

Physical and Mental Conditions With A High Probability of Resulting in Developmental Delay

Presented by: Laura Sorg, MD, FAAFP Medical Director Ohio Department of Developmental Disabilities





Participating in Today's Session

Communicating: Type questions/comments in Questions box.

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Tech tips:

- Close other apps (e.g. email, Word, etc.)
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- Make sure you're using your Computer Audio.
- Dial-in option- phone # on Control Panel.





Before We Begin....

-We do not expect you to become a doctor by the end of this presentation.

-This presentation is intended to help you increase your knowledge base on this important topic.





Why does Early Intervention Matter?

Earlier is better!

Intervention is **likely to be more effective** when it is provided earlier in life rather than later.

The connections in a baby's brain are most adaptable in the first three years of life. These connections, also called neural circuits, are the foundation for learning, behavior, and health. Over time, these connections become harder to change.

Intervention works!

Early intervention services can **change a child's developmental pat** improve outcomes for children, families, and communities.





What are these diagnoses and How do they qualify?

Newborn Conditions:

- Very low birth weight (i.e., less than 1,500 grams) diagnosed at birth or within thirty calendar days after birth with:
- (1) Intraventricular hemorrhage (grade III), and/or
- (2) Chronic lung disease (bronchopulmonary dysplasia), and/or
 (3) Retinopathy of prematurity (stage IV or stage V)

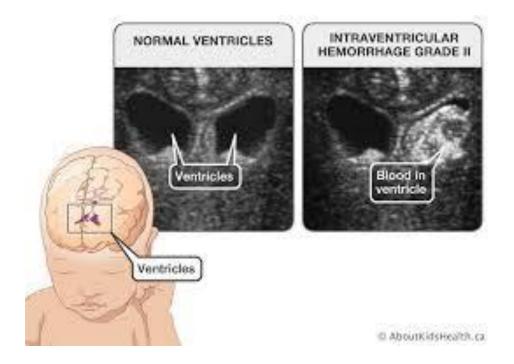




Eligibility Reason	SFY20	
	Number	Percent
Dx on List	2,632	11%
Dx on Form	629	3%
Total	3,261	14%



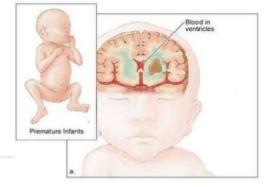
Intraventricular hemorrhage (grade III)



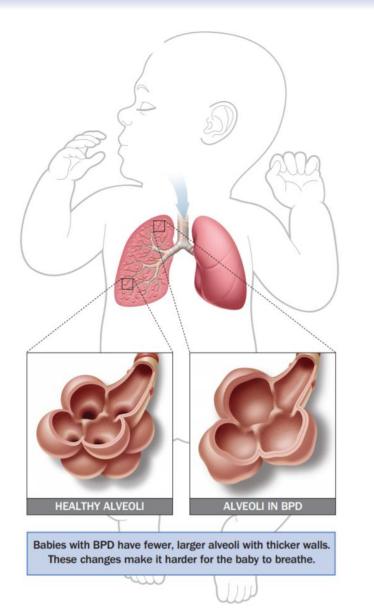
Intra-ventricular hemorrhage (IVH)

It is common in preterm infants due to:

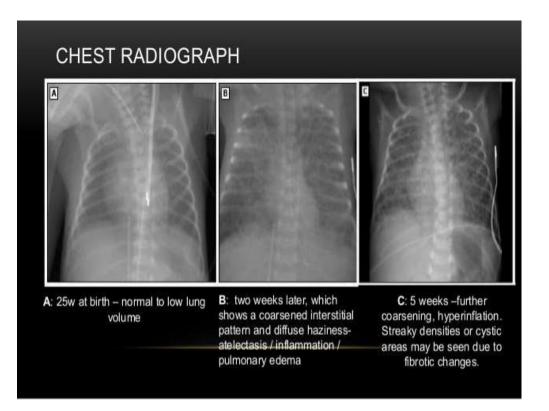
- Immature vasculature
- Disturbed cerebral auto-regulation of blood flow
- Clotting factor deficiency







Chronic lung disease (Bronchopulmonary Dysplasia)



Developed Retina Premature Retina

image Source: left - https://webeye.ophth.utowa.edu/eyeforum/allas/LARGE/Normal-fundus-LRG.jpg, Accessed 6/2/2018, right-https://www.esearchgate.net/figure/Normal-immedure-netice-not-dalawebsarteel_fig1_275627990, accessed 4/15/2018

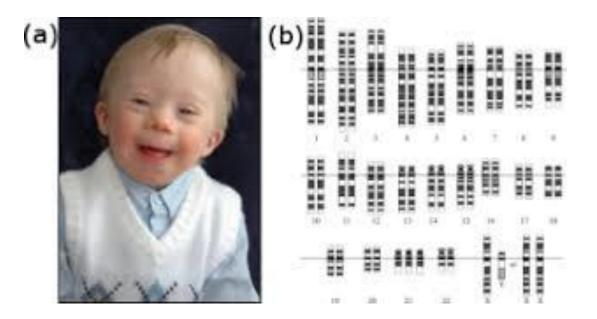
Retinopathy of prematurity (stage IV or stage V)





