

Glossary

Following is a list of common terms that you may come across in your search for information. Some of these terms are used to describe conditions that explain Rett syndrome. Other terms are used to rule out Rett syndrome.

A | B | C | D | E | F | G | H | I | J | K | L | M
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A

Abduction: movement of a limb or any other part away from the midline of the body.

Absence seizure: a generalized seizure in which consciousness is altered, formerly called “petit mal.”

Acetylcholine: a neurotransmitter chemical.

ADA (Americans with Disabilities Act): federal legislation enacted in 1990, intended to protect all people with disabilities from discrimination.

Adaptive behavior: skills needed by a child to function effectively and appropriately for her age in the school, family, and community settings.

Adduction: movement of a limb or any other part toward the midline of the body.

Aerophagia: swallowing of air.

Alternative communication: a method which replaces traditional forms or methods already used by someone to communicate.

Ambulatory: able to walk.

Amino acids: the building blocks or chief structure of proteins; several are essential in human nutrition.

Anecdotal: Report of clinical experiences based in individual cases, rather than an organized investigation with appropriate controls.

Angelman Syndrome (Happy Puppet Syndrome): AS is a neurodevelopmental disorder caused by loss of paternally derived gene(s) on chromosome 15. It has many features that are similar to Rett syndrome including ataxia, lack of speech, seizures and microcephaly.

Ankle-foot orthosis (AFO): an orthopedic apparatus, most commonly a splint or brace, used to support, align, or correct deformities or to improve the function of the ankle/foot.

Annual review: a review of a child's special education program to be held at least once a year to assess progress and determine whether any program changes are necessary in the following year.

Anorexia: a severe loss of appetite.

Anticonvulsant: any medication used to control seizures.

Apnea: episodic arrest of breathing, seen during waking and not usually during sleep

Appropriate education: a program that is capable of meeting the educational needs of a child who has an educational disability.

Apraxia: difficulty with the usually automatic planning done by the brain to execute voluntary movements.

Asphyxia: interference with circulation and oxygenation of the blood that leads to loss of consciousness.

Aspirated: inhaled.

Aspiration: entry of liquids (saliva or drink) or solids into the airway.

Aspiration pneumonia: a lung infection caused by inhaling a foreign body, such as food, into the lungs.

Assay: a method or way of measuring chemical or biological compounds.

Ataxia: imbalance or lack of coordination of voluntary and involuntary muscles; shakiness on reaching or moving the trunk that is associated with malfunction in the cerebellum; unbalanced gait and jerky, uncoordinated movements.

Atom: a particle composed of a nucleus (protons and neutrons) and electrons. Atoms differ from one another by having different numbers of protons, neutrons and electrons. Groups of atoms bonded together are called molecules.

Atonic seizures: generalized seizures in which body tone is suddenly lost and the child falls to the ground or her head slumps forward.

Atrophy: wasting, from disuse; implies normal tissue to begin with.

Atypical: not typical; different.

Auditory evoked potential: a test to evaluate the processing of sound by the brain stem.

Augmentative communication: all forms of communication that enhance, replace or supplement speech and writing.

Aura: the start of a seizure; a peculiar feeling, sense of fear or unusual sensation in one part of the body.

Autism: a developmental disability that results in impairments in social interaction, verbal, and nonverbal communication and unusual behaviors and responses to the environment.

Automatisms: purposeless automatic movements that accompany a complex partial seizure, such as smacking the lips, chewing, or picking at the clothes.

Autonomic nervous system: the part of the nervous system that regulates certain automatic functions of the body, for example, heart rate, temperature, and bowel movements.

Autonomic responses: body functions that happen involuntarily, such as breathing, sweating, blood pressure, heart rate, and flushing of the face.

Autosome: any of the first twenty-two pairs of chromosomes; all chromosomes are autosomes, except for the sex chromosomes, X and Y.

Autosomal dominant trait: a genetic trait carried on the autosomes. The disorder appears when one of a pair of chromosomes contains the abnormal gene; statistically, it is passed on from the affected parent to half of the children.

Autosomal recessive trait: a genetic trait carried on the autosome. Both asymptomatic parents must carry the trait to produce an affected child. This child has two abnormal genes. The risk of recurrence is 25 percent. Rett syndrome is not felt to be autosomal recessive.

Axon: a nerve fiber.

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Babinski reflex: extension of the big toe upon stimulation of the sole of the foot. This abnormal response is found in individuals with pyramidal tract damage.

Barrett's esophagus: damage to the normal skin-like lining of the esophagus, which is replaced with a lining that resembles that of the stomach. It is caused by persistent reflux of acid from the stomach. The new lining can resist the gastric reflux, but inflammation at the upper end of the new lining may narrow the interior passageway of the esophagus.

Basal ganglia: several large masses of gray matter embedded deep within the white matter of the cerebrum.

Base: a flat ring structure, containing nitrogen, carbon, oxygen and hydrogen, that forms part of one of the nucleotide links of a nucleic acid chain. The four different bases are adenine (A), thymine (T), guanine (G), and cytosine (C).

Base pair: two bases, one in each strand of a double-stranded nucleic acid molecule that are attracted to each other by weak chemical interaction. Only certain base pairs form: A-T (or T-A), G-C (or C-G), A-U (or U-A).

Behavior modification: the systematic application of the principles of learning theory to change behavior by modifying events that precede or follow the behavior.

Beta endorphins: the body's own morphine-like substance.

Binding: attaching.

Biogenic amines: neurotransmitters dopamine, norepinephrine (also known as noradrenaline), serotonin, and a few others.

