Geneticheck - Genetic Report

Gene

CACNA1A

Associated Diseases

Familial Paroxysmal Ataxia
Spinocerebellar Ataxia Type 6
Alternating Hemiplegia Of Childhood
Familial Or Sporadic Hemiplegic Migraine
Lennox-gastaut Syndrome
Benign Paroxysmal Torticollis Of Infancy
Migraine, Familial Hemiplegic, 1
Episodic Ataxia, Type 2
Non-specific Early-onset Epileptic Encephalopathy
Epileptic Encephalopathy, Early Infantile, 42
Spinocerebellar Ataxia 6

Phenotype

Feeding difficulties
Impaired ability to eat related to problems gathering food and getting ready to suck, chew, or swallow it.

Vertigo
An abnormal sensation of spinning while the body is actually stationary.

Oculomotor apraxia
Ocular motor apraxia is a deficiency in voluntary, horizontal, lateral, fast eye movements (saccades) with retention of slow pursuit movements. The inability to follow objects visually is often compensated by head movements. There may be decreased smooth pursuit, and cancellation of the vestibulo-ocular reflex.

Episodic hemiplegia
Transient episodes of weakness of the arm, leg, and in some cases the face on one side of the body.

Hypodontia
The absence of five or less teeth from the normal series by a failure to develop.

Hypsarrhythmia
Hypsarrhythmia is abnormal interictal high amplitude waves and a background of irregular spikes. There is continuous (during wakefulness), high-amplitude (>200 Hz), generalized polymorphic slowing with no organized background and multifocal spikes demonstrated by electroencephalography (EEG).

Personality disorder
An abnormality of mental functioning affecting the personality and behavioural tendencies of an individual and characterized by a rigid and unhealthy pattern of thinking and behavior. The definition of a personal disorder implies that the abnormality is not the result of damage or insult to the brain or from another psychiatric disorder.

Nausea and vomiting

Nausea is a commonly encountered symptom that has been defined as an unpleasant painless subjective feeling that one will imminently vomit. Vomiting has been defined as the forceful expulsion of the contents of the stomach, duodenum, or jejunum through the oral cavity. While nausea and vomiting are often thought to exist on a temporal continuum, this is not always the case. There are situations when severe nausea may be present without emesis and less frequently, when emesis may be present without preceding nausea.

Downbeat nystagmus
Downbeat nystagmus is a type of fixation nystagmus with the fast phase beating in a downward direction. It generally increases when looking to the side and down and when lying prone.

Status epilepticus
Status epilepticus is a type of prolonged seizure resulting either from the failure of the mechanisms responsible for seizure termination or from the initiation of mechanisms which lead to abnormally prolonged seizures (after time point t1). It is a condition that can have long-term consequences (after time point t2), including neuronal death, neuronal injury, and alteration of neuronal networks, depending on the type and duration of seizures.

Thin eyebrow
Decreased diameter of eyebrow hairs.

Distal upper limb muscle weakness
Reduced strength of the distal musculature of the arms.

Babinski sign
Upturning of the big toe (and sometimes fanning of the other toes) in response to stimulation of the sole of the foot. If the Babinski sign is present it can indicate damage to the corticospinal tract.

Mydriasis
Abnormal dilatation of the iris.

Downslanted palpebral fissures
The palpebral fissure inclination is more than two standard deviations below the mean.

Abnormal autonomic nervous system physiology
A functional abnormality of the autonomic nervous system.

Psychosis
A condition characterized by changes of personality and thought patterns often accompanied by hallucinations and delusional beliefs.

Autosomal dominant inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in heterozygotes. In the context of medical genetics, an autosomal dominant disorder is caused when a single copy of the mutant allele is present. Males and females are affected equally, and can both transmit the disorder with a risk of 50% for each child of inheriting the mutant allele.

Abnormality of coordination

EEG abnormality
Abnormality observed by electroencephalogram (EEG), which is used to record of the brain's spontaneous electrical activity from multiple electrodes placed on the scalp.

Dysphasia

Myotonia
An involuntary and painless delay in the relaxation of skeletal muscle following contraction or electrical stimulation.

Confusion
Lack of clarity and coherence of thought, perception, understanding, or action.

