
  - Parvovirus attacks erythroid progenitor cells → aplastic anemia
    - Thrombocytopenia also common and may be severe
- Severity based on timing of infection
- Evaluation: ultrasound, maternal titers, MCA dopplers, PUBS

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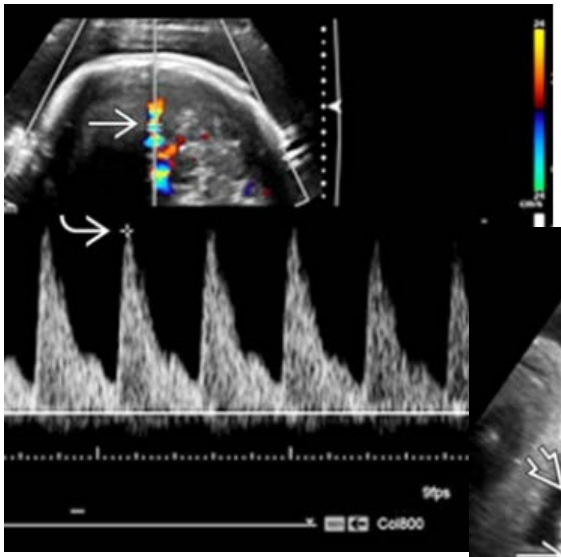
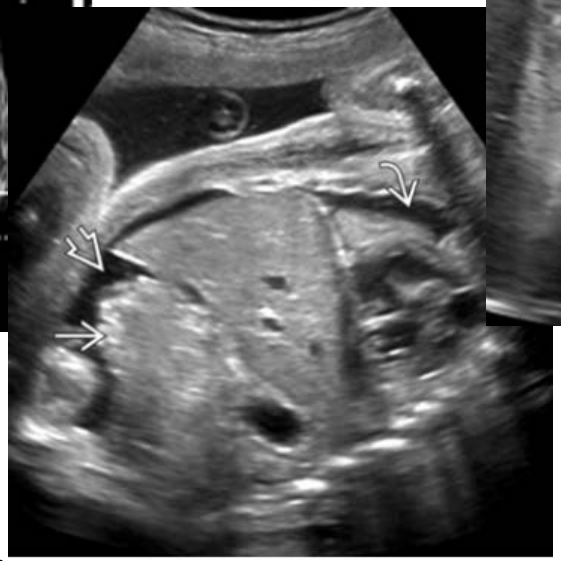
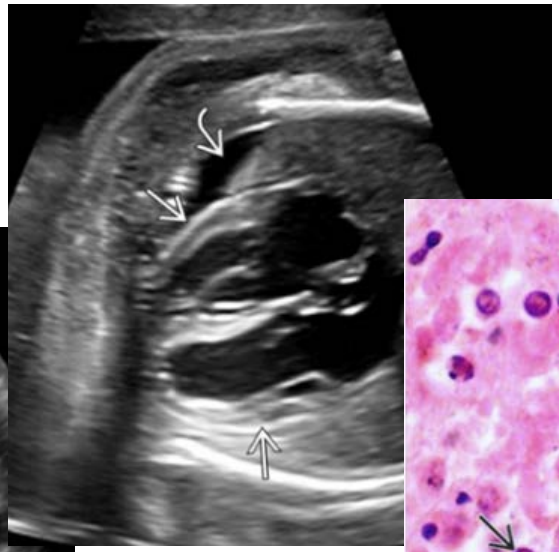
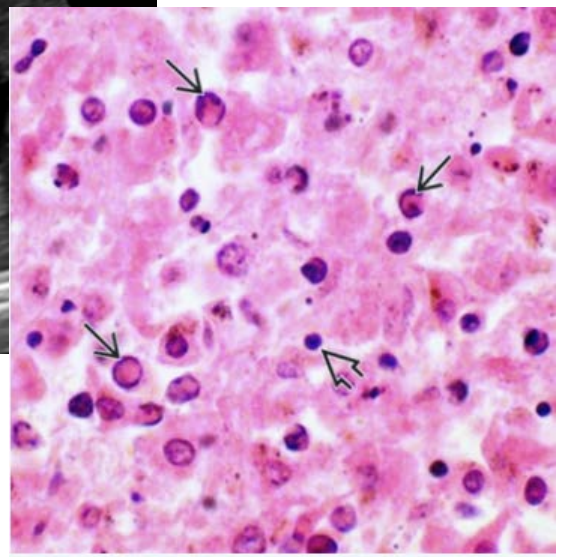
Infections: Parvovirus









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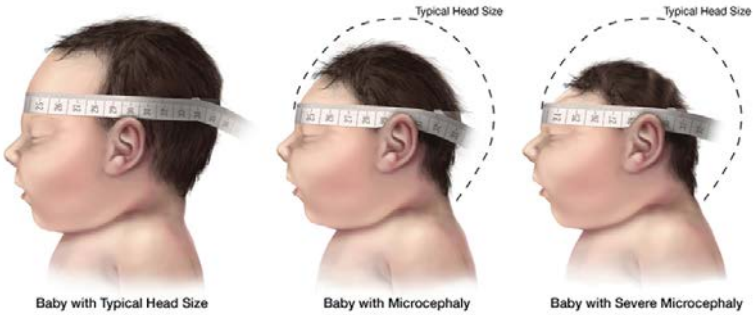
Congenital Infections: Other: Zika

