CHARGE SYNDROME: GLOSSARY

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A

Accutane: (isotretinoin, retinoic acid) prescription medication used to treat severe cystic acne. It is a synthetic derivative of Vitamin A. When taken during pregnancy, it can cause very serious birth defects, including hydrocephalus, microcephaly (very small head), mental retardation, small and malformed ears and other facial abnormalities, and heart defects. Although the ear malformations can be similar to those seen in CHARGE, the face is distinct.

amblyopia: "lazy eye" - poor vision in one eye without detectable cause. Often treated by patching the stronger eye.

anesthesia: 1. Loss of sensation resulting from pharmacologic depression of nerve function or from neurological dysfunction. 2. Broad term for anesthesiology as a clinical specialty. Sedation used during surgery

anophthalmia: absence of the eye or eyes. Anophthalmia can be considered the most severe form of a coloboma.

aorta: the large artery arising from the base of the left ventricle of the heart. The aortic arch is formed by the ascending aorta and the descending aorta.

aortic arch: The curved portion between the ascending and descending parts of the aorta. Aortic arch anomalies are common in CHARGE

apnea: A potentially life-threatening condition inn which breathing stops abnormally, usually during sleep. CPR is sometimes needed to start the breathing again.

arhinencephaly: a term used to refer to an absence of some or all areas of the forebrain, or certain areas of the anterior (front) portions of the brain, particularly the olfactory bulbs and nerves. The defect may occur relatively late in fetal differentiation, so that only a small area of the brain is involved. One result can be the lack of a sense of smell.

aspiration: The inspiratory sucking into the airways of fluid or foreign body, as in vomitus. Children with CHARGE are at risk for aspiration of food or liquid which is not adequately swallowed or which is refluxed up from the stomach.

aspiration pneumonia: A lung inflammation caused by inhaling a foreign body, such as food, into the lungs.

association: A connection of persons, things or ideas by some common factor. In genetics: the occurrence together in a population, more often than can be readily explained by chance, of two or more traits of which at least one is known to be genetic. For example VACTERAL association.

atresia: A general term for closure of a structure that should be open, such as a canal, passage or tube. See choanal atresia, esophageal atresia, and ear anomalies.

atrial septic defect (ASD): A hole between the two upper chambers (atria) of the heart. Surgery is usually required to close an ASD.

audiogram: The graphic record drawn from the results of hearing tests with audiometer. The audiogram charts the threshold of hearing at various frequencies (pitch) against sound intensity in decibels (loudness). Pure tone audiogram: a chart of the threshold for hearing acuity at various frequencies usually expressed in decibels above normal threshold and usually covering frequencies from 128 to 8000 Hz. Speech audiogram: the record of thresholds for spondaic word lists and scores for phonetically balanced word lists. See Ears and Hearing Section.

autistic disorder: Impaired development of social interaction and communication along with repetitive and stereotyped patterns of behavior, interests, and activities, with an onset prior to age three.

autistic behaviors may include withdrawal from social contact, avoidance of eye contact, and failure to develop friendships; delay or total lack of communication, or communication is abnormal; self-stimulation, preoccupation with objects or parts of objects, obsession with certain routines or rituals. Autistic-like behaviors are observed in some children with CHARGE. It is not clear whether to attribute them to central nervous system structural abnormalities, to dual sensory impairment, or whether a separate diagnosis of autism is appropriate.

B

BAER: Abbreviation for "brainstem auditory evoked response." Measurement of alteration in the electrical activity of the auditory system of the brain brought about by presenting sounds through earphones. This can be done with the child sedated and without the cooperation of the child.

bilateral Two-sided, or affecting both sides.

C

cartilage connective tissues found primarily in joints, the walls of the thorax, and tubular structures such as the larynx, air passages, and ears; comprises most of the skeleton in early fetal life, but is slowly replaced by bone. Cartilage is often weak in children with CHARGE, resulting in floppy ears, laryngomalacia and other complications.

