22q Glossary (Third Edition)

A dictionary of terms and definitions related to 22q11.2 differences

22q International 22q11.2 Foundation

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What does **22q11.2DS** mean?

22q11.2DS stands for **22q11.2 deletion syndrome.**

22	Chromosome 22 (the smallest of the human chromosomes, in every cell of the body)
q	The long arm of the chromosome
11.2	The position on the chromosome – like the GPS coordinates
Deletion	A piece missing from one of the pair of chromosome 22
Syndrome	A collection of features

Old Names of 22q11.2 deletion syndrome:

- Autosomal dominant Opitz G/BBB syndrome
- CATCH22
- Cayler cardiofacial syndrome
- Conotruncal anomaly face syndrome (CTAF)
- Deletion 22q11.2 syndrome
- DiGeorge syndrome
- Sedlackova syndrome
- Shprintzen syndrome
- VCFS
- Velocardiofacial syndrome
- Velo-cardio-facial syndrome

Now that researchers have found the unifying cause, we encourage everyone to use the name **22q11.2 deletion syndrome** (**22q11.2DS**)**.** In English-speaking communities, abbreviations that are also in use for 22q11.2 deletion syndrome include: 22q, 22qDS, and 22q11DS.

What does **22q11.2DupS** mean?

22q11.2DupS stands for **22q11.2 duplication syndrome.**

22	Chromosome 22 (the smallest of the human chromosomes, in every cell of the body)
q	The long arm of the chromosome
11.2	The position on the chromosome – like the GPS coordinates
Duplication	A piece that is repeated on one of the pair of chromosome 22
Syndrome	A collection of features

Disclaimer: The information in this glossary is provided for educational purposes only. It is not intended to be taken as medical advice. The needs for everyone are different, and not all information is applicable to everyone. If you have questions or concerns, please discuss them with your doctor or healthcare provider.

A

Aberrant Subclavian Artery – The subclavian arteries are two vessels that bring oxygenated blood to the head, neck, and arms. The left subclavian artery normally arises directly from the aorta (a larger vessel originating from the heart), while the right one arises from a branch of the aorta (brachiocephalic trunk). [See image of subclavian arteries on Wikipedia.] If they arise incorrectly from their origin, they are called aberrant. [See image of aberrant subclavian arteries on Wikipedia.] The condition may cause no symptoms, but it can also lead to difficulty swallowing (from compression on the esophagus), difficulty breathing, stridor or cough (from compression on the **trachea** or bronchi), or chest pain. Doctors will likely need several imaging studies to diagnose aberrant subclavian arteries. If they are asymptomatic, no treatment is necessary. If, on the other hand, they cause symptoms, one or more surgeries are needed to correct the problem.

Achalasia – [See **Esophageal Dysmotility**]

Adaptive Functioning – Adaptive functioning describes how a person manages common day-to-day demands in response to the environment and, most importantly, how independent they are relative to people of a similar age and background. Adaptive functioning includes daily living skills, communication, and social skills. There is a wide range in the level of adaptive functioning among individuals with chromosome 22q differences. Assessments are recommended every few years, as independent functional demands continually escalate at higher ages.

Adenoidectomy – The adenoids are a patch of tissue high on back of the throat and nose that trap harmful viruses and bacteria that we inhale or swallow. Sometimes, the adenoids get larger when they help the body fight off infections or when they themselves get infected. Enlarged adenoids can sometimes makie it difficult to breathe at night. In some situations, doctors may recommend an adenoidectomy, which means having the adenoids removed by surgery. Children with 22q who have adenoidectomy are at risk of developing **velopharyngeal dysfunction** after surgery. Adenoidectomy should be avoided in children with 22q unless it is medically necessary to remove them. The decision should be made with an interdisciplinary team. If adenoidectomy is necessary, a partial adenoidectomy is recommended if medically feasible.

Adenoids – Adenoids are a patch of tissue that sits at the back of the throat behind the nose and the top of the mouth. They are part of the immune system – they trap harmful bacteria and viruses that we breathe in or swallow.

Allergy – Our immune system usually fights off external things that are harmful to the human body. Allergy is a condition in which the immune system reacts to something that is harmless to most people. The **allergen** that triggers the allergic response can be food, animals, medications, plants, or other things in the environment. Depending on the allergen and the individual, different parts of the body may be affected. Mild allergies may not cause a lot of problems, but severe allergic reactions (called **anaphylaxis**) that are not treated right away can lead to death. Allergy is a condition that affects up to about 40% of children with 22q11.2 deletion syndrome and has also been reported in those with 22q11.2 duplication syndrome.

Amblyopia ("Lazy eye") – Amblyopia refers to vision which is reduced in one eye (or both eyes in rare cases) as a result of the visual pathway between the brain and the eye not being properly stimulated due to unequal need for glasses in either eye or misaligned eyes. This condition occurs in early childhood but can be corrected using eyeglasses or penalization (patching or atropine drops). If untreated, amblyopia can lead to permanent vision loss. Since parents may not realize that the symptoms indicate amblyopia, it is recommended that children have a vision screening in their early years.

Amniocentesis – Amniocentesis is a test done on pregnant women to check the health and/or genetic status of the fetus. It involves inserting a needle into the uterus to extract a small amount of amniotic fluid, which contains fetal cells and proteins. Families are strongly advised to learn about the risks of this procedure and be prepared for the results before this test.

Anesthesia – Anesthesia is the use of medications to help the person avoid pain and feelings during surgeries or medical examinations. There are many types of anesthesia, with different effects and risks. [See the website of the American Society of Anesthesiologists]

Anomaly – An anomaly is something that is different from what is expected.

Anotia – Anotia, a rare condition among individuals with 22q11.2DS, means that a baby is born without the outer part of the ear (visible on the side of the head). The inner parts of the ear may or may not be formed properly. Children with anotia often have difficulties with hearing and speech. They need surgeries to rebuild the ear, treatment for **hearing loss**, and speech therapy to help with speech development.

Antibodies – Antibodies (also called **immunoglobulins**) are Y-shaped proteins produced by plasma cells (mature **B cells**) of the immune system. They bind to a variety of targets, including bacteria, viruses, leading to clearance or inactivation. Rarely, antibodies get made to components of our own bodies, a process called **autoimmunity** [See **Autoimmune Disorders**]. The human immune system can make antibodies with a very wide variety of binding sites. Some children with 22q11.2 deletion syndrome experience a gradual decrease in antibody function and numbers. Antibody deficiencies have also been reported in those with 22q11.2 duplication syndrome.

Anticipatory Anxiety – Anticipatory anxiety means worrying excessively about what might happen, ahead of an event such as an appointment.

Anxiety Disorders – Most people get worried or feel stressed from time to time, and the feeling usually goes away after a while. A person may have an anxiety disorder if the fears are excessive, have been present over a period of time, and affect how the person functions with people, at home, and at school or work. Anxiety disorders are treatable conditions. Recognizing the presence, and the type, of anxiety disorder is important for deciding on the best treatment options. [See the Mental Health Series on the Health Conditions Explained page of our website.]

Aorta – The aorta is the largest blood vessel (artery) in the human body. It takes oxygen-rich blood from the heart to the rest of the body. [See "The Heart and Normal Blood Flow" in the Heart Series on the Health Conditions Explained page of our website.]

Aortic Arch Anomalies (**AAA**) – The **aorta** normally forms a left curved structure (called the "left-sided aortic arch") as it exits the heart. Aortic arch anomalies include diverse malformations that involve the aortic arch and the vessels that branch out from the aortic arch. There may be (1) changes in the way the aortic arch is curved or located (e.g., a right-sided aortic arch); (2) an interruption of the aortic arch; (3) a double aortic arch; and (4) abnormalities of the aortic vessels. AAA may happen on its own or together with other congenital heart diseases. It may lead to no symptoms or cause symptoms in the respiratory system or the esophagus. [For an example of AAA, please see "Interrupted Aortic Arch" in the Heart Series on the Health Conditions Explained page of our website.]

Aortic Root Dilation – The **aorta** is the large blood vessel that brings oxygenated blood from the heart to the rest of the body. An aortic root dilation is due to an increase in the normal diameter of the root (the beginning) of the aorta, which can sometimes prevent the aortic valve from fully closing. This bulging of the root allows some of the blood to flow back into the heart, resulting in reduced blood flow to the rest of the body. Severe cases of aortic root dilation can lead to more serious outcomes, including the formation of small blood clots at the bulge. The clots can flow into the blood stream and block vessels in the brain, causing a stroke. The bulge itself can rupture, and blood can quickly leak out of the aorta. If the dilation becomes very big, it is called an **aneurysm**, and surgery may be needed. People with an aortic root dilation may not have any symptoms, but doctors can discover the problem using X-ray, echocardiogram, or CT scan/MRI for a more detailed study of the structure of the heart and the blood vessels nearby. It is known that children and adults with 22q11.2 deletion syndrome can develop aortic root dilation, whether they have other existing congenital heart diseases or not. Therefore, it is important to get a heart checkup periodically.

Apraxia – [See **Motor Speech Disorders**]

Arrhythmia – Arrhythmia means having an irregular heartbeat. The heart may beat too fast (tachycardia), too slowly (bradycardia), or at an irregular rhythm. Some arrhythmias may be harmless and may not cause any signs or symptoms. However, other arrhythmias can cause troublesome, sometimes even life-threatening signs and symptoms. The treatment for arrhythmias may include medications, catheter procedures implanted devices or surgery.

Arteries – Arteries carry oxygen-rich blood from the heart to different parts of the body.

Articulation – Articulation is the ability to form the sounds (consonants and vowels) of your language to make words, phrases, and sentences. Articulation is a crucial part of speech formation, which is comprised of the following processes: (1) respiration (breath support for speech); (2) phonation (vibration of the vocal cords to form the voice); (3) articulation (making the sounds of speech in the mouth); and (4) resonation (the separation of air between the mouth and nose. All four parts require precisely coordinated movements. [See "Introduction to Speech" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Aspiration – When we swallow food and drinks, they normally go down the **esophagus**, which then brings them down to the **stomach**. Aspiration happens when the food or drinks down the wrong path, entering the windpipe or even reaching the lungs. The affected person can experience coughing, choking, difficulty breathing, and recurrent pneumonias (serious lung infections). Aspiration can also happen silently (without coughing), and parents and caregivers may not know. [See "Feeding Difficulties" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Asthma – Asthma is a condition associated with inflammation and narrowing of airways. The symptoms include shortness of breath, chest tightness, coughing, and wheezing. While airway muscles are relaxed in most people, they are sensitive and often inflamed in a person with asthma. When something irritates or triggers asthmatic airways, the muscles tighten even more, and the airways fill up with mucous. This makes it difficult to breathe – a situation commonly known as an **asthma attack**. Asthma cannot be cured but can be managed with medications. Scientists don't know why some people get asthma while others don't. Nevertheless, asthma is not infectious, so it cannot be caught from another person. Asthma has been reported in children with 22q11.2 deletion syndrome as well as those with 22q11.2 duplication syndrome.

Astigmatism – An astigmatism is a condition in which the cornea or the lens of the eye is curved irregularly. A normal eye is shaped like a round ball (like a basketball), while an eye with an eye with astigmatism is egg - or oval-shaped (like an American football). As a result, the light entering an eye cannot reach the brain properly (i.e., a **refractive error**), and the vision is blurry at both near and distance. Regular vision checks are recommended for all ages, and astigmatism can be corrected by eyeglasses or refractive surgeries.

Asymmetric Facies – [See **Hemifacial Paresis**]

Atopy – Atopy means having a genetic tendency to develop allergies. It is a common condition in 22q11.2 duplication syndrome. (see **Allergy**)

Atrium – An atrium is an upper chamber of the heart. The human heart has two of them: the right atrium receives oxygen-poor blood from different parts of the body, while the left atrium receives oxygen-rich blood from the lungs. [See "The Heart and Normal Blood Flow" in the Heart Series on the Health Conditions Explained page of our website.]

Attention Deficit Disorder (**ADD**) / **Attention Deficit Hyperactivity Disorder** (**ADHD**) – The official medical term is attention deficit hyperactivity disorder (ADHD). It is a common childhood condition, affecting approximately 10% of all schoolchildren, related to the brain's growth and development. The prevalence of ADHD is much higher (40-50%) among children with 22q11.2 deletion syndrome. Children with ADHD have difficulties paying attention and/or controlling impulsive behavior, or they may be overly active. The condition often lasts into adulthood, where difficulties with "executive functions" such as planning, organization, emotional dysregulation, etc. become more problematic.

Audiology – Audiology is a healthcare field that deals with hearing and balance. A healthcare provider who diagnoses and treat hearing and balance problems is called an audiologist.

Auricular anomalies – Auricular anomalies are ear problems. In 22q11.2DS these affect the structure of the external, middle, and inner ear, with ears possibly differing in size and function. Some individuals may develop **hearing loss**.

