Module Presentation

How to best use the Modules

To make the best use of this training we encourage you to complete each Module in order following the format below:

1. Read *Module Presentation*. Added explanations can be found in the HELPER Guidelines and in the extra information section if there is one.
2. Complete the *Extraction/Scenario* training exercises
   The extraction exercises use de-identified and altered patient medical records. The information is then entered into the provided section from the Birth Certificate Workbook.
   The Scenarios are situations you may encounter as you collect information from your patients' medical records.
3. Check your responses using the answer sheets in the "Answers" section.
4. Complete the Module specific *Evaluation*, faxing or emailing the completed evaluation to: rosemary_varga@urmc.rochester.edu.
   We will use these evaluations to identify areas where the training can be improved.
5. If not already done, read extra training materials, if available.

If you have questions about how to answer any of the requests for information in the NYS Certificate of Live Birth Training Modules,

Please, contact Rosemary Varga (585-275-8737).

*"Coding" is a convenient although slightly misleading term for entering the needed information in the Statewide Perinatal Data system. True "coding" is the entry of predetermined numbers into a system that can then rate the material. We do not use numbers rather we enter the requested information.

Module Two

Congenital Anomalies Screen

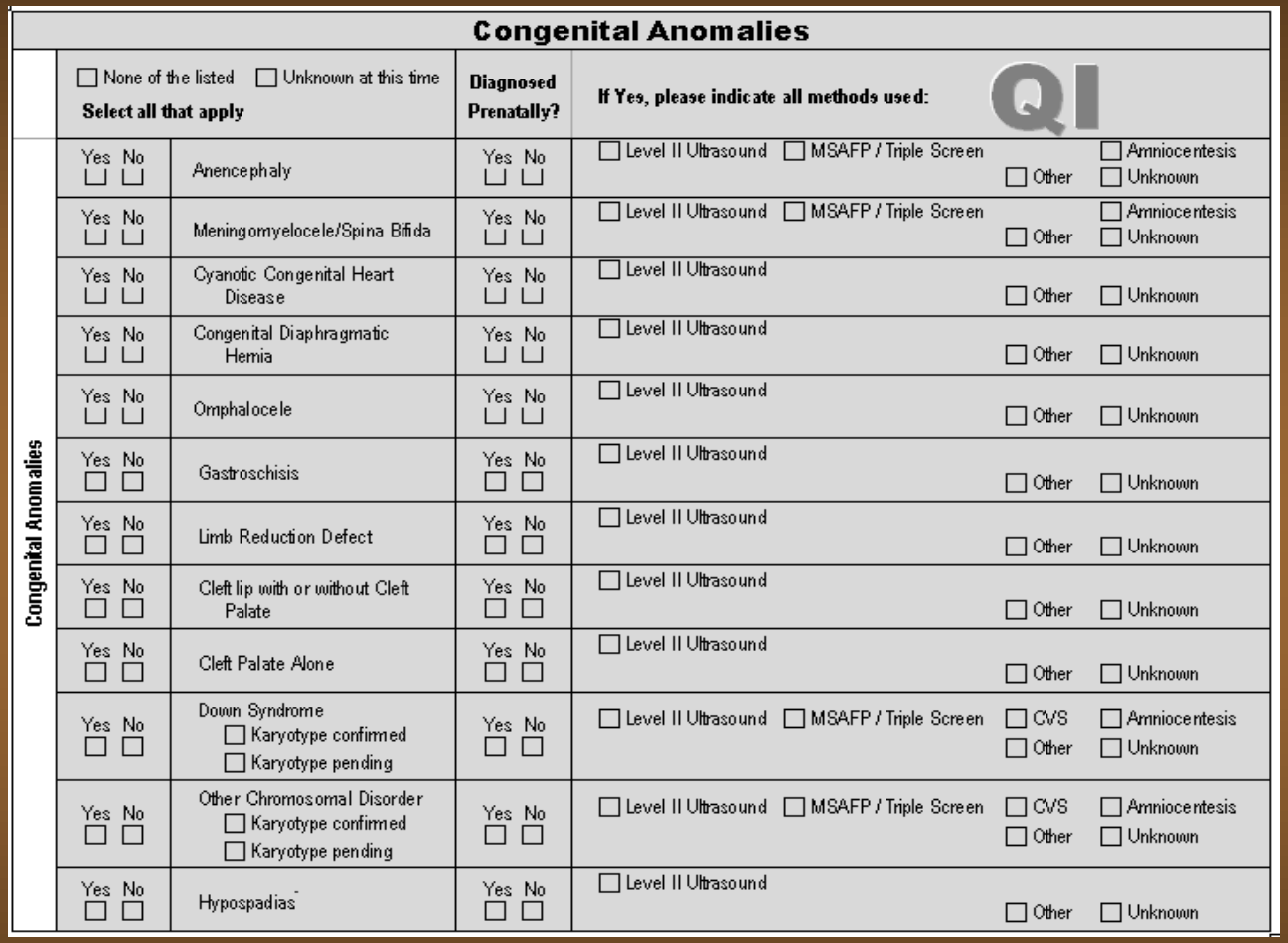
Why code Congenital Anomalies?

- Accurate coding of Congenital Anomalies provides information which can help establish the need for services, causal links, and changes in the incidence of the defect.
- And, while only those listed on the Birth Certificate are coded, ALL congenital anomalies are reported in the NYS Congenital Malformations

  Registry through the Health

  Commerce System (HCS)

Birth Anomaly Fields



Congenital Anomalies

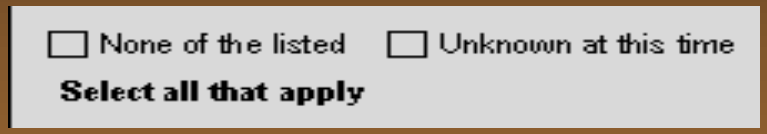
Congenital Anomalies	☐ None of the listed ☐ Unknown at this time **Select all that apply**		**Diagnosed Prenatally?**	**If Yes, please indicate all methods used:** QI
	Yes ☐ No ☐	Anencephaly	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP / Triple Screen ☐ Amniocentesis ☐ Other ☐ Unknown
	Yes ☐ No ☐	Meningomyelocele/Spina Bifida	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP / Triple Screen ☐ Amniocentesis ☐ Other ☐ Unknown
	Yes ☐ No ☐	Cyanotic Congenital Heart Disease	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Congenital Diaphragmatic Hernia	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Omphalocele	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Gastroschisis	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Limb Reduction Defect	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Cleft lip with or without Cleft Palate	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Cleft Palate Alone	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
	Yes ☐ No ☐	Down Syndrome ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP / Triple Screen ☐ CVS ☐ Amniocentesis ☐ Other ☐ Unknown
	Yes ☐ No ☐	Other Chromosomal Disorder ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP / Triple Screen ☐ CVS ☐ Amniocentesis ☐ Other ☐ Unknown
	Yes ☐ No ☐	Hypospadias	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown

- Information related to birth anomalies can be found in either infant's or mother's chart.
- If a diagnosis is made, be sure to check the prenatal record (in the mother's chart) to determine if testing & diagnosis was done prenatally.

CONGENITAL ANOMALIES OF THE CHILD

Indicate any of the specific conditions listed below. Information about other congenital anomalies is no longer being collected on the birth certificate. All congenital anomalies, both those listed below and any other significant anomaly, must be reported to the New York State Congenital Malformations Registry. Call (518) 402-7990 for further information about reporting.

Congenital Anomalies



- If a diagnosis is suspected or discussed (for one of the anomalies listed) but is not diagnosed, enter "Unknown at this time"
- If you are missing substantial amounts of prenatal and infant information and you have no way to get this, or you are not confident that the infant had no congenital anomalies, enter "Unknown at this time"
- If no anomalies listed are present enter "None."

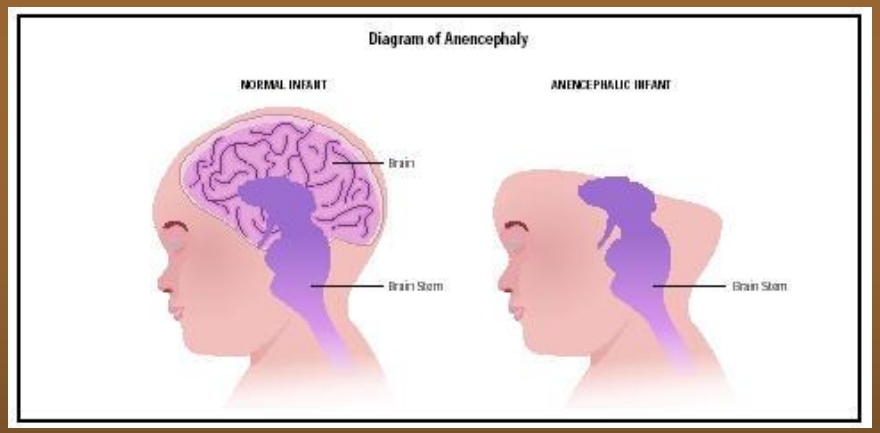
NYS GUIDELINES: The Registry was started in 1978 as a result of the Love Canal crisis

- **NONE** Select this item if the infant had none of the anomalies listed, even if he/she had other congenital anomalies. All significant congenital anomalies must be reported to the New York State Congenital Malformations Registry.
- **UNKNOWN AT THIS TIME**

Anencephaly

Yes	No	
☐	☐	Anencephaly

- Absence of a most of the brain and cranium
- Most are diagnosed by prenatal ultrasound
- Present in 1/1000 pregnancies
  - 1/10,000 live births



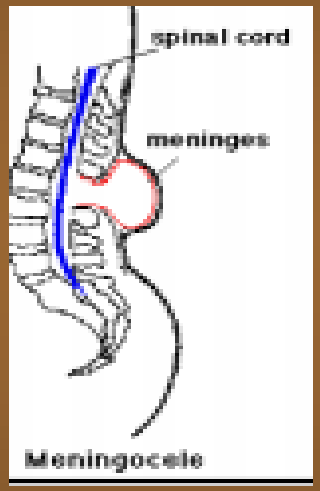
NYS GUIDELINES:

- **ANENCEPHALY Select this item if diagnosed by a physician. Synonyms include absent** brain, acrania, anencephalic, anencephalus, amyelencephalus, craniorachischisis, hemianencephaly, or hemicephaly.

