Glossary of Birth Anomaly Terms:

A

Anencephaly: A deadly birth anomaly where most of the brain and skull did not form.

Anomaly: Any part of the body or chromosomes that has an unusual or irregular structure.

Aortic valve stenosis: The aortic valve controls the flow of blood from the left ventricle of the heart to the aorta, which takes the blood to the rest of the body. If there is stenosis of this valve, the valve has space for blood to flow through, but it is too narrow.

Atresia: Lack of an opening where there should be one.

Atrial septal defect: An opening in the wall (septum) that separates the left and right top chambers (atria) of the heart. A hole can vary in size and may close on its own or may require surgery.

Atrioventricular septal defect (endocardial cushion defect): A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum. Together, these defects make a large opening (canal) in the middle part of the heart.

Aniridia (an-i-rid-e-a): An eye anomaly where the colored part of the eye (called the iris) is partly or totally missing. It usually affects both eyes. Other parts of the eye can also be formed incorrectly. The effects on children's ability to see can range from mild problems to blindness. To learn more about aniridia, go to the U.S. National Library of Medicine website.

Anophthalmia/microphthalmia (an-oph-thal-mia/mi-croph-thal-mia): Birth anomalies of the eyes. In anophthalmia, a baby is born without one or both eyes. In microphthalmia, one or both eyes are small because they do not develop all the way. Anophthalmia and microphthalmia happen as the baby is developing during pregnancy, and can happen with or without other anomalies. Children with anophthalmia are blind in the affected eye(s). Children with microphthalmia often have limited ability to see or are blind in the affected eye(s). To learn more about anophthalmia/microphthalmia, go to the CDC website.

Anotia/microtia (an-otia/mi-cro-tia): Birth anomalies of the ears. In anotia, the external ear, the part of the ear that you can see, is missing completely. In microtia, the external ear is small and not formed properly. Most of the time, anotia/microtia affects how the baby's ear looks, while the parts of the ear inside the head (the middle and inner ear) are not affected. However, some babies with this anomaly will also have a narrow or missing ear canal. This can mean that the baby has problems hearing with that ear or is deaf in that ear. To learn more about anotia/microtia, go to the CDC website.

B

Biliary atresia (bil-i-ar-y a-tre-si-a): When the tubes, known as bile ducts, inside or outside the liver do not develop normally. The reason why this happens is unknown. Bile is a liquid made by the liver. The bile ducts carry bile from the liver to the gallbladder, where it is stored. The bile ducts help remove waste from the liver and carry salts that help the small intestine break down (digest) fat from food. In babies with biliary atresia, bile flow from the liver to the gallbladder is blocked. This can lead to liver

damage and failure of the liver. To learn more about biliary atresia, go to the National Institute of Health's website.

Bladder exstrophy (blad-der ex-stro-phy): A birth anomaly of the bladder and the front bones of the pelvis. In most cases, the baby is born with the bladder outside of the body. Instead of being a normal balloon shape, the bladder can be in two halves that are inside-out. This makes the bladder unable to store urine (pee) made by the kidneys. The pubic bones at the front of the pelvis normally join to protect and support the bladder and the muscles of the outer belly wall. In children with bladder exstrophy, the pubic bones do not join, leaving a wide opening between the bones. The urethra, the tube from the bladder that allows urine to leave the body, also does not form properly. This means these babies also need their lower urinary tract and their external genitals repaired by surgery. For more information on bladder exstrophy, see the Children's Hospital of Philadelphia website: http://www.chop.edu/conditions-diseases/bladder-exstrophy .

Birth anomaly: A term that includes many conditions including physical malformations, sensory deficits, chromosomal abnormalities, metabolic defects, neurodevelopment disorders, and sometimes complications related to prematurity and low birth weight. Also known as a birth defect or congenital anomaly.

Birth defect: See Birth anomaly.

C

Cataract: An opacity (clouding) of the lens of the eye.