- 400-4,000 cases per year in USA with 750 deaths
- Transplacental infection with Zika virus
- Microcephaly, IUGR, optic nerve changes
- Severity based on timing of infection
  - 5-10% of confirmed infections associated with birth defects
- Evaluation: maternal titers, amniocentesis, serial ultrasound



www.cdc.gov

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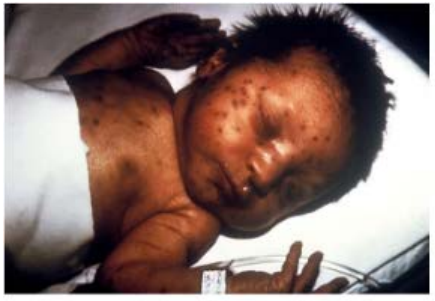
Congenital Infections: Rubella

- Rare in vaccinated populations
- Transplacental infection with Rubella virus
- Causes CHD, microcephaly, cataracts, deafness, IUGR, and liver and spleen damage.
- Severity based on timing of infection
- Evaluation: maternal titers, amniocentesis



www.cdc.gov,

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Congenital Infections: CMV

- 0.3-2.4/100 live births worldwide (most common infection)
- Transplacental infection with cytomegalovirus
- Multiple intracranial findings
  - Ventriculomegaly, Calcifications, Intraparenchymal cysts, intraventricular adhesions, Microcephaly
  - Hepatosplenomegaly, anemia, cardiomegaly, and IUGR
- Severity based on timing of infection
- Evaluation: maternal titers, amniocentesis

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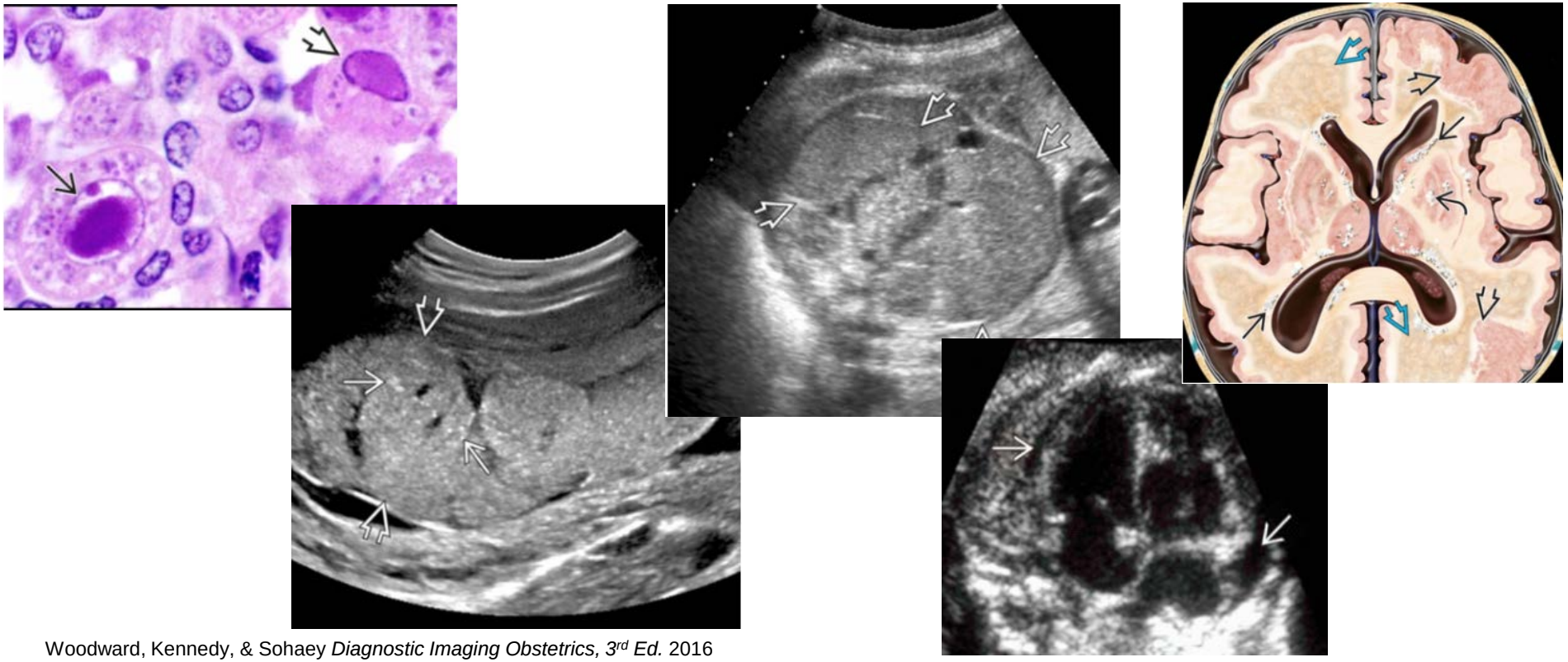
Infections: CMV