Meningomyelocele/Spina Bifida

Yes ☐ No ☐	Meningomyelocele/Spina Bifida

- An incomplete closing of the embryonic neural tube
- Most detected prenatally, otherwise diagnosed at birth
- Decreased incidence in recent years with folic acid supplementation
  - 1996: 10.5/ 10,000 live births
  - 2003: 5/10,000 live births



NYS GUIDELINES:

MENINGOMYELOCELE / SPINA BIFIDA Select this item if diagnosed by a physician. Synonyms include meningocele, myelocele, myelomeningocele, myelocystocele, syringomyelocele, hydromeningocele, rachischisis. Do NOT include spina bifida occulta detected on radiographs.

Cyanotic Congenital Heart Disease (CHD)

Yes ☐	No ☐	Cyanotic Congenital Heart Disease

- A group of congenital heart defects where the baby may appear cyanotic (blue)
- Congenital heart disease: 0.7% of all pregnancies, of which 15% are cyanotic
- Code this disease as "Yes" if the chart lists the infant as diagnosed with any of the abnormalities described in the NYS guidelines below.

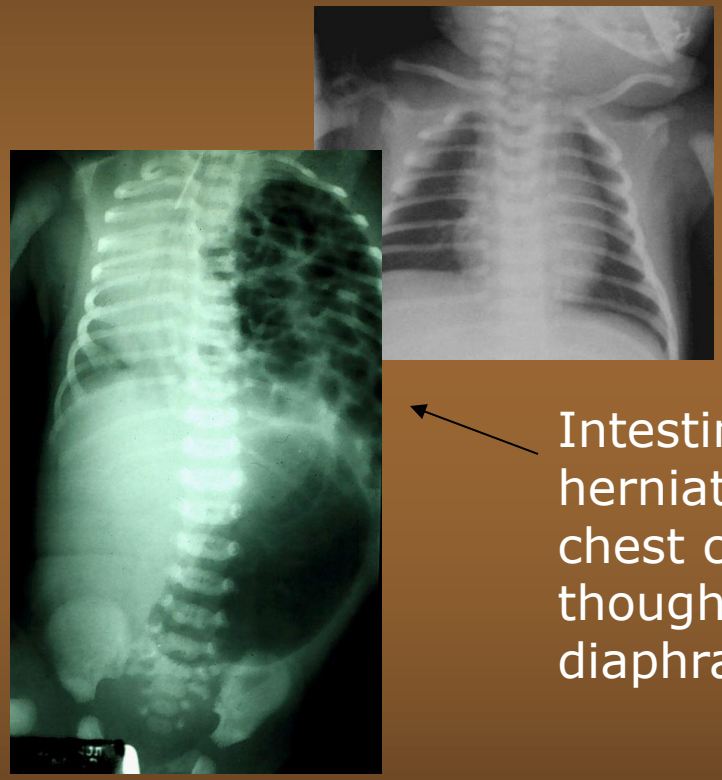
NYS GUIDELINES:

CYANOTIC CONGENITAL HEART DISEASE Select this item if any of the following conditions has been diagnosed by a physician: transposition of the great arteries (vessels), teratology of Fallot, pulmonary or pulmonic valvular atresia, tricuspid atresia, truncus arteriosus, total or partial anomalous pulmonary venous return with or without obstruction.

Congenital Diaphragmatic Hernia

Yes	No	Congenital Diaphragmatic Hernia
☐	☐	

- Usually detected prenatally, confirmed by chest x-ray
- Occurs in 1/2200 livebirths



Intestines herniating into chest cavity though hole in diaphragm

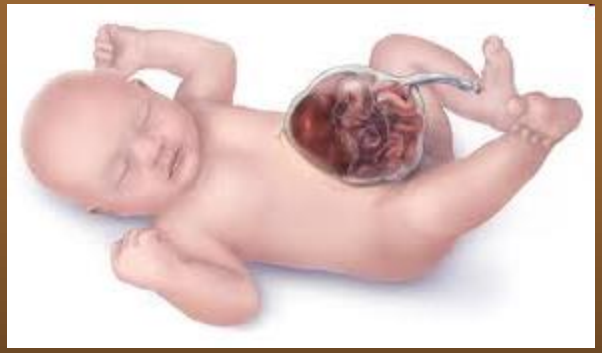
NYS GUIDELINES:

- **CONGENITAL DIAPHRAGMATIC HERNIA** Select this item if diagnosed by a physician.

Omphalocele

Yes	No	
☐	☐	Omphalocele

- Omphalocele is a birth defect in which the infant's intestine or other abdominal organs stick out of the belly button (navel). In babies with an omphalocele, the intestines are covered only by a thin layer of tissue and can be easily seen.
- Frequently infant has other anomalies (CHD, chromosomal anomalies)
- Frequently diagnosed prenatally by ultrasound
- Occurs in 1/4000 live births



NYS GUIDELINES:

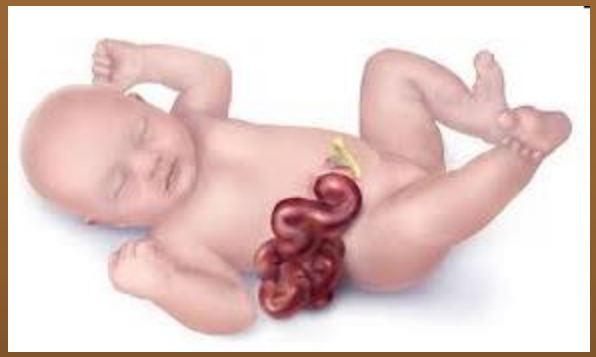
OMPHALOCELE Select this item if diagnosed by a physician. Synonyms include exomphalos. Do NOT include umbilical hernia (completely covered by skin) in this category.

Gastroschisis

Yes ☐ No ☐	Gastroschisis

- Isolated herniation of intestines through a defect in infant's abdominal wall
- Usually diagnosed prenatally by ultrasound

Rate of occurrence is increasing:
1997: 2.9/1000 live births
2001: 5/1000 live births



NYS GUIDELINES:

- **GASTROSCHISIS** Select this item if diagnosed by a physician. Synonyms include limb-body wall complex.

Limb Reduction Defect

Yes	No	Limb Reduction Defect
☐	☐	

- Frequently diagnosed prenatally by USG
- Occurs in 4-6/10,000 live births

Notice fingers on left hand

NYS GUIDELINES:

- **LIMB REDUCTION DEFECT** Select this item if diagnosed by a physician. This includes a missing hand, arm, foot, or leg, or any portion of it, excluding congenital amputation and dwarfing syndromes.

Congenital amputation is birth without a limb or limbs, or without a part of a limb or limbs.

Cleft Lip +/- Cleft Palate

Yes	No	
☐	☐	Cleft lip with or without Cleft Palate

- Cleft lip usually is diagnosed prenatally but may not be diagnosed until birth. Cleft palate often is not diagnosed until birth
- Birth defects in which there is an opening in the lip and/or palate (roof of the mouth) that is caused by incomplete development during early fetal formation
- Occurs in about 1 or 2 of every 1,000 babies born in the United States each year, making it one of the most common major birth defects.

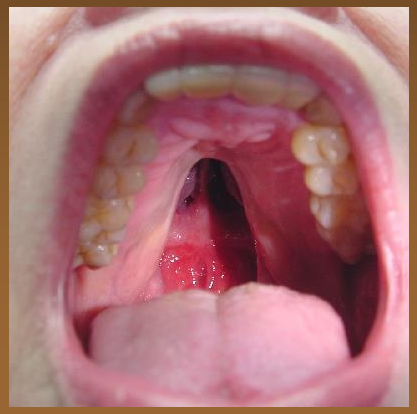
NYS GUIDELINES:

CLEFT LIP WITH OR WITHOUT CLEFT PALATE Select this item if diagnosed by a physician. Synonyms for cleft lip include harelip, cheiloschisis, and labium leporinum. Synonyms for cleft palate include cleft uvula, palate fissure, and palatoschisis.

Cleft Palate

Yes	No	
☐	☐	Cleft Palate Alone

- Usually diagnosed at birth
- Occurs in 1/5000 live births

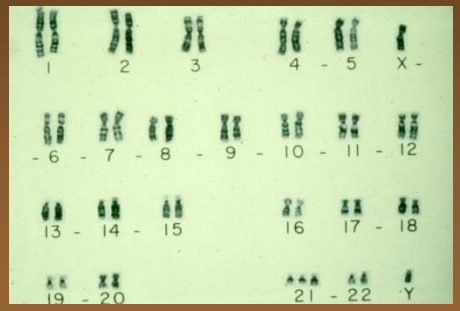


NYS GUIDELINES:

CLEFT PALATE ALONE Select this item if diagnosed by a physician. Synonyms include cleft uvula, palate fissure, palatoschisis. If cleft lip also present, record only under item above.