Biological marker: a scientific test which proves the presence of a condition.

Body jacket: a molded, padded thin plastic brace designed to apply gentle pressure in the proper areas to straighten the spine.

Bolus: clump; term used in tube feeding to indicate feeding given at one time.

Brain stem: the primitive portion of the brain that lies between the cerebrum and the spinal cord.

Brain stem auditory evoked response (BAER): a test to evaluate the processing of sound by the brain stem.

Breathholding: quick inspiration of a single full breath, followed by delayed expiration during which breathing stops. Also known as aerophagia.

Bruxism: tooth grinding, common in Rett syndrome.

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C

Carnitine: an amino acid which helps to transport long-chain fatty acids.

Cell: the smallest unit of living matter capable of self-perpetuation; an organized set of chemical reactions capable of reproduction. A cell is bounded by a membrane that separates the inside of the cell from the outer environment. Cells contain DNA (where information is stored), ribosomes (where proteins are made), and mechanisms for converting energy from one form to another.

Cell membrane: the membrane that contains the cell components and the substance in which they float.

Central apnea: cessation of breathing movement at the end of expiration.

Central nervous system (CNS): the portion of the nervous system that consists of the brain and spinal cord. It is primarily involved in voluntary movement and thought processes.

Cerebral cortex: the outer part of the cerebrum.

Cerebral palsy: a disorder of movement and posture due to a non-progressive defect of the immature brain.

Cerebrospinal fluid (CSF): the normally clear fluid that surrounds the brain and spinal cord.

Cerebrum: the largest region of the brain, made up of four lobes and connected by the corpus callosum.

Child Find: the continuous efforts of a school district to identify all children from birth to age twenty-one in its area who may have educational disabilities and need special education.

Chiropractic: a system of treating diseases by manipulation, mainly of the vertebrae of the backbone. It is based on the theory that nearly all disorders can be traced to the incorrect alignment of bones, with consequent malfunctioning of nerves and muscles throughout the body.

Chromatin: chromosomes in their less compact state. During this stage, they appear under a microscope as long tangled threads.

Chromosome: a subcellular structure containing a long, discrete piece of DNA plus the proteins that organize and compact the DNA.

Chromosome analysis: a study of the forty-six potential chromosome structures that consist of the genetic code for our physical and biochemical traits.

Chronological age: the child's actual age in years and months.

Classic: typical.

Clone: 1) noun, a group of identical cells, all derived from a single ancestor. 2) verb, to perform or undergo the process of creating a group of identical cells or identical DNA molecules derived from a single ancestor.

Code (genetic): the system in which the arrangement of nucleotides in DNA represents the arrangement of amino acids in protein.

Clinical diagnosis: a conclusion based on findings from the patient's history and physical exam, usually not as definitive as a laboratory-made diagnosis would be.

Clonus: alternate muscle contraction and relaxation in rapid succession.

Cognitive disability: an educational term usually synonymous with the medical term, mental retardation.

Cognitive skills: thinking abilities, most often represented by IQ scores; difficult to assess in Rett syndrome.

Complex partial seizure: a seizure which involves only part of the brain and which alters consciousness or awareness.

Computerized axial tomography (CAT scan): an X-ray which is used to examine soft tissues in the body. The technique involves the recording of "slices" of the body with an X-ray scanner and then integrating them by computer to give a cross-sectional image. Within the skull, it can be used to reveal the normal anatomy of the brain, and to distinguish pathological conditions such as tumors, abscesses, and hematomas.

Congenital: present at or before birth.

Constipation: a condition in which bowel evacuations occur infrequently, or in which the feces are hard and small, or in which passage of feces causes difficulty or pain.

Contracture: irreversible shortening of muscle fibers that causes decreased joint mobility.

Controls: study subjects; individuals without disease who are used to compare normal values.

Convulsion: older term for a seizure; it most commonly involves a series of involuntary contractions of voluntary muscles.

Corpus callosum: the bridge of white matter connecting the two hemispheres of the brain.

Cortex: the gray matter that lies at the outer portion of the cerebrum.

Cure: the complete eradication of symptoms so that no symptoms are discernible, even by a doctor and a battery of tests, and no impact will be felt in future generations.

Cyanosis: a bluish discoloration of the skin and mucous membranes resulting from an inadequate amount of oxygen in the blood.

Cytoplasm: the jellylike substance that surrounds the nucleus of a cell.

Cytological: relating to the study of cell structure, often using microscopy. A commercial cytology laboratory examines the structure of chromosomes for genetic disorders.

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D

Deceleration: reduced velocity. In Rett syndrome, deceleration often refers to slowing of the growth rate of the head (circumference).

Degenerative CNS disorder: a condition leading to progressive disability and death usually associated with significant, identifiable changes in the brain. Rett syndrome is no longer thought to be a degenerative CNS disorder.

Deletion: loss of genetic material from a chromosome.

Dementia: marked decline in intellectual ability; loss of cognitive skills. In Rett syndrome, loss of cognitive skills will have a different mechanism than aging-related dementias like Alzheimer's.

Dendrite: one of the shorter branching processes of the cell body of a neuron, which makes contact with other neurons at synapses and carries nerve impulses from them into the cell body.

Deoxyribonucleic acid (DNA): a fundamental component of living tissue; it contains the genetic code.

Developmental arrest: after normal development, a complete cessation of developmental progress.

Developmental delay: a delay in the development of skills and abilities which usually would have developed by a certain age.

Diagnostic test: a test that provides an in-depth assessment of a skill area, including strengths and weaknesses and error patterns.

