GLOSSARY

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Alleles: Alternative forms of a genetic locus; a single allele for each locus is inherited separately from each parent.

Articulation: The process of the structures of the speech mechanism (lips, tongue, teeth, etc.) coming into contact or close proximity to alter the sound emitted by the vocal cords, thus producing different speech sounds. An articulation disorder occurs when the wrong contact is made, or completely omitted. Misarticulations may include distortions, omissions, or even additions of sounds.

Attention deficit hyperactivity disorder (ADHD): A disorder characterized by symptoms of inattention, impulsivity, and hyperactivity.

Base Pairs(bp): Chemicals (e.g., adenine and thymine or guanine and cytosine) that make up a DNA molecule.

Bipolar mood disorder: Varying degrees of grandiosity, decreased need for sleep, pressured speech, flight of ideas, distractibility, increased inactivity, marked impairment in occupational functioning,
increase in activies resulting in painful consequences (e.g., spending huge amounts of money). Certain characteristics must be excluded; no delusion of hallucination during period when in abnormal mood state.

Catch 22 Sometimes used in the UK for VCFS, although this term is not felt to be appropriate by many, who feel it is in poor taste, and appears to trivialize a serious matter.

Chromosomes: The genetic structure of cells containing the DNA that bears in its nucleotide (chemical) sequence the linear array of genes.

Cleft palate (velum): The failure of embryonic (first 8 weeks of development) fusion of the hard and/or soft roof of the mouth and floor of the nose. See also submucous cleft and occult submucous cleft.

Conductive hearing loss: Hearing disorder caused bye a disruption in the sound-conducting mechanism of the outer or middle ear so that a reduced level of sound reaches the inner ear and the nerve conducting sound to the brain. See also sensori-neural hearing loss.

Conotruncal heart anomaly: Congenital heart defects particularly involving the ventricular (lower chambers) outflow tracts of the heart includes sub arterial ventricular septal defect, pulmonic valve atresia and stenosis, tetralogy of Fallot and truncus arteriosus.

Consultand: Individual seeking genetic counseling.

Deletion: 5 to 10 million missing base pairs of DNA allow for detection on high resolution karyotypes, while smaller changes, those involving less than 1 million and up to several million base pairs of DNA, need special molecular techniques to observe (e.g., FISH).

Depression: Chronic mood disorder.

DNA (deoxyribonucleic acid): The molecule that encodes genetic information.

Fluorescence *in c~itu* hybridization (FISH): An approach to mapping human genes using fluorescein radioactive tags.

Gene: The unit of heredity.

Gene expression: The process by which a gene's coded information is converted into the structures
present and operating in the cell.

Genetic code: The triplet coded sequence of the subunits of DNA (there are thousands of base pairs linked to form a DNA molecule).

Genome: All the genetic material in the chromosomes of a particular person.

Genotype: The ordered sequence of the genes of an individual.

Glottal stop: A type of articulation error, often confused for an omission, where a sound is articulated
in the larynx (sounding like a grunt) and substituted for a sound normally articulated in the oral cavity. In VCFS, glottal stops are often substituted for all other consonants except m, n, and ng. Glottal stop substitutions would make the word "puppy" sound like "uh-ee".

Human gene therapy: Insertion of normal DNA directly into cells to correct a genetic defect.

Hypernasality: Excessive nasal resonance during speech. In all languages, there are far more sounds produced without nasal resonance than with (there are only three nasal sounds in English: m, n, and ng). When nasality occurs on sounds which normally should have none (such as p, t, s, g, etc.), then this is referred to as hypernasality.

Language: A system for using of symbols, sounds, or signs to convey meaning to someone else and expressing ideas for the purpose of communication.

Karyotype: A picture of an individual's chromosomes arranged in a standard format showing the number, size and shape of each chromosome type.