Choanal atresia (cho-a-nal a-tre-si-a): A narrowing or blockage of the air passage of the nose that is present at birth. The condition is the most common nose-related birth anomaly in newborn infants. Girls are born with this condition about twice as often as boys. More than half of babies with choanal atresia also have other congenital problems. Choanal atresia is most often diagnosed shortly after birth while the infant is still in the hospital. To learn more about Choanal atresia, go to the National Institute of Health's website.

Cloacal exstrophy (clo-a-cal ex-stro-phy): Born with part of the large intestine outside of the body and with the two halves of the bladder out on either side of it. In males, the penis is either flat and short or sometimes split. In females, the clitoris is separated into a right half and left half. The intestine may be short and the anus, where normally poop passes out of the body, may not have an opening. With this condition there are often other birth defects, like spina bifida. To learn more about cloacal exstrophy, see the Children's Hospital of Philadelphia website.

Chromosome: A threadlike structure found in the nucleus of most living cells that carries genetic information in the form of genes.

Cloaca: A common chamber into which the intestine and urogenital tracts discharge in early development of the human embryo. In normal development, the common chamber changes into separate areas for the intestinal, genital, and urinary tracts.

Cloacal exstrophy: Congenital persistence of a common cloacal cavity into which gut, urethra, and reproductive tracts open with exstrophy of the cavity.

Clubfoot (Talipes equinovarus): A congenitally misshapen foot twisted out of position.

Coarctation: A stricture or narrowing especially of a blood vessel (such as the aorta)

Coarctation of the aorta: A congenital heart defect where a part of the aorta, the tube that carries oxygen-rich blood from the heart to the body, is narrower than usual.

Common truncus (truncus arteriosus): A rare congenital heart defect (the baby is born with it) in which a single shared tube for carrying blood comes out of the heart, instead of the usual two separate ones.

Common ventricle (single ventricle): See Single ventricle.

Congenital: Present at birth.

Congenital cataract (con-gen-i-tal cat-a-ract): When a baby is born with a clouded lens of one or both eyes. The lens of the eye normally should be clear. When there is a cataract, light that comes into the eye cannot shine well enough through the lens to be focused at the back of the eye, which is needed for clear sight. Cataracts in adults are common, and people often live with them for many years before getting repair. However, in infants, surgical repair as early as possible is critical to make sure that the vision centers of the brain develop normally. Congenital cataract is rare and often occurs as part of other birth anomalies. For more information on congenital cataracts, go to the National Institute of Health website.

Congenital hearing loss: A partial or full deafness present at birth. It can affect one or both ears. Congenital hearing loss can be due to one or more parts of the outer or middle ear not having formed normally, or due to the nerves of the inner ear not having formed or not working properly. Congenital hearing loss can be caused by genes and hereditary (runs in the family) or by other factors present either while the baby is growing in the womb or at the time of birth.
For more information about congenital hearing loss, visit the American Speech-Language-Hearing Association's website.

Congenital posterior urethral valves: A birth anomaly of the urethra in boys. The urethra is the tube that connects the bladder, which stores urine, or pee, to the tip of the penis. Urethral valves are small flaps on the inside of the urethra. In this condition, the valves partly block the tube, and urine can't flow normally from the bladder. The urine backs up and can damage all the urinary tract organs including the urethra, bladder, ureters, and kidneys. To learn more about congenital posterior urethral valves, please visit the Stanford Children's Hospital website.

Craniosynostosis: Craniosynostosis is a birth defect in which the bones in a baby's skull join too early. This happens before the baby's brain is fully formed. As the baby's brain grows, the skull can become more misshapen. To learn more about Craniosynostosis, go to the CDC website.
https://www.cdc.gov/ncbddd/birthdefects/craniosynostosis.html

D

Deletion 22q11 (22q deletion syndrome): A birth anomaly that happens when a small piece of chromosome 22 is missing. The genes of the human body, which are the instruction set of how the body is built, come in 26 pairs of chromosomes. 22q deletion syndrome ("deletion" means "missing") can affect almost any part of the body and how it works. Many children with 22q deletion syndrome have developmental delays, including slow physical growth, a hard time learning to speak well, and learning disabilities. For more information about 22q deletion, go to the U.S. National Library of Medicine website.