Differential: a list of conditions that could possibly cause the patient's presenting problem. Differential diagnoses for Rett syndrome include cerebral palsy and autism.

Diffuse: widely spread, as in diffuse cerebral impairment, which means that many areas on both sides of the brain are affected, not a localized problem.

Distal: situated away from the organ or point of attachment or from the median line of the body.

DNA: Deoxyribonucleic acid. DNA is a spiraling, ladder-like(helical) molecule that is the carrier for the genetic code. It is usually found as two complementary chains and is often hundreds to thousands of times longer than the cell in which it resides (it is tightly wrapped to fit inside). The links or subunits of DNA are the four nucleotide (deoxyadenylate, deoxycytidylate, deoxythymidylate, deoxyguanylate). The precise arrangement of these four subunits is used to store all information necessary for life processes. DNA is also found in mitochondria, the rod-like structures outside the nuclei of cells that function as a primary source of cellular energy.

Dominant: a “strong” gene whose effect will appear whether its partner gene is of the same type or different.

Dorsiflexion: backward flexion of the foot or hand or the toes and fingers.

Due process: a system of procedures designed to ensure that individuals are treated fairly and have an opportunity to contest decisions made about them. The due process requirements of Public Law 94-142 and Section 504 are intended to safeguard the right of children who have disabilities to a free, appropriate, public education.

Duodenum: upper part of the small intestine.

Dysmotility: impaired movement, usually used in reference to parts of the body that work automatically, e.g., gastric dysmotility refers to impaired emptying of the stomach.

Dyspraxia: partial impairment of the ability to perform coordinated motor movements.

Dystonia: alteration in muscle tone, usually referring to muscle cramps/spasms of muscles close to the midline of the body (neck, shoulders, hips).

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E

Early intervention: the provision of educational services at an early age for children with learning difficulties to avoid more serious problems later in life.

Electrocardiogram (EKG or ECG): the tracing made by an instrument for recording the changes of electrical potential occurring during the heartbeat.

Electroencephalogram (EEG): a test to measure and record brain waves. In Rett syndrome, the EEG is almost always

abnormal, although clinical seizures are not always present.

Electromyogram (EMG): a test in which wire probes which detect muscle contraction are inserted into a muscle. The muscle is then stimulated electrically to initiate a contraction and the response is recorded. The test allows monitoring of muscle contractions, timing, and efficiency of the response to a known stimulus.

Electronic communication device: a communication system which is computer chip based and contains messages which are words or phrases designed for the user of the device. Pictures, small objects, or words are placed on the device as cues to where messages are located.

Empirical: Pertaining to, or founded upon, experiment or experience.

Encephalopathy: an indication that something is wrong in the brain.

Encode: to contain a nucleotide sequence specifying that one or more specific amino acids be incorporated into a protein.

Endometrial ablation: a laser procedure to remove the lining of the uterus, which results in total or partial cessation of menstrual bleeding.

Endorphins: the body's natural opiates, probably involved in the perception of pain and pleasure.

Endoscopy: the use of an instrument to visualize the interior of a hollow organ, such as the esophagus; used to detect gastroesophageal reflux (GER).

Enzyme: a protein molecule specialized to accelerate a biological chemical reaction without itself being consumed. Generally, enzyme names end in -ase.

Epiglottis: a lid-like structure that hangs over the entrance to the windpipe and prevents aspiration into the lungs during swallowing.

Epilepsy: recurrent seizures, excluding ones caused solely by fever.

Epileptiform patterns: patterns which resemble those of a seizure.

Equilibrium: balance.

Equinus: involuntary extension of the foot downward, due to a tight or overactive heelcord.

Esophagitis: inflammation of the esophagus.

Esophagus: tube extending from the pharynx to the stomach.

Etiology: cause.

Evaluation: the process by which a team of professionals gathers information about a child's skills, deficits, aptitudes, interests, and personality variables from a variety of sources including testing, observation, and other procedures to guide decisions about the child's educational program. Often used interchangeably with "assessment."

Exon: a region of RNA that encodes a portion of a protein.

Expressive language: communication that is given out to others.

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F

Fine motor coordination: the ability to use the small muscles to accomplish tasks requiring precision.

Flexion: the bending of a joint so that the bones forming it are brought toward each other.

Flexor: a muscle whose primary function is flexion at a joint.

Formal test: a standardized evaluative measure which has explicit methods for administration and scoring and for which norms are available.

Forme fruste: incompletely expressed (a frustrated form), as in a Rett variant who has most but not all of the classical characteristics.

Fortunate activation: females usually randomly inactivate one of the two copies of the X- chromosome in each cell. Fortunate activation refers to a situation where a female who carries an abnormal gene on one X-chromosome silences the abnormal copy, leaving the normal copy to function. It protects her from symptoms of the X-linked disease, but does not prevent her from passing it on.

Frontal lobe: front part of the cerebrum; important for voluntary muscle movement and memory.

Fundoplication (fundal plication): an operation in which the opening from the esophagus to the stomach is closed.

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G

Gagging: voluntary or involuntary rhythmic movements of the back of the mouth near the epiglottis; protects airway from a bolus; usually triggered by food near the back of the tongue that does not trigger the swallowing reflex.

Gait: manner or style of walking.

Gastroesophageal reflux disease (GER): the backward flow of stomach contents from the stomach back into the esophagus.

Gastroenterologist: a doctor who specializes in disorders of digestion.

Gastrostomy (G-tube): a surgical creation of an artificial opening into the stomach through the wall of the abdomen.

Gel electrophoresis: a method for separating molecules based on their size and electric charge. Molecules are driven through a gel by placing them in an electric field. The speed at which they move depends on their size and charge.