Cardiologist: physician who specializes in the heart.

central auditory processing A central hearing loss that may prevent sound from being interpreted meaningfully by the brain. Sound may be picked up by the ears and signals transmitted to the brain, but the brain has difficulty making sense of the input.

central nervous system (CNS) abnormalities: CNS abnormalities seen in children with CHARGE Syndrome include: structural brain abnormalities (diagnosed by CT scan or

MRI), microcephaly, seizures, apnea, and central processing problems (including a central hearing loss). See also: cranial nerve abnormalities and Brain Section.

cerebellum The large back portion of the brain. It consists of the two lateral hemispheres united by a narrow middle portion, the vermis.

cerebrum Originally referred to the largest portion of the brain, including practically all parts within the skull except the medulla, pons, and cerebellum; it now usually refers only to the parts derived from the telencephalon and includes mainly the cerebral hemispheres (cerebral cortex and basal ganglia.

cerumen: ear wax

CHARGE facial features: Children with CHARGE Syndrome resemble their parents, but they may also have some of the following features that make them look similar to other children with CHARGE Syndrome: square shape of the face and head, flat cheekbones, facial asymmetry (with or without facial palsy), wide nose with high bridge, and unusual ears (see Ear anomalies). The facial features do not cause any health problems, but can be very helpful in making the diagnosis of CHARGE Syndrome. See Diagnosis Section and photos with stories.

choroid (eye): The portion of the eye lying between the retina and the sclera.

chromosomes: Microscopic structures found in the nucleus of cells which contain the genetic information (DNA). Possible chromosome tests include routine (g-banded, including most prenatal chromosome testing), fragile X (to diagnose a particular cause of mental retardation), and FISH (to look for specific microdeletions or other abnormalities). No specific chromosome abnormalities have yet been shown to cause CHARGE. In VCF, there is a small deletion (microdeletion) of chromosome 22.

choanal atresia or stenosis: The choanae are the passages from the back of the nose to the throat which allow breathing through the nose. Choanal atresia is a birth defect in which this passage is completely blocked; stenosis refers to a narrowed passage. The atresia or stenosis can be on one or both sides. If a baby is born with bilateral choanal atresia, immediate surgery is needed to permit the baby to breathe through the nose. Choanal stenosis can lead to problems with breathing or increased nasal stuffiness. See Choanal atresia Section.

cleft lip and/or cleft palate: Many children with CHARGE Syndrome have a cleft lip (hare lip) and/or cleft palate (an opening in the roof of the mouth). Children with a cleft palate may have problems with ear infections and speech even after surgical correction. Submucous cleft palate may be difficult to diagnose in children with CHARGE. See Cleft lip and palate Section.

cochlea: part of the inner ear, responsible for transmitting sound to the auditory nerve.

cochlear implant A device that changes sound into electrical signals. These signals are then sent through the skin to an electrode array surgically implanted within the cochlea. Some individuals with severe to profound sensorineural hearing loss can get some hearing through a cochlear implant. Experience with cochlear implants in children with

CHARGE is limited.

coloboma: A cleft or keyhole-shaped defect of the eyeball. Colobomas can occur anywhere in the eye (any combination of iris, retina, or disc) and can affect one or both eyes. A coloboma of the iris (colored part of the eye) will result in a keyhole-shaped pupil, but probably will not affect vision. A coloboma of the retina or disc in the back of the eye can only be detected by an eye exam done by an experienced ophthalmologist. Retinal or disc colobomas can cause significant vision loss, both by restriction of the visual field (large blind spots across the top of normal vision) and by decreased acuity (blurred vision). See Eye Section.

columella: the part of the nose between the nostrils. It is sometimes prominent (sort of long) in children with CHARGE. See CHARGE face photos in Diagnosis Section.

conductive hearing loss: Hearing loss due to abnormalities of the middle ear bones (ossicles) and/or to fluid accumulation in the middle ear. If the loss is due to fluid accumulation, it may get better with use of PE tubes and/or decongestants. If the loss is due to malformed ossicles, surgery might be considered, but this is controversial.

conotruncal anomalies of the heart: Most common heart defects in CHARGE, including, double outlet right ventricle, tetralogy of Fallot, interrupted aortic arch, and VSD.

corpus callosum: the plate of nerve fibers which connect the two cortical hemispheres of the brain. Some individuals with CHARGE have absent or hypoplastic (underdeveloped) corpus callosum.

cranial nerves those nerves that emerge from, or enter, the cranium or skull, in contrast to the spinal nerves, which emerge from the spine or vertebral column. The twelve paired (one on each side) cranial nerves are the I - olfactory, II- optic, III - oculomotor, IV - trochlear, V - trigeminal, VI - abducent, VII - facial, VII - vestibulocochlear, IX - glossopharyngeal, X - vagal, XI - accessory, and XII - hypoglossal. Cranial nerve abnormalities are very common in CHARGE.

cryptorchidism: undescended testicles. If the testes do not descend into the scrotum, they can be lowered surgically.