Diagnosed Conditions in Neonates, Infants, or Toddlers:

Chromosomal Conditions

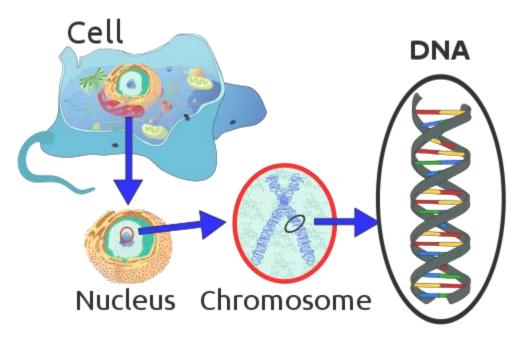


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What is a chromosome?

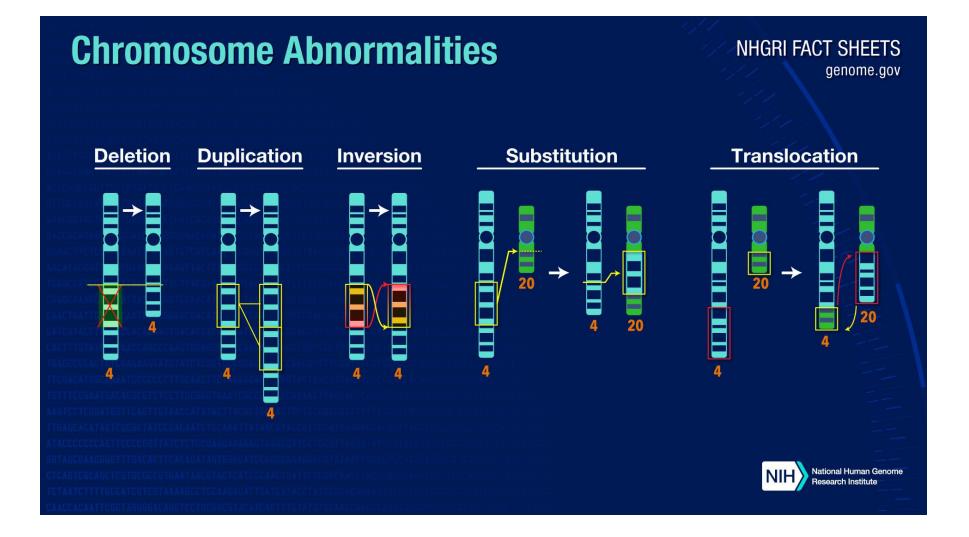
- Chromosomes are thread-like structures located inside the nucleus of animal and plant cells. Each chromosome is made of protein and a single molecule of deoxyribonucleic acid (DNA).
- Passed from parents to offspring, DNA contains the specific instructions that make each type of living creature unique.
- The term chromosome comes from the Greek words for color (chroma) and body (soma).
 Scientists gave this name to chromosomes because they are cell structures, or bodies, that are strongly stained by some colorful dyes used in research.





WHAT ARE CHROMOSOMAL DISORDERS

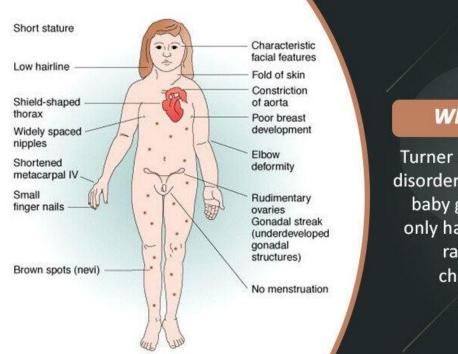
- A chromosomal disorder occurs when there is a change in the number or structure of the chromosomes.
- This change in the amount, or arrangement of, the genetic information in the cells may result in problems in growth, development and/or functioning of the body systems.





Examples of Chromosomal Disorders:

Turner's Syndrome



What is Turner Syndrome?

Turner syndrome is a female-only genetic disorder that affects about 1 in every 2,000 baby girls. A girl with Turner syndrome only has one normal X sex chromosome, rather than the usual two. This chromosome variation happens randomly when the baby is conceived in the womb.