Autism Spectrum Disorder (**ASD**) – Autism Spectrum Disorder (ASD) is a condition in which a person's brain development affects his/her ability to socialize, communicate, and behave. There is a wide range of symptoms, which is the reason for having "spectrum" in the name of the condition. ASD is diagnosed by meeting a minimum number of impairments in the three domains of social interaction, communication, and restricted repetitive and stereotyped patterns of behavior, interests, and activities. Signs of ASD are typically obvious by the time a child is 2 years old, and each child with ASD has his/her own set of symptoms and level of severity. Among children confirmed to have 22q11.2 duplication syndrome (22q11.2DupS), perhaps 14-25% have autism spectrum disorder (ASD). Among children with 22q11.2 deletion syndrome (22q11.2DS), ASD estimates range from 10-40%. Many children with 22q11.2DupS or 22q11.2DS have some features of ASD, but do not meet full diagnostic criteria. Further, the true rate of ASD in 22q11.2DupS or 22q11.2DS may not be known because some individuals may not be diagnosed.

Autoimmune Disorders – Our immune system can usually distinguish between foreign entities and our own body. Autoimmune disorders happen when our immune system attack components of our own healthy body by mistake. There are over 100 autoimmune disorders. The overall frequency of autoimmune disease may be as high as 25% in 22q11.2 deletion syndrome.

Autoimmune Enteropathy/Enteritis – Autoimmune enteropathy/enteritis is a condition in which the immune system attacks the inside lining of the intestines. Symptoms such as having very frequent and severe diarrhea and poor weight gain can start soon after birth. [See "Autoimmune Issues in the GI System" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

B

B Cells (**B Lymphocytes**) – B cells are a type of white blood cells in the immune system. The letter "B" comes from the fact that these cells were first discovered in the Bursa of Fabricius of chickens. In humans, B cells mature in the bone marrow. The surface of B cells displays receptors that are basically **antibodies** (also called **immunoglobulins**) tethered to the surface of B cells, and the human genetic make-up allows for many possible types of antibodies. During B cell development, these antibodies are "checked" to make sure they do not accidentally react to components of the healthy human body. Once B cells are ready to function, their surface antibodies survey for proteins and pathogens around them. Unlike T cells, B cells do not need the proteins and pathogens to be broken down for recognition and response. If the surface antibodies bind successfully to a foreign item, the B cell will generate many copies of itself, so that there are more of the same B cells to bind to the invaders. Some B cells will mature into plasma cells that makes many more of the same antibodies—this time not attached to cell surfaces. These free-flowing antibodies will go around the body to bind to the targets and trigger a response from the immune system to remove them. Some individuals with 22q11.2 deletion syndrome have a deficiency in antibody-mediated immune responses, but scientists are still trying to find out the exact details. B cell deficiencies have also been reported in children with 22q11.2 duplication syndrome.

Bernard-Soulier Syndrome (**BSS**) – Individuals with BSS have platelets (an important component for forming blood clots) that are large and do not work properly. The reason they don't work as well is because the platelets lack certain proteins (including GPIBB) that are necessary for the platelets to come together and form a blood clot. In patients with BSS, because of the poor platelet function, it can be difficult to stop bleeding when there is injury to a blood vessel and individuals can have too much bleeding (bruising, nosebleeds, bleeding with surgery and with injuries and heavy menstrual bleeding) because it can take longer for bleeding to stop. The gene that encodes the GPIBB protein is located between LCR22A and LCR22B on chromosome 22, so individuals with any of the A-B, A-C, or A-D 22q11.2 deletions are in fact missing one of the two copies of the GPIBB gene. Most patients with 22q11.2DS have a functional copy of the GPIBB gene on the undeleted chromosome and are unlikely to have BSS. However, in rare cases where there is something wrong with the remaining GPIBB gene, the person will have both 22q11.2DS and the bleeding disorder BSS.

Bifid Uvula – The **uvula** is the small, bell-shaped piece of flesh hanging at the back of the soft palate (roof of the mouth). A bifid uvula is one that is split into two halves. It may not be easy to see a bifid uvula very well in a newborn baby, but the condition may be more obvious as the uvula grows and develops. Having a bifid uvula may mean that the child has **submucous cleft palate**, which is when the muscles in the roof of the mouth are split. He/she may struggle with feeding, or milk may leak out from the nose. Later, in some cases, speech may be nasal. Since palate problems are very common in babies with 22q11.2DS, it is recommended that they receive a thorough check by a doctor specialized in these conditions. A surgery may be needed to repair the palate to avoid further speech complications.

Bile – [See **Gallbladder**]

Biliary Colic – Biliary colic is an intense severe pain in the upper right area of the abdomen. It happens when the **gallbladder** contracts to release bile after a large meal. [See "**Gallstones**" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Bipolar Disorder (previously called **Manic Depression**) – Bipolar disorder is a brain condition that involves episodes of major mood changes, intense emotional highs and lows, often with a normal phase separating episodes. These extreme moods affect how they think, behave, and function. If left untreated, there may be serious complications. There are various subtypes of bipolar disorder based on the severity and types of symptoms. Some other physical and mental health conditions may worsen bipolar disorder.

Blood Vessels – Blood vessels are channels that allow blood to flow through. There are three main types: **arteries** carry oxygen-rich blood from the heart to different parts of the body; **veins** carry oxygen-poor blood back from different parts of the body back to the heart; and **capillaries** are the smallest blood vessels that connect arteries and veins.

Bradycardia – [See **Arrhythmia**]

Bruise – When an area of our body gets bumped, one or more blood vessels breaks. A small amount of blood leaks out into the skin and appears as the dark mark – which is the bruise. Usually, the body eventually reabsorbs the blood, and the bruise gradually disappears. However, frequent large bruising, or bruises that are raised and can be felt, may indicate a more severe problem (such as an issue with blood clotting).

C

Caffeine – Caffeine is a stimulant that is present in coffee, tea, cocoa, and some other plants. It is also found in energy drinks and a variety of supplements, such as meal replacements and pre or post workout supplements. Excessive caffeine can lead to jitteriness, anxiety, and trouble sleeping. Having too much caffeine or mixing caffeine with alcohol/medications is unsafe, especially for children and youth.

Cardiology – Cardiology is the branch of medicine that deals with the heart as well as the blood vessels and structures around it. A medical doctor that specializes in cardiology is called a **cardiologist**.

Cataract – The lenses inside the eyes are normally clear. A cataract is the clouding of the lenses which can reduce vision. This condition has been reported in individuals with 22q11.2DS and 22q11.2DupS.

Catatonia – Catatonia is a severe condition in which individuals can stop talking, stop eating, display extreme physical agitation and/or extreme slowness, have odd movements or postures, and sometimes require management in an intensive care unit. Psychotic illnesses that are not treated can increase the risk of catatonia. [See **Psychosis**.]

Celiac Disease – Celiac disease is a condition in which gluten in food causes the immune system to attack and damage the lining of the small intestines, making it difficult to absorb nutrients. [See "Autoimmune Issues in the GI System" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Choanal Atresia – Choanal atresia is a condition in which the back of the breathing passages of one or both sides of the nose are blocked at birth. It is rare among individuals with 22q11.2DS. If it happens on one side, it may not be noticed until the child is older. However, if it affects both side of the nose, the baby will have trouble breathing (since babies mainly breathe through their noses except when crying). Choanal atresia affecting both sides of the nose is an emergency that may require doctors to insert a breathing tube into the windpipe immediately in order to let the newborn baby breathe before planning for a surgery to open the blockage.

Cholelithiasis – [See **Gallstones**]

Chronic Infection – An infection is an illness caused by pathogens such as viruses, bacteria, or yeasts. Usually, the immune system of the human body can battle these pathogens and eliminate them. If the pathogens cannot be cleared from the body for a prolonged period, the person is said to have a chronic infection. This condition can occur in people with 22q11.2 deletion syndrome as well as those with 22q11.2 duplication syndrome.

Cleft Palate – The **palate** is the roof of the mouth. The front of the palate is the hard palate and the part of the palate in the back is the soft palate. A cleft palate is a split in the roof of the mouth, leaving an opening between the mouth and the nose, allowing food or liquids to come through the nose with feeding and air to come through the nose with speech. A cleft palate happens if the two halves of the palate do not fuse together properly in utero (before birth). An overt cleft palate is less common in 22q than a submucous cleft palate. A submucous cleft palate is a split in the muscle that is covered by the membrane of the palate. There are specific visible characteristics of a submucous cleft palate including: a notch on the posterior border of the hard palate, a bifid (split) uvula, a bluish or translucent area on the palate known as a zona pellucida. Surgery is necessary to repair an overt cleft palate but repair of a submucous cleft palate is determined by its impact on speech. If a submucous cleft palate does not affect speech, it does not have to be repaired. Cleft palate occurs in about 11% of babies with 22q11.2DS, but about 70% of patients with 22q11.2DS have a different palate disorder called Velopharyngeal Dysfunction. [See also: **Submucous Cleft Palate**, **Bifid Uvula**, and **Velopharyngeal Dysfunction**]

Club Foot (**Pas equinovarus**) – Club foot is when a baby is born with one or both feet turned downward and inward instead of straight. Club foot happens in about 1 in every 150 to 1000 individuals in the general population but in about 1 in every 9 individuals with 22q11.2DS. Club foot can be diagnosed using ultrasound at about 20 weeks of pregnancy. Treatment is possible using casts and braces, sometimes over several years, and/or surgery. If not corrected, club foot can cause significant problems with walking, arthritis, and poor self-image.

Cognitive Behavioral Therapy (**CBT**) – CBT is now the most common form of psychotherapy ("talk" therapy). CBT helps people find out how their thoughts, attitudes, and beliefs affect their emotions and behavior, and potentially make changes to aspects that cause difficulties. Professionals who can provide CBT include psychiatrists, psychologists, family doctors, and some counsellors. Some health insurance policies (but not all) may cover CBT. [See also **Psychiatry** and **Psychology**]

Cognitive Impairment – Cognitive impairment is a broad term describing either a failure to develop or a decline in previously intact abilities, such as memory, attention, reasoning, judgement, the ability to plan and perform complex tasks, etc.

Coloboma – A coloboma is a condition in which the eye is missing some tissue at birth. There are different types of colobomas, depending on the exact type of tissue that is missing. A coloboma can result in a loss of vision as well as sensitivity to light. Coloboma has been reported in individuals with 22q11.2DS and 22q11.2DupS.

Colon – [See **Large Intestines**]

Comorbidity – Comorbidity is the state of having more than one illness at a time in the same person. Most individuals with 22q differences have multiple health conditions.

Compensatory Articulation Disorders – Compensatory articulation disorders consist of abnormal articulation patterns that result from alterations in the structure and/or function of the speech mechanism. These are most often errors in the place of articulation of sounds, such as sounds made in the throat or nose, instead of in the mouth. Examples of compensatory articulation errors include glottal stops as well as pharyngeal fricatives/stops, nasal fricatives, clicks and others. Compensatory articulation is common (~26-56%) in 22q11.2DS and occurs in the presence of cleft palate and/or **velopharyngeal dysfunction**.

Conductive Hearing Loss – [See **Hearing Loss**]

Congenital – Congenital means present at birth. Some features in 22q11.2 deletion and duplication syndromes are congenital, while others develop later in life.

Congenital Heart Disease (**CHD;** also called **congenital heart defects** or **congenital heart anomaly**) – A congenital heart disease is a defect in the structure of the heart or great vessels that is present at birth. There are many types of heart defects that may involve the interior walls of the heart, the heart valves, or the large blood vessels that lead to and from the heart. CHDs are present in about 75-80% of patients with 22q11.2DS . The percentage of CHDs in 22q11.2DupS is not completely well established, but recent studies reported it as about 20-33%.

Conotruncal Heart Defects – Conotruncal heart defects are a group of congenital anomalies (problems present from birth) involving the cardiac outflow tract, which is the path that allows blood to flow out of the heart to the rest of the body. Conotruncal heart defects that commonly affect babies with 22q11.2 deletion syndrome include **tetralogy of Fallot** (**ToF**), **pulmonary atresia with ventricular septal defect** (an extreme type of ToF), **truncus arteriosus** and **interrupted aortic arch**. [See "Tetralogy of Fallot", "Truncus Arteriosus" and "Interrupted Aortic Arch" in the Heart Series of the Health Conditions Explained page of our website.]

Constipation – Constipation means releasing feces less frequently than normal, or having feces that are hard and difficult to pass. This condition is common among individuals with 22q11.2DS. It is recommended that affected individuals adopt a healthy diet with lots of fruits and vegetables as well as good hydration to avoid constipation. Physical activity is also beneficial. It is also recommended that individuals with 22q differences check with their healthcare providers before using any stool softeners or **laxatives**, as each has its own side effects. [See "Constipation" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Continuous Positive Airway Pressure Therapy (**CPAP**) – CPAP involves applying low levels of air pressure through a mask at night to prevent the upper airway from collapsing, thereby allowing uninterrupted sleep. It is a standard treatment for **obstructive sleep apnea**.

Craniosynostosis – Craniosynostosis is a condition of premature skull fusion. When a baby is born, the skull contains some gaps (called **sutures**), which allow space for the brain to grow. The many sutures between the skull normally close around 10 months to 2 years of age. In a baby who has craniosynostosis, one or more sutures closes too early in the skull, and the brain does not have enough space to grow. The shape of the head may become abnormal with a raised edge, there may be no "soft spot" on the baby's skull, and the baby's head size may not increase normally over time. A possible serious consequence of craniosynostosis is an increased pressure inside the brain, which can lead to headaches, blindness, **seizures**, and even brain damage. Craniosynostosis has been reported in babies with 22q11.2 deletion syndrome. It is often treated with surgery to separate the sutures when severe.