CT scan: computerized tomography. A special X-ray of the head used to look at the structure of the brain.

D

Dandy-Walker malformation or cyst: A developmental anomaly of the fourth ventricle of the brain. It can result in cerebellar hypoplasia, hydrocephalus, and posterior fossa cyst formation. Occasionally seen in children with CHARGE.

developmental delay: Most children with CHARGE Syndrome will have delayed or retarded development. Vision loss causes delays in motor development. hypotonia (low muscle tone) and balance problems also delay motor development. Hearing loss can cause delays in speech and language. As developmental depends on the combination of intelligence, hearing, and the ability to see, early developmental delay does not always

mean mental retardation (see mental retardation). Many children with CHARGE have normal intelligence.

DiGeorge sequence: congenital absence of the thymus and parathyroid glands in combination with heart defects. The thymus problems lead to increased infections. Common in VCF syndrome (22q deletion), rare in CHARGE.

dysfunction: difficult or abnormal function

E

ear anomalies: Children with CHARGE Syndrome often have misshapen ears. Often the ears have characteristics which are unique to CHARGE. Typical "CHARGE ears" are small and wide, with little or no ear lobe. Often the outer fold of the ear (helix) is missing or may appear clipped-off. The child's two ears often look different. The unusually shaped ears do not cause hearing loss unless there is stenosis (narrowing) or atresia (collapse) of the ear canal, which is rare in CHARGE Syndrome. If a hearing aid is required, the ear anomalies may make it difficult to fit the ear mold properly. The ear anomalies can be very helpful in making the diagnosis of CHARGE Syndrome because they look different from ear anomalies seen in any other syndrome. See EARS and Hearing and Diagnosis Sections.

echocardiogram the ultrasonic record obtained by echocardiography. Sound waves are used to get an image of the heart to diagnose heart defects.

EEG (electroencephalogram): brain wave test used to look for seizure activity.

Endocrinologist: physician who specializes in treatment of hormone abnormalities.

esophageal atresia: the esophagus (food pipe) ends in a pouch instead of connecting to the stomach. Babies with this abnormality spit up all their food until it is surgically corrected. Even after surgery, feeding may be difficult for some time due to other swallowing problems which are seen in CHARGE Syndrome. See TEF/EA Section.

Eustachian tube: the tube leading from the tympanic cavity (behind the eardrum) to the nasopharynx (back of the throat). It enables equalization of pressure within the tympanic cavity with ambient air pressure, referred to commonly as “popping of the ears.” Blocked Eustachian tubes predispose to ear infections.

evoked potentials auditory screening for newborns using auditory evoked potential measures, including auditory brainstem response (ABR, BAER)

exotropia: outward turning of the eyes.

F

facial palsy or paralysis: The facial nerve (cranial nerve VII, which controls facial muscles) does not work in many children with CHARGE. This can cause a lopsided smile and trouble blinking the eye on the affected side. The affected eye may not produce tears very well. It may be unilateral or bilateral. If bilateral, it leads to a very blank expression. Facial

nerve palsy can affect both eating and speaking.

FISH Fluorescent in-situ hybridization. A specialized chromosome test using fluorescent dyes to identify very small deletions or other abnormalities of chromosomes. Many children with CHARGE will have chromosome tests done including FISH for deletion of chromosome 22 seen in VCF.

FM Trainer FM (frequency-modulated) signal transmission, represents the most successful and largest market of assistive listening devices for children. FM systems have an advantage of an improved signal-to-noise ratio over hearing aids.

fundoplication An operation in which the opening from the esophagus to the stomach is tightened.

G

G-tube gastrostomy tube (see below).

Gastroenterologist: physician who specializes in the esophagus, liver, spleen, bowel and pancreas.

gastrostomy An operation in which an artificial opening is made into the stomach through the wall of the abdomen to place a g-tube.

gastrostomy tube: a tube which is surgically placed in the stomach through the abdominal wall for feeding. This is done when the child cannot eat by mouth because of a severe cleft palate, tracheo-esophageal (T-E) fistula, or swallowing problems. Often, a gastrostomy button or Mic-key is used.