Diaphragmatic hernia (di-aphrag-matic h-er-nia): A birth anomaly where there is a hole in the diaphragm (the large muscle that separates the chest from the abdomen). Organs in the abdomen (such as intestines, stomach, and liver) can move through the hole in the diaphragm and upwards into a baby's chest. When an organ pushes through the hole, it is called a hernia. A diaphragmatic hernia can prevent the baby's lungs from developing completely. This causes breathing difficulties for the baby at birth. To learn more about Diaphragmatic hernia, go to the CDC website.

Double Outlet Right Ventricle (DORV): A rare congenital heart defect (the baby is born with it) involving the "great arteries" (the aorta and the pulmonary artery). In a normal heart, the aorta exits from the left ventricle and pumps blood to the body, while the pulmonary artery exits from the right ventricle and pumps blood to the lungs (where it picks up oxygen). In DORV, the aorta and the pulmonary artery both come out of the right ventricle. Children with DORV also have a ventricular septal defect. For more information on DORV, visit the Boston Children's Hospital website tinyurl.com/y9ruxllb.

Down syndrome: A genetic disorder. It is also called trisomy 21. People who have Down syndrome have learning difficulties, intellectual impairment, a characteristic facial appearance, and poor muscle tone in infancy. A child with Down syndrome may have heart anomalies and problems with vision and hearing. How severe or mild these problems is different for each child.

E

Early Hearing Detection and Intervention (EHDI): An Oregon state program that assures all Oregon newborns receive a hearing screening by one month of age. Infants who are referred to newborn screening receive a diagnostic evaluation by three months of age. Infants diagnosed with loss are enrolled into early intervention services by six months of age. For more information, go to the EHDI website.

Ebstein anomaly: A birth anomaly of the heart's tricuspid valve. The tricuspid valve separates the right upper chamber (the right atrium) and the right lower chamber (the right ventricle) of the heart. In a normal heart, the tricuspid valve closes all the way when the right ventricle squeezes to push blood to the lungs. Being closed keeps the blood from going backward. With Ebstein's anomaly of the tricuspid valve, some blood "leaks" back into the right atrium because the valve doesn't close completely. The atrium can start getting too big because of the extra volume of blood. Some children with Ebstein's anomaly of the tricuspid valve also have an atrial septal defect (ASD), a hole in the wall between the two upper chambers of the heart, or other heart defects.

Edwards syndrome: A chromosomal disorder in which there is an extra chromosome 18, that is usually deadly within 2-3 years. It is characterized by intellectual disability, abnormal skull shape, low set and malformed ears, small lower jaw, heart anomalies, short sternum, hernias of the diaphragm or the groin, and other anomalies.

Encephalocele (en-sef-a-lo-seal): A rare type of birth anomaly that happens very early in pregnancy. The neural tube, which ends up forming the baby's brain and spinal cord, starts as a group of special cells that make a "C" shape. Normally, this C-shape of special cells then closes to make a long tube during the third and fourth weeks of pregnancy. Encephalocele happens when the neural tube does not close completely. When this happens part of the brain and the membranes that cover it stick out through an opening in the baby's skull. This usually happens at the back or top of the head or between the forehead and the nose. To learn more about Encephalocele, go to the CDC website.

Esophageal atresia/tracheoesophageal fistula (EA/TEF) (e-soph-a-ge-al a-tre-si-a/tra-che-o-e-soph-a-ge-al fis-tu-la): A birth anomaly of the swallowing tube (esophagus) that connects the mouth to the stomach. In a baby with EA, the esophagus has two separate sections—the upper and lower esophagus—that do not connect. A baby with this birth anomaly is unable to pass food from the mouth to the stomach, and sometimes has difficulty breathing. EA often occurs with TEF, a birth anomaly in which part of the esophagus is connected to the windpipe (trachea). The trachea is the tube that connects the mouth and nose to the lungs. To learn more about EA/TEF, go to the CDC website.