Cricopharyngeus Muscle – [See **Inferior Pharyngeal Constrictor Muscle**]

Crohn's Disease – [See **Inflammatory Bowel Disease** (**IBD**)]

Cyclic Vomiting Syndrome (**CVS**) – Cyclic vomiting syndrome is a condition in which the individual throws up suddenly, severely, and repeatedly without any known reason usually in the early hours of the morning. It usually starts in children 3-7 years of age and is closely related to migraine. In between bouts of **vomiting**, individuals have no symptoms. [See "**Nausea** and Vomiting" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Cytopenia – Cytopenia means "low blood cell counts." Some individuals can have low blood counts, and this may affect any of the blood cells. Patients may have a mildly low platelet count, especially as they get older (**thrombocytopenia**), a low red blood cell count (anemia – which is most often due to iron deficiency, but can be because of break down of red blood cells [See **Hemolytic Anemia**]) or a low white blood cell count (leukopenia) that might be because of low neutrophil count (neutropenia – white blood cells that help fight bacterial infection, rarely low because of an autoimmune process) or low lymphocyte count (with low T cell counts; [See **T Cells**]).

D

Dehydration – Dehydration is a condition in which the body does not have enough fluids to perform its normal functions, and it happens when we lose more fluids than we take in. While dehydration is common in individuals both with and without 22q differences, some health conditions (such as **vomiting** or **gastroparesis**) make the individual more likely to be dehydrated. [See "**Nausea** and Vomiting" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Deletion – In genetics, deletion means a piece of DNA is missing. In individuals with 22q11.2 deletion syndrome, a segment of DNA from one of the two copies of chromosome 22 is missing. There are four main sizes of deletions for this syndrome: The A-D deletion is by far the most common, involving over 40 different genes, and happens in 85 to 90% of patients. The A-B deletion involves a smaller number of genes and accounts for 5 to 10% of patients. The B-D and C-D deletions are much smaller and rarer than the common deletion.

Delusion – A false belief that cannot be changed with reasoning, e.g., the person believes that they are being followed.

Dentistry – Dentistry is the healthcare field that deals with the teeth, the gum, and their supporting structures. A professional who specializes in dentistry is a dentist.

De Novo – In genetics, a de novo variant is a new one that was not passed on from the parents. Most individuals affected by 22q11.2DS have a de novo deletion – their parents do not have it. On the other hand, about 70% of patients with the 22q duplication inherit it from a parent.

Dental Caries – Dental caries, also known as tooth decay or cavities, are holes in the teeth. The damage usually happens when acid (produced by bacteria) erodes the enamel surface (the hard part that you see) of the teeth. [See "Let's Talk 22q Teeth – Info for Families" in the Dental Series on the Health Conditions Explained page of our website.]

Depression (**Major Depressive Disorder**) – A depressive episode is when a person feels sad, depressed, or uninterested, and has multiple other symptoms that persist for a period of time, and these changes affect how the person functions. This is a common but treatable condition.

Dermatology – Dermatology is a branch of medicine that deals with the skin, hair, and nails. A medical doctor who specializes in dermatology is a dermatologist.

Developmental Age – Developmental age is an assessment of someone's intellectual, social, cultural, physical, and emotional level compared to others in the same age range. A person's developmental age in a specific aspect may be younger or older than their chronological age. Determining a child's developmental age (e.g., through neurocognitive testing) may help parents and educators set realistic expectations.

Developmental Articulation Disorder – Children who have developmental articulation disorders may show substitutions, omissions, and distortions of the sounds of speech. These errors may persist longer than expected. [See "**Speech Sound Disorders**" in the Speech Series of the Health Conditions Explained page of our website for more information and examples.]

Developmental Coordination Disorder (**DCD**) – DCD is a neurodevelopmental condition that impacts the person's motor skills and coordination (their ability to convert their mental intentions into physical action accurately). It is common among children with 22q11.2DS. The affected individuals appear clumsy and tend to perform less well than their peers in sports and fine motor skills (e.g., writing or fastening buttons). There is no cure for DCD, but occupational therapy is often helpful in improving function.

Developmental Disabilities – Developmental disabilities are a group of conditions that affect a person's day-to-day functioning. These disabilities are a result of mental or physical problems which are present at birth, and last throughout the person's life. Individuals with 22q11.2 deletion or duplication syndromes have developmental disabilities, but they may vary widely among these individuals.

Diabetes Type 2 – Diabetes Type 2 is a complex long-term condition in which the body struggles to regulate and use glucose properly as a source of energy. As a result, the level of sugar in the blood is too high, which then leads to many possible problems that affect the heart and blood vessels, nerves, kidneys, eyes, skins, immune system etc. There are many possible risk factors, but in general, eating healthily and getting active are lifestyle choices that can prevent or slow down the progression of diabetes. Individuals with 22q11.2 deletion syndrome are more likely to develop type 2 diabetes compared to the general population and develop it at a younger age.

Digestive System (**Gastrointestinal Tract**) – The digestive system is a collection of organs in the abdomen that extract the nutrients and energy from the food we eat and removes the remaining waste. [See "The Digestive System (Gastrointestinal Tract)" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Down Syndrome – Down syndrome is a genetic condition that happens when a person has 3 (instead of 2) copies of chromosome 21. It happens in about 1 in 700 babies. Individuals who are affected have many possible physical and developmental problems, but each person is affected differently.

Duodenum – [See **Small Intestines**]

Duplication – In genetics, duplication means a piece of DNA is repeated, so that there is an extra piece of DNA. In individuals with 22q11.2 duplication syndrome, a segment of DNA from one of the two copies of chromosome 22 is repeated. In most cases, the duplication involves over 40 different genes.

Dysmenorrhea – (Females) Dysmenorrhea means having severe cramping pain during the menstrual periods, especially if missing school or work. This is a treatable condition with several treatment possibilities.

Dysarthria – Dysarthria is a condition in which the muscles that are needed for the function of speech systems (breathing, voice, resonation) are weak, paralyzed, or poorly-coordinated. It leads to slurred or slow speech that can be difficult to understand. [See "**Motor Speech Disorders**" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Dysphagia – Dysphagia means difficulty swallowing, and it is a problem that has been reported in children with 22q differences. The food and medications for the affected individuals may need to be processed for easy swallowing. It is important to talk to your doctor to find out the cause of the dysphagia so that the correct treatment can be prescribed. [See "Swallowing and Dysphagia" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Dysphonia – Dysphonia is a condition in which the voice sounds abnormal, such as soft voice, hoarseness, breathiness, tense, strained, and/or with abnormal pitch. [See "Voice Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Dystonia – Dystonia is a movement disorder that causes muscles to contract involuntarily, which leads to unintended repetitive or twisting movements. There are many types of dystonia and multiple possible causes. The current available treatments include medications and surgeries, but they only provide relief from symptoms and do not cure or slow down the progression of dystonia.

E

Echocardiogram (**an "echo"**) – An echocardiogram is a non-invasive method for studying the size, shape, structure, and function of the heart, the nearby blood vessels exiting the heart, and the blood flow through the heart valves. During the test, the technician ("sonographer") puts a hand-held tool ("transducer") with some cool gel on the patient's chest. The ultrasound (high frequency sound waves that humans cannot hear) from the tool generates pictures of the heart valves and chambers, so that the technician can see how well the heart is working.

Edema – Edema means swelling because of fluids that build up, most commonly in the legs and feet, but that can happen in any part of the body. It is important to check with a healthcare provider about this swelling, especially if there is shortness of breath, irregular heartbeat, or chest pain, as these may be signs that the lungs are filled with fluids.

Electroencephalogram (**EEG**) – Cells inside the brain communicate using little bursts of electricity, and EEG is a way to measure these electrical activities. During EEG, electrodes are attached to the person's head to detect the electricity. The information collected can be used to check for any brain injuries, tumors, and even the cause of **seizures**.

Enamel (**Dental**) – Enamel is the hard covering of the tooth. It protects the tooth when people chew, bite, crunch, and grind. It is what you see when you look at a tooth. [See "Let's Talk 22q Teeth – Info for Families" in the Dental Series on the Health Conditions Explained page of our website.]

Endocarditis – Endocarditis is the inflammation of the endocardium, which is the inner lining of the heart's chambers and valves. Some individuals with 22q11.2 deletion syndrome (such as those with specific congenital heart disease or those with prosthetic valve) may need **endocarditis prophylaxis**, which means taking antibiotics before certain dental (or other interventional) procedures to prevent bacteria from causing infections in the heart.

Endocrinology – Endocrinology is a branch of medicine that deals with the **hormones** that help the body function and grow. This is the specialty that manages diabetes. A medical doctor who specializes in endocrinology is an endocrinologist.

Endoscopy – Endoscopy is a procedure that uses a tiny camera (an **endoscope**) to look inside the body to check or treat organs or structures. Usually the tiny camera is attached to the end of a long thin tube, and the endoscopist inserts this tube into the body. Different types of endoscopies can check different parts of the body. For example, in upper endoscopy, a tube is inserted through the mouth to look at the inside of the digestive tract (food pipe, **stomach**, and beginning of the **small intestine**). [See **Digestive System**]

Epiglottis – Epiglottis is a small, thin, and movable piece of cartilage that covers the windpipe. It prevents food and drink from going into the windpipe.

Epilepsy – Epilepsy is a brain disorder in which the person had at least two **seizures**. The person may also show unusual behavior or an experience loss of sensation and awareness.

Epistaxis – Epistaxis means nosebleeding, which is a common condition even among the general public. The typical cause is dry air, but other causes include infections, allergies, chemical irritants, certain medications, and structural abnormalities of the nose etc. The recommended way to stop a nosebleed is to sit upright, lean forward, breath through the mouth, and pinch the soft part of the nose continuously for at least 5 minutes (use a timer). If nosebleeding happens often, please check with the doctor. [See also: **Hemostasis**]

Esophageal Dysmotility (**Esophageal Motility Disorders**) – Usually, the muscles in the **esophagus** (the pipe that connects the throat to the **stomach**) tighten and relax to bring liquid and food into the stomach, and this is called esophageal motility. Esophageal dysmotility is a group of swallowing disorders (including **achalasia**) in which the esophagus does not work properly, so that liquid and food get stuck in the pipe without arriving at the stomach. There are a variety of causes and symptoms. [See "Esophageal Dysmotility" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Esophagus – The esophagus is the muscle tube that brings food from the throat (**pharynx**) to the **stomach**. [Note: The British spelling of "esophagus" is "oesophagus".]

Eustachian Tube – The Eustachian tube is the channel that connects the middle ear to the back of the throat. It allows air pressure in the ears to be maintained and fluids in the ear to be drained. **Eustachian tube dysfunction** (**ETD**) happens if the tube is blocked. This leads to the collection of fluids in the ear, popping noises in the ear, a plugged feeling, decreased hearing, and pain. ETD can happen with a cold, sinus infections, or allergies. Children are at higher risk of ETD because their Eustachian tubes are very narrow. Multiple treatments are available. Medications may be used to manage allergies or reduce inflammation, and surgeries can be performed to insert a tube to equalize the pressure in the ears. [See also Eustachian Tube Dysfunction from the website of the Stanford Ear Institute]

Executive Functions – Executive functions are the mental skills that allow us to respond to situations appropriately, complete tasks successfully, plan, and make good judgments. These include starting a task, switching between tasks, planning for the next step, stopping one's automatic reactions, and retaining information. Individuals who have difficulties with executive functions may need step-by-step instructions and help with decision-making.

Exophoria – Exophoria is a condition in which one or both eyes turn can be induced to turn outward. [Contrast with **Intermittent Exotropia** and **Exotropia**.] **Ophthalmologists** and **optometrists** can use various techniques to test how a child's eyes are aligned. They can then recommend lenses, exercise, or surgeries to correct the problem. [See this video excerpt from orthobook.com for illustrations of the eye misalignment, tests for diagnosis, and treatment options.]

Exotropia – Exotropia is a condition in which one or both eyes turn outward, and this misalignment is constantly present. [Contrast with **Exophoria** and **Intermittent Exotropia**.] **Ophthalmologists** and **optometrists** can use various techniques to test how the child's eyes align. They can then recommend lenses or surgeries to correct the problem. [See this video excerpt from orthobook.com for illustrations of the eye misalignment, tests for diagnosis, and treatment options.]

Exploitation – Exploitation is the process in which someone takes advantage of others. For example, financial exploitation happens when someone uses illegal or unethical means to gain control of the victim's money or other assets. Sexual exploitation happens when someone takes advantage of the victim's vulnerability to obtain sexual favors. For an interaction to be designated sexual exploitation, there needs to be a power difference between the perpetrator and the victim (an example would be where the victim genuinely depends on the perpetrator for housing, money, or even survival). Some individuals with 22q11.2DS have decreased abilities for safe and appropriate social judgment and decision making, which can increase risk of both financial and sexual exploitation.

F

Fatigue – Fatigue is an overall state of being tired or lacking energy. It is common in both children and adult with 22q11.2 deletion syndrome. Although it can be a direct result of the body dealing with known health issues, fatigue can be an early sign of additional problems (such as having low calcium levels or a mental health problem).