GE Reflux gastroesophageal reflux, the backward flow of food from the stomach into the esophagus, possibly into the pharynx where they can be aspirated between the vocal cords and down into the trachea; symptoms of burning pain and acid taste result; pulmonary complications of aspiration are dependent upon the amount, content, and acidity of the aspirate. This is very common in CHARGE.

growth deficiency small stature or size. In CHARGE, this can be due to multiple medical problems, growth hormone deficiency, or some other cause

growth hormone deficiency: Growth hormone (GH) is produced in the pituitary gland. It promotes body growth, fat mobilization, and the inhibition of glucose utilization. Most children with CHARGE are small. Some have documented GH deficiency. The frequency of GH deficiency in CHARGE is not yet known.

H

hearing loss: About 85% of children with CHARGE Syndrome have some hearing loss. The loss can be conductive, sensorineural (nerve), or mixed (both) and can range from a mild hearing loss to profound deafness. The loss may be progressive. A CHARGE syndrome hearing loss is typically a mixed loss with a large conductive component in the low frequencies and a sensorineural loss or mixed loss for high frequency sounds. The

losses can be severe and very difficult to measure completely and accurately, especially in infants and young children. As a result, repeated testing may be required before a satisfactory set of results is obtained. See Ears and Hearing Section.

heart disease, congenital (CHD): About 2/3 of children with CHARGE are born with some kind of heart defect. Some have only a murmur, while others may have a life-threatening heart defect which requires surgery. Heart abnormalities described in children with CHARGE include tetralogy of Fallot, ASD, VSD, PDA, aortic arch anomalies, double outlet right ventricle, pulmonic stenosis, and others. All children in whom a diagnosis of CHARGE is suspected should have a cardiac evaluation.

hockey-stick crease: crease on the palm of the hand which bends up to form a deep crease between the index and middle fingers. Although this has no medical significance, it is very common in children with CHARGE and can help confirm the diagnosis. See photo in Diagnosis Section.

holoprosencephaly: failure of the forebrain or prosencephalon to divide into hemispheres or lobes; cyclopia occurs in the severest form. It is often accompanied by a deficit in midline facial development. Often diagnosed by MRI or CT. Rare in CHARGE.

horseshoe kidney: union of the lower or occasionally the upper extremities of the two kidneys by a band of tissue extending across the vertebral column, resulting in a single horseshoe-shaped kidney. See Renal Section for illustration.

hydrocephalus :a condition marked by an excessive accumulation of fluid in the brain, resulting in dilation of the ventricles of the brain and raised intracranial pressure; may also result in enlargement of the cranium and atrophy of the brain. Treated by surgical placement of a shunt. Rare in CHARGE.

hydronephrosis ("water on the kidneys"): Dilation (enlargement) of the pelvis and calices of one or both kidneys resulting from obstruction or backflow of urine.

hypogonadism: underdeveloped genital system. In boys, there may be a small penis or cryptorchidism (undescended testicles). In girls, the labia (external skin folds) may be small or absent. In children of both sexes, hypogonadism may prevent puberty unless hormones are given.

hypoplasia: underdevelopment of a tissue or organ.

hypospadias: a genital problem in males. The urethral opening is not at the end of the penis and needs to be corrected surgically.

hypotonia: low muscle tone. This can lead to a "floppy" baby. Many children with CHARGE have hypotonia, especially of the trunk (upper body), contributing to delay of some motor milestones such as sitting and walking and predisposing the children to scoliosis.

IEP Individualized Education Plan

imperforate anus: the anus is closed over and needs to be opened surgically. Sometimes only a thin membrane needs to be opened. More often the blind end of the large bowel needs to be connected to the skin on the belly as a colostomy. Later the end of the bowel is put down through an artificial opening created where the anus ought to be. Not the same as choanal atresia. This is not usually seen in CHARGE.

incontinence: inability to prevent the discharge of any of the excretions, especially of urine or feces. Includes bedwetting. Common in CHARGE.

infections: Children with CHARGE Syndrome tend to be very sickly, especially in the early years. The most common illnesses are recurrent ear infections and pneumonia. The pneumonias tend to become less frequent after two to three years of age. The ear infections and ear drainage may last well into the teens.

inguinal hernia: also known as a "rupture". This is a lump noted in the groin and is actually a small loop of intestine sticking out a small hole connecting the inside of the abdomen to the groin. If the intestine gets stuck there, gangrene can occur, so preventive surgery is necessary.

iris: the colored part of the eye, with the pupil in the center. A coloboma of the iris results in a keyhole-shaped pupil. Common in CHARGE.