Multi-view videofluoroscopy: A diagnostic procedure using X-rays to obtain a three dimensional viev ofvelopharyngeal function (see below) and record them on video tape. Barium contrast is used to help see the movements of the speech musculature. At least two different views must be used for adequate diagnosis (frontal view and lateral view).

Mutation: Any heritable change in DNA sequence.

Nasopharyngoscopy: A diagnostic procedure used to assess velopharyngeal function during speech (see below).

Nasopharyngoscope: A fiber optic instrument (an endoscope) which can be inserted through the nose to see the pharynx and larynx(voice box). The examination is video taped through the endoscope.

Occult submucous cleft palate: Not all submucous clefts have a bifid uvula as a clue to diagnosis. Or, findings may be normal, yet the balate may still be missing muscle tissue which results in abnormal function. This is referred to as an "occult" cleft (meaning mysterious). Occult clefts can be diagnosed with nasopharyngoscopy because the nasal surface of the palate is always abnormal in occult clefts.

Occult submucous cleft palate: Basically the same anomaly as in submucous cleft palate, but there is no bifid uvula, no zona pellucida, and no notch in the hard palate. In other words, the palate looks normal on oral examination, but speech is hypernasal. Occult submucous cleft palate is diagnosed by nasopharyngoscopy.

Pharynx: The inside of the throat, i.e., the tube consisting of the region behind the nasal cavity, behind the tonsils and palate, and just above the larynx. The pharynx is surrounded by an intricate set of muscles which are active during speech and are also used for swallowing, but using a different physiological mechanism than speech.

Pharyngeal flap: Soft tissue layer of mucous membrane and its underlying muscle, elevated from the back wall of the throat and then inserted into the top surface of the palate.

Phenotype: The observable physical and behavioral characteristics of a person. "Observable" refers to the ability to assess them either by physical examination, or by use of special diagnostic tests, such as lab tests, X-rays, IQ tests, or physical measurements. Height, ear size, head shape, learning ability, and temperament are all phenotypic features.

Polymorphism: Difference in DNA sequence among individuals.

Proband: First family member coming to medical attention with a specific known genetic condition.

Psychosis: A severe form of ps ychiatric disorder characterized by a disorganization of normal mental processes, such as thinking, personality and temperament.

Sensori-neural hearing loss: Hearing dissorder caused by an abnormality in the sound sensing mechanism (the inner ear) or the nerve which conducts sound to the brain.

Submucous cleft palate: In cleft palate, there is an observable opening between two separated halves of what should normally be one solid palate (roof of the mouth). In submucous cleft, the skin of the palate is intact, but the muscle and perhaps even the bony tissue underneath the skin is separated. Submucous cleft palate is easily detectable on oral examination because of the presence of a split in the uvula (bifid uvula), a notch in the bone of the hard palate (which can be palpated with the finger), and even an
observable groove down the center of the soft palate, often called a "zona pellucida" or translucent zone caused by an absence of muscle tissue.

Submucous cleft palate: Refers to a cleft which is "submucous," meaning underneath the mucous
membrane. This refers to a separation of the muscles in the soft palate and perhaps the bone of the hard palate, even though the skin envelope (the mucous membrane) is intact. Submucous lceft palate is often signalled by a cleft of the uvula, known as a bifid uvula and is usually easily diagnosed by experienced clinicians by oral examination.

Velopharyngeal valve: The area behind the palate, in the pharynx, which opens and closes during speech to modulate the flow of air through the mouth and nose during speech. The overwhelming majority of speech occurs with the velopharyngeal valve closed.

Velopharyngeal insufficiency (VPI): Not to be confused with hypernasalit, though VPI may result in hypernasalit, VPI is the failure for the muscular portion of the soft palate (velum) and the throat (pharynx) to close completely during normally nonnasal speech. If ari leaks into the nose during speech, this is VPI. However, it is possible to have VPI without resulting hypernasality, but it is not possible to have hypernasality without VPI.

Voice: Sound usually produced by the vocal cords which then resonates in the throat above the larynx.