Endocardial cushion defect: A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.

Epispadias: A congenital defect in boys in which the outlet of the urethra (urinary tract) opens at the top of the penis instead of at the tip. There may also be abnormal urinary sphincters, which results in incontinence.

F

Folic acid: One of the B vitamins especially important for a woman to take before conceiving a baby to help prevent neural tube defects. It is obtained from fortified food or from a multivitamin containing at least 400 micrograms each day. It is also found in natural sources including liver, beans, and leafy green vegetables. While folate and folic acid are both forms of water-soluble B vitamins, folic acid refers to the man-made vitamin used in vitamin supplements, whereas folate is the form found in foods.

G

Gastroschisis (gas-troh-skee-sis): A birth anomaly of the abdominal (belly) wall. There is a hole in the muscle and skin next to the belly button. The baby's intestines come out through this hole. The hole can be small or large and other organs, such as the stomach and liver, can also be found outside of the baby's body. Gastroschisis occurs early during pregnancy when the muscles that make up the baby's abdominal wall do not form correctly. To learn more about Gastroschisis, go to the CDC website.

Genetic: Relating to genes or heredity. Some health conditions may be caused by genes or inherited.

H

Hernia: When part of an organ inside the body sticks out through connective tissue or through a wall of the body.

Holoprosencephaly (HPE) (hol-o-pros-en-ceph-a-ly): A birth anomaly in which the developing brain in very early pregnancy doesn't separate normally into separate left and right halves. This condition can also affect development of the head and face. Holoprosencephaly signs can include intellectual disability and problems with the brain's pituitary gland, which is a master control system for many hormones in the body. Holoprosencephaly can be caused by problems with genes, exposure of the mother to certain substances during pregnancy, or the cause can be unknown.

Hypoplasia: See Renal agenesis.

Hypoplastic left heart syndrome (HLHS): A congenital heart condition where the left side of the heart, which normally pumps oxygen-rich blood to the whole body, is very underdeveloped.

Hypospadias: A birth anomaly in boys where the tube that drains urine from the bladder to exit the body, called the urethra, opens on the underside of the penis instead of at the tip. With hypospadias, the opening of the urethra may be located anywhere along the underside of the penis. This can be treated by surgery. When hypospadias is not treated, it can result in problems learning to use the toilet, urine spraying when using the toilet, and a curved penis during erection. More severe forms of hypospadias can interfere with sexual intercourse later in life. In most cases, the exact cause of hypospadias is unknown. Sometimes hypospadias runs in families, but environment also may play a role.

I

Interrupted aortic arch: A rare congenital heart defect (the baby is born with it) that happens when a baby's aorta, the main blood vessel (tube) coming out of the heart to carry oxygen-rich blood to the whole body, doesn't develop correctly. In interrupted aortic arch, the part of the aorta that directs oxygen-rich blood to the lower part of the body does not connect to the heart.

J

K

L

Limb deficiencies: Also called reduction deformities. These include any partial or complete absence of the upper arm, thigh, lower arm, lower leg, wrist, ankle, hand, foot, fingers, or toes.

M

Major anomaly: A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development, or has significant cosmetic impact.

Microcephaly: The congenital smallness of the head, corresponding with smallness of the brain.

Microphthalmia/Anophthalmia (mi-croph-thal-mia / an-oph-thal-mia)**:** See Anophthalmia/microphthalmia.

Microtia/anotia (mi-cro-tia an-otia): See Anotia/microtia

Minor anomaly: A congenital abnormality that does not require medical or surgical treatment, does not seriously affect health and development, and does not have a significant cosmetic impact.

Multiple anomalies: Two or more defects affecting an infant and occurring in different organ-systems or body sites, that are not part of a known embryological sequence or syndrome, and do not have a common primary defect.