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Examples of Chromosomal Disorders: Down Syndrome

Range for Children with Down Syndrome	Typical Range	
6 - 30 Months	5 - 9 Months	
8 - 22 Months	6 - 12 Months	
1 - 3.25 Years	8 - 17 Months	
1 - 4 Years	9 - 18 Months	
1 - 4 Years	1 - 3 Years	
2 - 7.5 Years	15 - 32 Months	
1.5 - 5 Months	1 - 3 Months	
10 - 24 Months	7 - 14 Months	
12 - 32 Months	9 - 17 Months	
13 - 39 Months	12 - 20 Months	
2 - 7 Years	16 - 42 Months	
3.5 - 8.5 Years	3.25 - 5 Years	
	with Down Syndrome 6 - 30 Months 8 - 22 Months 1 - 3.25 Years 1 - 4 Years 2 - 7.5 Years 1.5 - 5 Months 10 - 24 Months 12 - 32 Months 13 - 39 Months 2 - 7 Years	



Examples of Chromosomal Disorders: <u>Rett Syndrome</u>

- Most children diagnosed with Rett syndrome have a gene (MECP2) that changes (or mutates) and interrupts typical brain development.
- This mutation causes the brain to either make too little of an essential protein, or to make damaged protein the body can't use.
- As a result, the brain cannot develop typically. Most cases of Rett syndrome aren't inherited.
- The condition, which can produce symptoms similar to those of autism, is linked to mutations in a gene on the X chromosome.
- For this reason, approximately 95 percent of children diagnosed with Rett syndrome are girls. Rett syndrome in boys is extremely rare.



Rett Syndrome Stages and Interventions.

What Is Rett Syndrome?

Rett syndrome is a rare condition that affects the brain's growth and development.

Early Stages

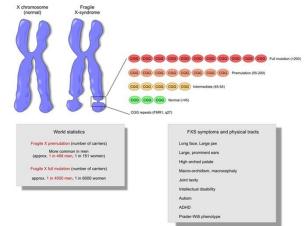
Symptoms typically develop between 6 and 18 months of age, and can include:

- Crying or irritability.
- Difficulty crawling or walking.
- Loss or decline of speech skills.
- Loss of eye contact. 🎸
- Weak or floppy muscle tone.
- Movement disorders.
- Slowed head growth.
- Epilepsy.



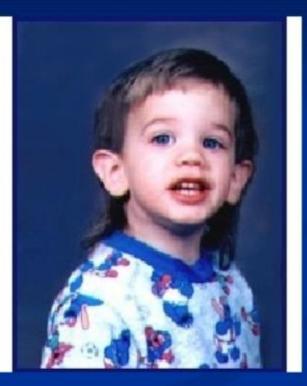
Fragile X Syndrome:

- A genetic condition causing intellectual disability.
- Fragile X syndrome causes mild to severe intellectual disability. It affects both males and females, but females usually have milder symptoms. Males are more frequently affected.
- Symptoms include delays in talking, anxiety, and hyperactive behavior. Some people have seizures. Physical features might include large ears, a long face, a prominent jaw and forehead, and flat feet.
- Therapy can be used to treat learning disabilities. Medications may be used to treat anxiety and mood disorders.



CHARACTERISTICS OF FRAGILE X SYNDROME FOR MALES

- prominent or long ears
- a long face
- delayed speech
- large testes (macroorchidism)
- Hyperactivity
- tactile defensiveness
- gross motor delays
- autistic-like behaviors.





Inborn errors of metabolism

- Inborn errors of metabolism are rare genetic (inherited) disorders in which the body cannot properly turn food into energy. The disorders are usually caused by defects in specific proteins (enzymes) that help break down (metabolize) parts of food.
- A food product that is not broken down into energy can build up in the body and cause a wide range of symptoms.
- Several inborn errors of metabolism cause developmental delays or other medical problems if they are not controlled.



There are many different types of inborn errors of metabolism.

A few of them are: •Fructose intolerance •Galactosemia •Maple sugar urine disease (MSUD) •Phenylketonuria (PKU)

<u>Newborn screening tests</u> can identify some of these disorders.





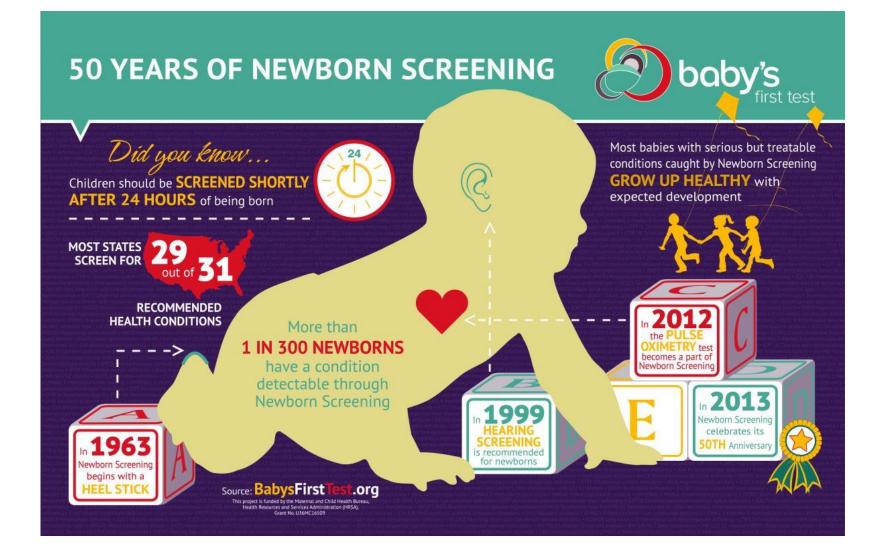
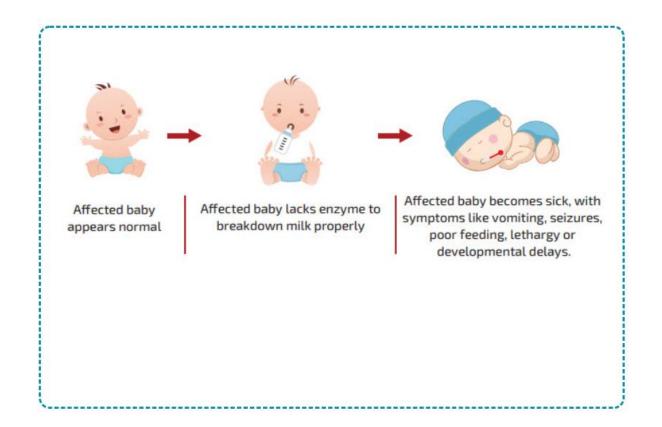