Gene: a small section of DNA that contains information for construction of one protein molecule.

Gene cloning: a way to use microorganisms to produce millions of identical copies of a specific region of DNA.

Gene expression: the process of making the product of a gene; information is transferred, via messenger RNA, from a gene to ribosomes, where a specific protein is produced.

Generalized seizure: a seizure involving the whole brain.

Gene therapy: a process of altering genes in living persons through the administration of specific regions of DNA.

Genetic engineering: the manipulation of the information content of an organism to alter the characteristics of that organism. Genetic engineering may use simple methods such as selective breeding or complicated ones such as gene cloning.

Genetic map: a representation of DNA in which the relative position of regions is determined by the frequency of genetic recombination between observable traits.

Genome: the genetic information of an organism or virus; for organisms with two pairs of each chromosome, the genome refers to the information in one set.

Genotype: genetic profile or makeup of an individual.

Germ cells (gametes): reproductive cells. Male gametes are called sperm. Female gametes are called eggs.

GI: gastrointestinal.

Global: affecting all areas, e.g., global developmental delays.

Grand mal: old term for tonic-clonic; a form of seizure in which there is a sudden loss of consciousness immediately followed by a generalized convulsion.

Gray matter: the parts of the brain that contain the cell bodies of nerve cells (neurons).

Gross: on a large scale; not to be confused with the common term for “yucky.” “Grossly normal,” means “after less than in-depth inspection, this appears to be normal.” “Gross anatomy” refers to the study of organs as they appear to the naked eye, without the benefit of a microscope.

Gross motor coordination: the ability to use the large muscles in a coordinated, purposeful manner to engage in such activities as walking and running.

Grossly normal: usually implies a less than detailed evaluation of the body part or function being described.

Gyri: convolutions of the surface of the brain.

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Habilitation: teaching of new skills.

Handicapped child: the term used in federal and state law to designate a child who has a specific cognitive, physical, or emotional disability to the extent that specially designed instruction is necessary for her to learn effectively.

Head circumference: the size of the head, usually plotted on a growth curve to compare with other children of the same age; the increase in the head circumference in infancy and early childhood is a reflection of normal brain growth.

Heredity: the genetic transfer of characteristics from parents to offspring.

Hip dislocation: occurs at the ball and socket joint of the hip, when the ball is completely pulled out of the socket.

Hippotherapy: therapeutic horseback riding.

Hip subluxation: occurs at the ball and socket joint of the hip, when the ball is partially pulled out of the socket.

Histones: members of a small class of eukaryotic chromosomal proteins that wrap DNA into ball-like structures called nucleosomes.

Homebound instruction: temporary instruction at home, provided if a child is unable to attend school for medical reasons or if the school is in the process of arranging a special education placement.

Human genome: the information content of one set of human chromosomes.

Hydrotherapy: water therapy, such as provided in a whirlpool bath or warm pool.

Hypertonia: high muscle tone, muscle tightness, or spasticity.

Hyperventilation: exaggerated inspirations followed immediately by equally exaggerated expirations (fast, deep breaths), contributing to a central apnea at the end.

Hypoplasia: underdeveloped tissue. Both atrophy and hypoplasia can lead to smaller than normal amounts of tissue, but atrophy implies that normal tissue was once present and hypoplasia implies that normal tissue never developed. Both processes appear to occur in the brains of girls with Rett syndrome.

Hypotonia: low muscle tone, not to be confused with muscle strength, implies a certain amount of looseness or floppiness of joints.

Hypothesis: A supposition that appears to explain a group of phenomena and is advanced as a basis for further investigation, a proposition that is subject to proof or to an experimental or statistical test.

Hypoxia: reduction of oxygen content in body tissues.

Hypsarrhythmia: an abnormality of the EEG, a wildly chaotic pattern with multiple spikes and slow waves.

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I

Ictal (ictus): an event. A seizure of any type is referred to as an ictus.

IDEA (Individuals with Disabilities Education Act): passed in 1997, this law was written to strengthen academic expectations and accountability for the nation's 5.4 million children with disabilities, and bridge the gap that has too often existed between what those children learn and the regular curriculum.

Idiopathic: of unknown cause.

Ileum: lower portion of the small intestine.

Incidence: the number of cases of something in a given population over a specific time (e.g., the number of cases of lung cancer in the United States in one year).

Inclusion: the placement of a child who has an educational disability in an instructional setting in which most students do not have disabilities, in a manner that is educationally and socially beneficial to the child.

Incontinence: absence of bowel or bladder control.

Independent evaluation: evaluation of a child by one or more professionals who have no formal relationship with the school district. Parents can request this evaluation if they disagree with the school's evaluation.

Independent living skills: skills needed to care for oneself and to function effectively in a community setting (including for example, personal hygiene, money management, cooking, and use of public transportation).

Individualized educational program (IEP): a written plan that a team composed of school staff, parents, and the child, if appropriate, develops for every special education student. Must include, at a minimum, the child's current educational strengths and weaknesses, goals and objectives, educational services, start-up dates for those services, and procedures for program evaluation.

Individualized habilitation plan (IHP): a written plan for someone over school age; used in adult programs.

Intelligence quotient (IQ): score on an intelligence test for which 100 is the mean. Indicates a child's test performance relative to other children of the same age.

Intelligence test: a test used to measure overall capacity for learning. In Rett syndrome, an adequate intelligence test has not been devised that can accurately measure understanding.

Intussusception: the slipping of a length of intestine into an adjacent portion, usually producing obstruction.

Intubation: the insertion of a tube through the nose or mouth into the trachea to provide artificial ventilation.

