

**Appendix: Useful definitions for the Geneticist (adapted from American-Speech-Language-Hearing Association definitions, [www.asha.org](http://www.asha.org))**

Speech: the verbal means of communicating. This is made up of articulation (making sounds), voice (producing sounds using the vocal folds and breathing) and fluency (rhythm of speech).

Language consists of socially shared rules that include what words mean, how to make new words, how to put words together and what word combinations are best in what situations.

Speech and language disorders can occur in the same person:

Receptive language disorder: difficulties understanding others (receptive language)

Expressive language disorder: difficulties sharing thoughts, ideas, and feelings

Speech disorder: Difficulties to produce speech sounds correctly or fluently, or voice problems.

Receptive language delay: difficulties understanding the meaning of gestures, following directions, answering questions, identifying objects and pictures and taking turns when talking with others.

Expressive language delay: difficulties with asking questions, naming objects, using gestures, putting words together into sentences, learning songs and rhymes, using correct pronouns or knowing how to start a conversation and keep it going.

Childhood apraxia of speech (CAS): a motor speech disorder characterized by difficulties saying sounds, syllables, and words. The individual knows what he or she wants to say, but the brain has difficulty coordinating the muscle movements necessary to say those words.

Dysarthria: a motor speech disorder caused by impaired movement of the muscles used for speech production, including the lips, tongue, vocal folds, and/or diaphragm. The type and severity of dysarthria depend on which area of the nervous system is affected.

Speech sound disorder: difficulties with articulation (making sounds) and phonological processes (sound patterns) leading to reduced intelligibility (degree to which speech can be understood).

Language-based learning disabilities: problems with age-appropriate reading, spelling, and/or writing which can occur in children with normal intelligence and in children with intellectual disability. For example dyslexia.

Selective mutism: a child who does not speak in certain situations such as at school. The child speaks at other times, such as at home or with friends.