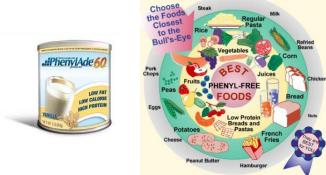
Table 1. Signs and Symptoms of Inborn Errors of Metabolism

- Poor feeding/refusal to feed
- Periods of difficulty consoling
- Periods of difficulty awakening
- Vomiting
- Failure to thrive
- Hypotonia
- Hypertonia
- Tachypnea
- Tachycardia
- Hypoglycemia
- Acidosis
- Dehydration, especially if recurrent
- Apnea
- Seizures
- Altered mental status
- Sudden unexplained death
- Cardiomegaly
- Arrhythmia
- Shock
- Loss of thermoregulation





- Phenylketonuria (**PKU**) is an inborn error of metabolism that results in decreased metabolism of the amino acid phenylalanine.
- Untreated, PKU can lead to intellectual disability, seizures, behavioral problems, and mental disorders.
- Babies with PKU are missing the enzyme Phenylalanine hydroxylase.
- Without this enzyme, levels of Phenylalanine and two closely related substances build up in the baby's body.
- Those substances are bad to the central nervous system and can cause brain damage.
- PKU is inherited or passed down through the family. Both parents must pass on the 'defective' gene in order for the baby to have the condition, this is called "autosomal recessive trait."









*ADAM



Neurological conditions:

- (a) Blindness, including visual impairments
- (b) Cerebral palsy
- (c) Deafness, including hearing impairments
- (d) Epilepsy/seizure disorder
- (e) Hydrocephalus
- (f) Hypoxic ischemic encephalopathy
- (g) Intraventricular hemorrhage (grade IV)
- (h) Microcephaly
- (i) Muscular dystrophy
- (j) Spina bifida
- (k) Spinal muscular atrophy
- (I) Traumatic brain injury



(4) Other:

- (a) Acquired immune deficiency syndrome (AIDS)
- (b) Attachment disorder
- (c) Autism spectrum disorder
- (d) Blood lead level of five micrograms per deciliter or greater
- (e) Cranio-facial anomalies
- (f) Cyanotic congenital heart disease
- (g) Fetal alcohol syndrome
- (h) Infant of untreated mother of phenylketonuria
- (i) Infection, fetal/neonatal (herpes, syphilis, cytomegalovirus, toxoplasmosis, and rubella)
- (j) Neonatal abstinence syndrome
- (k) Sickle cell anemia



• Cerebral palsy (CP) is the most common motor disability in childhood.

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opmental Disabilities

- About 1 in 345 children has been identified with CP according to estimates from the CDC.
- **CP** is a group of disorders that affect a person's ability to move and maintain balance and posture.
- CP is more common among boys than girls, and more common among black children than among white children.
- Most (about 75%-85%) children with CP have spastic CP. This means that their muscles are stiff, and as a result, their movements can be awkward.
- Over half (about 50%-60%) of children with CP can walk independently.
- Many children with CP have one or more additional conditions or diseases along with their CP, known as co-occurring conditions. For example, about 4 in 10 children with CP also have <u>epilepsy</u> and about 1 in 10 have <u>autism</u> <u>spectrum disorder</u>.



- Most CP is related to brain damage that happened before or during birth and it is called congenital CP. Some of these factors include:
 - 1. Being born too small

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opmental Disabilities

- 2. Being born too early
- 3. Being born a twin or other multiple birth
- 4. Being conceived by IVF
- 5. Having a mother who had an infection during pregnancy
- 6. Having <u>kernicterus</u> (a type of brain damage that can happen when severe newborn jaundice goes untreated)
- 7. Having complications during birth

•A small percentage of CP is caused by brain damage that happens more than 28 days after birth. This is called acquired CP. Examples include meningitis or suffering a serious head injury.

- •The specific cause of CP in most children is unknown.
- •CP is typically diagnosed during the first or second year after birth.

•If a child's symptoms are mild, it is sometimes difficult to make a diagnosis until the child is a few years older.