In utero: before birth.

Invasive test: a procedure or examination that requires that the body be entered in some way, either through a needle or with a tube.

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J

Joy: what you experience when your daughter lights up the room with a smile.

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K

Karyotyping: photograph of the chromosomal makeup of a cell. In humans, there are twenty-three pairs of chromosomes in the sex chromosomes, in this case, female. A few girls with Rett syndrome have had abnormal results. Karyotyping is a chromosome analysis, which looks for visibly missing or extra pieces of whole chromosomes; not the same as looking for a specific gene.

Ketogenic diet: a diet which provides the minimal amount of protein necessary for growth, no carbohydrates, and most of the calories from fats. The diet is used in seizure control.

Ketosis: the buildup of acid in the body due to starvation; important in the ketogenic diet.

Kindred cases: recurrences of Rett syndrome in a family, for example, sisters, cousins, etc.

Kinesthesia: the unconscious awareness of body parts in relation to movement.

Kyphosis: a spinal curvature as seen from the side, often termed "hunchback".

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L

Least restrictive environment (LRE): a standard established by Public Law 94-142 for special education placement. A child who has an educational disability must be allowed to participate in as much of the regular education program as is appropriate in view of her educational needs. The law holds that children with special needs must not be separated from students who do not have disabilities any more than is educationally necessary.

Lumbar puncture (spinal tap): the tapping of the subarachnoid space to obtain cerebrospinal fluid from the lower back region for examination.

Lysosomal enzymes: enzymes normally involved in the process of digestion.

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M

Magnetic resonance imaging (MRI): imaging procedure that uses the magnetic resonance of atoms to provide clear images on interior parts of the body.

Mediation: a process for settling disputes between parents and school districts through the intervention of a neutral third

party who tries to negotiate an agreement acceptable to all.

Medulla: the brain stem.

Melatonin: hormone which determines skin pigmentation. In humans, it is believed to play a role in the establishment of circadian rhythms.

Meiosis: the form of cell division that occurs during formation of reproductive cells to reduce the number of chromosomes from two to one of each type.

Meninges: the three connective tissue membranes that line the skull and vertebral canal and enclose the brain and spinal cord.

Mental age: a form for expressing a child's performance on an intelligence test. A child who receives an MA of 8-4 has achieved a score comparable to an "average" child of eight years, four months.

Mental retardation: a significant delay in the development of cognitive (problem-solving) skills that is associated with a significant delay in adaptive (use of intelligence in daily living) skills that occurs during childhood. The term "significant" refers to "statistically significant" when compared to other children of the same age. Usually measured by intellectual functioning at least two standard deviations below the mean, or average.

Messenger RNA: RNA used to transmit information from a gene on DNA to a ribosome, where the information is used to make protein.

Metabolites: the products of metabolism; in Rett syndrome articles, often seen in the phrase "biogenic amine metabolites," which refers to the normal breakdown products of the neurotransmitters dopamine, norepinephrine and serotonin. These can be measured in the cerebrospinal fluid directly.

Metabolic disorder: also referred to as an "inborn error of metabolism," these conditions are caused by impairment in a person's ability to process the breakdown products of protein, fat or carbohydrates. Your daughter may have had a metabolic workup when she first regressed or developed seizures.

Microcephaly: head circumference below normal for age (two standard deviations below the average); usually reflects lack of brain growth.

Mitochondria: a specialized intracellular structure that converts the chemical energy stored in food into a more useful form as molecules called ATP.

Mitosis: the type of cell division that results in exact duplicates of the original cells.

Molecule: a group of atoms tightly joined together.

Mucopolysaccharides: product of metabolism that may accumulate in cells and cause a progressive neurological disorder known as mucopolysaccharidosis; usually ruled out before the diagnosis of Rett syndrome is given.

Music therapy: the use of music by someone (music therapist) who is trained to use musical activities to teach non-music skills, such as communication, socialization, choice-making, and motor skills.

Mutation: a change in a gene, such as loss, gain, or substitution of genetic material that alters its function or expression by an incorrect amino acid sequence. This change is passed along with subsequent divisions of the affected cell. Gene mutations may occur randomly for unknown reasons or may be inherited.

Myelination: the production of a coating called myelin around an axon, which quickens neurotransmission.

Myoclonic: repetitive contraction of muscles; occurs in infantile spasms.

Myopathy: condition affecting the muscles.

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N

Nasogastric tube (Ng tube): a temporary plastic feeding tube placed through the nose down the esophagus and into the stomach for introduction of high caloric foods.

Negative test result: the test was normal (usually perceived by the patient to be a “positive” event).

Neurologist: a physician skilled in the diagnosis and treatment of disease of the nervous system.

Neurons: the nerve cells of the brain.

Neuropathy: any disease of the peripheral nerves, causing weakness and numbness.

Neurotransmitters: the chemicals used by nerves to “talk” to each other; released at the synapse that permits transmission from one nerve to another.

Neurotrophic: involved in the nutrition or maintenance of neural (brain) tissue. A classic example is nerve growth factor.

Nociception: the perception of pain, impaired in girls with Rett syndrome.

Nonspecific findings: those physical or laboratory results that can be seen in a variety

of conditions, for example, seizures are a nonspecific finding because they occur in many neurologic diseases, not just Rett syndrome.

Nucleotide: one of the building blocks of nucleic acids. A nucleotide is composed of three parts: a base, a sugar, and a phosphate.

Nucleotide pairs: two nucleotides, one in each strand of a double-stranded nucleic acid molecule, that are attracted to each other by weak chemical reactions between the bases.