Fatty Liver – A fatty liver is one that has too much fat built up. The excessive fat can lead to liver inflammation and damage the liver. Fatty liver can be seen in patients who are overweight or obese. [See "Nonalcoholic Fatty Liver Disease" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Feeding Difficulties – Feeding difficulties happen in newborn babies with 22q11.2 deletion syndrome who fail to consume enough breastmilk or formula to receive the necessary nutrients. The difficulties can have many causes, such as palate issues, swallowing disorders, and gastrointestinal problems. The condition can even be caused when malformed blood vessels compress the airway and the esophagus. Therefore, babies with 22q11.2 deletion syndrome who have feeding difficulties should be assessed by a pediatrician who is familiar with this genetic condition and specializes in feeding.

Flow Cytometry – Flow cytometry is a method of counting cells based on specific characteristics, such as whether the cells are expressing a certain protein on their surface. It is a useful method to check the cell distribution and numbers in individuals who have **immunodeficiencies**.

Focal Cortical Dysplasia – Focal cortical dysplasia is a condition in which the organization of the layers of the brain and the appearance of neurons (nerve cells) are abnormal. Individuals with this condition have it since they are born, and there may be an association between **seizures** and focal cortical dysplasia. Doctors can use imaging techniques to check for this brain abnormality and correct it using surgery.

Foramen Ovale – The foramen ovale is a special hole in the wall between the left and right atria of every human fetus [See **atrium**]. This hole allows blood to bypass the fetal lungs, which cannot work until they are exposed to air. When a newborn takes its first breath, the foramen ovale closes start to close, and within a few months it is sealed completely in about 75% of people. If it remains open, it is called a **patent foramen ovale** (**PFO**; patent means open). For the vast majority of people with a PFO, it is not a problem, and treatment is required only if there is a risk of stroke.

Functional Voice Disorder – Functional voice disorder happens when a voice disorder occurs for no identifiable reason. [See "Voice Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Fundoplication (**Nissen Fundoplication**) – Fundoplication is a surgery that strengthens the **lower esophageal sphincter** to reduce **stomach** acid reflux. It is only performed in severe cases when medications and lifestyle changes cannot control the problem. [See "**Gastroesophageal Reflux Disease** (**GERD**)" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

G

Gallbladder – The gallbladder is a small organ that sits on the right side of the abdomen and is part of the digestive system. It stores and releases **bile**, which is a digestive fluid (made by the liver) that breaks down fat. [See "Gallstones" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Gallstones (also called **Cholelithiasis**) – Gallstones are like little pebbles of bile in your gallbladder. **Bile** is a digestive fluid that is stored in the **gallbladder** and released into the small intestine that helps to break down fat. If the gallstones block the gallbladder outlet and cause pain (**biliary colic**), a surgery is done to remove the gallbladder containing the stones. If the stones block the common bile duct, there is another procedure (called ERCP) to remove the stones. [See "Cholelithiasis (Gallstones)" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Gastric Tube (**G-Tube**) – A gastric tube is a feeding tube that is inserted directly into the **stomach** or intestines to deliver nutrients there. This feeding method is used when there are severe problems in the mouth or esophagus that prevent the individual from eating the regular way. [See "**Feeding Difficulties**" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Gastroenterology – Gastroenterology is the branch of medicine that deals with the **digestive system**, which includes the gastrointestinal tract (or GI tract) that consists of the **esophagus**, **stomach**, **small intestine**, colon, and rectum, as well as additional organs like the pancreas, **gallbladder**, and **liver**. A medical doctor who specializes in gastroenterology is called a gastroenterologist.

Gastroesophageal Reflux Disease (**GERD**) – Acid in the **stomach** is an important tool to digest food. Gastroesophageal reflux (also called acid reflux) happens when the **lower esophageal sphincter** is weak or opens at the wrong time, allowing stomach acid to flow back up into the **esophagus**. Stomach acid can irritate the lining of the esophagus, causing a burning feeling in the chest ("**heartburn**"). Gastroesophageal reflux is a common problem in the general population, whether they have 22q differences or not. Treatment is recommended when children have pain, irritability, excessive vomiting/spitting up that leads to poor weight gain, food refusal, swallowing problems, spiting up blood, recurrent choking or pneumonias, and interrupted breathing or apnea. [See "Gastroesophageal Reflux Disease (GERD)" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Gastrointestinal Tract – [See **Digestive System**]

Gastroparesis (**Gastric Stasis**) – Gastroparesis is a condition in which food takes too long to be emptied out of the **stomach** into the intestines, even when there are no blockages. The delay in stomach emptying is due to damaged nerves and/or muscles. The affected individuals may have low appetite, bloating, reflux, nausea, and pain, and they may not be able to eat enough to meet their nutritional needs. Gastroparesis is a condition that cannot be cured but can be managed. [See "Gastroparesis" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Gastroscopy – [See **Endoscopy**]

Generalized Anxiety Disorder – Generalized anxiety disorder is a treatable condition where there are worries about many things that are hard to control and come with multiple other symptoms, e.g., muscle tightness, sleep difficulties, trouble concentrating, and the symptoms are present most days for over 6 months.

Genetic – A genetic condition is one that is determined by a particular sequence of DNA. Genetic conditions are not always inherited and can arise for the first time when a baby is conceived. For example, most 22q11.2 deletions and some duplications occur "out of the blue" without being inherited from either parent. However, once a person is born with the deletion or duplication, he/she can pass it onto future generations.

Genetics – Genetics is a branch of biology that is concerned with the study of hereditary units called **genes**. It studies how traits are passed through generations, and how variations in genes lead to different health conditions and physical features.

Genetic Counseling – Genetic counseling is a process in which trained professionals (usually genetic counselors) advise individuals about genetic conditions that affect them and their families. The counselors can help individuals understand the conditions that affect them and the risk of passing the conditions to future generations. For individuals who have family histories or are at risk of developing certain conditions, the counselors can help them assess the risks and make plans.

Genitourinary Abnormalities – Genitourinary abnormalities are those affecting the genital tract (reproductive organs) and/or the urinary tract (kidneys, ureters, and bladder). This group of problems affect about 15% of individuals with 22q11.2 deletion syndrome, but sometimes the diagnosis may be delayed until adulthood.

Germline Mosaicism (also called **Gonadal Mosaicism**) – Germline mosaicism is a condition in which some of a person's eggs or sperms have a genetic change that is not present in the cells (e.g., blood cells) in the rest of the body. If an affected sperm or egg goes on to form a baby, the baby will have the genetic change. Germline mosaicism is a very rare finding in families where there is more than one child with 22q11.2DS (or 22q11.2DupS) but the parents' blood tests show no 22q deletion (or duplication). This is one of the reasons that parents with children with 22q11.2DS or 22q11.2DupS are recommended to receive genetic counselling. [See Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions]

Glottic Web – A glottic web is a membrane that forms between the vocal cords. It narrows the windpipe and makes breathing more difficult and noisier. [See **Stridor**] This web is often present from birth, but long-term intubation can also cause it to form. A glottic web may be misdiagnosed as asthma, and imaging techniques will reveal the obstruction. Surgery is needed to remove the web and open the airway.

Graves' Disease – Graves' disease is an autoimmune disorder that leads to the overproduction of thyroid hormone. [See **Hyperthyroidism**] Because thyroid hormone normally regulates the use of energy in many organs, this disorder affects large parts of the body. There are many possible symptoms, including bulging eyes, enlargement of the thyroid gland, unexplained weight loss, irregular heart rhythm, etc. The cause of the disorder is not understood, but the body's immune system somehow produces antibodies that target a type of cells in the thyroid gland and override the normal regulation process. To diagnose Graves' disease, doctors may perform a blood test to check for thyroid hormone and/or use imaging strategies to observe the size and function of the thyroid. There are several strategies to treat Graves' disease, including the selective killing of the problematic thyroid cells, the reduction of the effects of thyroid hormone on the rest of the body, the decrease of the swelling around the eyes, and the surgery to remove the thyroid.

Growth Hormone Deficiency – The human body produces factors called growth hormones to direct normal growth and development. Growth hormone deficiency is a condition in which there are not enough growth hormones, leading to reduced growth and short stature. This condition can be diagnosed by an **endocrinologist** and a growth hormone stimulation test.

Gynecology – Gynecology is the branch of medicine that deals with the female reproductive system, which includes the uterus (womb), fallopian tubes, ovaries, vagina, vulva, breasts, and all aspects of female sexual health, including preventive care, cancer screening, and physical exam. Very often, gynecology is paired with **obstetrics**, which deals with childbirth. A common short form for both is OB/GYN. A medical doctor who specializes in gynecology is called a gynecologist. [Note: The British spelling of gynecology is gynaecology]

H

Hallucinations – A mistaken perception, e.g., the person hears noises or voices but there is no sound coming into the ear.

Hammer Toe – Hammer toe is when the second, third, or fourth toe bends (from the middle joint) downward permanently, making it look like a hammer. Changing footwear or using special devices can help with the problem, but surgeries may be needed in severe cases.

Hearing Loss – Hearing loss is the decreased ability to perceive sound. Often the hearing loss is in the higher frequency range (trouble hearing higher pitched sounds). **Conductive hearing loss** happens when sound cannot get through the outer and/or middle ear through to the inner ear and nerves. **Sensorineural hearing loss** happens when the inner ear is damaged, or when the nerves cannot deliver the sound to the brain. Individuals with 22q11.2 deletion can have either conductive or sensorineural hearing loss or both.

Heartburn – [See **Gastroesophageal Reflux Disease** (**GERD**)]

Hematology – Hematology is the branch of medicine that deals with blood and blood-forming organ such as the bone marrow. A doctor who specializes in hematology is a hematologist.

Hemifacial Paresis (**Asymmetric Facies**) Asymmetric facies in 22q11.2 deletion syndrome means that one side of a person's face looks a little different or doesn't move the same way as the other side. This can happen because of differences in how the muscles and nerves in the face developed. For example, one side of the mouth might not move as much when smiling, or one eye might appear slightly lower than the other. It's a common feature in people with 22q11.2 deletion syndrome, but it doesn't usually cause serious problems.

Hemolytic Anemia – Hemolytic anemia happens when red blood cells are destroyed either because of an autoimmune disorder or because of a problem within the red blood cells themselves, so that the body does not have enough working red blood cells. Since red blood cells are responsible for bringing oxygen to all parts of the body, having too few of them means the body cannot work well. Some types of hemolytic anemia are short term (such as an autoantibody), but others last a lifetime (like sickle cell disease). Hemolytic anemia can be inherited, as in the cases of sickle cell anemia and thalassemia. However, the condition can also happen because of cancers, heart valve problems, medication side effects, or, in some individuals with 22q11.2DS, **autoimmune disorders**. The symptoms of hemolytic anemia include an abnormally pale color on the skin, yellowish eyes, dark urine, dizziness, weakness, increased heart rate, etc. To diagnose hemolytic anemia, doctors will likely check the red blood cell count, the urine, and rarely, even the bone marrow. Strategies in treating the condition will depend on the cause.

Hemostasis – Hemostasis is the normal process of forming a blood clot, which stops blood from leaking out of a damaged blood vessel. Usually, platelets first gather around the injury to form a temporary plug to stop the bleeding. Then, coagulation factors in the blood arrive to form a more stable clot. Some individuals with 22q11.2DS have impaired hemostasis, which means they have trouble stopping bleeding. They tend to have nosebleeds (**Epistaxis**), heavy menstrual bleeding , and bleeding after surgeries.

Hernia – A hernia happens when a part of an organ (e.g., the small intestine) pushes through a weak area of the muscles and connective tissues that normally hold the organ in place. For hernias that push outward, a lump is usually visible on the surface of the body. Hernias may or may not lead to pain or other symptoms but can lead to life-threatening complications if not repaired by surgery. Hernias that are common in 22q11.2DS include:

- Diaphragmatic – an organ in the abdomen pushes up through the diaphragm (the muscle that separates the chest from the abdomen)
- Umbilical – an organ in the abdomen pushes outward near the belly button.
- Inguinal – an organ in the abdomen pushes outward near the groin

See “Inguinal Hernias” in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.

Heterotopia - A brain heterotopia is when some brain cells end up in the wrong spot in the brain. Normally, brain cells have a specific place they need to be to help the brain work properly. But in a brain heterotopia, some of those cells don't move to the right place while the brain is forming. One example is periventricular ("around the ventricles") nodular ("clumps") heterotropia ("out of place"). Heterotopia can sometimes cause problems like **seizures** or trouble with learning because the brain isn't organized the way it should be.

Hippocampal Malrotation (**HIMAL**; or **Incomplete Hippocampal Inversion, IHI**) – A human brain has two hippocampi (singular = hippocampus), one on the left and one on the right, and they are important in learning and memory. HIMAL is a condition in which the hippocampus failed to fold normally during fetal development, resulting in an abnormal round shape. **Seizures** in some individuals may be related to HIMAL, but the relationship is still unclear.

Hormones – Hormones are little messenger molecules that are produced in one part of the body and travel to another part to control certain processes. For example, the thyroid produces the hormone thyroxine to control the body's temperature, heart rate, and growth, while the pancreas produces insulin to control blood sugar levels.

Hypernasal Speech – [See **Velopharyngeal Dysfunction**]

Hyperopia (**Farsightedness**) – Hyperopia is a condition in which a person can see distant objects more clearly than near ones. It is a type of **refractive error** which can often be compensated for by accommodation or thickening of the lens inside the eye. It occurs because of the shape or size of the eye rather than just an increase in age. Usually the problem can be corrected using prescription eyeglasses.