Down Syndrome

Yes	No	Down Syndrome
☐	☐	☐ Karyotype confirmed ☐ Karyotype pending

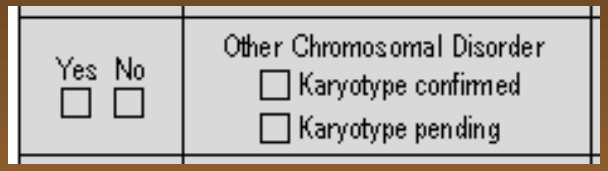


- High diagnostic sensitivity of prenatal ultrasound plus first or second trimester screening help with diagnosis.
- Confirmed by chromosomes from amniocentesis, chorionic villus sampling, cell-free DNA, or after delivery
- Occurs in 1/800 live births

NYS GUIDELINES:

DOWN SYNDROME Select this item if diagnosed by a physician. Synonyms include Trisomy 21. Indicate "Karyotype Confirmed" if chromosomal studies have been completed. Indicate "Karyotype Pending" if chromosomal studies have been initiated, but final results are not in.

Other Chromosomal Disorder



- Could be various disorders listed below (in Guidelines)
- May be suspected on ultrasound
- Diagnosis is confirmed by Karyotype testing (chromosomal studies) from Chorionic Villus Sampling (CVS), amniocentesis, or after birth
- Monosomy X (Turners) and Trisomy 13 and 18 are most common

NYS GUIDELINES:

OTHER CHROMOSOMAL DISORDER Select this item if diagnosed by a physician. Examples include Trisomy 13, Trisomy 18, Klinefelter syndrome, Edwards syndrome, Patau syndrome. Indicate "Karyotype Confirmed" if chromosomal studies have been completed. Indicate "Karyotype Pending" if chromosomal studies have been initiated, but final results are not in.

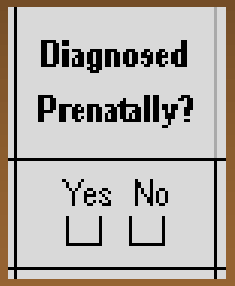
Hypospadias

- Urethra ends before the tip of penis
- Most often detected on physical exam, rarely prenatally

NYS GUIDELINES:

HYPOSPADIAS Select this item if diagnosed by a physician.

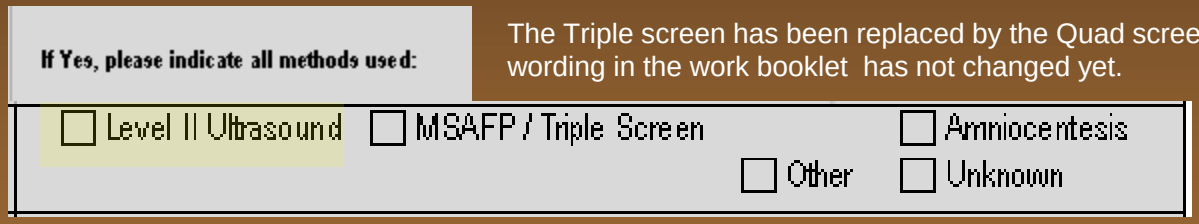
Diagnosed Prenatally?



- If a congenital anomaly is diagnosed, look through the prenatal record to see if it was diagnosed prenatally

If diagnosed prenatally, look for the method(s) used to make the diagnosis (can be more than one)

Methods used – Level II Ultrasounds



The Triple screen has been replaced by the Quad screen. The wording in the work booklet has not changed yet.

- Level I Ultrasound looks to see if there is a baby and if there a heartbeat
  - e.g.: quick scan
- Level II Ultrasound checks to see if all the necessary organs are present and look normal
  - e.g.: 16 week anatomic scan in the OB's office
- A Level II ultrasound can be used to identify most of the congenital anomalies listed in work booklet which can be diagnosed prenatally
- Level III Ultrasound looks at the organs to see if they are normal and if they are not to see what exactly is wrong with them
  - e.g.: referral to Strong for fetal ECHO by Pediatric Cardiologist

Methods Used – MSAFP/Triple Screen

Term: MSAFP= maternal serum alpha fetoprotein

- MSAFP is used prenatally to diagnose: Anencephaly, Meningomyelocele/ Spina Bifida, Down Syndrome, and Other Chromosomal Disorders

MSAFP/ triple screen tests involves drawing blood from the expectant mother and looking at the levels of various proteins and then comparing these levels with the established norms

- Levels are low in trisomy 21 (T21), aneuploidy
- Levels are elevated in neural tube defects, ventral wall defects, tumors, dermatologic disorders, congenital nephrosis

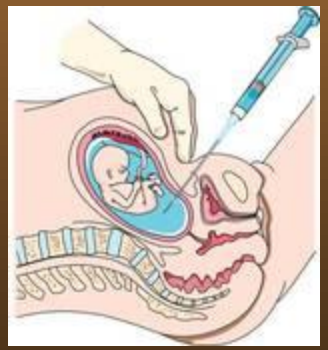
Remember - The Triple screen has been replaced by the Quad screen. The wording in the work booklet has not changed yet.

Methods Used - Amniocentesis

If Yes, please indicate all methods used:

☐ Level II Ultrasound ☐ MSAFP / Triple Screen ☐ Amniocentesis
☐ Other ☐ Unknown

- Amniocentesis, also referred to as Amniotic Fluid Testing (AFT) is used prenatally to diagnose: Anencephaly, Meningomyelocele/ Spina Bifida, Down Syndrome and Other Chromosomal Disorders
- This test uses chromosomes taken from cells in amniotic fluid in second/ third trimester

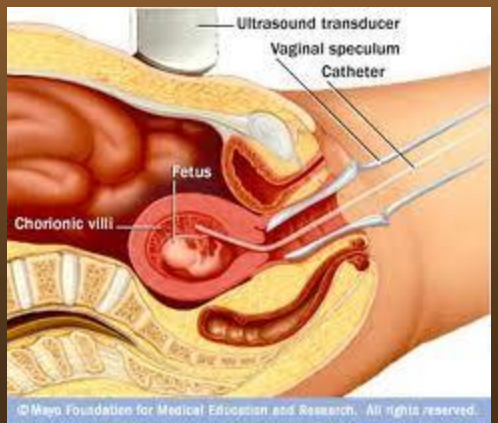


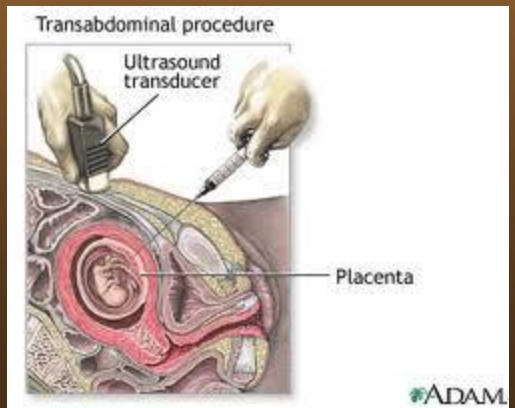
Methods Used - CVS

If Yes, please indicate all methods used:

☐ Level II Ultrasound ☐ MSAFP / Triple Screen ☐ Amniocentesis
☐ Other ☐ Unknown

- Chorionic Villus Sampling (CVS) may be used to diagnose Down Syndrome and Other Chromosomal Disorders
- This test uses chromosomes from cells taken from chorionic tissue early in pregnancy
- Cells can be obtained through the abdomen or through the cervix
- This test is coded as "other"





Methods Used – "NIPT"

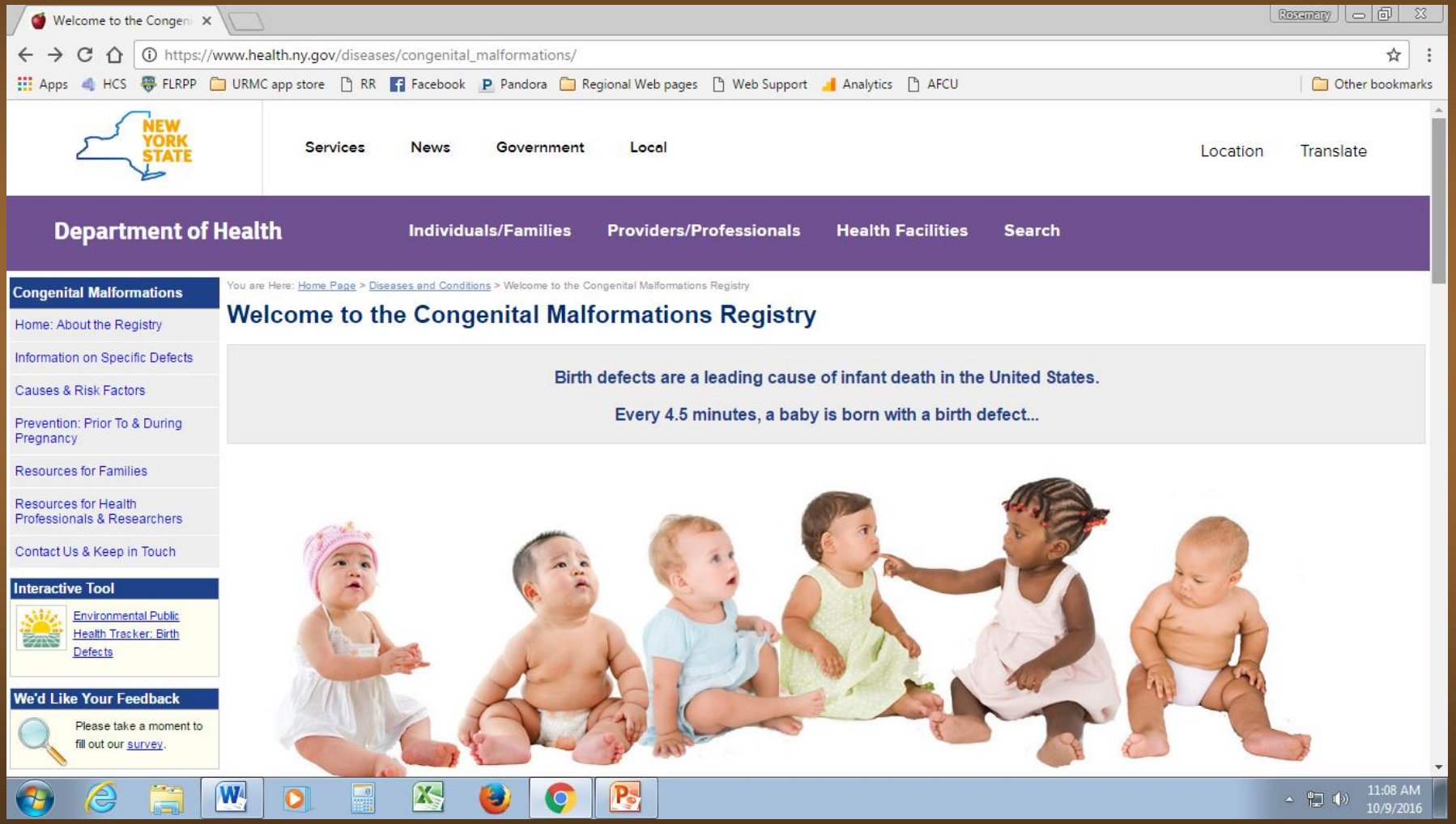
- Cell-free DNA
  - Non-Invasive Prenatal Testing, AKA NIPT
  - An expensive but more accurate *screening* test
    - Very good for Down syndrome
    - Pretty good for trisomy 13 and 18
    - Not as accurate as amnio or CVS
- Should not be coded as "Genetic Testing"