IUGR: Intrauterine growth retardation. Slower than expected growth of a baby before birth. At birth, it will show up as low birth weight. Rare in CHARGE.

K

kidney abnormalities: 40% of children with CHARGE have kidney abnormalities. Kidney abnormalities seen in CHARGE include hydronephrosis, small or absent kidney, posterior urethral valves, and kidney reflux. See Renal Section.

kidney reflux: back flow of urine into the kidney. This can result in eventual damage to the kidney. IVP is often needed to confirm reflux in the kidneys.

L

labia: female genital folds. May be smaller than normal in girls with CHARGE.

laryngomalacia or laryngotracheomalacia: floppy airway. The presence of soft laryngeal cartilage, most often seen in the epiglottis of young children. Very common in CHARGE, and contributes to surgical risks of anesthesia, breathing problems and swallowing problems.

larynx: the organ of voice production; the part of the respiratory tract between the pharynx and the trachea; it consists of a framework of cartilages and elastic membranes housing the vocal folds and the muscles which control the position and tension of these elements. May be weak in CHARGE.

limb defects abnormalities of arms, legs, hands, or feet. Occasional limb defects seen in CHARGE include thumb and forearm abnormalities. See Muscles and Bones Section.

M

macula: the center of the retina of the eye. The macula is responsible for seeing details. Coloboma of the retina can result in legal blindness.

Medical Geneticist: physician with medical training (most often in pediatrics or OB/Gyn) with subspecialty training in Medical Genetics or (more recently) physician with residency training in medical genetics and certification by the American Board of Medical Genetics of the AMA.

mental retardation: The intelligence in children with CHARGE syndrome ranges from normal to severe mental retardation. Intelligence is very hard to estimate and in fact is often underestimated, especially when children are young. Vision and hearing problems can delay speech and development. Special testing methods are needed to evaluate children with sensory deficits (See Assessment Sections). In addition to the sensory deficits, so many children with CHARGE spend a large portion of their early years in the hospital. Nevertheless, it is important to have your child evaluated early and often to help set up the most appropriate educational program possible. Evaluations should be done by specialists with experience testing children with sensory deficits (hearing and vision loss).

microcephaly: unusually small head

micrognathia: small jaw or chin

micropenis: abnormally small penis. See Genital Section.

microphthalmia/microphthalmos: abnormal smallness of the eye. A severe coloboma can result in microphthalmia or anophthalmia.

midface: the middle of the face, especially the cheekbone area. May be flattened or small in CHARGE.

Mondini defect: an abnormal opening from the semicircular canal into the middle ear. This is a potentially treatable (with surgery) cause of balance problems.

MRI: magnetic resonance imaging; a diagnostic form of imaging (of the brain or other body parts) Unlike conventional radiography or CT, MRI does not expose patients to ionizing radiation. In addition, it can provide superior 3-D images of the body's interior, delineating muscle, bone, blood vessel, nerve, organ, and tumor tissue. Often used to describe the exact choanal atresia and/or inner ear abnormalities in CHARGE.

N

nares: nostrils. May be small or appear pinched in CHARGE.

Neurologist: physician who specializes in the nervous system.

NG tube: nasogastric tube. Feeding tube which is put through the nose, down the throat into the stomach for feeding.

nystagmus: involuntary movements of the eyeball, most often side to side or in circles.

O

olfactory nerve: cranial nerve I, responsible for smell. Often absent or abnormal in children with CHARGE.

omphalocele: failure of the abdominal wall to close properly around the umbilical cord during fetal development. Often some of the intestines are outside the body. This can usually be corrected surgically. An umbilical hernia is a very mild omphalocele.