Exaggerated cupid's bow
More pronounced paramedian peaks and median notch of the Cupid's bow.

Progressive gait ataxia
A type of gait ataxia displaying progression of clinical severity.

Metamorphopsia
A visual anomaly in which images appear distorted. A grid of straight lines appears wavy and parts of the grid may appear blank.

EEG with focal sharp waves
EEG with focal sharp transient waves of a duration between 80 and 200 msec.

Diplopia
Diplopia is a condition in which a single object is perceived as two images, it is also known as double vision.

Abnormal involuntary eye movements
Anomalous movements of the eyes that occur without the subject wanting them to happen.

Flushing
Recurrent episodes of redness of the skin together with a sensation of warmth or burning of the affected areas of skin.

Fever
Body temperature elevated above the normal range.

Emotional lability
Unstable emotional experiences and frequent mood changes; emotions that are easily aroused, intense, and/or out of proportion to events and circumstances.

Spasticity
A motor disorder characterized by a velocity-dependent increase in tonic stretch reflexes with increased muscle tone, exaggerated (hyperexcitable) tendon reflexes.

Seesaw nystagmus

Seesaw nystagmus is a type of pendular nystagmus where a half cycle consists of the elevation and intorsion of one eye, concurrently with the depression and extortion of the fellow eye. In the other half cycle, there is an inversion of the ocular movements.

Apathy

Optic atrophy
Atrophy of the optic nerve. Optic atrophy results from the death of the retinal ganglion cell axons that comprise the optic nerve and manifesting as a pale optic nerve on fundoscopy.

Hyporeflexia
Reduction of neurologic reflexes such as the knee-jerk reaction.

Myoclonus
Very brief, involuntary random muscular contractions occurring at rest, in response to sensory stimuli, or accompanying voluntary movements.

Cardiomyopathy
A myocardial disorder in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality.

Progressive neurologic deterioration

Abnormal corpus callosum morphology
Abnormality of the corpus callosum.

Abnormality of eye movement
An abnormality in voluntary or involuntary eye movements or their control.

Encephalopathy
Encephalopathy is a term that means brain disease, damage, or malfunction. In general, encephalopathy is manifested by an altered mental state.

Tinnitus
Tinnitus is an auditory perception that can be described as the experience of sound, in the ear or in the head, in the absence of external acoustic stimulation.

Auditory hallucinations
The false perception of sound.

Progressive cerebellar ataxia

Hypertonia
A condition in which there is increased muscle tone so that arms or legs, for example, are stiff and difficult to move.

Constipation
Infrequent or difficult evacuation of feces.

Incoordination

Gastroesophageal reflux
A condition in which the stomach contents leak backwards from the stomach into the esophagus through the lower esophageal sphincter.

Gaze-evoked nystagmus
Nystagmus made apparent by looking to the right or to the left.

Seizure
A seizure is an intermittent abnormality of nervous system physiology characterised by a transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain.

Abnormality of the gastrointestinal tract
An abnormality of the gastrointestinal tract.

Involuntary movements
Involuntary contractions of muscle leading to involuntary movements of extremities, neck, trunk, or face.

Amaurosis fugax
A transient visual disturbance that is typically caused by a circulatory, ocular or neurological underlying condition.

Decreased vigilance
A reduction in the ability to maintain sustained attention characterized by reduced alertness.

Chorea
Chorea (Greek for 'dance') refers to widespread arrhythmic involuntary movements of a forcible, jerky and restless fashion. It is a random-appearing sequence of one or more discrete involuntary movements or movement fragments. Movements appear random because of variability in timing, duration or location. Each movement may have a distinct start and end. However, movements may be strung together and thus may appear to flow randomly from one muscle group to another. Chorea can involve the trunk, neck, face, tongue, and extremities.

Autistic behavior
Persistent deficits in social interaction and communication and interaction as well as a markedly restricted repertoire of activity and interest as well as repetitive patterns of behavior.

High forehead
An abnormally increased height of the forehead.

Torticollis
Involuntary contractions of the neck musculature resulting in an abnormal posture of or abnormal movements of the head.