Congenital Malformations Registry

A significant birth defect that is not currently coded on the birth certificate will be reported by the hospital to the NYS Congenital Malformations Registry through the Health Commerce System (HCS) or 518-402-7990

For more information on Birth Defects go to

https://www.health.ny.gov/diseases/congenital_malformations/



The
End

Scenario Exercise(s)

Module 2 – Congenital Anomalies

Scenario Exercise #1

Enter the correct information

21 yo G3 P2 at 25.6 wk. – fetal gastrochisis diagnosed at 17 wk. by Level II U/S.
U/S today – EFW 1.41 lb. 9% nl amniotic fluid volume. Doppler 3.75
Plan is for repeat C/S

Congenital Anomalies

☐ None at this time ☐ Unknown at this time

Select all that apply		Diagnosed Prenatally? If Yes, please indicate all methods used:	
Yes ☐ No ☐	Gastroschisis	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☐	Limb Reduction Defect	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☐	Cleft lip with or without Cleft palate	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☐	Cleft Palate Alone	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☐	Down Syndrome ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes ☐ No ☐	Other Chromosomal Disorder ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes ☐ No ☐	Hypospadias	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown

See next page for answers

Module 2 – Congenital Anomalies

Scenario Exercise #1 Answers

21 yo G3 P2 at 25.6 wk. – fetal gastrochisis diagnosed at 17 wk. by Level II U/S.
U/S today – EFW 1.41 lb. 9% nl amniotic fluid volume. Doppler 3.75
Plan is for repeat C/S

Congenital Anomalies

☐ None of the listed ☐ Unk at this time **Select all that apply**:		**Diagnosed Prenatally?**	**If Yes, please indicate all methods used:**
Yes ☒ No ☐	Gastroschisis	Yes ☒ No ☐	☒ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Limb Reduction Defect	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Cleft lip with or without Cleft palate	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Cleft Palate Alone	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Down Syndrome ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes ☐ No ☒	Other Chromosomal Disorder ☐ Karyotype confirmed ☐ Karotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes ☐ No ☒	Hypospadias	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown

Module 2 – Congenital Anomalies

Scenario Exercise #2

Enter the correct information

Pt is a 32 yo G5 P2022 who presents to BC for induction of labor. Fetus w/ Tetralogy of Fallot
Dating criteria: _____ U/S 19.5 wks. confirms

Congenital Anomalies			
☐None of the listed ☐Unknown at this time **Select all that apply**		**Diagnosed Prenatally?**	**If Yes, please indicate all methods used:**
Yes No ☐ ☐	Gastroschisis	Yes No ☐ ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes No ☐ ☐	Limb Reduction Defect	Yes No ☐ ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes No ☐ ☐	Cleft lip with or without Cleft palate	Yes No ☐ ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes No ☐ ☐	Cleft Palate Alone	Yes No ☐ ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes No ☐ ☐	Down Syndrome ☐ Karyotype confirmed ☐ Karyotype pending	Yes No ☐ ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes No ☐ ☐	Other Chromosomal Disorder ☐ Karyotype confirmed ☐ Karyotype pending	Yes No ☐ ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes No ☐ ☐	Hypospadias	Yes No ☐ ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown

AND / OR

___ Enter into the NYS Congenital Malformations Registry through the Health Commerce System

See next page for answers

Module 2 – Congenital Anomalies

Scenario Exercise #2 Answers

Pt is a 32 yo G5 P2022 who presents to BC for induction of labor. Fetus w/ Tetralogy of Fallot
Dating criteria: _____ U/S 19.5 wks. confirms

Congenital Anomalies			
☒ None of the listed ☐ Unknown at this time **Select all that apply**		**Diagnosed Prenatally?**	**If Yes, please indicate all methods used:**
Yes ☐ No ☒	Gastroschisis	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Limb Reduction Defect	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Cleft lip with or without Cleft palate	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Cleft Palate Alone	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown
Yes ☐ No ☒	Down Syndrome ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes ☐ No ☒	Other Chromosomal Disorder ☐ Karyotype confirmed ☐ Karyotype pending	Yes ☐ No ☐	☐ Level II Ultrasound ☐ MSAFP/ ☐ CVS ☐ Amniocentesis ☐ Triple Screen ☐ Other ☐ Unknown
Yes ☐ No ☒	Hypospadias	Yes ☐ No ☐	☐ Level II Ultrasound ☐ Other ☐ Unknown

AND / OR

_X_ Entered into the NYS Congenital Malformations Registry by the billing coders

*Only those listed on the Birth Certificate are coded.
ALL other congenital anomalies are reported in the
NYS Congenital Malformations Registry
(Slide 2)

Module Evaluation

MODULE TWO EVALUATION

(Please check the appropriate response)

1. **Which of the following congenital malformations is least likely to be identified prenatally?**
   - Anencephaly
   - Meningomyelocele/Spina Bifida
   - Congenital Diaphragmatic Hernia
   - Omphalocele
   - Gastroschisis
   - Limb Reduction Defect
   - Cleft Palate

2. **For a congenital malformation that is likely to be identified prenatally, which of the records below would be the best source of information related to testing & diagnosis?**
   - Infant's record
   - Hospital record (L&D)
   - Mother's prenatal record

3. **If a diagnosis is suspected or discussed for one of the anomalies listed in the Birth Certificate work book, but is not diagnosed what should you enter?**
   - 'Unknown at this time'
   - 'None'
   - 'Yes' next to the suspected anomaly
   - 'No' next to the suspected anomaly

4. **If the anomaly is not listed in the Birth Certificate work book you should enter all 'None of the listed'**
   - True
   - False

5. **A Level II ultrasound can be used to identify any of the congenital anomalies listed in work book.**
   - True
   - False

6. **Accurate entry of congenital malformations provides which of the following information (check all that apply)**
   - Establish the need for services
   - Identify causal links
   - Track changes in incidence over time

7. **The MSAFP/ triple screen testing involves getting urine from the expectant mother and looking at levels of various proteins**
   - True
   - False

8. **Which of the genetic tests listed below can be done within the 1st trimester of pregnancy?**
    - Chorionic Villus Sampling (CVS)
    - Amniocentesis

9. **If a significant birth defect <u>not listed</u> on the birth certificate is identified, is it reported?**
    - It is not reported
    - It is reported by the hospital to the NYS Congenital Malformations Registry
    - It is identified on the birth certificate

10. **Downs syndrome is often diagnosed prenatally**
    - True
    - False

See next page for answers

MODULE TWO EVALUATION *ANSWERS*

1. **Which of the following congenital malformations <u>are likely</u> to be identified prenatally? (check all that apply)**
   - o Anencephaly
   - o Meningomyelocele / Spina Bifida
   - o Congenital Diaphragmatic Hernia
   - o Omphalocele
   - o Gastroschisis
   - o Limb Reduction Defect
   - ● Cleft Palate

   Answer: Cleft palate may be diagnosed prenatally but is most often diagnosed after the infant is born. (Slides 5, 6, 8, 9, 10, 11, 12, 13)

2. **For a congenital malformation that is likely to be identified prenatally, which of the records below would be the best source of information related to testing & diagnosis?**
   - o Infant's record
   - o Hospital record (L&D)
   - ● Mother's prenatal record

   Answer: Check the mother's prenatal record for tests and diagnoses made during pregnancy (Slides 3, 17)

3. **If a diagnosis is suspected or discussed for one of the anomalies listed in the Birth Certificate work booklet, but is not diagnosed what should you enter?**
   - ● Unknown at this time
   - o None
   - o 'Yes' next to the suspected anomaly
   - o 'No' next to the suspected anomaly

   Answer: If a diagnosis is suspected or discussed (for one of the anomalies listed) but is not diagnosed, enter 'Unknown at this time' (Slide 4)

4. **If the anomaly is not listed in the Birth Certificate work booklet you should enter all 'No's"**
   - ● True
   - o False

   Answer: If no anomalies listed are present enter 'None'. (Slide 4)