Early Signs of CP

In a baby 3 to 6 months of age:

•Head falls back when picked up while lying on back

•Feels stiff

•Feels floppy

•Seems to overextend back and neck when cradled in someone's arms

•Legs get stiff and cross or scissor when picked up

In a baby older than 6 months of age:

- •Doesn't roll over in either direction
- •Cannot bring hands together
- •Has difficulty bringing hands to mouth

•Reaches out with only one hand while keeping the other fisted

In a baby older than 10 months of age:

•Crawls in a lopsided manner, pushing off with one hand and leg while dragging the opposite hand and leg

•Scoots around on buttocks or hops on knees, but does not crawl on all fours











Autism Spectrum Disorder:







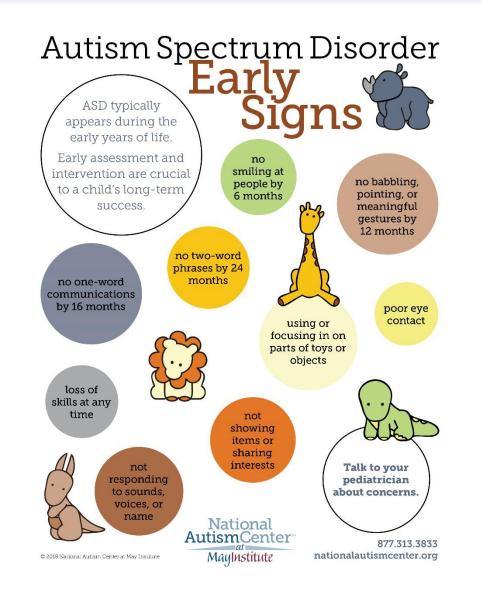


- Autism spectrum disorder (ASD) is a developmental disability caused by differences in the brain.
- Scientists do not know yet exactly what causes these differences for most people with ASD. However, some people with ASD have a known difference, such as a genetic condition.
- There are multiple causes of ASD, although most are not yet known.
- About 1 in 54 children has been identified with autism spectrum disorder (ASD) according to estimates from CDC
- ASD is reported to occur in all racial, ethnic, and socioeconomic groups.
- ASD is more than 4 times more common among boys than among girls.
- About 1 in 6 (17%) children aged 3–17 years were diagnosed with a developmental disability, as reported by parents, during a study period of 2009-2017. These included autism, attention-deficit/hyperactivity disorder, blindness, and cerebral palsy, among others.



- ASD begins before the age of 3 and lasts throughout a person's life, although symptoms may improve over time.
- Some children with ASD show hints of future problems within the first few months of life. In others, symptoms may not show up until 24 months or later.
- Some children with ASD seem to develop normally until around 18 to 24 months of age and then they stop gaining new skills, or they lose the skills they once had.
- Studies have shown that one third to half of parents of children with ASD noticed signs before their child's first birthday, and nearly 80%–90% noticed by 24 months of age.







Possible "Red Flags"

- •Not respond to their name by 12 months of age
- •Not point at objects to show interest (point at an airplane
- flying over) by 14 months
- •Not play "pretend" games (pretend to "feed" a doll) by 18 months
- •Avoid eye contact and want to be alone
- •Have trouble understanding other people's feelings or talking about their own feelings
- •Have delayed speech and language skills
- •Repeat words or phrases over and over (echolalia)
- •Give unrelated answers to questions
- •Get upset by minor changes
- •Have obsessive interests
- •Flap their hands, rock their body, or spin in circles
- •Have unusual reactions to the way things sound, smell, taste, look, or feel



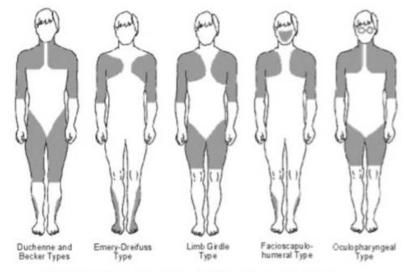


Muscular Dystrophy:

- **Muscular dystrophy** is a group of inherited diseases that damage and weaken your **muscles** over time.
- This damage and weakness is due to the lack of a protein called **dystrophin**, which is necessary for normal **muscle** function.
- The absence of this protein can cause problems with walking, swallowing, and **muscle** coordination.
- In muscular dystrophies, abnormal genes (mutations) lead to muscle degeneration. Most forms begin in childhood.
- Damaged muscles become progressively weaker. Most people who have the condition eventually need a wheelchair. Other symptoms include trouble breathing or swallowing.
- Medications, therapy, breathing aids, or surgery may help maintain function, but life span is often shortened.



Types of Muscular Dystrophy



Main areas of muscle weakness in different types of dystrophy

Duchenne's Impact on the Body

• DECREASED HEART FUNCTION

CARDIOMYOPATHY LEADS TO HEART FAILURE

• WEAKENS DIAPHRAGM • REQUIRES VENTILATOR • LEADS TO PNEUMONIA

LOSS OF MUSCLE MASS
 WEAKNESS
 INFLAMMATION
 FOR THE SECOND



Source: CureDuchenne™





Cranio-facial anomalies

- What is a craniofacial anomaly?
- Craniofacial anomalies are deformities that affect a child's head and facial bones. These disorders are typically present at birth (congenital) and can range from mild to severe.