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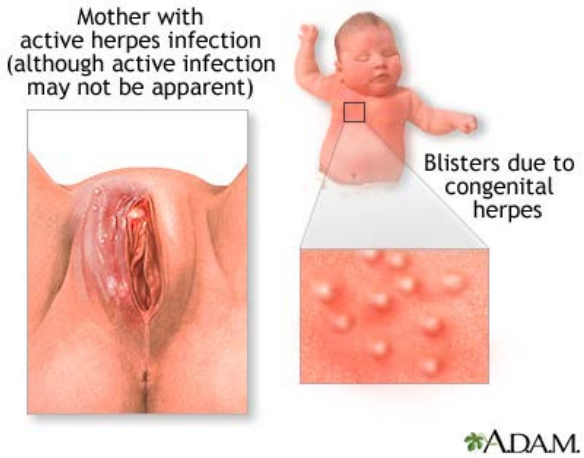
Congenital Infections: Herpes

- 1/3-20,000 live births, usually acquired in genital tract
- Transplacental infection with *Herpes Simplex Virus*
- Usually present within a week of life
  - Seizures, apnea, jaundice, shock
- Outcomes based on severity and initiation of treatment
- Evaluation: maternal cultures, neonatal cultures



www.medline.gov

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Outline

- Background
- Common congenital anomalies
- Common congenital infections
- **Steps for evaluation of possible anomalies**
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Evaluation Steps: Ultrasound

- Anatomy can be examined in any trimester
  - Limited, often transvaginal in first trimester
- Detailed anatomic survey
  - Focus on Brain, Heart, Spine, Diaphragm, Kidneys
- Follow the fetal growth and fluid

- For IUFD, evaluate for hydrops, any anatomic “clues”
  - Offer amniocentesis for genetics and infectious evaluation

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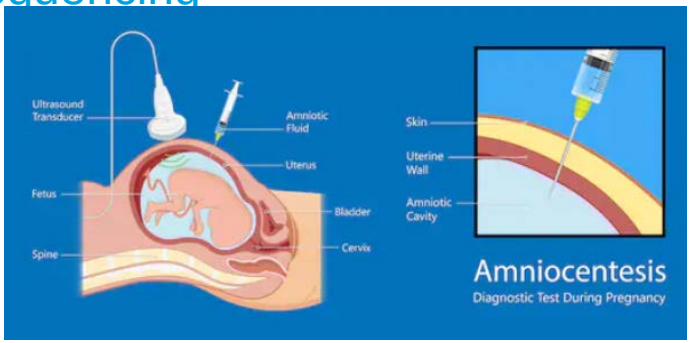
Evaluation Steps: Genetics

- Tests guided by history, suspected etiologies
- Anniocentesis typically yields the best results
  - Fascia lata or placenta can be used as alternatives
- Send for karyotype
  - Microarray detects additional 6% of abnormalities
  - Consider whole exome sequencing



www.genome.gov, www.shutterstock.comc

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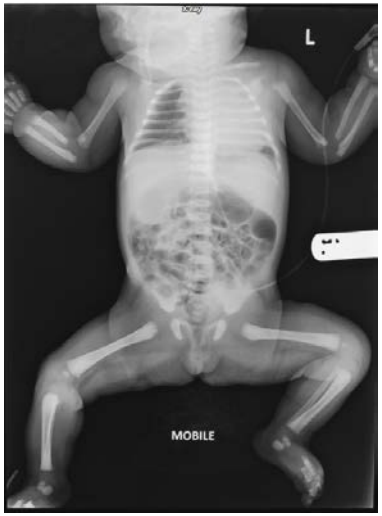
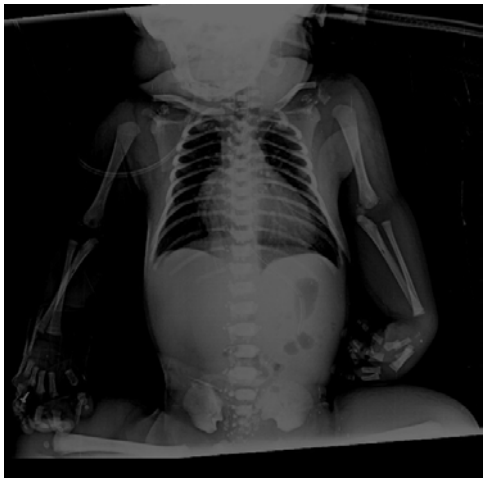
~~Evaluation Steps: Clinical~~ Exam and Babygram

- Thorough evaluation of the external anatomy
  - Including weight, length, head circumference
- Babygram can evaluate skeletal structure
  - Especially helpful if autopsy declined





https://radiopaedia.org/

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Evaluation Steps: Placenta and Autopsy