Nucleus: the cell control center. It houses the chromosomes containing the genes.

Nystagmus: rapid involuntary movements of the eyes.

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O

Obstipation: severe and obstinate constipation; leads to fecal impaction.

Occipital lobe: one of the major divisions of each cerebral hemisphere.

Occupational therapy: treatment by an occupational therapist to improve an individual's ability to integrate different mental and motor processes in a purposeful and efficient manner. The occupational therapist concentrates on promoting, maintaining or restoring use of the body for daily living skills.

Opthamalogical: anything related to the eye.

Organic acids: error in metabolism of organic acids that can cause the child to develop symptoms of acidosis, coma, developmental and psychomotor retardation. Some forms of this disorder have been successfully treated by vitamin therapy both before and after birth.

Organomegaly: enlarged internal organs, not seen in Rett syndrome.

Oropharyngeal dysfunction: improper function of the mouth and pharynx.

Orotic acids: excessive excretion of orotic acids is indicative of a genetic metabolic disorder, characterized by physical and mental retardation; ruled out in Rett syndrome.

Orthopedic: relating to bones or joints.

Orthosis: an orthopedic appliance used to support, align, prevent, or correct deformities or to improve the functioning of movable parts of the body.

Osteotomy: surgical realignment of the bone; used for hip subluxation.

Osteoarthritis: inflammation of joints characterized by degenerative changes and sometimes increasing bulk in the bone and cartilage.

Osteopenia: deficient mineralization of bone.

Osteoporosis: loss of bone minerals.

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P

Palmar grasp: immature hand movement in which the palm rather than the fingertips makes contact with an object.

Paresis: weakness, e.g., quadriparesis means weakness in four extremities. Technically, quadriplegia means paralysis of all four extremities, but often the terms are used interchangeably.

Parietal lobe: one of the major divisions of each cerebral hemisphere, lying behind the frontal lobe, above the temporal lobe, and in front of the occipital lobe. It contains the sensory cortex and association areas of the brain.

Pathologic fractures: when minor trauma results in broken bones.

Pedigree: chart showing how members of various generations are related. Also known as a family tree.

Peripheral nervous system: the parts of the nervous system that are outside the brain and spinal cord.

Petit mal: old term for absence seizures.

Pharynx: a muscular tube lined with mucous membrane which extends from the beginning of the esophagus up to the base of the skull.

Phenotype: the clinical appearance of a patient, her historical and/or physical findings; in contrast to a patient's genotype, which is the genetic profile of a patient. Patients with the same phenotype can have different genotypes. Or, just because patients share the same clinical findings doesn't mean that they were caused by the same thing.

Phenylketonuria (PKU): Congenital absence of phenylalanine hydroxylase (an enzyme that converts phenylalanine into tyrosine). Phenylalanine accumulates in the blood and seriously impairs early neuronal development. The defect can be controlled by diet and is not serious if treated in this way.

PhProbe: a test for gastroesophageal reflux (GER), in which a probe is placed near the esophageal sphincter, and acid reflux is measured.

Physical therapy: Treatment by a physical therapist to improve an individual's motor skills and increase the strength and endurance of body parts.

Pincer grasp: refined, mature hand movement in which the thumb and first finger are used to pick up a small object.

Plantar flexion: bending of the toes or fingers downward toward the sole or palm.

Placement: the educational setting in which a student receives instruction.

Pneumonia: An acute infection of lung parenchyma including alveolar spaces and interstitial tissue.

Polymorphism: a normal variation in a gene.

Prenatal: before birth.

Probe: a DNA or RNA molecule, usually radioactive, that is used to locate a complementary RNA or DNA by hybridizing to it.

Protein: a class of long, chainlike molecules often containing hundreds of links called amino acids. Twenty different amino acids are used to make proteins. The thousands of different proteins serve many functions in the cell. As enzymes, they control the rate of chemical reactions, and as structural elements they provide the cell with its shape. Proteins are also involved in cell movement and in the formation of cell walls, membranes and protective shells. Some proteins also help package DNA molecules into chromosomes.

Positioning: physical management of posture and body alignment for daily living skills such as eating and standing up.

Positive findings: an abnormal test or finding (although usually not a positive event from the patient's point of view, a consistently positive test result in girls with Rett syndrome would lead to a diagnostic test for this disorder).

Positron emission tomography (PET scan): imaging study which uses radioactive labeled chemical compounds to study the metabolism of an organ.

Prevalence: the number of cases of something in a population at any given time (the number of girls with Rett in a country at any given time).

Procedural safeguards: legal regulations intended to safeguard t free, appropriate, public education, and to ensure that both child and parents receive the due process of law.

Prognosis: what is likely to happen.

Progressive: the problem gets worse with time. In Rett syndrome, this means that symptoms continue to develop over

time; it is not clear whether or not the underlying disease process is actively getting worse with time or whether the new symptoms represent the result of previous involvement of the brain.

Prolonged Q-T syndrome: when the resting period between heart beats is elongated. See Q-T interval.

Pronation: turned inward.

Prone stander: a piece of equipment that holds a person upright in a standing position. The support is under the front of the body and the individual is tipped forward slightly.

Proprioceptive input: unconscious information from the muscles and joints about position, weight or pressure, stretch, movement, and changes of position in space.

Pseudobulbar palsy: weakness of muscles coordinated by nerves originating in the brainstem; generally refers to coordination of mouth/throat muscles.

Public Law 94-142: in the USA, the primary federal legislative act involving the education of children who have educational disabilities. Called the Education for All Handicapped Act of 1975, this law aims to assure the availability of a "free appropriate public education" for every eligible child. It sets forth a range of school and parental responsibilities as well as procedural safeguards to ensure the due process of law.