N

Neural Tube: In an embryo, the hollow tube from which the brain and spinal cord form.

Neural tube defect: The failure of the neural tube to close in the first month of pregnancy. Anomalies in this category include anencephaly, spina bifida, and encephalocele.

Normal variant: A minor anomaly that occurs in approximately 4% or more of the population.

O

Obstructive genitourinary defect: Stenosis or atresia of the urinary tract at any level.

Omphalocele (uhm-fa-lo-seal): A birth anomaly of the abdominal (belly) wall. The infant's intestines, liver, or other organs stick outside of the belly through the belly button. These organs are covered in a thin, clear sac. Normally, as a baby develops during 6-10 weeks of pregnancy, the intestines get longer and push out from the belly into the umbilical cord. By the eleventh week of pregnancy, the intestines normally go back into the belly. However, if this does not happen, an omphalocele occurs. The omphalocele can be small, with only some of the intestines outside of the belly, or it can be large, with many organs outside of the belly. For more information about Omphalocele, go to the CDC website.

P

Patau syndrome: A chromosomal disorder with an extra copy of chromosome 13. It is usually deadly by 2 years of age. It is characterized by intellectual impairment, malformed ears, cleft lip or palate, small eyes, small jaw, extra fingers, and heart, kidney, and other anomalies.

Patent ductus arteriosus: An opening that connects the two major blood vessels leading out from the heart. The opening, called the ductus arteriosus, is a normal part of a baby's circulatory system before birth that usually closes shortly after birth.

Pulmonary valve atresia: A rare congenital heart defect where the valve that controls blood flow from the heart to the lungs doesn't form at all. In babies with this defect, blood has trouble flowing to the lungs to pick up oxygen for the body. This condition may be diagnosed during pregnancy or soon after a baby is born.

Q

R

Rectal and large intestinal atresia: A group of rare birth anomalies of the lower intestines in which they are narrowed, blocked, or disconnected. If they are "rectal", that means it occurs in the anus, from which poop passes out of the body. If they are "large intestinal", that means it occurs in the large bowel, which is the structure in which poop is formed before passing through the anus. "Atresia" means that part of the rectum or large intestine did not form at all. To learn more about rectal and large intestinal atresia, please visit the Minnesota Department of Health website.

Rectal and large intestinal stenosis: A group of rare birth anomalies of the lower intestines in which they are narrowed, blocked, or disconnected. If they are "rectal", that means it occurs in the anus, from which poop passes out of the body. If they are "large intestinal", that means it occurs in the large bowel, which is the structure in which poop is formed before passing through the anus. "Stenosis" means that the part is too narrow. These conditions are sometimes detected before birth. To learn more about rectal and large intestinal stenosis, please visit the Minnesota Department of Health website.

Reduction deformities of the brain: A birth anomaly in which a portion of the brain does not develop.

Renal agenesis: A birth anomaly in which one or both kidneys did not form or formed incompletely.

Reportable disease: A health condition or diagnosis that is required to be reported to a public health agency due to its public health significance. Birth anomalies are not reportable in Oregon.

S

Single ventricle (common ventricle): Single ventricle anomalies of the heart occur when one of the two pumping chambers in the heart, called ventricles, isn't large enough or strong enough to work correctly. In some cases, the chamber might be missing a valve. There are several types of single ventricle defects, and each has a unique name.

Small intestinal atresia/stenosis: A birth anomaly in which the small intestine, the digesting tube after the stomach, is narrowed, blocked, or disconnected. "Atresia" means that part of the small intestine did not form at all. "Stenosis" means that part of the small intestine is too narrow. This anomaly prevents the baby from absorbing nutrition properly. They are often called "bowel obstructions." To learn more about small intestinal atresia/stenosis, please visit the Stanford Health website.