- Placenta, including membranes and cord
  - Reveals cause in up to 30% of cases
  - Useful for determining infection, esp in preterm cases
- Autopsy
  - Single most useful evaluation
  - Provides information in 30% of cases

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Tips for Effective Review: Family History

- Take detailed family history
  - Recurrent miscarriages or early pregnancy losses
  - Childhood deaths or early adult deaths
  - Surgeries as a child
  - Race/Ethnic background

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Tips for Effective Review: Chart Review

- Personal medical history in the patient
- Travel
- Medication exposures
- Co-morbid conditions
- Any recent illnesses
- All prenatal records

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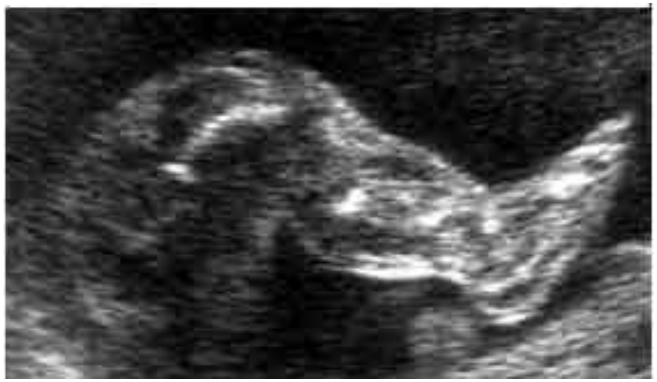
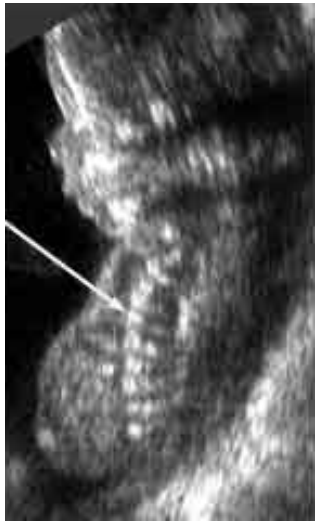
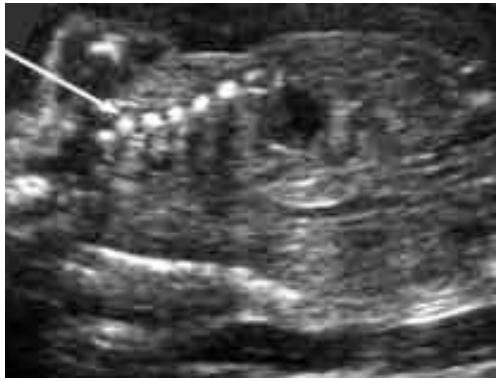
Case 1

- 31yo G3P1011 at 13 weeks
- Short long bones with narrow chest and frontal bossing noted







www.sonoworld.com

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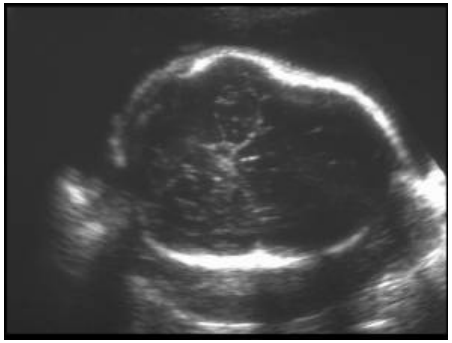
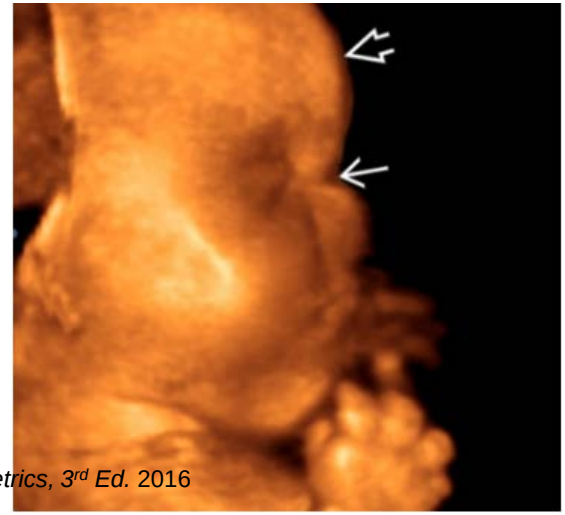
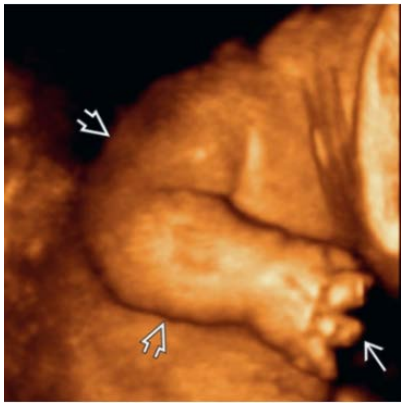
Case 1