Hypertension (**High Blood Pressure**) – Hypertension happens when the blood pushes against the walls of the blood vessels with a force that is always too high, and the heart must work harder to pump blood. Uncorrected high blood pressure can lead to damage on multiple organs in the body as well as other complications. Increased blood pressure values are not always accompanied by the appearance of symptoms. In any case, the symptoms associated with high blood pressure are not specific, which is why they are often underestimated. The most common symptoms can include: headache, lightheadedness and dizziness, ringing in the ears, vision changes. Therefore, it is recommended that adults with 22q11.2DS get cardiac checkups periodically.

Hyperthyroidism – Hyperthyroidism means having an overactive **thyroid gland** that produces too much of the hormone **thyroxine**. Symptoms include unintended weight loss as well as an irregular or very fast heart rate. This condition affects a minority of children and adults with 22q11.2 deletion syndrome. Usually, the doctor would use medication to regulate hormone production in the thyroid. However, in some cases, the thyroid gland may need to be removed by surgery or treated with radioactive iodine therapy.

Hypocalcemia – The human body typically maintains a steady level of **calcium** in the blood. Hypocalcemia, a common condition in individuals with 22q11.2 deletion syndrome, happens when the level of the calcium in the blood is too low. This can be due to vitamin D deficiency or when the parathyroid glands are not working [See **Hypoparathyroidism**]. Symptoms include muscle spasms, tingling in fingers and toes, cramps, numbness around the mouth and **seizures**. Hypocalcemia is usually treated with calcium supplements, vitamin D supplements, and medications that adjust hormone levels. [Note: The British spelling of "hypocalcemia" is "hypocalcaemia".]

Hypoparathyroidism – Hypoparathyroidism happens when the **parathyroid gland** produces too little **parathyroid hormone (PTH)**, whose job is to regulate the levels of calcium and phosphorus in the body. When there is too little PTH, the calcium level becomes too low, and the patient becomes affected by **hypocalcemia**. Individuals who have hypoparathyroidism may need to take calcium supplements for life. Some of them may need a replacement of PTH as well. Hypoparathyroidism is very common in children with 22q11.2 deletion syndrome.

Hypothyroidism – Hypothyroidism means having an underactive **thyroid gland** that cannot produce enough of certain important hormones. It affects a minority of children and adults with 22q11.2 deletion syndrome. Because the symptoms, such as fatigue and weight gain, may not be obvious, a blood test may be needed to assess thyroid function. The condition can be treated with thyroid hormones.

Hypotonia – Hypotonia means low or decreased muscle tone, or the muscles are "floppy". When a baby with hypotonia is carried, he/she may feel like a rag doll. Hypotonia may be without consequence when it is mild but can have more serious effects when more severe. Among children with 22q11.2 deletion syndrome, hypotonia can be more severe and can affect the throat (the **pharynx**), contributing to a floppy airway and **obstructive sleep apnea**. It can also cause problems in feeding and swallowing.

I

Iatrogenic – An iatrogenic condition is one that is caused by medical activities. For example, iatrogenic **hypercalcemia** (having a blood calcium level that is too high) can be caused by medical treatment that overcorrects **hypocalcemia** (having a blood calcium level that is too low).

Idiopathic – A condition that is idiopathic arose spontaneously or without known causes.

Ileum – [See **Small Intestines**]

Immune – If a person is immune to something, he/she is protected from it.

Immunization – Immunization means receiving a **vaccine** to teach the body's immune system to recognize invading viruses/bacteria and make antibodies against them. Once immunized, if the body encounters the same invaders in the future, the immune system can get rid of them quickly, and the person will be less likely to get severely sick. It is very important to get accurate, evidence-based information about vaccines from health authorities. Here is the Vaccination and Immunization page from the World Health Organization.

Immunodeficiencies – Our immune system normally fights off things that are foreign (e.g., viruses, bacteria, fungi, etc.). Immunodeficiencies (or immune deficiencies) happen when the immune system is weakened. The patient may have too many infections, infections that are difficult to cure [see **Chronic Infection**], unusually severe infections, or infections with unusual organisms. Many individuals with 22q11.2 deletion syndrome are immunodeficient. The condition has also been reported in those with 22q11.2 duplication syndrome.

Immunoglobulins – [See **Antibodies**]

Immunology – Immunology is the branch of medicine and biology that deals with an organism's reaction to antigenic challenge and the way the organism figures out what belongs to its own body versus what does not. A doctor or scientist that specializes in immunology is called an immunologist.

Imperforate Anus (**Anorectal Malformation**) – The anus is a ring of muscles that lets a person holds stool (poop) inside the rectum and then release it during a bowel movement (BM). An imperforate anus is a birth defect in which the person is either missing the anus or does not have a hole in it. Depending on the structure of the rest of the system, the stool may cause painful BM, exit at the same place as the urine, or even enter other areas of the abdomen and get trapped inside the body. Typically, when a baby is born, they have a BM within 48 hours, and doctors can notice malformations early on. Babies with an imperforate anus will very likely need reconstructive surgeries to build a proper opening for the stool to pass.

Individualized Education Plan (**IEP**) – An IEP is a written plan that includes realistic goals designed to allow children with disabilities to access a Free and Appropriate Public Education (FAPE). Crucial to this process, an IEP must include 1) specific support services that will enable the student with special needs to learn at a suitable pace, and 2) clearly defined 'progress monitoring' to periodically assess whether the student is in fact making suitable progress. Almost all children with 22q differences, learning difficulties, and/or intellectual disabilities benefit from an IEP if they are struggling in school. For more information about the referral and assessment as well as the development of an IEP, please check with your child's school district, or with the Department of Education for your state.

Inferior Pharyngeal Constrictor (**Cricopharyngeus Muscle**) – The inferior pharyngeal constrictor is a muscle at the top of the esophagus. When it relaxes, food and drinks can go from the throat into the esophagus. [See "Swallowing and **Dysphagia**" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Inflammatory Bowel Disease (**IBD**) – Inflammatory bowel disease is a group of **autoimmune disorders** that involve long-term damage of the digestive tract. The two main types are **ulcerative colitis** (ulcers in the colon including rectum) and **Crohn's disease** (inflammation in any part of the digestive tract). Inflammatory bowel disease (IBD) is not the same as irritable bowel syndrome (IBS). [See "Autoimmune Issues in the GI System" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Intellectual Disabilities (**ID**) – Individuals with ID have significant limitations in (1) intellectual functioning (issues in learning, reasoning, and problem solving), and (2) adaptive behavior (language and math concepts, social skills, and daily living skills), originating before age 22. Many individuals with 22q11.2DS have mild ID. Severe ID in 22q11.2DS is rare. It is recommended that individuals with 22q11.2DS be assessed for both intellectual functioning and adaptive behavior every few years so that support can be provided if necessary.

Intermittent Exotropia – One or both eyes turn outward, and this misalignment is intermittent and can occur without being induced as in the case of an **exophoria**.

Intestinal Malrotation – Intestinal malrotation means that the **small intestines** did not turn the correct way while the baby was still in the womb. The malrotated intestines twist in ways that may block off their own blood supply and prevent food from moving along in the digestive tract. Intestinal malrotation may be one of the many possible causes of feeding difficulties. [See "Feeding Difficulties" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

J

Jejunum – [See **Small Intestines**]

Juvenile Rheumatoid Arthritis (**JRA**, or **Juvenile Idiopathic Arthritis**, **JIA**) – JRA is a form of arthritis in children. It causes joints to be inflamed and stiff, and any joint can be affected. In severe cases, JRA can limit the movement of the joints. JIA is thought to occur in 2% of children with 22q11.2 deletion syndrome.

K

L

Large Intestines (**Large Bowel**) – The large intestines form the last section of the digestive tract, and have four main parts: cecum, colon, rectum, and anus canal. Partially digested food moves from the **small intestines** into a little pouch called the cecum before entering the colon, where water, nutrients, and electrolytes are absorbed. The remaining waste go through the rectum and exits through the anus as feces (also called stool or "poop").

Laryngeal Web – A laryngeal web is a congenital (born with it) deformity and is a membrane-like tissue in the **larynx** (voice box) which partially blocks the space between the two **vocal cords**, making it hard to breathe and changing the quality of the voice. [See "Voice Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Laryngomalacia – Laryngomalacia is a condition in newborns in which the tissues of the **larynx** (voice box) above the baby's **vocal cords** are soft and "floppy". It is a common cause for noisy breathing (**stridor**). To diagnose laryngomalacia, doctors use a tiny camera (and potentially other imaging techniques) to check the affected area. Laryngomalacia usually goes away on its own within one year. In severe cases, doctors may recommend anti-reflux medications or surgery to correct the problem.

Larynx (**Voice Box**) – The larynx sits at the front part of the throat. It contains the vocal cords, which vibrate to create the voice. [See also: **Vocal Cords**]

Laxative – Laxatives are medications that help with bowel movements, i.e., they relieve **constipation**. There are various types of laxatives, each with its own way of helping the body release the feces from the bowels, but each has its own side effects. The first line of management for constipation is usually an increase in the intake of liquids and fiber-rich food and the adoption of an active lifestyle. Even though laxatives are usually available without prescriptions, it is recommended that individuals with 22q differences discuss their situation with their healthcare provider before using them. [See "Constipation" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Liver – The liver is a multi-function organ located in the top right area of the abdomen. It makes many chemicals that the body needs – e.g., it makes bile, which is needed in the **small intestines** to digest food, and it makes components needed to form blood clots. It also clears away toxins from the blood and helps maintain normal blood glucose levels.

Lower Esophageal Sphincter – The lower esophageal sphincter is located at the junction of the esophagus and the **stomach**. It opens to allow food to enter the stomach and shuts to prevent stomach acid and partially digested food from going up to the esophagus. [See "**Gastroesophageal Reflux Disease (GERD)**" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Lymphedema – Usually the body's lymphatic system is made up from vessels like blood vessels, but they move lymph, also known as serum or plasma around the body. This lymph is rich in cells and proteins of the immune system. If the system is blocked, the fluids collect and leads to lymphedema, which is the swelling of tissues. This condition usually happens on an arm or a leg, but can also happens in the chest, abdomen, neck, and the genital area. Lymphedema can make it hard to move the arm or leg and can lead to frequent infections. Multiple treatment options (stockings, bandages etc.) are available, mostly aiming to compress the arm or leg to release the fluids. The causes are many and varied.

M

Malocclusion – Malocclusion is a condition in which the upper and lower teeth do not align properly when the mouth is closed. It happens when teeth are crooked or too large, leading to crowding in the mouth; or sometimes when teeth are missing or very narrow, leading to too much space between the teeth. Misaligned teeth can affect the chewing of food, gum health, and even social and mental wellbeing. Usually, malocclusion can be corrected using dental braces.

Manic Depression – [See **Bipolar Disorder**]

Meningomyelocele (MM; or **myelomeningocele**) – [See **Spina Bifida**]

Mental Health – Mental health includes our emotional, psychological, social, and behavioral well-being – how we think, feel, act, handle stress, relate to others, and make decisions. Currently, "mental health" is used an umbrella term often used to mean mental or **psychiatric illness**. [See Mental Health Series on the Health Conditions Explained page of our website.]

Mental Health Disorders – [See **Psychiatric Illnesses**]

Microcephaly – Microcephaly means having a head that is smaller than other children of the same age and sex. There are several possible causes, including genetic conditions, complications during pregnancy/delivery, and **craniosynostosis**. Some children with microcephaly can still reach their milestones, but others have developmental delays and additional complications.

Motor Speech Disorders – Motor speech disorders include impairments in planning, sequencing, reproducing speech, and/or moving the structures needed to form speech. Types of motor speech disorders include dysarthria and apraxia. Children with **dysarthria** may exhibit weakness, paralysis, or incoordination of the musculature responsible for respiration, phonation, articulation, and resonation. **Apraxia** of speech includes difficulties in planning and executing oral speech movements. Some features of apraxia include groping, searching, or effortful volitional oral movements, difficulty sequencing or transitioning from one sound to another, difficulty imitating speech, inconsistency in articulation with repeated productions of the same utterance, errors in vowels as well as consonants, and abnormalities of prosody, including rate, rhythm, intonation, and stress patterns. Motor speech disorders are often seen in 22q with prevalence estimates from 32-86%.

Motor Tics – Motor tics are sudden movements that a person can't control. It's like when your body does something on its own, like a quick blink, a shrug of the shoulders, or a twitch. These movements happen over and over again, and the person usually can't stop them even if they want to. Motor tics can come and go, and sometimes they get better or worse. They're not usually dangerous, but they can be annoying or distracting.

Multimorbidity – Multimorbidity means having 2 or more long term health issues that need treatment. It is a common situation for individuals with 22q deletion or duplication syndrome.

Myelomeningocele (MM; or **meningomyelocele**) – [See **Spina Bifida**]

Myoclonus – Myoclonus are uncontrolled, sudden, startle-like movements that occur in one part of the body or all over the body. Some types of myoclonus happen in healthy people and do not cause concern – these include hiccups and jerking that happen just before falling asleep. However, problems with the brain and nerves can lead to myoclonus, and these phenomena have been reported in adults with 22q11.2DS. Treatment or the strategy for symptom control for myoclonus depends on the underlying cause.