5. **A Level II ultrasound can always be used to identify any of the congenital anomalies listed in work booklet.**
   - o True
   - ● False

   Answer: <u>Only those anomalies that can be identified prenatally</u> will benefit from ultrasound. Cleft palate is often identified at birth (Slides 5, 6, 8, 9, 10, 11, 12, 13)

6. **Accurate entry of congenital malformations provides which of the following information (check all that apply)**
   - ● Establish the need for services
   - ● Identify causal links
   - ● Track changes in incidence over time

   Answer: Accurate coding of Congenital Anomalies provides information which can help establish the need for services, causal links and changes in the incidence of the defect. (Slide 2)

7. **MSAFP/ triple screen testing involves getting urine from the expectant mother and looking at the levels of various proteins .**
    - True
    - False

   Answer: MSAFP/ triple screen testing involves drawing blood from the expectant mother and looking at the levels of various proteins and then comparing these levels with the established norms. (Slide 19)

8. **Which of the genetic tests listed below can be done within the 1st trimester of pregnancy?**
    - Chorionic Villus Sampling (CVS)
    - Amniocentesis

   Answer: Amniocentesis uses chromosomes taken from cells in amniotic fluid in second/ third trimester (Slide 20) while CVS uses chromosomes from cells taken from chorionic tissue early in pregnancy (Slide 21)

9. **If a significant birth defect not listed on the birth certificate is identified, is it reported?**
    - It is not reported
    - It is reported by the hospital to the NYS Congenital Malformations Registry
    - It is identified on the birth certificate

   Answer: A significant birth defect that is not currently coded on the birth certificate will be reported by the hospital to the NYS Congenital Malformations Registry by the Health Information Departmant(Slide 22)

10. **Downs syndrome is often diagnosed prenatally?**
    - True
    - False

   Answer: A suspected diagnosis of Downs can result from abnormal prenatal ultrasound and triple marker screen which can be confirmed by amnio or CVS (Slide 14)

Extra Information

Birth Registrars

Calling a Spade a Spade

The Whys and Hows of Coding Birth Anomalies

Patricia R Chess MD
Associate Professor
Departments of Pediatrics (Neonatology)
and Biomedical Engineering
University of Rochester

Overview

- Why code?
- What to code
- Description of coded anomalies, how they are diagnosed
- Review of diagnostic screens used for coding
- Incidence of coded anomalies
- Using coded data for research

The ultimate goal is to optimize maternal-infant health and well-being

Why Code Anomalies

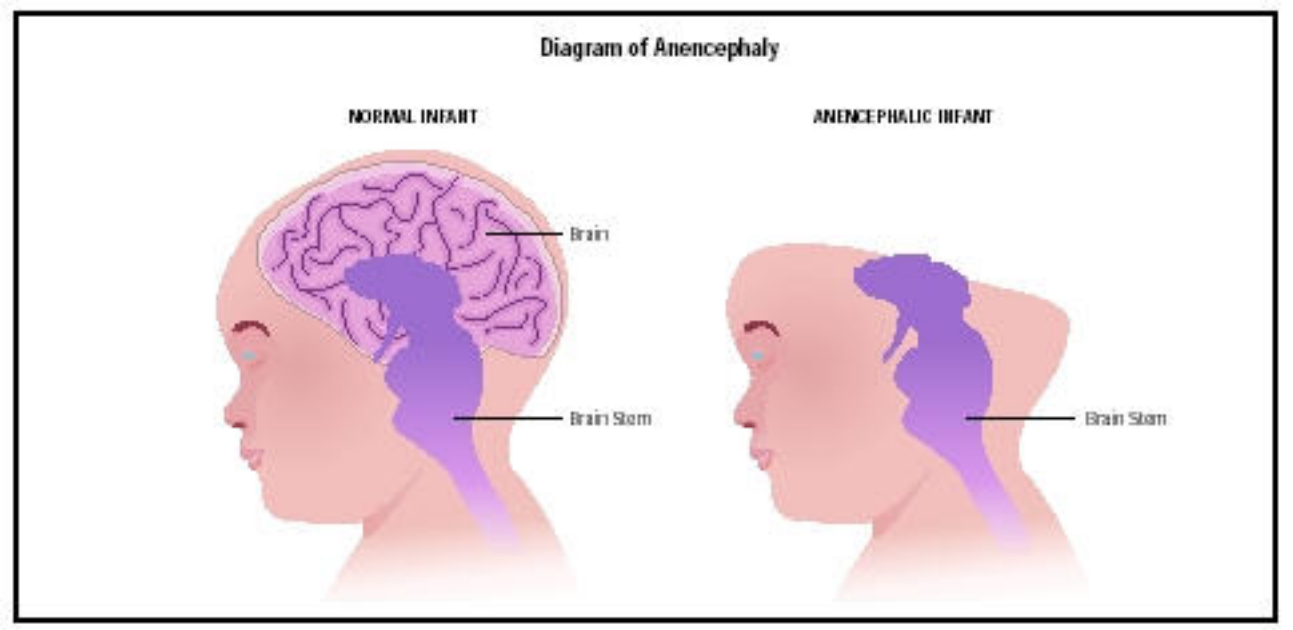
- Determine incidence
  - Is incidence changing, and if so, why
  - Establish patterns (demographics/ epidemiology)
  - Identify possible environmental, ethnic (genetic) or cultural links
- Assess necessary resources
- Project future needs

Which Anomalies to Code

- Anencephaly
- Meningomyelocele (aka spina bifida)
- Cyanotic congenital heart disease
- Congenital diaphragmatic hernia
- Gastroschisis
- Omphalocele
- Limb reduction defects
- Cleft lip +/- cleft palate
- Down Syndrome (+/- karyotype)
- Other Chromosomal Disorder (+/- karyotype)
- Hypospadias

Anencephaly

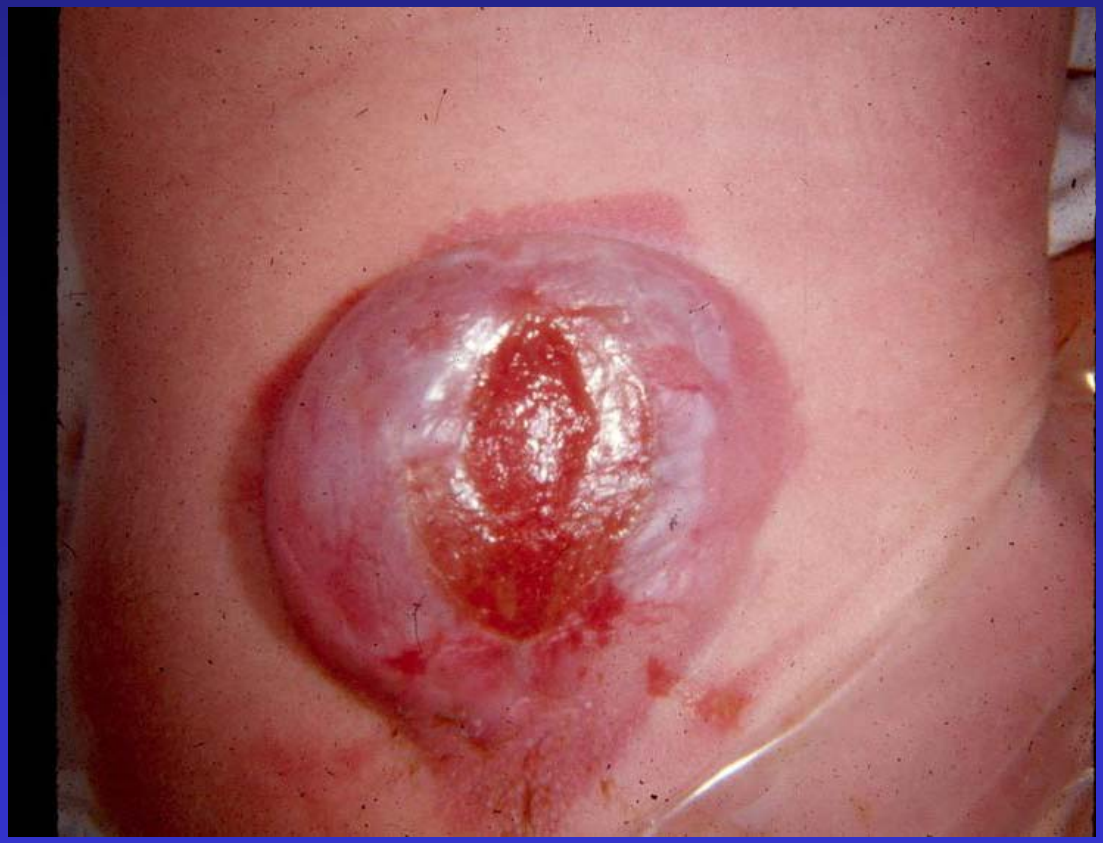
- Lacking the cortex (thinking, motor part of the brain) brainstem present (controls reflex neurologic functions such as breathing)
- Diagnosed prenatally if USG performed



Neural Tube Defects

Spina Bifida

- Most detected prenatally, otherwise at birth
- Decreased incidence with folic acid supplementation