- Cloverleaf skull, frontal bossing, and trident hand with micromelia
- Amniocentesis performed







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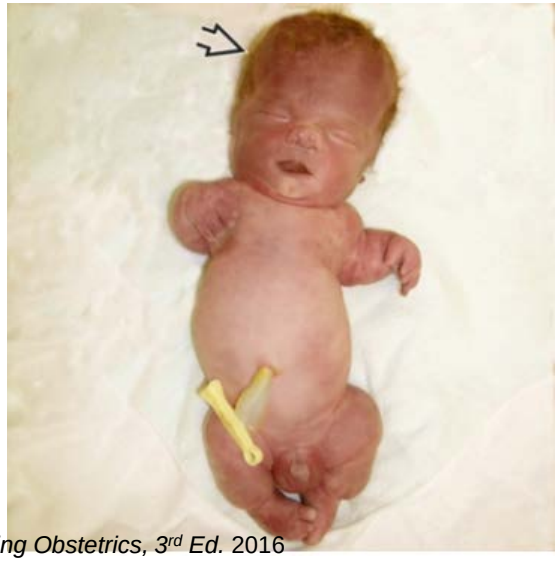
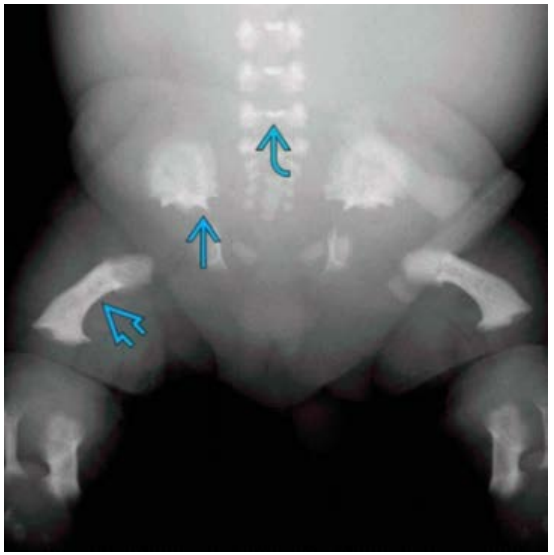
Case 1

- Tiny chest and disproportionately large head and long
- Rhizomelic shortening with curved femora is typical with severe micromelia

- Thanatophoric dysplasia





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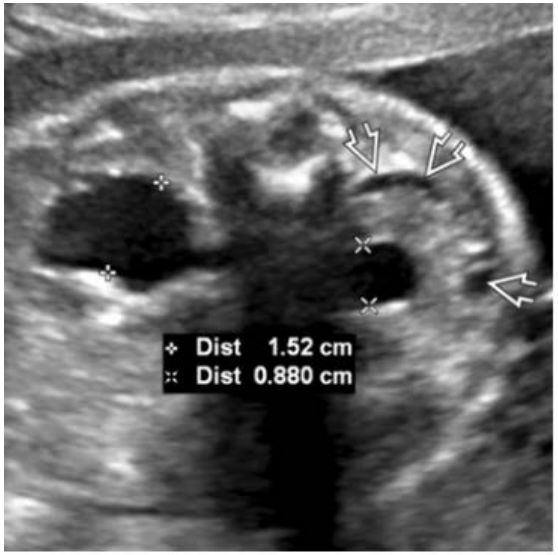
Case 2

- 24yo G1P0 presenting for her anatomy scan
- No significant history
- Smokes, no other drug use

- Bilateral pyelectasis noted



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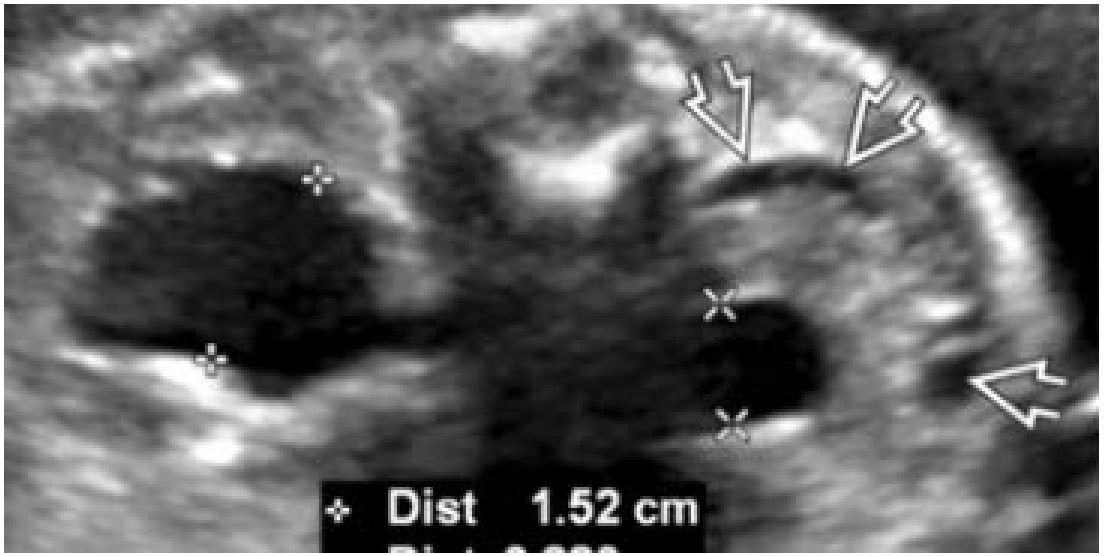
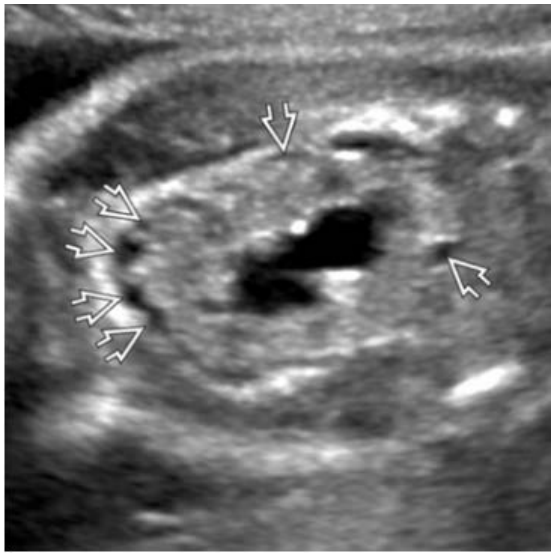
Case 2