Ophthalmologist: MD who specializes in the eyes. Children with CHARGE should be evaluated by a Pediatric Ophthalmologist.

optic nerve: cranial nerve VII. Abnormalities of cranial nerve VII result in facial palsy.

optometrist: technician who specializes in detecting problems of visual acuity.

ossicles: tiny bones in the middle ear: anvil, stapes and hammer.

ossicular malformation: malformation of the small bones of the middle ear (hammer, anvil and stapes). Ossicular malformations are very common in CHARGE and result in conductive hearing loss.

OT: occupational therapist. A specialist in the development of fine motor, social, and adaptive skills.

otitis media: ear infections. These can occur when fluid accumulates in the middle ears, behind the eardrums. If the fluid is infected, the child will complain of pain and there will be hearing loss. If the fluid is not infected, the child usually will have no pain, but may still have significant hearing loss. In CHARGE Syndrome, otitis media often lasts into the teenage years and requires constant medical care (see PE tubes).

Otolaryngologist: specialist in hearing, either MD (ENT specialist) or PhD (Audiologist)

P

PAX2: group of genes responsible for orchestrating development of eyes, ears, and kidney. No PAX2 gene abnormalities have yet been detected in children with CHARGE.

palmar crease: referring to the palm of the hand. Children with CHARGE often have an unusual “hockey-stick” palmar crease. See photos in Diagnosis Section.

parathyroid gland: adjacent to the thyroid gland; one of two small paired endocrine glands. They secrete parathyroid hormone that regulates the metabolism of calcium and phosphorus.

PE Tubes: tiny polyethylene (plastic) tubes which can be surgically placed in the eardrum to drain the excess fluid from behind the drum. This will help prevent hearing loss caused by recurrent otitis media. Often several sets are needed over many years in children with CHARGE.

perseverative behavior: repetitive, repeated behavior, often seen in children with sensory deficits.

photophobia: intolerance to light, especially bright lights

pinna: external ear. Often very unusual in CHARGE.

pituitary abnormalities: The pituitary is a gland at the base of the brain which produces several important hormones which help control growth, thyroid and sex gland function, and steroid production. The pituitary gland does not function properly in some children with CHARGE. This can result in deficiencies in growth hormone and/or in the sex hormones which cause puberty. If left untreated, these children will be short and will not develop secondary sex characteristics. Hormone therapy is available for these problems.

polyhydramnios: excess amount of amniotic fluid. In pregnancies where the baby has CHARGE, polyhydramnios can related to choanal atresia and/or swallowing abnormalities.

postnatal: occurring after birth

posterior urethral valves: small pieces of tissue which prevent urine from flowing out of the bladder through the urethra to the outside. This can cause back-up of urine and damage to the kidneys. Surgery is usually necessary.

prenatal: preceding birth; antenatal

proprioception: A sense or perception, usually at a subconscious level, of the movements and position of the body and especially its limbs, independent of vision; this sense is gained primarily from input from muscles and the vestibular (balance) apparatus.

ptosis: droopy eyelids, caused by a facial nerve problem in CHARGE syndrome.

R

renal: nephric; relating to the kidney.

retarded development: see Developmental delay and Mental retardation.

retina: The part of the eye which receives light and transmits electrical signals to the brain, resulting in vision. Colobomas of the retina will result in blind spots and/or visual acuity (sharpness) abnormalities.

retinal detachment: separation of the retina from the choroid. This can result in blindness.

retinoic acid Vitamin A acid; used topically in the treatment of acne. See Accutane.

S

sclera: whites of the eye

scoliosis: curvature of the spine. Common in CHARGE at any age.

sedation: medication given to calm and/or quiet a child to make testing easier and/or more accurate. Children with CHARGE can have unusual (more or less extreme) reactions to sedation.

semicircular canals: part of the inner ear involved in balance

sensorineural hearing loss (nerve deafness): hearing impairment due to disorders of the cochlear division of cranial nerve VIII (auditory nerve), the cochlea, or the mitochondrial nerve tracts, as opposed to conductive hearing loss. Very common in CHARGE.

sensory deficit: vision loss and/or hearing loss.

sonogram: ultrasound. These terms can be used interchangeably

short stature: can be caused by a lack of growth hormone (see pituitary abnormalities). Some children with CHARGE Syndrome have normal levels of growth hormone but still have short stature from an unknown cause.

stapes: the smallest of the three auditory bones in the middle ear.