Cyanotic Congenital Heart Disease

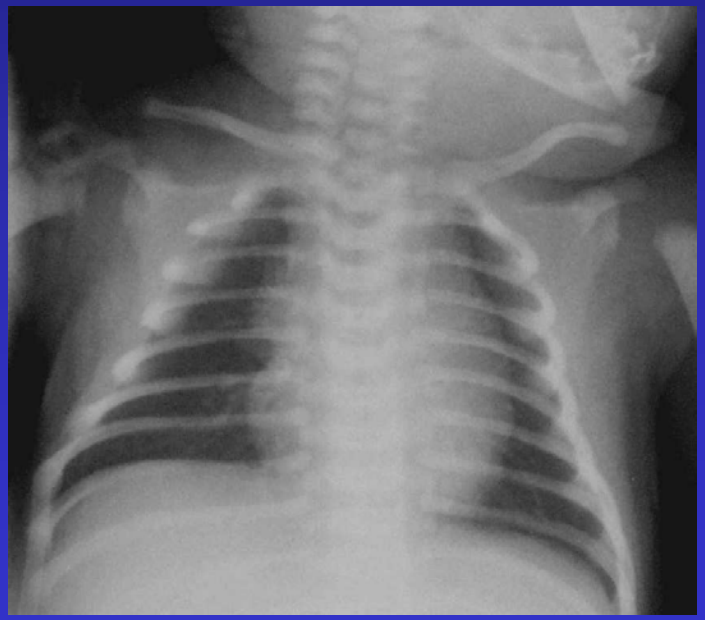
blue baby, diagnosed by fetal or postnatal cardiac ECHO, usually needs prostaglandins to survive

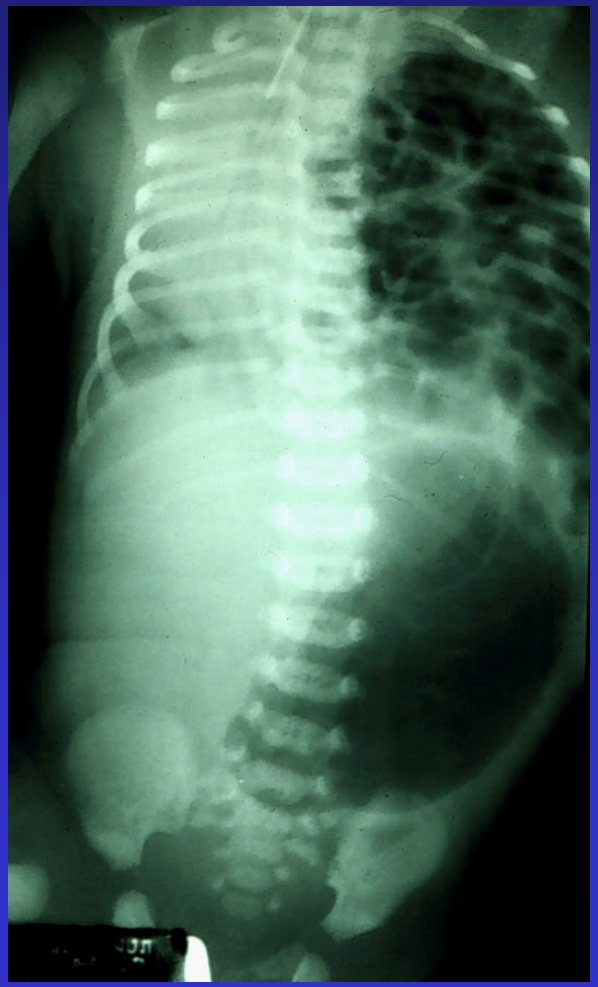
- Tetralogy of Fallot with significant pulmonic stenosis
- Total Anomalous Pulmonary Venous Return
- Transposition of the Great Vessels
- Tricuspid Atresia
- Truncus
- Hypoplastic Left heart

Congenital Diaphragmatic Hernia

Usually detected prenatally, confirmed by chest x-ray

Normal

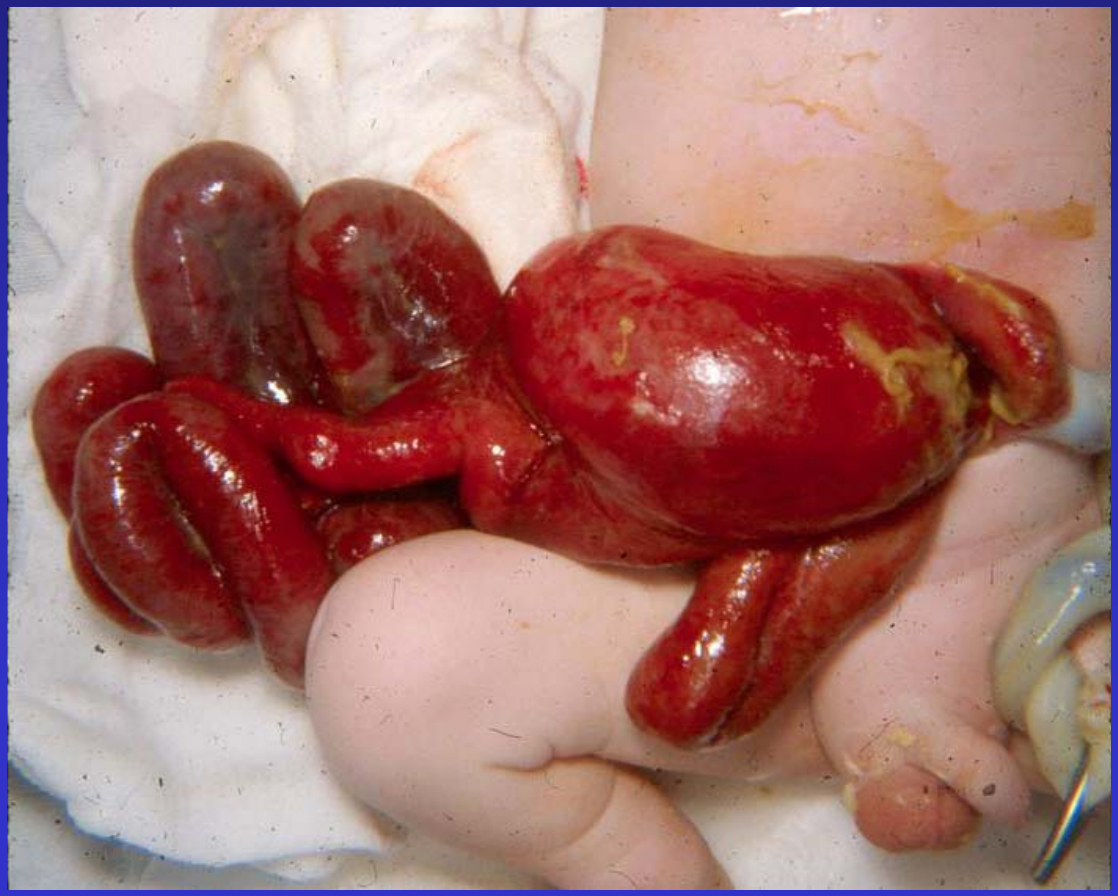




Intestines herniating into chest cavity though hole in diaphragm

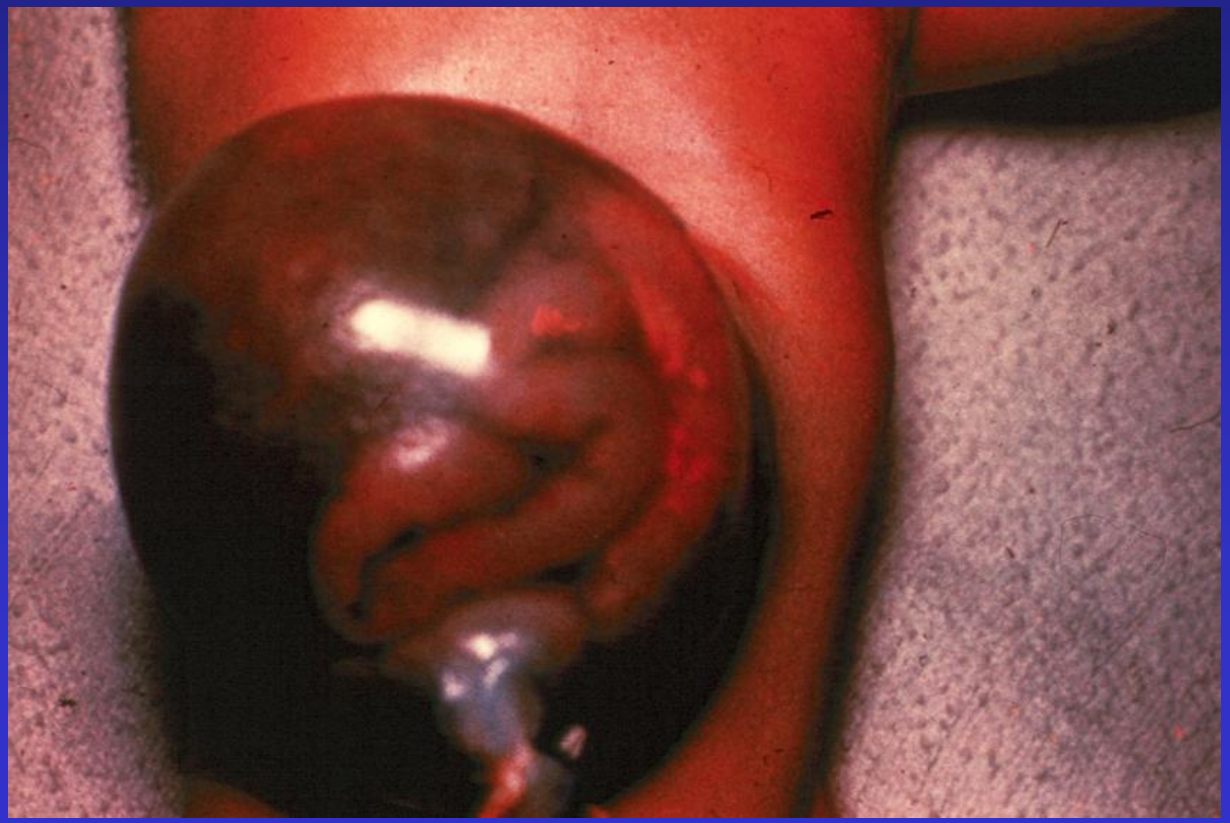
Gastroschisis

Isolated herniation of intestines through defect in abdominal wall, usually diagnosed prenatally by USG