- Subtle subcortical cysts are seen, suggesting the obstruction has caused renal dysplasia.





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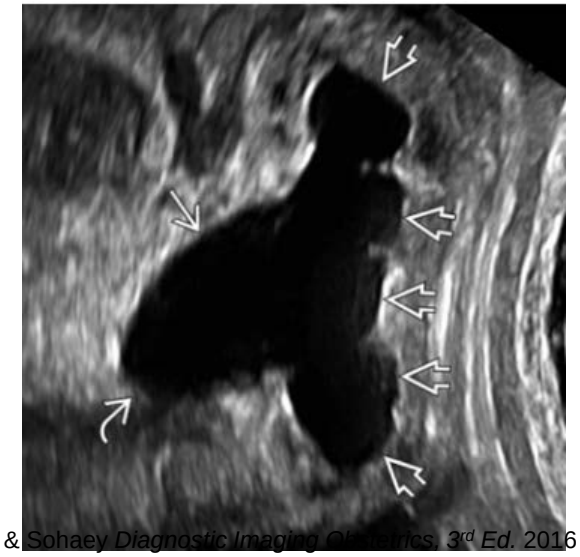
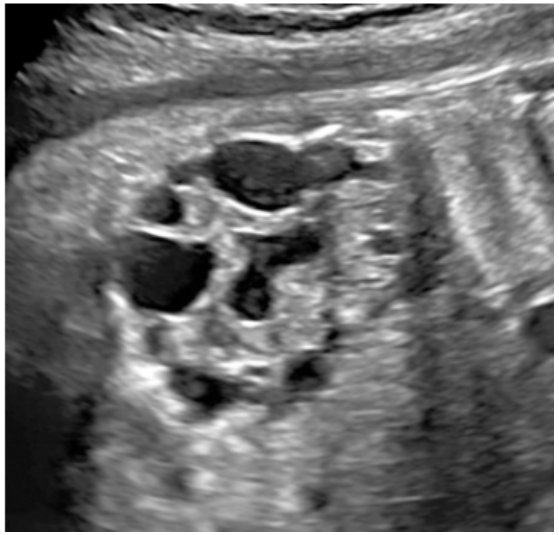
Case 2

- Later, obstruction is noted at the ureteral-pelvic junction
- Cortical cysts have increased in size and further parenchymal cysts have formed





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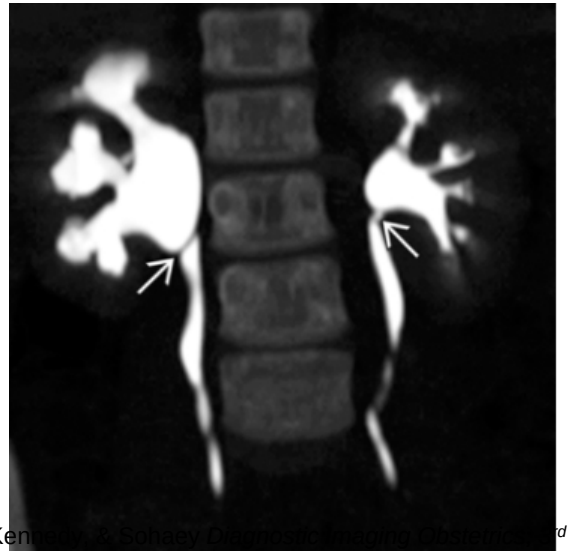
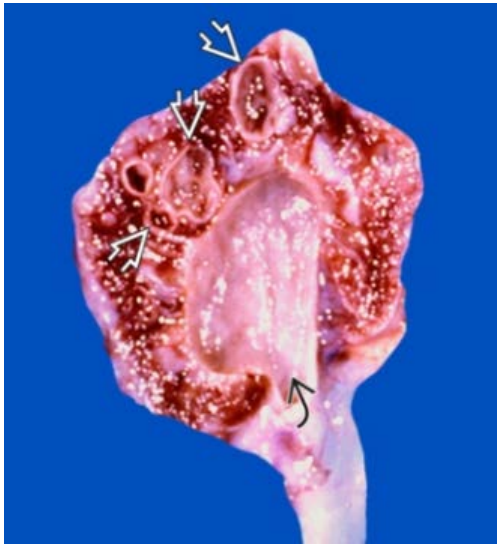
Case 2