N

Nasopharyngeal Reflux – Nasopharyngeal reflux is the abnormal movement of swallowed food from the mouth or food pipe up into the throat and nose areas. It is a common problem in children with 22q11.2DS and happens because the defective soft palate cannot close the area between the mouth and the nose. Problems with muscles, nerves, breathing, and hormone levels are possible contributing factors, but the exact medical cause may or may not be known. The condition tends to improve over time, but a small number of patients may need an esophagus surgery to prevent the reflux. [See "Swallowing and **Dysphagia**" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Nausea – Nausea is the uncomfortable feeling that you need to throw up. There are many causes, ranging from harmless smells to serious brain injury. Some children with 22q11.2DS experience severe nausea and **vomiting**. [See "Nausea and Vomiting" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Nephrology – Nephrology is a branch of medicine that deals with the kidneys. A medical doctor who specializes in nephrology is called a nephrologist.

Neurology – Neurology is the branch of medicine that deals with the brain and the nerves. A doctor that specializes in neurology is a neurologist.

Neuropsychiatric Disorders – Neuropsychiatric disorders are a broad group of symptoms that may be associated with a medical condition such as 22q11.2DS. It is important to ensure that these symptoms did not result from medical causes such as thyroid dysfunction or low vitamin levels. Neuropsychiatric disorders that are often seen in adults with 22q11.2 deletion syndrome include (but are not limited to) **schizophrenia**, **anxiety disorders**, and **depression**. Some children with the syndrome have **attention deficit hyperactivity disorder**, **mood disorders**, and **autism spectrum disorder**. Similar neuropsychiatric disorders are also seen in individuals with 22q11.2 duplication syndrome.

Neuropsychological Testing – Neuropsychological assessment is designed to measure how well the person's brain is functioning. The specific tests used assess many aspects, including the person's ability to think, remember, learn, perceive, problem-solve, and coordinate different parts of the body, etc. Neuropsychological testing is much more extensive than the routine psychoeducational evaluations offered through school districts. It is strongly recommended for all children with 22q differences, and should be repeated periodically. Early assessment and interventions have been medically proven to be beneficial to young children with 22q differences. Repeated evaluations are critical for monitoring progress, and for determining whether students are continuing to make progress as academic demands escalate in higher grades.

Nissen Fundoplication – [See **Fundoplication**]

Nonalcoholic Fatty Liver Disease (**NAFLD**) – [See **Fatty Liver**]

Nonverbal Learning Disability (**NLD**) – Nonverbal learning disability refers to a pattern of test results and behaviors found among up to 70% of children with 22q11.2 syndromes. Individuals with NLD display higher verbal than nonverbal IQ scores, difficulty learning math skills, fine motor delays such as poor writing and drawing, attention problems, and deficient social skills.

O

Obligatory Articulation Disorders (**Passive Articulation Errors**) – Children with an obligatory articulation disorder put their speech structures in the right place when they talk, but their speech is distorted because of structural problems such as **cleft palate** and **velopharyngeal dysfunction** (**VPD**). Obligatory errors may include weak pressure on consonant sounds, nasal emission of air, nasal sounding speech sounds (such as /m/ or /n/ for /p/ and /b/: example: "maymee"/baby). This type of speech disorder is common in 22q11.2DS and occurs as a result of cleft palate or velopharyngeal dysfunction. This type of speech disorder is common in 22q11.2DS. [See "**Speech Sound Disorders**" in the Speech Series of the Health Conditions Explained page of our website for more information and examples.]

Obsessive Compulsive Disorder (**OCD**) – OCD is an illness where there are unwanted and distressing thoughts and fears (obsessions) the person had over and over that may lead the person to feel an uncontrollable urge to repeat certain behaviors (compulsions) over and over, and these symptoms affect functioning. Attempts to ignore these may make them worse.

Obstetrics – Obstetrics is the branch of medicine that deals with pregnancy, childbirth, and the post-partum (after birth) period. Very often, obstetrics is paired with **gynecology**, which deals with a woman's reproductive health, and a common short form for the two fields is OB/GYN. A medical doctor who specializes in obstetrics is called an obstetrician.

Obstructive Sleep Apnea (**OSA**) – OSA occurs when the upper airway becomes blocked, or obstructed, during sleep. It may be caused by enlarged tonsils and adenoids, by relaxation of the muscles of the tongue and throat during sleep, by obesity, or by surgery, such as posterior pharyngeal flap surgery. Persons with OSA wake up multiple times throughout the night to restore normal breathing, leading to very fragmented sleep. Untreated, longstanding OSA can bring about multiple other health problems such as high blood pressure and heart disease. The treatment of OSA depends upon its cause, and may include tonsillectomy and adenoidectomy or **continuous positive airway pressure** (**CPAP**).

Oncology – Oncology is the branch of medicine that deals with cancers, which are cell masses that are growing out of control. A medical doctor who specializes in oncology is called an oncologist.

Optician, **Optometrist**, and **Ophthalmologist** – These are all eyecare professionals, but their level of training and their specific roles are different.

Opticians are eyecare professionals who fit people with glasses (or contact lenses). They do not prescribe them, and they do not perform eye exams. If you go to an optician to get your eyeglasses, please bring your prescription.

Optometrists ("eye doctors") check the health of your eyes, such as your retinal health and the pressure inside your eyes. They will also examine how well you can see (ability to see far away and up close, ability to see colors, etc.) and can prescribe corrective eyeglasses if needed.

Ophthalmologists are medical doctors who specialize in the health and diseases (e.g., cataracts, glaucoma, **strabismus**) of the eye. Their medical field is called ophthalmology. Typically, a person will only need to see an ophthalmologist if an optometrist has found a concern during a regular eye exam. Ophthalmologists can perform eye exams and prescribe glasses/contacts, but they can also perform surgeries and many other procedures on the eyes.

Orthopedics (or **Orthopedic Surgery**) – Orthopedics is the branch of medicine that deals with bones, joints, ligaments, tendons, and muscles. A medical doctor that specializes in orthopedics is called an orthopedic surgeon. [Note: The British spelling of "orthopedics" is "orthopaedics".]

Otitis Media with Effusion, Chronic – Otitis media with effusion is when liquid gathers in the space in the middle ear. Symptoms may or may not include a temporary decrease in hearing, and the liquid drains on its own in a few weeks. This condition can happen repeatedly and is then called **recurrent otitis media**. In **chronic otitis media with effusion**, the liquid persists in the space, and a small procedure (**Myringotomy**) can be done to drain it. Many children with 22q11.2 deletion syndrome have recurrent or chronic otitis media, with or without effusion.

Otolaryngology – Otolaryngology is the branch of medicine that deals with the head and neck region. Other names for this specialty include "Head and Neck Surgery" or "Ear, Nose, Throat (ENT)". A medical doctor who specializes in this field is called an otolaryngologist or an ENT.

Overt Cleft Palate – [See **Cleft Palate**]

P

Pancreas – The pancreas is an organ located behind the **stomach**. It makes digestive enzymes that helps break down proteins and carbohydrates.

Panic Disorder – Panic disorder is a treatable condition where the person has repeated panic attacks (intense fear including pounding heart, sweating, trembling, etc.) for no reason, plus at least one month of worries about having more panic attacks and significant behavior changes due to this fear of having more panic attacks.

Paresthesia – Paresthesia is a tingling feeling on the skin. For example, the hand and the arm may have this sensation after a person sleeps on the arm, but this sensation lasts a short time and goes away on its own. Paresthesia that are constantly present may indicate a medical issue that affect the nerves, thyroid, parathyroid, or immune system, or it can be a sign of infectious diseases or toxicity.

Parkinsonism – Parkinsonism is an umbrella term for movement disorders similar to those seen in **Parkinson's disease** (**PD**). Individuals with parkinsonism have symptoms such as slowed movement, muscle stiffness, tremors, and trouble with balance and coordination. PD and certain medications (especially antipsychotics) may lead to parkinsonism, but there are multiple other possible causes. Management of parkinsonism depends on both the underlying cause and the severity of the symptoms.

Parkinson's disease (**PD**) – PD is a neurodegenerative disease, characterized by a progressive loss of structure or function of brain cells. Symptoms of PD include slowed movement, muscle stiffness, tremors, and trouble with balance and coordination. Adults with 22q11.2DS are at increased risk of developing PD, especially early-onset PD. A neurological assessment can help with early diagnosis and effective treatment. [See also **Parkinsonism**]

Pediatrics – Pediatrics is the branch of medicine that deals with the health, growth, and diseases of babies, children, and teenagers. A medical doctor that specializes in pediatrics is a pediatrician. [Note: The British spelling of pediatrics is paediatrics]

Peristalsis – Peristalsis is the wave-like muscle movement that the esophageal muscles make. The repeated contraction and relaxation action moves food down the esophagus into the **stomach**. [See "Swallowing and Dysphagia" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Periventricular Nodular Heterotopia (**PNVH**) – [See **Heterotopia**]

Pharyngeal Hypotonia – Pharyngeal means related to the throat, and hypotonia means muscle weakness. Causes of the muscle weakness in the throat area include genetic conditions and nerve problems. The weakness can lead to feeding and swallowing problems as well as **obstructive sleep apnea**. Treatments will depend on the cause and the extent of the problem.

Pharynx – The pharynx is an area in the throat behind the mouth and the nasal cavity, and above the **esophagus** and **trachea** (windpipe). It is important in swallowing, breathing, and in speech. [See "The **Digestive System** (Gastrointestinal Tract)" in the Gastrointestinal (GI) Series and "Introduction to Speech" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Phenotypic Variability – In medical genetics, phenotypic variability (also known as **variable expressivity**) happens when people with the same genetic makeup show different clinical features. Individuals (even those in the same biological family) who have the exact 22q deletion can have very different symptoms and severities. This happens for all the standard 22q11.2 deletions and 22q11.2 duplications.

Phonation – Phonation is the production of sound in the **larynx**. [See "Introduction to Speech" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Phonological/Developmental Speech Sound Disorder – A developmental speech sound disorder is one in which a child is following a typical pattern of speech development but continues to make errors that should have disappeared when they were younger. Phonological disorders are predictable, rule-based errors that may affect more than one sound. They are common in children with 22q. [See "**Speech Sound Disorders**" in the Speech Series of the Health Conditions Explained page of our website for more information and examples.]

Platelets (**Thrombocytes**) – Platelets are colorless, disc-shaped blood cells that help form blood clots, which slow down bleeding and help wounds heal. [See **Thrombocytopenia**]

Polyhydramnios – When a baby is inside the uterus, he/she is surrounded by amniotic fluid. Polyhydramnios is the condition in which too much amniotic fluid is present. In severe cases of polyhydramnios, the pregnant mother may have shortness of breath, swelling, and contractions, and the position of the baby may be affected. This condition can cause complications for both the mother and the baby during the birth process. Polyhydramnios can happen while a fetus with 22q11.2 deletion is developing in the uterus.

Polymicrogyria – The human brain usually has some folds on the surface. In an individual with polymicrogyria, there are too many folds, and the folds are small compared to normal. This happens when nerve cells in the brain do not migrate to the right places when the baby is developing in the womb. The area of the brain affected by polymicrogyria often does not function properly, and sometimes the function of other parts of the brain may also be affected. **Seizures** in some individuals may be related to polymicrogyria. This malformation cannot be reversed, but doctors can help the patient manage the seizures.

Polypharmacy – Polypharmacy means using several medications (prescribed by one or more healthcare professionals) at the same time. It is a common situation for individuals with 22q differences as they try to manage multiple health conditions. When a person takes multiple medications, it is best to minimize side effects that arise from the combination. Medical professionals also need to ensure that one medication does not interfere with the functioning of another.

Post-Traumatic Stress Disorder (**PTSD**) – Post-traumatic stress disorder is a condition where the person has flashbacks, nightmares, and anxiety for several months or longer after a scary event.

Prophylaxis – Prophylaxis is an action that is done to prevent disease. For example, some individuals with 22q11.2DS who were born with certain heart defects may need antibiotics for "**endocarditis** prophylaxis" before dental appointments to prevent bacteria from the mouth from causing endocarditis (inflammation of the inner lining of the heart). [See Heart Series in Health Conditions Explained]

Prosody – Prosody of speech is the variation in speech rate, intonation, rhythm, and stress patterns. It helps convey meaning, emotions, and social cues. Abnormal speech prosody has been observed in some children with **motor speech disorders**, which is common with 22q differences. [See "**Speech Sound Disorder**" and "Motor Speech Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Psychiatric Illnesses – Psychiatric illnesses, also called mental health disorders, are a wide range of conditions that affect people's mood, thinking, behavior, and often physical state. Most respond well to available treatments. For the best outcome, it is important to diagnose and treat psychiatric illnesses as early as possible. [See Mental Health Series on the Health Conditions Explained page of our website.]

Psychiatry – Psychiatry is the branch of clinical medicine that deals with **psychiatric illnesses** and mental health. These include conditions that affect how a person thinks, feels, acts, handles stress, relates to others, makes decisions, etc. A medical doctor who specializes in psychiatry is a psychiatrist, able to provide treatments, including prescribing medications.

Psychoeducation – Psychoeducation is a learning process in which healthcare providers help the person (sometimes a child), parents or other caregivers/family members understand the person's diagnosis, symptoms, abilities, limits, the impact of health condition, and the potential treatments available. Psychoeducation often increases the effectiveness of treatment.

Psychology – Psychology is the study of the mind, emotions, and behavior, and used to be a branch of philosophy. A psychologist usually has a graduate degree (MA, or PhD). He/she performs neurocognitive and psychoeducational assessments and uses various therapies (but not medications) to treat patients' mental health issues.