Pseudo arthritis: failure of bone to fuse, sometimes requiring bone graft.

Pulmonary: pertaining to the lungs.

Pyramidal tract: a collection of nerve tracts in the brain stem.

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Q-T interval: a measurement from an EKG (electrocardiogram) that tells how long electricity takes to get through a portion of the heartbeat; a prolonged Q-T interval means that the impulse takes longer than expected and may be a factor in sudden, unexpected death. A Qtc interval means that the time has been corrected for how fast the heart is beating.

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R

Range of motion: the amount of movement available in a joint, measured in degrees. May be reduced in Rett syndrome, as seen in spasticity (as in tight heel cords) or by the rigidity or fixing (as in the shoulders and elbows). The motion may be temporarily reduced. If motion is limited for a long time, the limitation becomes structural because muscles and tendons become shortened.

Receptive language: communication that is received.

Recessive: a “weaker” gene whose effect will only appear when its partner gene is the same type.

Recombination: the breaking and rejoining of DNA strands to produce new combinations of DNA molecules. Recombination is a natural process that generates genetic diversity. Specific proteins are involved in recombination.

Related services: support services needed by a child to benefit from special education.

Residential placement: a placement, usually arranged and paid for by a state agency or the parents, where an individual with special needs resides.

Restriction mapping: a procedure that uses restriction endonucleases to produce specific cuts in DNA. The positions of the cuts can be measured and oriented relative to each other to form a crude map.

Retinopathy: disease of the retina (the back of the eye that registers visual signals); NOT seen in Rett syndrome.

Righting response: ability to return to upright after tilting.

Rigidity: abnormal stiffness of muscle.

RNA (ribonucleic acid): long, thin chainlike molecules in which the links or subunits are the four nucleotides adenylate, cytidylate, uridylate, and guanylate (A,C,U,G). The precise arrangement of these four subunits is used to transfer and sometimes store genetic material.

Rotation: turning of a body part about its long axis as if on a pivot; i.e. of the head to look over the shoulder.

Rumination: after swallowing, the regurgitation of food followed by chewing another time.

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S

Salivation: the secretion of saliva by the salivary glands of the mouth, increased in response to the chewing action of the jaws or to the thought, taste, sight, or smell of food.

Scoliosis: curvature of the spine that includes a twisting component; in Rett syndrome, usually due to neurologic factors.

Section 504: a federal civil rights law passed in 1973 to eliminate discrimination against people with disabilities in federally funded programs. Requires that children with disabilities receive educational services and opportunities equal to those provided other children.

Segmental rolling: rolling over where there is rotation (twisting) between the shoulders and hips, often difficult for the child with Rett syndrome.

Seizure: an episodic electrical discharge of nerve cells in the brain resulting in alteration of function or behavior.

Self-help skills: skills related to the care of oneself such as eating, dressing, and grooming.

Sensory modalities: specific channels through which a person receives information about the environment, including sight, sound, touch, taste, and smell.

Serial casting: a series of casts on the feet to correct shortened heel cords.

Sequence: the order of; in reference to DNA or RNA, the order of nucleotide.

Sex chromosomes: those chromosomes that determine gender; the X and Y chromosomes.

Short-term instructional objective: precise statement, described in terms of overt behavior, of what a child is expected to accomplish over a short period in a specific educational area. An intermediate step between the student's current skill level and the annual goal.

Siblings: the other kids who live at your house. Before you had a daughter with Rett, they were called brothers and sisters.

Side lie: a piece of equipment that supports and stabilizes the child on her side. It may be used to provide change of position for the very inactive child or may be used as part of a therapy program to correct scoliosis.

Simple partial seizures: local seizures involving a single area of the brain.

Sleep myoclonus: sudden massive jerks of the body when going to sleep. These are normal.

Somatic: pertaining to the body. When referring to a type of cell, somatic means body cell rather than a germ (sperm or egg producing) cell. Somatic cells contain two pairs of each chromosome, while germ cells contain only one.

Sporadic: occurrence by chance with little chance of recurrence.

Spatial disorientation: when a person's perception of where she is in space is distorted and not accurate. This is observed in Rett when the girl's perception of midline is disturbed, causing her to lean.

Spastic: increased muscle tone so that muscles are stiff and movements are difficult.

Special education: specialized instruction for children who have educational disabilities based on a comprehensive evaluation. The instruction may occur in a variety of settings, but must be precisely matched to their educational needs and adapted to their learning styles.

Splint: a material or device used to protect or immobilize a body part.

Sporadic: occurring by chance, with little risk of recurrence.

SSI (Supplemental Security Income): federal and state funded program that provides money to offset expenses for children with disabilities who come from low-income families under the age of 18, and all individuals with disabilities after the age of eighteen.

Static: unchanging; a static encephalopathy refers to a brain disorder that does not get worse with time. It does not mean or imply that the child will remain static or unchanging. A child with a static encephalopathy, such as cerebral palsy, can continue to learn and develop.

Stereotypies: repetitive, patterned movements; in Rett, usually referring to the hands.

Storage diseases: a number of metabolic diseases in which some material, usually a breakdown product of normal tissue, cannot be further metabolized and is stored within nerve cells of the brain, which produces malfunction.

Strabismus: inability of one eye to attain binocular vision with the other because of imbalance of the muscles of the eyeball.

Stem cell: a cell type that has not specialized to carry out particular functions and retains the ability to divide and differentiate to form a variety of cell types.

Subluxation: partial dislocation.