- CT urogram after birth shows near complete UPJ obstruction, leading to renal failure
- Pathology reveal renal dysplasia





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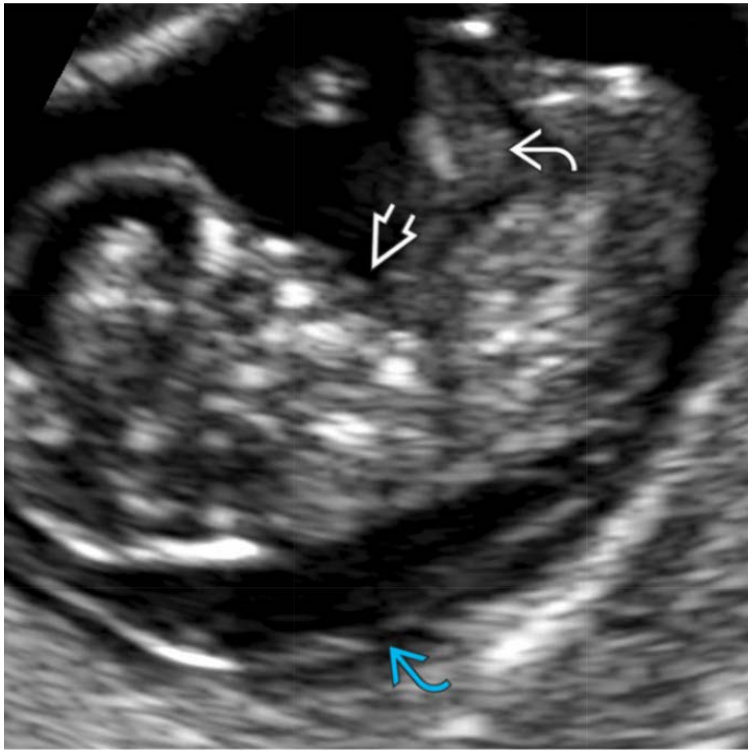
Case 3

- 41yo G4P2012 at 12 weeks
- Planning genetic screening

- Large cystic hygroma
- Micrognatha

- CVS performed



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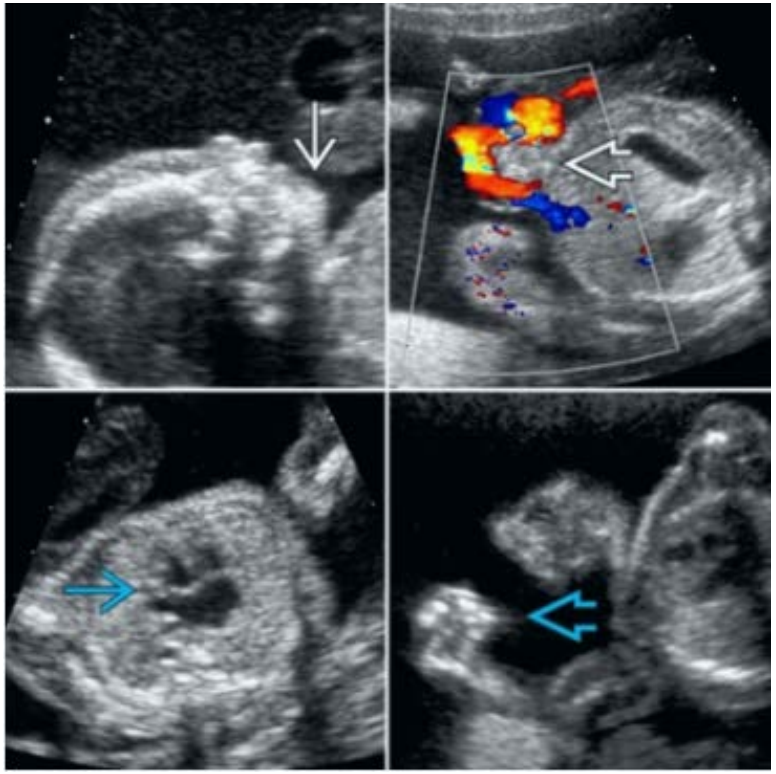
Case 3

- At 18w:
  - Micrognathia
  - Omphalocele
  - AV canal defect
  - Clenched fist
- CVS: trisomy 18