Psychosis – Psychosis is a symptom in which the person becomes out of touch with reality, that is, has difficulty telling apart what is real and what is not. This can include believing that someone is out to harm them when this is not the case, or hearing voices or seeing things that are not there. They may Thinking may seem confused or mixed-up. Psychiatric illnesses that include psychosis as a symptom are called **psychotic disorders**. These are treatable conditions. An example is **schizophrenia**, which develops in about 1 in 4 to 5 adults with 22q11.2 deletion syndrome. Individuals with 22q11.2DS are known to have a higher risk of developing schizophrenia than others in the general population. It is important to identify changes in thinking, emotions, behavior, and functioning in order to intervene as early as possible. [See Mental Health Series on the Health Conditions Explained page of our website.]

Ptosis – Ptosis refers to a droopy eyelid and happens in about 4% of children with 22q11.2 deletion syndrome. The eyelid may droop a little or droop enough to affect vision if it blocks light entering the pupil. Most ptosis occurs as people age, but some infants are born with this condition. Ptosis can be corrected by surgery to lift the eyelid.

Puberty – Puberty is the period in which children's bodies develop and mature into adults' bodies that are capable of sexual reproduction. Usually, girls start puberty between ages 8 to 13, while boys start between ages 9 to 14. The puberty experience and the speed at which changes come about are different from person to person.

Q

R

Refractive Errors – Refractive errors happen when the shape of the eye does not allow incoming light to focus properly on the retina, leading to blurry vision. Refractive errors such as **hyperopia** and **astigmatism** are common in individuals with 22q11.2DS and may be corrected using eyeglasses.

Resonation – Resonation is the balance of air between the mouth and nose during speech. The build-up of air pressure in the mouth is needed for most speech sounds. This oral pressure requires that air be sealed in the mouth by closing off the space to the nose in the back of the throat. [See "Introduction to Speech" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Respiration – Respiration is the continuous cycle of breathing in and out.

Respirology (or **Respiratory Medicine**) – Respirology is the branch of medicine that deals with diseases of the respiratory system, which is the collection of body parts (nose, **trachea**, bronchi, lungs, etc.) that helps a person breathe. A medical doctor who specializes in respirology is a respirologist.

Restless Legs Syndrome (**RLS** or **Willis-Ekbom disease**) – RLS is a neurological disorder that primarily affects the legs, causing an uncontrollable urge to move them due to uncomfortable sensations. These sensations can be relieved temporarily by movement. The symptoms are typically worse during the evening or at night, leading to sleep interruptions and subsequent daytime fatigue. The cause of RLS is unknown, but it can sometimes happen together with more nerve problems, kidney problems, or iron deficiency. The treatment may include lifestyle changes or medications.

Retrognathia – Retrognathia is a condition in which the lower jaw sits farther back compared to the upper jaw. It may be a feature of several genetic syndromes. Retrognathia can make it difficult for a baby to latch onto the nipple during breastfeeding, and he/she can also have **obstructive sleep apnea**. Severe cases of retrognathia in babies may require surgical intervention to improve breathing. Less severe cases may not be treated until childhood or adolescence.

Rheumatology – Rheumatology is the branch of medicine that deals with inflammation of the muscles, joints, and connective tissues. Examples of inflammatory (rheumatic) conditions include arthritis, osteoporosis, and tendonitis, but there are many more. A medical doctor who specializes in rheumatology is a rheumatologist.

S

Sacral Dimple – A sacral dimple is a small indentation or pit that is present on the lower back, just above the crease of the buttocks, and it is visible at birth. In many cases, sacral dimples are harmless and do not cause any issues. They are quite common, occurring in approximately 2-4% of newborns. Most sacral dimples are considered benign and are not a cause for concern. However, in some instances, a sacral dimple may be associated with underlying spinal cord or spinal canal abnormalities, which are more serious conditions like **tethered cord** and **spina bifida**.

Salivary Glands – The salivary glands are located in the bottom and back of the mouth. They produce saliva, which have important roles in swallowing, digestion, teeth protection, and mouth lubrication. [See "The Digestive System (Gastrointestinal Tract)" in the Gastrointestinal (GI) Series and "Let's Talk 22q Teeth" in the Dental Series of the Health Conditions Explained page of our website for more information.]

Schizophrenia – Schizophrenia is a serious but treatable psychotic disorder. This illness involves psychotic symptoms (e.g., **delusions** and/or **hallucinations**) and a decline in the ability to function with respect to relationships, work/school, and/or self-care/basic hygiene. Schizophrenia affects about 1 in 25 individuals with 22q11.2DS. Although schizophrenia cannot be prevented, it can be treated with medications, psychoeducation, and rehabilitation. Early diagnosis and treatment can lead to better outcomes. [See Mental Health Series on the Health Conditions Explained page of our website.]

Sclerocornea – Sclerocornea is a birth defect of the eye in which the cornea (front clear part) is cloudy. This condition, which is rare among individuals with 22q11.2DS, does not involve any inflammation and does not get worse. The cloudiness of the cornea may decrease the amount of light that enters the eye and therefore reduce vision, but some affected individuals have near-normal eyesight. An **optometrist** or **ophthalmologist** can diagnose this condition and check for any additional (potentially serious) conditions by performing a complete eye exam. If the sclerocornea is the only problem, a cornea transplant may be recommended.

Scoliosis – Scoliosis happens when the spine (backbone) curves sideways, and often the cause is unknown. Mild scoliosis may not lead to a lot of issues and can be monitored. Severe scoliosis can lead to problems with breathing, back pains, and an uneven appearance. Some patients with scoliosis need to wear braces to prevent the backbone from curving more, while other people may need surgery to correct it. The prevalence of scoliosis is about 48% in 22q11.2 deletion syndrome. The curve pattern resembles adolescent idiopathic scoliosis (the routine type of scoliosis) in most patients, and treatment is similar. Children with 22q11.2DS should be evaluated with physical exam of the spine at their annual physical and referred to orthopedics if a curve is detected. Curves of the spine can begin as early as age 5 years.

Seizure – A seizure is like a sudden "storm" in the brain. Normally, the brain sends out tiny electric signals that help us think, move, and feel. But during a seizure, there's a sudden, unexpected burst of these signals, like a big surge of electricity. This can make someone do different things, like shake all over, stiffen up, stare off into space, or even fall down. Seizures can happen for lots of reasons, and some are more serious than others. Some people might need medicine or surgery to help control them, while others might not need as much help.**Sensorineural Hearing Loss** – [See **Hearing Loss**]

Separation Anxiety Disorder – This childhood condition involves feeling excessive distress about being away from home or loved ones. Young children usually experience separation anxiety as a part of normal development, but most outgrow it by about 3 years of age.

Small intestines – The small intestines are part of the digestive tract between the **stomach** and the **large intestines**. The three main sections are the **duodenum**, **jejunum**, and **ileum**. In the small intestines, food become further digested and absorbed before the remaining waste go to the large intestines. [See "The **Digestive System** (Gastrointestinal Tract)" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Social Anxiety Disorder (**Social Phobia**) – Social anxiety disorder involves having excessive worries about being judged in social situations, with the anxiety causing great distress and/or avoidance of situations, lasting more than 6 months, and impairing day-to-day functioning.

Specific Phobia – Specific phobias involve having intense, irrational, beyond-usual fears of things like certain animals, environments, situations, blood, etc.

Speech Development – Speech development is the complex but predictable process in which children learn to speak. They need to figure out how to move their muscles in order to produce speech that other people can understand. Hearing sounds and words are important in this process. [See "Introduction to Speech" and "Consonants and Vowels in English" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Speech Intelligibility – Speech intelligibility refers to how well someone's speech is understood. Those closest to a child with poor speech intelligibility may understand their speech best. The listener's knowledge of context (what the child is speaking about) can often aid speech intelligibility.

Speech Motor Delay (**SMD**) – Speech motor delay is a recently proposed diagnostic category that was previously called "motor speech disorder – not otherwise specified (MSD-NOS)". It includes speech issues that do not meet the criteria of dysarthria or childhood apraxia of speech. [See "**Motor Speech Disorders**" in the Speech Series of the Health Conditions Explained page of our website for more information]

Speech Sound Disorders – Speech sound disorders are common in children with 22q differences. These children have difficulty producing sounds correctly, and their speech can be difficult to understand. [See **Compensatory Articulation Disorder**, **Obligatory Articulation Disorders**, **Phonological Speech Sound Disorder**, and **Developmental Articulation Disorders**. See also "Speech Sound Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Speech Therapy – Speech therapy is treatment that helps improve a person's ability to make speech sounds and communicate with language. However, while speech therapy is essential to the treatment of speech errors in children with 22q differences, velopharyngeal surgery is necessary to correct **velopharyngeal dysfunction** (**VPD**). [See "How Parents and Professionals Can Help" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Speech Language Pathology – Speech language pathology is the healthcare field that deals with communication and swallowing disorders. A healthcare provider who diagnoses and treats these problems is called a speech-language pathologist (SLP). Most children with 22q differences benefit from speech therapy from SLPs. [See the Speech Series of the Health Conditions Explained page of our website for more information.]

Spina Bifida – Spina bifida is a birth condition that affects how the spine and spinal cord develop in a baby during early pregnancy. It happens when the neural tube, which is supposed to form the brain and spinal cord, doesn't close all the way. This can leave an opening in the bones of the spine, and sometimes part of the spinal cord and its coverings stick out through the gap. Spina bifida is rare in people with 22q11.2DS. Spina bifida can be mild, where it's hidden (called spina bifida occulta) and might not cause any problems. Sometimes, it's only found by accident during imaging. But in the most severe cases (called **meningomyelocele** or **myelomeningocele**), the nerves and tissues stick out in a sac on the baby's back. This can cause serious, life-long problems with movement, feeling, and control over parts of the body. To check if a baby might have spina bifida, doctors can do a blood test and an ultrasound during pregnancy. Babies with spina bifida might need surgery before or after they are born. Some common issues they might face include trouble moving their legs, problems with using the bathroom, a buildup of fluid in the brain (called hydrocephalus), and learning difficulties. The exact cause of spina bifida isn't known, but doctors recommend that women who could get pregnant take folic acid (a type of vitamin) every day to help prevent it.

Strabismus – Strabismus refers to any form of misaligned eyes and can be the result of various causes. The eyes are not aligned properly and are looking in different directions at the same time. If untreated, the brain can learn to ignore the image from one eye, leaving that eye with permanently-reduced vision due to amblyopia. Treatment options for strabismus include eyeglasses, prism lenses, vision therapy, and eye surgery. Strabismus happens in about 18% of children with 22q11.2 deletion syndrome.

Stress – A person feels stressed when their capacity does not match what the environment requires, e.g., a child being required to stay at a social situation longer than they can handle.

Stomach – The stomach is an organ in the **digestive system**. It produces acids and enzymes that break down food into smaller components that can be absorbed in the **small intestines** and used by the body. [See "The **Digestive System** (Gastrointestinal Tract)" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Stridor – Stridor is a high-pitched sound heard with breathing that results from turbulent air flow when the airway is partially blocked or narrowed. About 20% of children with 22q11.2 deletion syndrome have airway anomalies which can lead to stridor. In rare cases, malformed blood vessels (**vascular ring**) compressing the airway can also lead to this type of noisy breathing. Any patient with stridor should get medical attention.

Subglottic Stenosis – Subglottic stenosis is a narrowing of the airway in the section of the **larynx** (voice box) below the **vocal cords**. The affected child often breathes noisily (**stridor**) and, in severe cases, may struggle to breathe. Subglottic stenosis may be present at birth or may occur after birth because of infection or as a complication of intubation (breathing tube placement). Treatment depends upon the severity of the narrowing and may include simple observation, dilation (widening) of the airway using a balloon, or surgery.

Submucous Cleft Palate – The palate is the roof of the mouth, and it usually separates the mouth from the nose. A **cleft palate** happens if the tissues there did not fuse together properly before birth, leaving an opening between the mouth and the nose. In a submucous cleft palate, there is a split of the muscles that make the palate work normally, but the mucous membrane covering the palate is intact. The child with a submucous cleft palate may have difficulty swallowing, reflux of liquids or foods out of the nose, a nasal speaking voice, and chronic ear infections. Since palate problems are very common in babies with 22q11.2DS, it is recommended that they receive a thorough check by a doctor specialized in these conditions. A surgery may be needed to repair the palate to avoid further complications.

Suggestibility – Highly suggestible individuals tend to be easily influenced. They may readily accept other people's ideas or actions without thinking them through. This is often a sign of weak 'executive functioning' skills, and can leave individuals vulnerable to physical or emotional abuse, scams, etc. Individuals with 22q-related difficulties typically have reduced executive functioning skills.

Surgical Complications – Surgical complications are unpleasant and/or potentially life-threatening health conditions that happen during or after surgeries. There is a somewhat increased risk of surgical complications for individuals with 22q11.2DS, and they include bleeding, infections, **seizures**, and difficult intubation. These individuals may need close monitoring of calcium levels, platelets, and oxygen saturation, and some may need smaller intubation equipment.