stapedius tendon: stapedius muscle; dampens vibration of the stapes by drawing head of stapes backward as a result of a protective reflex stimulated by loud noise. The stapedius tendon is often abnormal in CHARGE.

strabismus: crossed eyes

stridor: a high-pitched, noisy respiration, like the blowing of the wind; a sign of respiratory obstruction, especially in the trachea or larynx.

swallowing difficulties: Many children with CHARGE Syndrome have trouble coordinating the muscles used for normal sucking and swallowing, even in the absence of other problems such as esophageal atresia or T-E fistula. This may be due to abnormalities in cranial nerves IX and X. The incoordination can lead to gagging, apnea (breathing stops), and pneumonia (food inhaled into the lungs, causing infection). In some cases, tube feeding (gastrostomy) is used until the child is able to learn to swallow. Children with CHARGE may not learn to swallow effectively until they are 5 or 6 years old. See Feeding Section.

syndrome: the aggregate of signs and symptoms associated with any morbid process, and constituting together the picture of the disease. As in CHARGE syndrome.

T

teratogen: a drug or other agent that causes abnormal fetal development, for example, Accutane. There are no teratogens known to cause CHARGE.

thymus: a gland in the lower part of the neck, that is necessary in early life for the normal development of immunological functions. It reaches its greatest relative weight shortly after birth and its greatest absolute weight at puberty; it then begins to involute, and much of the lymphoid tissue is replaced by fat. It is abnormal in children with DiGeorge sequence.

ToF: Tetralogy of Fallot. Complex heart defect often seen in CHARGE.

tracheo-esophageal (T-E) fistula: an abnormal connection between the trachea (wind pipe) and esophagus (food pipe). If it is not corrected surgically, food will get into the lungs and cause choking and/or pneumonia.

tracheomalacia: floppy airway due to weakness of elastic and connective tissue of the trachea. Common in CHARGE.

tracheostomy/tracheotomy: the operation of opening into the trachea to permit breathing directly into the throat, usually intended to be temporary. Often necessary in CHARGE.

U

ultrasound: imaging procedure which uses high-frequency sound waves to get an image of a fetus during pregnancy. Echocardiography also uses ultrasound to image the heart.

umbilical hernia: see omphalocele.

unilateral: affecting one side only.

URI: upper respiratory infection

urinary tract malformations: The urinary tract malformations seen in CHARGE Syndrome include posterior urethral valves, abnormal kidney shape or location, and backup of urine from the bladder into the kidney (reflux).

UTI: urinary tract infection

V

VACTERAL/VATER association Acronym for Vertebral defects, Anal atresia, Cardiac defects, TracheoEsophageal fistula and/or esophageal atresia, Renal anomalies and Limb defects. Many features overlap with those of CHARGE, but can usually be distinguished by a Medical Geneticist.

VCF: velocardiofacial syndrome; Shprintzen syndrome. Syndrome consisting of cleft palate, heart defects, learning disabilities, and distinct physical features. The overwhelming majority of children with VCF have a microdeletion of chromosome 22 detectable by FISH. Although many features overlap, it is distinct from CHARGE. The shapes of the ears, face, and hands are distinct from CHARGE. To date, no one with a definite diagnosis of CHARGE has been positive for the chromosome 22 deletion associated with VCF.

velopharyngeal: pertaining to the soft palate (velum palatinum) and the posterior nasopharyngeal wall.

ventricles (head, heart) A normal cavity or space. In the heart, the spaces through which blood is pumped (left and right ventricle). In the brain, fluid-filled spaces (lateral, third, and fourth ventricles - see hydrocephalus).

vestibular: relating to a vestibule, especially the vestibule of the ear. The vestibule of the ear is responsible for one component of balance. Vestibular abnormalities are common in CHARGE and contribute to delay in motor milestones such as sitting and walking.

VER: visual evoked responses; used to measure electrical activity of the parts of the brain used in vision.

videofluoroscopy: examination x-ray, using the fluoroscope, using an image intensifier and television camera for image detection and a video monitor for display to get live action pictures of , e.g. swallowing.

visual field: the portion of side view, and top and bottom view, in which a child can see or has functional vision. Colobomas cause visual field defects (blind spots).

webbed neck: wide or broad neck, often somewhat short. Many children with CHARGE have a webbed neck or just a short neck.