Common craniofacial anomalies include:

•<u>cleft lip and palate</u> - a separation in the lip and the palate

<u>craniosynostosis</u> - premature closure of the soft spots in an infant's skull
<u>hemifacial microsomia</u> - a condition in which the tissues on one side of the face are underdeveloped
<u>vascular malformation</u> - an abnormal growth

composed of blood vessels

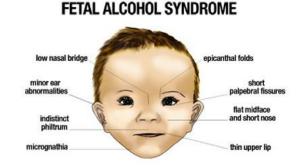
•<u>hemangioma</u> - a benign tumor that causes a red birthmark





Fetal Alcohol Syndrome

- **Fetal alcohol syndrome** is a condition in a child that results from **alcohol** exposure during the mother's pregnancy.
- Fetal alcohol syndrome causes brain damage and growth problems. The problems caused by fetal alcohol syndrome vary from child to child, but defects caused by fetal alcohol syndrome are not reversible.
- Often, FAS children may have distinct features including:
- small head.
- a smooth ridge between the upper lip and nose, small and wide-set eyes, a very thin upper lip, or other abnormal facial **features**.
- below average height and weight.





An infant or child with an FAS might also have:

- Low body weight
- Poor coordination
- •Hyperactive behavior
- •Difficulty with attention
- •Poor memory
- •Difficulty in school (especially with math)
- •Learning disabilities
- •Speech and language delays
- Intellectual disability or low IQ
- •Poor reasoning and judgment skills
- •Sleep and sucking problems as a baby
- •Vision or hearing problems
- •Problems with the heart, kidneys, or bones



<u>Neonatal Abstinence Syndrome (NAS)</u> <u>Neonatal Opioid Withdrawal Syndrome (NOWS)</u>

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DRAMATIC INCREASES IN MATERNAL OPIOID USE AND NEONATAL ABSTINENCE SYNDROME

THE USE OF OPIOIDS DURING PREGNANCY CAN RESULT IN A DRUG WITHDRAWAL SYNDROME IN NEWBORNS CALLED **NEONATAL ABSTINENCE SYNDROME** (NAS), WHICH CAUSES **LENGTHY** AND **COSTLY** HOSPITAL STAYS. ACCORDING TO A NEW STUDY, AN ESTIMATED **21,732 BABIES** WERE BORN WITH THIS SYNDROME IN THE UNITED STATES IN 2012, A **5-FOLD INCREASE** SINCE 2000.



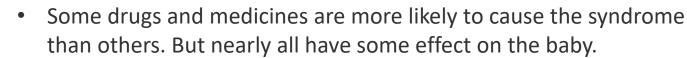


What is neonatal abstinence syndrome/neonatal opioid withdrawal syndrome?

- Neonatal abstinence syndrome is what happens when babies are exposed to drugs in the womb before birth.
- Babies can then go through drug withdrawal after birth.
- The syndrome most often applies to opioid medicines.

What causes neonatal abstinence syndrome/neonatal opioid withdrawal syndrome?

- Almost every drug and medicine passes from the mother's bloodstream through the placenta to her unborn baby.
- If the mother uses substances that affect her nervous system, they will also affect the baby's.
- At birth, the baby has become used to getting the drug. But because the drug is no longer available, the baby may have symptoms of withdrawal.



- When more than one drug has been used, the symptoms are often worse.
- These include:

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- Opioids like heroin and prescribed medicines such as codeine and oxycodone
- Stimulants such as amphetamines or cocaine. For these drugs, the effects on a baby are more likely from the drug itself instead of withdrawal.
- Antidepressant medicines such as selective serotonin reuptake inhibitors (SSRIs)
- Depressants such as barbiturates, or alcohol, or marijuana
- Nicotine from cigarette smoking

Finnegan Neonatal Abstinence Scoring Tool (FNAST)															
atient ID: Name:							Tod	ay's	s W	eig	ht:			DC	DB: Date:
Signs & Symptoms	Time	Sc	Т	Т		1						PM			Comments
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Mild Tremors: Undisturbed Mod-Severe Tremors Undisturbed		3 4													
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Excoriation (Specific Area)		1													
Myoclonic Jerk		3													
Generalized Convulsions		5													
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Sweating		1													
Fever < 101 (37.2-38.3c) Fever > 101 (38.4c)		1 2													
Frequent Yawning (> 3)		1		Т	Γ										
Mottling		1		Т	Γ										
Nasal Stuffiness		1		Т	Γ										
Sneezing (>3)		1													
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Score	_		,	,	1	,	, ,								
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Average Daily Score			+		\vdash										
Inter-Observer Reliability %			+	+	\vdash		H		H		H				
Initials Of Scorer 1			+	+	\vdash										
Initials Of Scorer 2		-	+	+	+	+	\vdash								



Blood lead level of five micrograms per deciliter or greater:

• In Ohio, a child who has a confirmed elevated blood lead level (BLL) of five micrograms per deciliter or greater is automatically eligible for EI.