T

T Cell (T Lymphocytes) – T cells are a type of white blood cells in the immune system. They come from the **bone marrow** but mature in the **thymus** (which is why they are called T cells). On the surface of the T cells are **T cell receptors (TCRs)**, and there are thousands of different TCRs. TCRs checks the broken-down proteins that **antigen presenting cells (APCs)** show them. If the broken-down proteins come from harmful sources, the T cells will trigger a response from the immune system to remove the offender. The exact way of accomplishing this action depends on the subtype of T cells. About 80% of babies with 22q11.2 deletion syndrome have T cell levels that are too low (**T cell lymphopenia**, also called **T cell lymphocytopenia**). The level gradually increases as they develop, but the T-cells that patients eventually end up with may not function as well or live as long. Problems with T cells are also related to **autoimmune disorders** and **allergies** in patients with 22q11.2DS. It is strongly recommended that babies with T cell problems be assessed by an immunologist to determine the precautions they need regarding the administration of live vaccines, the use of blood products, and the need for thymus transplants.

Tachycardia – [See **Arrhythmia**]

Tetany – Tetany is an uncontrolled movement of muscles. Tetany can be triggered by a toxin from *Clostridium tetani* bacteria, but this type of tetany can be prevented by receiving the tetanus vaccine every 10 years. Tetany can also be triggered by **hypocalcemia** (having a low level of calcium ions in the blood), which happens in about 60% of children with 22q11.2 deletion syndrome. Tetany due to hypocalcemia should be treated with calcium through slow infusion but not through the mouth. The aim is to maintain calcium levels in the blood in the low-normal range to avoid other complications.

Tethered cord – A tethered cord is a condition where the spinal cord is abnormally attached to tissues around the spine, usually at the lower back. Normally, the spinal cord floats freely within the spinal column, allowing it to move as the body grows and bends. However, in tethered cord syndrome, the spinal cord is anchored, or "tethered," to the surrounding structures. This can cause the spinal cord to stretch as a person grows, leading to a range of symptoms such as back pain, leg weakness, numbness, and difficulties with bladder or bowel control. Tethered cord can be present at birth (congenital) or develop later in life. It is often associated with other spinal abnormalities, like **spina bifida**. Treatment typically involves surgery to release the cord, preventing further damage to the spinal nerves and relieving symptoms.

Tetralogy of Fallot – Tetralogy of Fallot (TOF) is a congenital heart disease that occurs in approximately 20 to 45% of individuals with a 22q11.2 deletion, while 10 – 15% of patients with TOF have a 22q11.2 deletion. TOF is characterized by 4 features: (1) a hole in the wall between the left and right lower pumping chambers (**ventricles**); (2) a narrowing of the pulmonary valve which regulates the blood flow from the heart to the lungs; (3) an **aorta** that incorrectly gets blood from 2 lower heart chambers instead of just 1; and (4) a thickening of the muscle of the wall of the lower right chamber. In babies with TOF, blood that exits the heart and goes to the rest of the body does not carry enough oxygen, which leads to a bluish-purple color to the skin (**cyanosis**). Surgery at a very young age is usually required and performed very successfully. [See "Tetralogy of Fallot" in the Heart Series on the Health Conditions Explained page of our website.]

Tonsillectomy – The tonsils are part of the immune system that guard against viruses and bacteria that enter the child's body through the mouth. Tonsillectomy is a surgical procedure to remove the tonsils. Tonsillectomy may be recommended for the treatment of frequent infections or as a treatment for **obstructive sleep apnea** in children. In some cases, tonsillectomy may be recommended prior to posterior pharyngeal flap surgery to lessen the risk of post-operative obstructive sleep apnea.

Thrombocytopenia – Thrombocytopenia is a condition in which there are too few **platelets** in the blood. Platelets are blood components that help blood clot and stop bleeding, so having too few of them can increase the risk of bleeding. Thrombocytopenia may get better on its own or may require treatment. Some individuals with 22q11.2 deletions have immune thrombocytopenia, in which the body's immune system mistakenly destroy platelets. In these cases, doctors may prescribe medications to improve the platelet count. This problem occurs in about 4% of children with 22q11.2 deletion syndrome.

Thymic Hypoplasia (**Hypoplastic Thymus**) – Thymic hypoplasia means that the **thymus** is not fully developed. As the schoolhouse required for **T cell** development and education, this leads to low T cell counts and **immunodeficiencies**. About 80% of babies with 22q11.2 deletion syndrome have this condition.

Thymus – The thymus is a small organ that is part of the immune system. It is located at a spot that is in front of and above your heart, between your lungs. The thymus is the place for the maturation and specialization of **T cells**, which play essential roles in the immune response against foreign pathogens such as viruses and bacteria. Babies with severe **immunodeficiencies** that result in very low T cell counts may require a thymus transplant.

Trachea – The trachea, commonly known as the windpipe, is a tube that connects the **larynx** (voice box) to the lungs. It brings oxygen-rich air to the lungs and carries oxygen-poor air back up. The trachea is split into two bronchi, which go onto the left and right lungs.

Tremors – Tremors in people with 22q11.2 deletion syndrome are involuntary shaking or trembling movements that they can't control. These tremors can affect different parts of the body, like the hands, arms, or head. In 22q11.2DS, tremors may occur due to neurological differences, and they can vary in intensity. Some people might experience mild tremors that are barely noticeable, while others may have more significant shaking that affects daily activities like writing or holding objects. Tremors can sometimes be managed with medications, physical therapy, or other treatments depending on their severity and impact on the person's life. It's important for individuals with 22q11.2DS and their families to work with healthcare professionals to find the best way to manage these symptoms.

U

Ulcerative Colitis – [See **Inflammatory Bowel Disease** (**IBD**)]

Ultrasound Sonography (or "**Ultrasound Imaging**") – Ultrasound means sounds that are at such a high frequency that humans cannot hear. Sonography means using sound waves to generate images. Ultrasound sonography can be used to check internal organs (such as the bladder, the kidneys, the liver etc.) and the fetus in the womb.

Urology – Urology is branch of medicine that deals with the urinary tract in both men and women, as well as the reproductive organs in men. A medical doctor who specializes in urology is a urologist.

V

Vaccine – Vaccines are generally made with one of the following: (1) Purified components from dead viruses or dead bacteria; or (2) weakened viruses or bacteria (These are "live attenuated vaccines"). The purpose of vaccination (receiving a vaccine) is to teach the immune system about viruses and bacteria that should be removed from the body. [See **Immunization**] All individuals benefit from standard vaccinations, including COVID-19 and influenza, though some may have reduced responses. At the same time, it is strongly recommended that individuals with 22q11.2 deletion syndrome be evaluated by an immunologist to determine if they can safely receive live attenuated vaccines.

Vagus Nerves (Vagal Nerves) – The vagus nerves are long nerves that run from the brain to the **large intestines**. They are an important part of the nervous system that allow brain messages to be sent to different parts of the body. These brain messages direct body functions such as digestion, heart pumping, and immune defence. If the vagus nerves are damaged, the brain cannot communicate with some of the important organs in the body, resulting in symptoms such as fainting, problems swallowing and delayed **stomach** emptying (**gastroparesis**). [See "Gastroparesis" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.]

Veins – Veins are blood vessels that carry oxygen-poor blood from different parts of the body to the heart.

Velocardiofacial Syndrome (**VCFS**) – Velocardiofacial syndrome is one of the old names of 22q11.2 deletion syndrome. "Velo" comes from the Latin word "velum", which means the palate; "cardia" means the heart; and "facies", which means the face. The term velocardiofacial syndrome was coined by Dr. Robert Shprintzen in 1978.

Velopharyngeal Dysfunction (**VPD**; also called **Velopharyngeal Incompetence** (**VPI**) or **Velopharyngeal Insufficiency** (**VPI**)) – During normal speech, the soft palate (the back part of the roof of the mouth) closes against the back of the throat, so that air cannot come out through the nose. Velopharyngeal dysfunction happens when the back of the palate and the throat cannot close the space that connects the mouth and the nose during speech and swallowing. This can lead to nasal emission of air during speech production, hypernasal speech, weak pressure on consonants, **speech sound disorders**, and difficulty swallowing. VPD can happen as a result of any or all of the following: anatomical (structural) and/or physiological (functional) abnormalities, motor disorders, mislearning. VPD occurs in about 70% of patients with 22q11.2 deletion syndrome. Although speech therapy is essential to the treatment of articulation errors in 22q11.2DS, velopharyngeal surgery is necessary to treat velopharyngeal dysfunction.

Velum – Velum is Latin for the palate, which is the roof of the mouth. It separates the mouth and the nose.

Ventricles –Ventricles are lower pumping chambers of the heart. The right ventricle pumps oxygen-poor blood towards the lungs, while the left ventricle pumps oxygen-rich blood towards the rest of the body. [See "The Heart and Normal Blood Flow" and "Ventricular Septal Defect" in the Heart series on the Health Conditions Explained page of our website.]

Ventricular Septal Defect (**VSD**) – A ventricular septal defect is a hole in the heart wall separating the two lower chambers of the heart (ventricles). Of all patients with **conoventricular VSD**, 5% have 22q11.2 deletion syndrome (22q11.2DS). (Conoventricular VSD is a type of VSD where the hole is in the upper portion of the septum just before the pulmonary valve and aortic valve.) 10 to 50% of children with 22q11.2DS are born with conoventricular VSD. [See "Ventricular Septal Defect" in the Heart Series on the Health Conditions Explained page of our website.]

Vertebrae – The vertebrae are the 33 small bones that stack up to make up your backbone (also called spine). In the middle of the vertebrae is a tunnel which houses the spinal cord and nerves. Most of the vertebrae can move around to allow for a range of motion. The vertebrae in the cervical spine (neck) frequently have a congenital abnormality of shape or formation. The congenital abnormalities may in some cases place the patient at an increased risk of injury because of potential instability of the cervical spine. X-ray examination of the cervical spine, including flexion and extension views of the lateral cervical spine, should be considered as a screening exam in all patients with 22q deletion syndrome regardless of symptoms. [See **Scoliosis**]

Vocal Cords (**Vocal Folds**) – Vocal cords are two bands of muscles in your **larynx** (voice box). Our voice is generated when the vocal cords vibrate the outgoing breath stream and make sound. The faster the vibration, the higher the voice. The vocal cords also close the windpipe when a person swallows. [See "Introduction to Speech" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Vocal Cord Paralysis – Vocal cord paralysis is a condition in which the nerves that control the **vocal cords** are damaged (by trauma or neurologic disease), resulting in the failure of one or both vocal cords to move. The affected person may therefore have a hoarse, breathy, strained and/or soft voice. If the vocal cords are stuck in an open position, they cannot close the top of the windpipe, and food and liquids may enter the windpipe when the person swallows. [See "Voice Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Vocal Nodules – Vocal nodules are little bumps, or callouses, on the inner edge of the **vocal cords**. They affect the way the vocal cords contact each other and vibrate, leading to a voice that is hoarse, breathy, and strained. They result from overuse or straining the voice. [See "Voice Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Vomiting – Vomiting, also called throwing up, happens when the **stomach** contracts and pushes its contents up the **esophagus** and out of the mouth. It is a common problem in children with 22q11.2DS. See "**Nausea** and Vomiting" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.

W, X, Y, Z

See also: **Health Conditions Explained**

https://22q.org/symptoms-care/health-conditions-explained/

To help our patients and families navigate through their medical journeys, the International 22q11.2 Foundation started a section called "**Health Conditions Explained**" on our website in 2022. The information sheets are generated by individuals who are highly involved in 22q-related education and communication, and the content is carefully reviewed by 22q experts.

In addition to the 22q Glossary (this document), we have published the following series as of September 2024:

HEART SERIES

- The Heart and Normal Blood Flow
- Ventricular Septal Defect (VSD)
- Tetralogy of Fallot (ToF)
- Truncus Arteriosus (TA)
- Interrupted Aortic Arch (IAA)

DENTAL SERIES

- Let's Talk 22q Teeth – Info for Families
- Dental Health in Children with 22q – Info for Dentists

PALATE SERIES

- The Velopharynx
- Cleft Palate & Submucous Cleft Palate
- Velopharyngeal Dysfunction: Introduction and Causes
- Velopharyngeal Dysfunction: Diagnosis
- Velopharyngeal Dysfunction: Surgery
- Velopharyngeal Dysfunction: Speech Issues

MENTAL HEALTH SERIES

- Mental Health and 22q11.2 Deletion Syndrome
- Mental Health and 22q11.2 Duplication Syndrome
- What Parents Can Do for Their Children
- Attention Deficit Hyperactive Disorder
- Autism Spectrum Disorder
- Anxiety Disorders
- Psychotic Disorders [Including schizophrenia]
- Mood Disorders
- Mental Health Q&A (Spring 2023)

(Continued on the next page)

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GI SERIES

- GI Issues in 22q11.2 Deletion Syndrome
- The Digestive System
- Feeding Difficulties
- Swallowing and Dysphagia
- Constipation
- Gastroesophageal Reflux Disease (GERD)
- Esophageal Dysmotility
- Gastroparesis
- Cholelithiasis (Gallstones)
- Nausea and Vomiting
- Autoimmune Issues in the GI System
- Inguinal Hernias
- Nonalcohol Fatty Liver Disease (NAFLD)

SPEECH SERIES

- Introduction to Speech
- Consonants and Vowels in English
- Speech Disorders in Individuals with 22q11.2 Deletion Syndrome – an Overview
- Speech Disorders in Individuals with 22q11.2 Duplication Syndrome – an Overview
- Voice Disorders
- Speech Sound Disorders
- Motor Speech Disorders
- How Parents and Professionals Can Help

We will add more contents on various topics as they become available.

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