Omphalocele

Intestines within umbilical cord, frequently has other anomalies (CHD, chromosomal anomalies), frequently diagnosed prenatally by USG

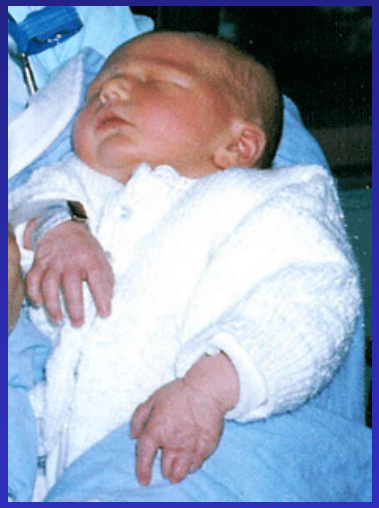


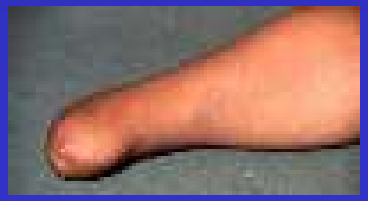
Limb Reduction Defect

frequently diagnosed prenatally by USG

Thalidomide
Chicken Pox
TAR Syndrome

Amniotic Banding
(not coded)

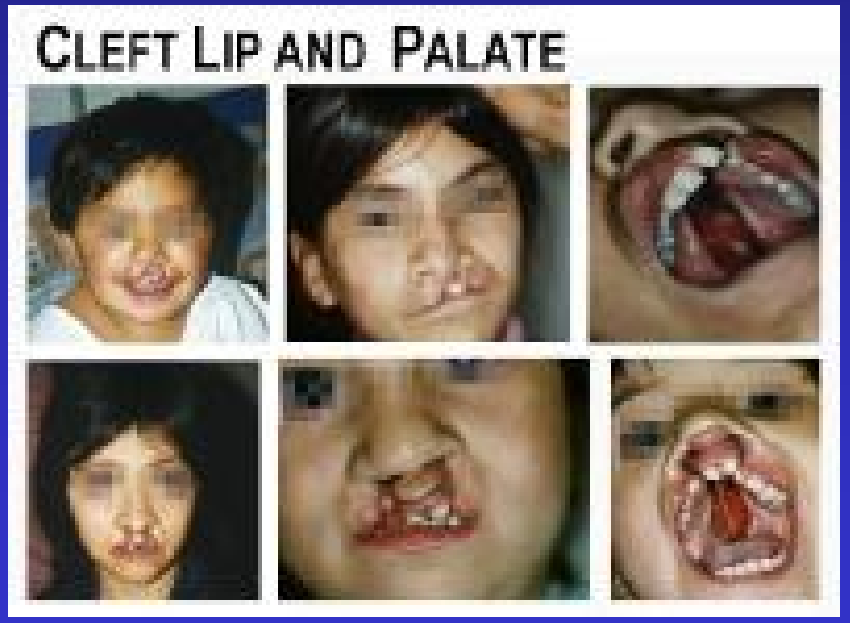




Dwarfism
(not coded)
shortened limbs,
fingers/toes present

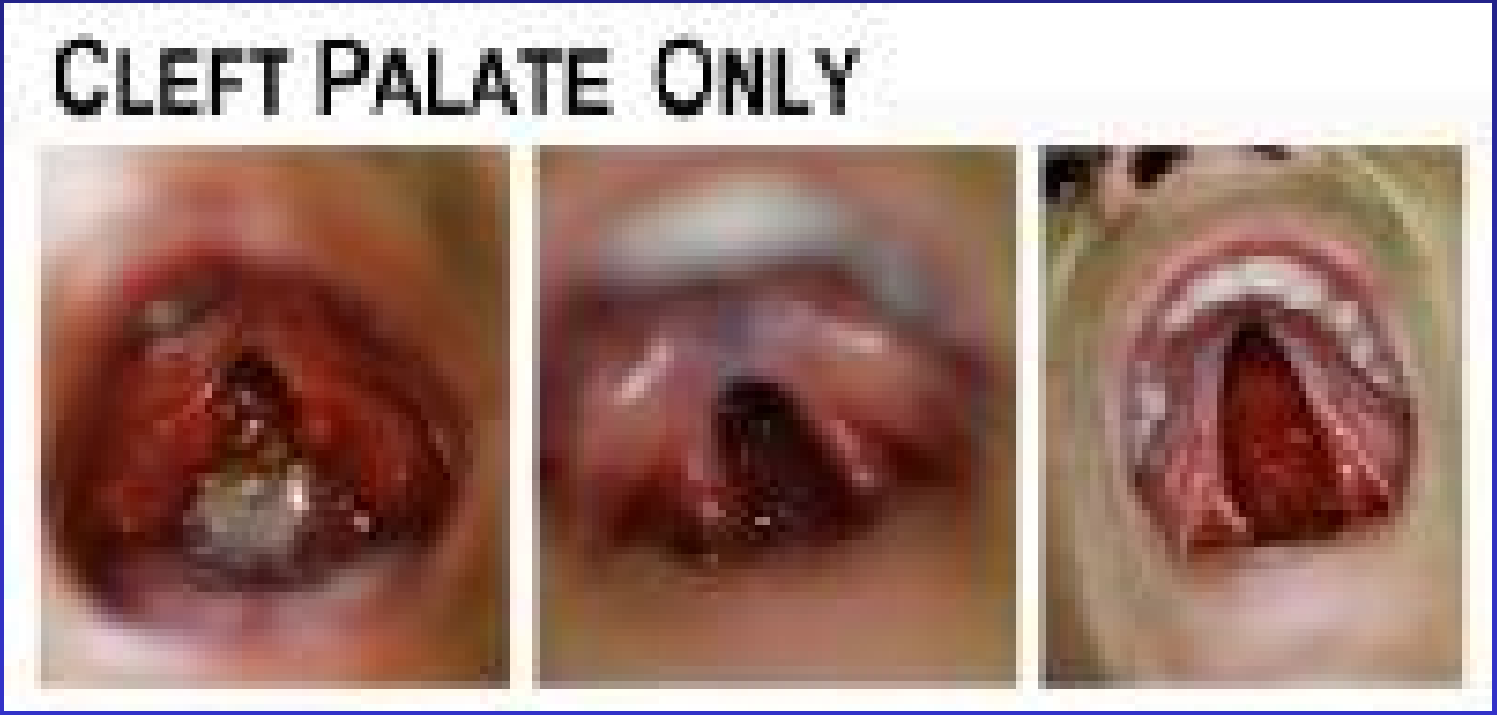
Cleft lip +/- Cleft palate

Usually diagnosed at birth



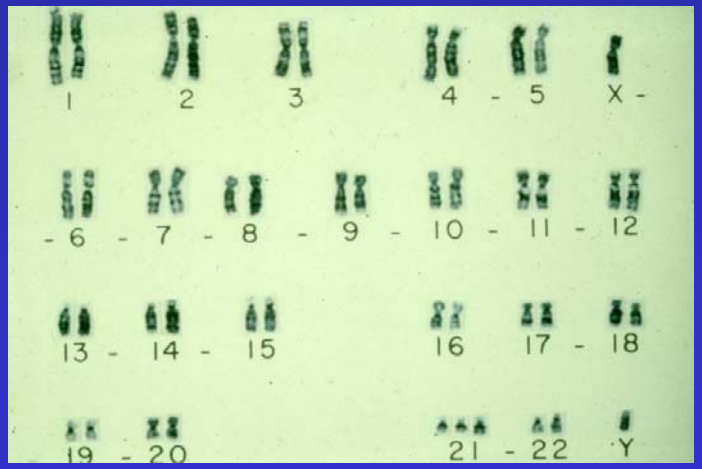
Cleft Palate Alone

Usually diagnosed at birth



Down Syndrome

- High diagnostic sensitivity of prenatal USG and triple screen
- Confirmed by chromosomes from amnio or post delivery



Other Chromosomal Anomalies

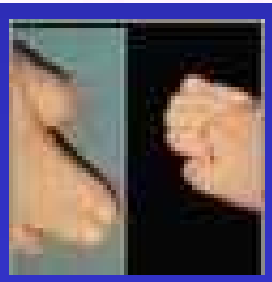
Trisomy 13, 18 most common

Could be suspected on USG, diagnosed by chromosomes from CVS, amniocentesis or after birth (exam leads to clinical diagnosis, chromosomes confirm)