Lead Exposure and Child Development

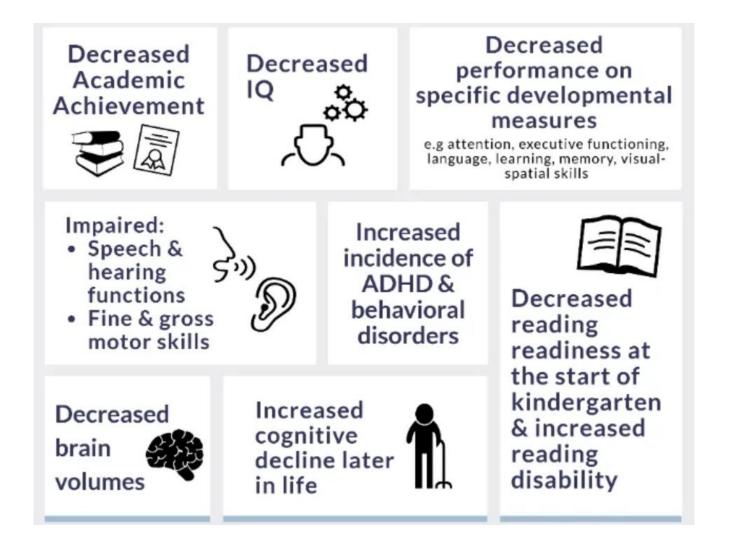
•Children in Ohio are exposed to lead primarily through chipping and peeling paint and dust in homes built before 1978.

•Children under the age of 3 are at the greatest risk of lead poisoning because their brains and bodies are developing quickly, they absorb lead more easily than adults, and they are more likely to be crawling on floors and putting their hands and other objects in their mouths.

•Children who are exposed to lead can have learning issues, such as a lower attention span, behavioral concerns like hyperactivity, or even hearing loss.

A Third Of The World's Children Active and the second seco

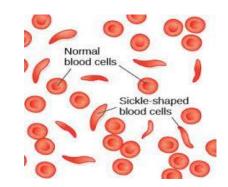


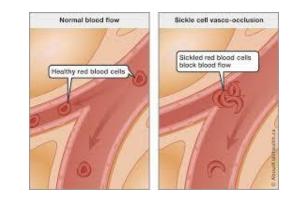




Sickle Cell Anemia:

- A group of disorders that cause red blood cells to become misshapen and break down.
- With sickle cell disease, an inherited group of disorders, red blood cells contort into a sickle shape.
- The cells die early, leaving a shortage of healthy red blood cells (sickle cell anemia), and can block blood flow causing pain (sickle cell crisis).
- Infections, pain, and fatigue are symptoms of sickle cell disease. Long term disease can cause poor growth, organ failure, stroke and even death in children.
- Treatments include medications, blood transfusions, and rarely a bone-marrow transplant.
- Sickle Cell Anemia is genetic and most commonly found in African Americans.
- Most states check newborn babies for abnormal hemoglobin as part of routine newborn screening tests including Ohio.







• Most children with SCD will start to have symptoms during the first year of life, often around 5 months. Each child's symptoms may vary. They may be mild or severe.

•Anemia. This is the most common symptom. Having fewer red blood cells causes anemia. Anemia can make a child pale and tired.

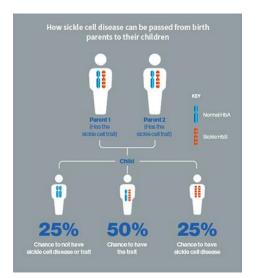
•Yellowing of the skin, eyes, and mouth (jaundice).

Sickle cells don't live as long as normal red blood cells. They die faster than the liver can filter them out. The yellow color is caused by a substance (bilirubin) that is released when the red blood cells die.

•Pain crisis, or sickle crisis. When sickle cells move through small blood vessels, they can get stuck. This blocks blood flow and causes pain. This sudden pain can happen anywhere, but most often occurs in the chest, arms, and legs.
•Splenic sequestration (pooling). The spleen becomes enlarged and painful when sickle cells get stuck and build up there. Fewer red blood cells are able to move. This can cause a sudden drop in hemoglobin. It can be deadly if not treated at once.



SCD affects approximately 100,000 Americans.
SCD occurs among about 1 out of every 365 Black or African-American births.
SCD occurs among about 1 out of every 16,300 Hispanic-American births.
About 1 in 13 Black or African-American babies is born with sickle cell trait (SCT).