Small intestinal stenosis: See Small intestinal atresia

Spina bifida: A condition in which the neural tube, a layer of cells that ultimately develops into the brain and spinal cord, fails to close completely. The spinal cord is the bundle of nerves that come down from the brain to control and to send signals back from the entire body. Spina bifida can occur anywhere on the back from the upper neck to the tailbone area. As a result, when the spine forms, the bones of the spinal column do not close completely around the developing nerves of the spinal cord. The effect on the child depends upon if the spinal cord itself is formed correctly. It also depends upon where the spina bifida opening is on the neck or back, how long it is, and whether all or only some of the layers have an opening. For more information about spina bifida, see tinyurl.com/ya4eowbo.

Surveillance: In public health, surveillance is the tracking of information on health issues in the population.

Syndrome: A grouping or pattern of abnormalities which, when found together, form a specific diagnosis.

T

Talipes equinovarus: See Clubfoot.

Tests: A medical test is a kind of medical procedure performed to find, diagnose, or keep track of a disease, see what the disease is doing in the body, find out if a person might be able to get a certain disease, and figure out how best to treat it.

Tetralogy of Fallot: A rare congenital heart defect that affects normal blood flow through the heart. It causes the amount of oxygen in the blood to be reduced. Tetralogy of Fallot is considered a critical congenital heart defect, as most babies need surgery or other procedures soon after birth.

Total anomalous pulmonary venous return (TAPVR): A rare congenital heart defect in which oxygen-rich blood does not return from the lungs to the left atrium. Instead, the oxygen-rich blood returns to the right side of the heart. This causes the baby to get less oxygen than is needed to the body.

Tracheoesophageal fistula: See Esophageal atresia.

Transposition of the great arteries (TGA): A rare congenital heart defect in which the two main arteries carrying blood out of the heart are switched in position. Babies with TGA may need surgery or other procedures soon after birth.

Transposition of the great vessels: See Transposition of the great arteries.

Tricuspid valve atresia: A rare congenital heart anomaly where the valve that controls blood flow from the right upper chamber of the heart to the right lower chamber of the heart doesn't form at all. In babies with this anomaly, blood can't flow correctly through the heart and to the rest of the body.

Tricuspid valve stenosis: A rare congenital heart anomaly where the valve that controls blood flow from the right upper chamber of the heart to the right lower chamber of the heart doesn't form correctly and has a very narrow opening. In babies with this anomaly, blood can't flow correctly through the heart and to the rest of the body.

Trisomy: A chromosomal abnormality in which cells contain three copies of a particular chromosome, instead of the usual two.

Trisomy 13: Having three #13 chromosomes, instead of the usual two. See also Patau syndrome.

Trisomy 18: Having three #18 chromosomes, instead of the usual two. See also Edwards syndrome.

Trisomy 21: Having three #21 chromosomes, instead of the usual two. See also Down syndrome.

Truncus arteriosus: See Common truncus.

Turner Syndrome: A chromosomal condition that affects development in girls. The most common feature of Turner syndrome is being short, which is noticed by about age 5. It is also very common for the ovaries to not work. Many girls with Turner Syndrome do not go through puberty unless they get treatment to replace the missing female hormones. Most of them are unable to conceive children in later life. Most girls and women with Turner syndrome have normal mental development. However, sometimes they can have developmental delays, learning disabilities, and behavior problems. For more information about Turner syndrome, go to the U.S. National Library of Medicine website.

U

V

Ventricular septal defect (VSD): A heart anomaly where there are one or more holes in the wall (septum) that separates the two lower chambers (ventricles) of the heart. This forces the heart and lungs to work harder and can increase the risk for other health problems.

Ventricle: One of the two lower chambers of the heart that perform most of the heart's pumping action. The right ventricle pumps blood into the lungs to receive oxygen. The left ventricle pumps oxygen-rich blood to all parts of the body.

W

X

Y

Z

Zika virus: A disease spread to people usually through the bite of an infected mosquito. It can also be spread between people by having sex with a partner infected with Zika virus. The illness is usually mild with symptoms lasting up to a week, and most people do not have symptoms at all. However, any infection with Zika virus infection during pregnancy can cause a serious birth defect called microcephaly and other severe brain defects.