Trisomy 13

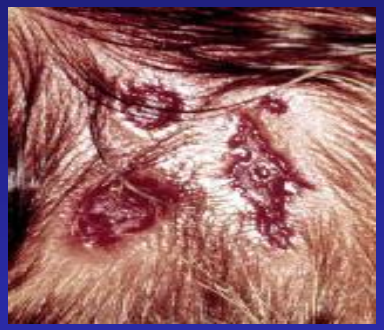
Midline defects common

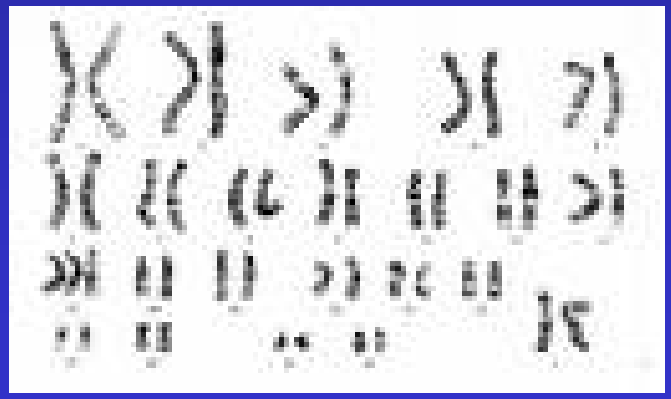
Cleft lip, small for gestational age



Syndactyly- fingers, toes not normally separated

Cutis aplasia- skin defect on scalp

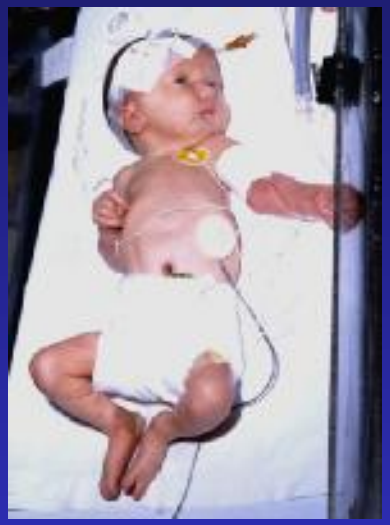


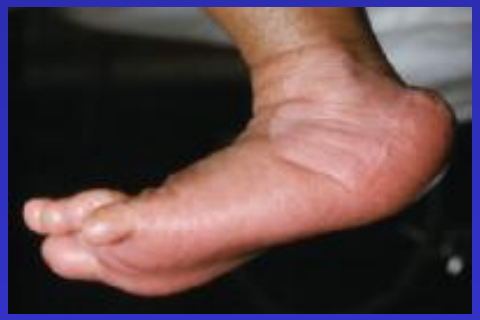


Extra chromosome 13

Trisomy 18

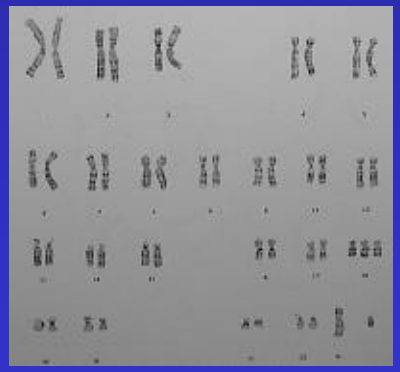
Small for gestational age





Rocker bottom feet

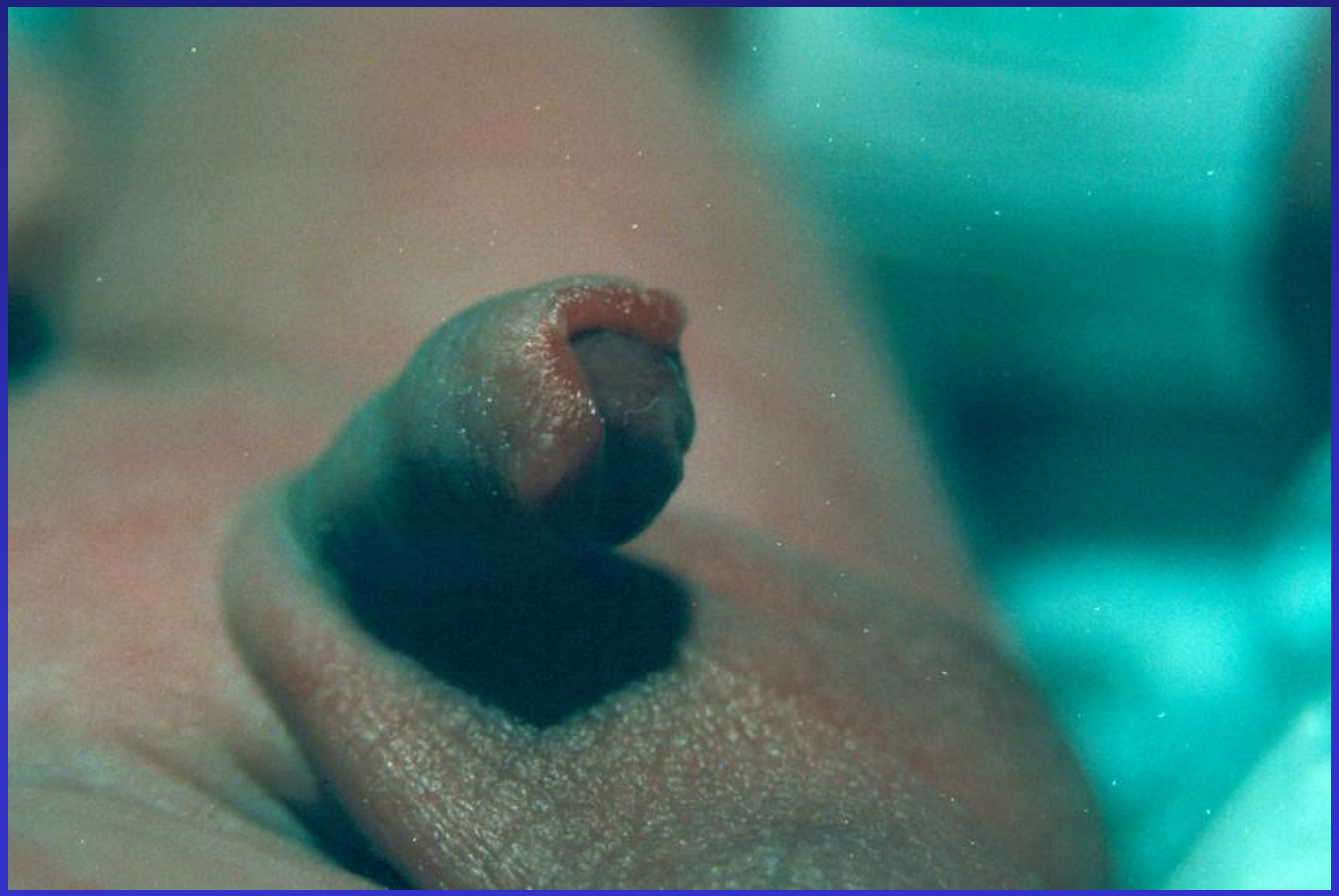
Overriding fingers



Extra chromosome 18

Hypospadias

End of urethra does not end at tip of penis.
Detected on physical exam, not prenatally



What is a level II Ultrasound?

- Level I- Is there a baby and is there a heartbeat?
  - eg: quick scan in the ED
- Level II- Are there all the necessary organs and do they look normal?
  - eg: 16 week anatomic scan in the OB's office
- Level III- If the organs don't look normal, what exactly is wrong with them?
  - eg: referral to GCHaS for fetal ECHO by Pediatric Cardiologist

Other Screening Tests Coded

- MSAFP/ triple screen: Maternal blood work with norms established
  - MSAFP: maternal serum α- feto protein
    - Low in trisomy 21 (T21), aneuploidy
    - Elevated in neural tube defects, ventral wall defects, tumors, dermatologic disorders, congenital nephrosis
  - hCG: human chorionic gonadotropin
    - Elevated in T21
  - uE 3: unconjugated estriol
    - Low in T21
  - IH-A: inhibin A (quad screen)
    - Elevated in T21
- Chorionic Villous Sample (CVS): chromosomes determined on cells from chorionic tissue early in pregnancy
- Amniocentesis: chromosomes determined from cells in amniotic fluid in second/ third trimester

Incidence of Coded Anomalies

- Anencephaly: 1/68,000 births
- Meningomyelocele (aka spina bifida)
  - 1996: 10.5/ 10,000 livebirths
  - 2003: 5/10,000
- Congenital heart disease: 8/1000, 15% cyanotic
- Congenital diaphragmatic hernia: 1/2200
- Gastroschisis
  - 1997: 2.9/1000
  - 2001: 5/1000
- Omphalocele: 1/4000

Incidence (continued)

- Limb reduction defects: 659/1.2 million
- Cleft lip + cleft palate: 1/1000
- Cleft palate alone: 1:500
- Down Syndrome: 1/800
- Other Chromosomal Disorder
  - Trisomy 13: 1/5000
  - Trisomy 18: 1/3000
- Hypospadias:
  - 1970's: 1/250 boys
  - 1990's: 1/125 boys

Research Using Database Data

- Uses de-identified data
- Each research project reviewed by hospital-based institutional review board
- Results often provide information that can help identify trends and improve health care for future patients
- Critical to have accurate data

Example of using database to determine population statistics

Risk of RSV-related Hospitalization Among Infants 32-35 Weeks' Gestation					
	N	**RSV-hosp Rate (%)**	**Lower 95% CI**	**Upper 95% CI**	**NNT**
Overall	652	2.91	.66	4.21	45
Mat tobacco during preg.	151	4.64	1.24	8.03	28
Exclusively formula fed at discharge	227	3.96	1.10	6.52	33
One or more living siblings	364	3.85	1.86	5.83	34
Multiple gestation	138	3.62	0.47	6.78	36
Maternal tobacco & $\geq$ 1 sib	98	6.12	1.29	10.95	21
Exclusively formula fed & $\geq$ 1 sib	152	4.61	1.24	7.98	28
Mat tobacco and exclusively formula fed	75	5.33	0.13	10.54	25

T Stevens et al

If there is a significant birth defect present that is not currently entered into the Statewide Perinatal Data System (SPDS), each hospital is required to report to NYS Congenital Malformations Registry.

This is done throught the hospital Health Information Management System. It is entered into another section on the NYS Health Commerce System

Accurate coding of birth registry data provides information which can help establish the need for services and can be used to identify potential causal links to suboptimal outcomes and suggest future interventions to optimize health in newborns.