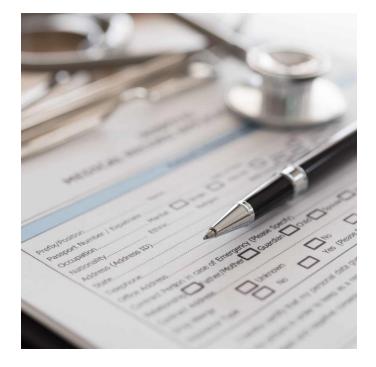


Documentation of a Diagnosed Condition on the List

Hospital record or other documentation from a medical professional which includes:

A diagnosed condition listed in Appendix C of Rule 5123-10-02
The child's name
A professional's signature or authorization (a professional licensed to diagnose and treat mental or physical conditions)

Case notes should reflect the date the EISC received the documentation – this is the date the diagnosis was confirmed



Developmental Disabilities ODH Referrals for NAS or EBLLs

A **system referral** from ODH can serve as documentation of Neonatal Abstinence Syndrome (NAS) or Elevated Blood Lead Levels (EBLL) if:

The referral came from ODH

AND

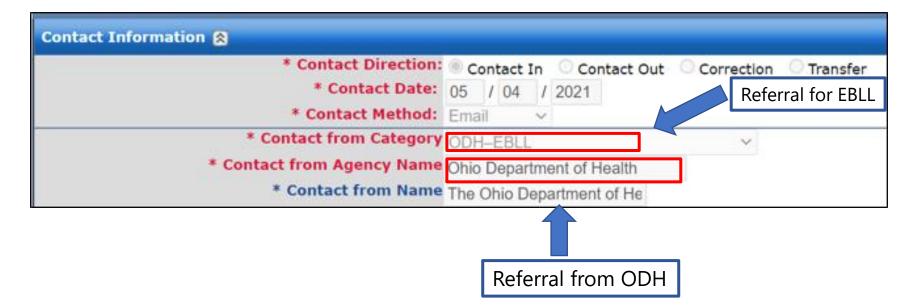
NAS or EBLLs listed as the reason for referral in at least one of these places on the referral

- Contact from Category dropdown
- Diagnosed Physical or Mental Conditions box
- Consolidated Referral Information box
- Referral notes

Date of the program referral = date diagnosed condition was confirmed ODH Referral for NAS or EBLL

Department of Developmental Disabilities





ODH Referral for NAS or **EBLL**

	Is there concern about the child's development?: Yes No
Diagnosed Physical or Mental Conditions:	
Available	Selected
Acquired Immune Deficiency Syndrome Attachment Disorder Autism Spectrum Disorders Blindness, including visual impairments Cerebral Palsy Chromosomal conditions Chronic lung disease (bronchopulmonary dysplasia) Cranio-facial anomalies Cyanotic congenital heart disease	Blood lead level of five micrograms per decliter or greater
4	

Referral Notes:

Department of Developmental Disabilities

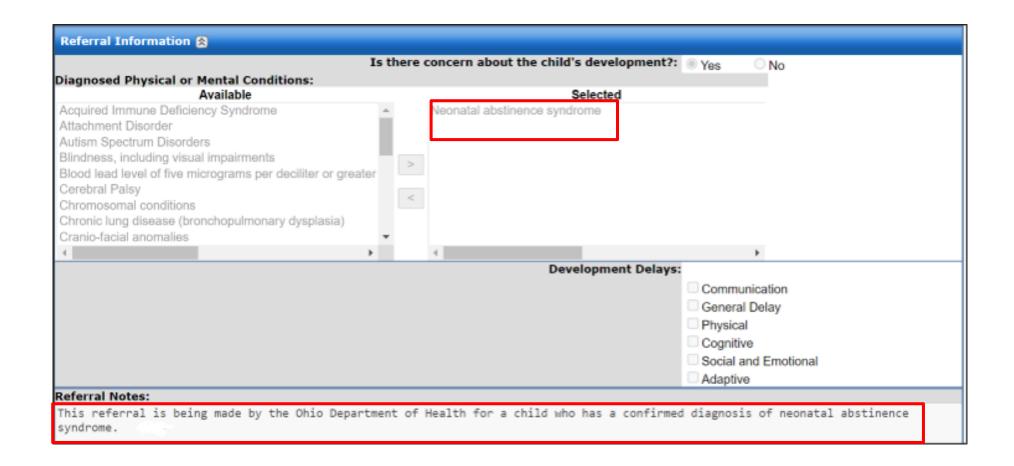
Ohio

"The ODH has referred this child to EI because the child has a confirmed blood lead level of at least five micrograms per deciliter.

ODH Referral for **NAS** or EBLL

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Developmental Disabilities





Referral Information 😫	
Is there co	ncern about the child's development?: OYes ONo
Referral Notes:	
Not interested-DA	
Consolidated Referral Information as of: 2021-05-11 07:32 AM Diagnosed Medical Conditions	Development Delays
Blood lead level of five micrograms per deciliter or greater	
Contact Date: 2020-10-06 08:39 AM - Contact Name: ODH - Diagnose greater,	d Medical Conditions: Blood lead level of five micrograms per deciliter or

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Certificates: Please type your name, role and county in the Questions box. If participating as a group, please include names of all participants.

Webinar was recorded. Link will be posted to the El website, Trainings page within the course description.

Activation code is

DIA0512

Certificate in Control Panel – Handouts section For help accessing certificate, contact Shakila Dixon, Shakila.Dixon@dodd.ohio.gov