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Case 3

- Induced for IUGR
- Discharged on day of life #3
- Survived for 31 days
- Passed away peacefully at home with her parents

https://www.trisomy18.org/story/bridget-noras-story/

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Contra Costa FIMR Program
Folic Acid Community Campaign

Special thanks to the current and past members of the Contra Costa FIMR program, California. This information is presented with the permission of Christina Boothman, Natalie Berbick, and Dawn Dailey

Contra Costa FIMR case review findings:

- A large proportion of Latina women in the catchment area experienced a loss due to a neural tube defect.
- Many of the women were not aware of recommendations related to folic acid intake.
- Some women received folic acid intake education from their health care provider yet did not take the next step to purchase supplements.
- There was a lack of evidence that folic acid information was consistently and comprehensively provided to women during encounters with health care providers.

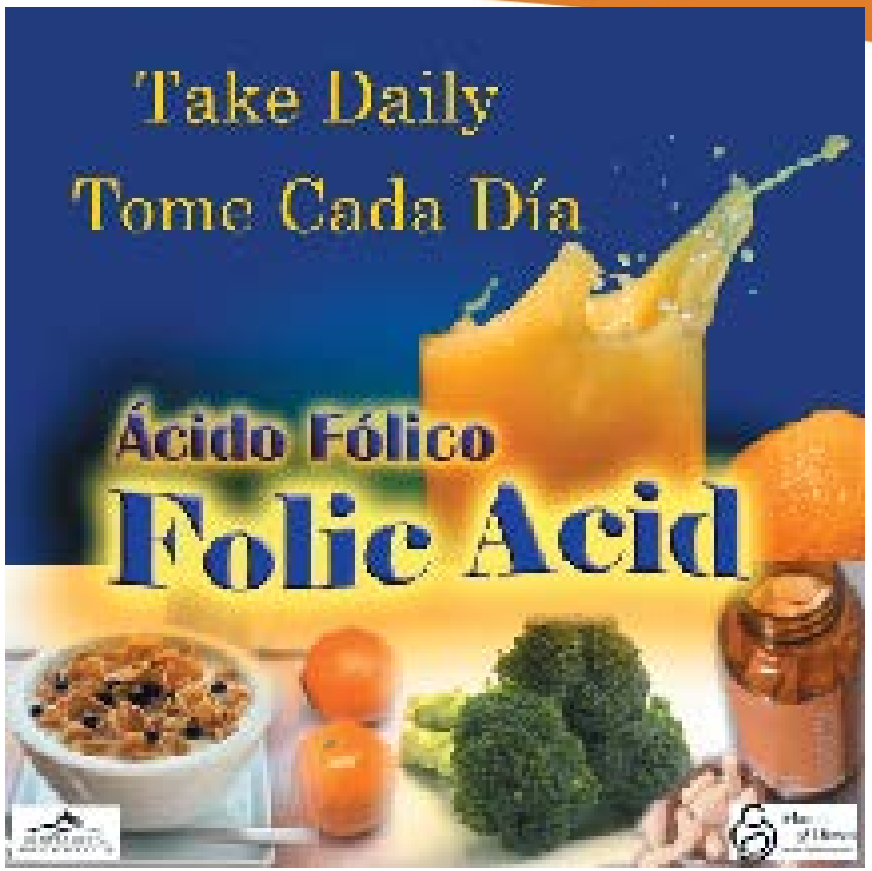
Folic Acid Community Campaign: A public Health Approach

- Collaborative efforts: The Contra Costa FIMR Program collaborated with several programs and community-based agencies on the design and implementation of the campaign.
- Sponsors:
  - Family, Maternal & Child Health Programs of Contra Costa Health Services, March of Dimes, USDA, and California Nutrition Network.

https://cchealth.org/folic-acid/community-campaign.php

Folic Acid Community Campaign: A public Health Approach

Media activities included on-screen theater advertising in three local movie theaters, project website, mailing to food stamp recipients, local cable TV shows and advertising, and transit advertising.



Folic Acid Community Campaign: A public Health Approach

Regional trainings: Folic acid trainings were conducted for public health nurses, community health workers, home visiting program staff, nutrition staff, public health interpreters, health educators and other health care providers.

Folic Acid Community Campaign: A public Health Approach

Educational materials and incentives: A series of educational materials were developed, including folic acid brochures in English, Spanish, Vietnamese, Russian, Farsi and Lao and a magnet, mailer/bookmark and fact sheet in English and Spanish

The National Center for Fatality Review and Prevention

NATIONAL CFRP Saving Lives Together

Questions

- As a reminder:
  - Questions can be typed into the “Questions” pane
  - Due to the large number of attendees, we may not be able to get to all questions in the time allotted
  - All unanswered questions will be posted with answers on the NCFRP website
  - **Recording of webinar and copy of slides will be posted within 2 weeks on the NCFRP website: www.ncfrp.org**

NCFRP is on Social Media: NationalCFRP

THANK YOU!

Additional questions can be directed to:
info@ncfrp.org

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