Substrate: the molecules on which an enzyme acts.

Sulci: crevices on the surface of the cerebrum.

Synapse: the minute spacing separating one neuron from another; neurochemicals breach this gap.

Syncope: fainting; dizziness, pallor, sweating, and loss of consciousness.

Systemic illness: illness affecting the body as a whole instead of one part.

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T

Temporal lobe: one of the main divisions of the cerebral cortex in each hemisphere of the brain, lying at the side within the temple of the skull and separated from the frontal lobe by a cleft (sulcus).

Tonic: continuous increased muscle tone.

Tonic-clonic seizures: once called grand mal seizures; seizures associated with stiffening followed by rhythmic jerking.

Tongue thrust: oral-motor feeding problems; voluntary tongue motions are not controlled; tongue extends in front of the lips when touched with spoon or food; interferes with moving food from the front of the mouth to the back for swallowing.

Transcription: the process of converting information in DNA into information in RNA. Transcription involves making an RNA molecule using the information encoded in the DNA. RNA polymerase is the enzyme that executes this conversion of information.

Transgene: DNA integrated into the germ line from Transgenic organisms.

Transgenic organisms: organisms that have integrated foreign DNA into their germ line as a result of the experimental introduction of DNA.

Transitional movements: movements which allow us to change position. For example rolling, pulling to stand.

Translocation: the transfer of a fragment of one chromosome to another chromosome.

Treatment: interventions that cause a reduction in clinical manifestations that improve the medical/cognitive outcome of a disorder.

Tropic foot disturbances: poor growth of the feet, likely resulting in poor circulation.

Truncal ataxia: ataxia is poor coordination resulting most often from poor function of a part of the brain called the cerebellum. Truncal ataxia describes limitation or exaggeration of the symptoms to the muscles of the torso. If the torso is unsteady and poorly balanced, the limbs have to work overtime to maintain posture.

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U

Unconditional love: love with no conditions, as seen through your daughter's eyes.

Unfortunate activation: females usually randomly inactivate one of the two copies of the X- chromosome in each cell. Unfortunate activation refers to a situation when a female who carries an abnormal gene on one X-chromosome silences the normal copy of the gene. This allows females to manifest symptoms of X-linked recessive diseases like Duchenne muscular dystrophy or hemophilia.

Upper motor neuron: refers to the nerve cell that starts in the cerebral cortex, winds its way down through the brain and then into the spinal cord and that carries information about movement to the lower motor neuron. Damage to the upper motor neuron results in spasticity and deep tendon reflexes (like the knee jerk) that are too brisk, while damage to the lower motor neurons results in weakness and decreased reflexes.

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V

Vacant spells: may be confused with seizures, but are usually associated with irregular breathing patterns instead.

Vagal tone: impulse from the vagus nerve.

Vagus nerve: the tenth cranial nerve which supplies motor nerve fibers to the muscles of swallowing and parasympathetic fibers to the heart and organs of the chest cavity and abdomen.

Valgus: turned outward, usually referring to the ankle in Rett syndrome.

Valsalva's manoeuvre: long inspirations (breath holds) capable of raising blood pressure and heart rate changes.

Varus: turned inward, usually referring to the ankle in Rett syndrome.

Vasomotor disturbance: relating to the nerves or the centers from which they arise that supply the muscle fibers of the walls of blood vessels, which regulate the amount of blood passing to a particular body part or organ. In Rett syndrome, used to describe cold, bluish hands and feet.

Vestibular: movements which give the body input about posture and movements in space which allow coordination and balance.

Vestibular input: unconscious information from the inner ear about equilibrium (state of balance), gravity, movement, and changes of position in space.

Video EEG: the use of video cameras to capture visually the onset and characteristics of seizures while simultaneously monitoring the EEG to see electrical changes.

Volvulus: a twisting of the intestine upon itself which causes obstruction.

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W

Weight-for-height: a term that refers to the appropriateness of a child's weight compared to her height; one measure of nutritional status. "Low weight-for-height" would mean "underweight."

Weight shift: the movement of the body's center of gravity in any direction. Most movement sequences are initiated by this and it is a significant concern because this may be very difficult for the girl with Rett syndrome to do.

White matter: the parts of the brain made up of axons, the long "extension cords" of the nerve cells that carry messages from one cell to another. These extensions are wrapped in an insulating substance called myelin and when the axons are grouped together, they appear white.

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X

X-inactivation/random: because females have two X chromosomes, yet only one functioning copy is necessary, a percentage of X chromosomes with the mutated gene may not be expressed. As a result, females with X-linked disorders often have less severe symptoms than affected males. In contrast, because males have one X chromosome from the mother and one Y chromosome from the father, those who inherit an X-linked dominant disease trait typically fully express the mutated gene on the X chromosome, causing a more severe form of the disorder that may result in lethality before or shortly after birth. Fathers with an X-linked dominant trait transmit the gene to their daughters but not to their sons. Mothers with a single copy of an X-linked dominant gene have a 50 percent risk of transmitting the gene to their daughters as well as their sons.

X-linked dominant disorder: a disorder caused by a gene located on the X chromosome; also called sex-linked; passed on by one parent.

X-linked dominant trait: human traits, such as an individual's specific blood group, eye color, or expression of certain diseases, result from the interaction of one gene inherited from the mother and one from the father. In X-linked dominant disorders, the gene mutation for the disease trait is transmitted as a dominant gene on the X chromosome and therefore may "override" the instructions of the normal gene on the other chromosome, resulting in expression of the disease.

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Y

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Z

Zero reject: all children are to be provided a free appropriate education.

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