Ataxia
Cerebellar ataxia refers to ataxia due to dysfunction of the cerebellum. This causes a variety of elementary neurological deficits including asynergy (lack of coordination between muscles, limbs and joints), dysmetria (lack of ability to judge distances that can lead to under- or overshoot in grasping movements), and dysdiadochokinesia (inability to perform rapid movements requiring antagonizing muscle groups to be switched on and off repeatedly).

Nystagmus
Rhythmic, involuntary oscillations of one or both eyes related to abnormality in fixation, conjugate gaze, or vestibular mechanisms.

Abnormal vestibulo-ocular reflex
An abnormality of the vestibulo-ocular reflex (VOR). The VOR attempts to keep the image stable on the retina. Ideally passive or active head movements in one direction are compensated for by eye movements of equal magnitude.

Irritability
A proneness to anger, i.e., a condition of being easily bothered or annoyed.

Downturned corners of mouth
A morphological abnormality of the mouth in which the angle of the mouth is downturned. The oral commissures are positioned inferior to the midline labial fissure.

Drowsiness
Excessive daytime sleepiness.

Decreased fetal movement
An abnormal reduction in quantity or strength of fetal movements.

Brain atrophy
Partial or complete wasting (loss) of brain tissue that was once present.

Hyperactivity
Hyperactivity is a state of constantly being unusually or abnormally active, including in situations in which it is not appropriate.

Facial paralysis
Complete loss of ability to move facial muscles innervated by the facial nerve (i.e., the seventh cranial nerve).

Apnea
Lack of breathing with no movement of the respiratory muscles and no exchange of air in the lungs. This term refers to a disposition to have recurrent episodes of apnea rather than to a single event.

Focal sensory seizure
A focal sensory seizure is a type seizure beginning with a subjective sensation.

Blepharospasm
A focal dystonia that affects the muscles of the eyelids and brow, associated with involuntary recurrent spasm of both eyelids.

Reduced tendon reflexes
Diminution of tendon reflexes, which is an invariable sign of peripheral nerve disease.

Scotoma
A regional and pathological increase of the light detection threshold in any region of the visual field surrounded by a field of normal or relatively well-preserved vision.

Epileptic encephalopathy
A condition in which epileptiform abnormalities are believed to contribute to the progressive disturbance in cerebral function. Epileptic encephalaopathy is characterized by (1) electrographic EEG paroxysmal activity that is often aggressive, (2) seizures that are usually multiform and intractable, (3) cognitive, behavioral and neurological deficits that may be relentless, and (4) sometimes early death.

Bradyopsia
Difficulty in seeing moving objects.

CSF lymphocytic pleiocytosis
An increased lymphocyte count in the cerebrospinal fluid.

Nuchal rigidity
Resistance of the extensor muscles of the neck to being bent forwards (i.e., impaired neck flexion) as a result of muscle spasm of the extensor muscles of the neck. Nuchal rigidity is not a fixed rigidity. Nuchal rigidity has been used as a bedside test for meningism, although its sensitivity for this purpose has been debated.

Generalized tonic seizure
A generalized tonic seizure is a type of generalized motor seizure characterised by bilateral limb stiffening or elevation, often with neck stiffening without a subsequent clonic phase. The tonic activity can be a sustained abnormal posture, either in extension or flexion, sometimes accompanied by tremor of the extremities.

Hemiparesis
Loss of strength in the arm, leg, and sometimes face on one side of the body. Hemiplegia refers to a complete loss of strength, whereas hemiparesis refers to an incomplete loss of strength.

Bilateral tonic-clonic seizure
A bilateral tonic-clonic seizure is a seizure defined by a tonic (bilateral increased tone, lasting seconds to minutes) and then a clonic (bilateral sustained rhythmic jerking) phase.

Episodic ataxia
Periodic spells of incoordination and imbalance, that is, episodes of ataxia typically lasting from 10 minutes to several hours or days.

Strabismus
A misalignment of the eyes so that the visual axes deviate from bifoveal fixation. The classification of strabismus may be based on a number of features including the relative position of the eyes, whether the deviation is latent or manifest, intermittent or constant, concomitant or otherwise and according to the age of onset and the relevance of any associated refractive error.

Esotropia
A form of strabismus with one or both eyes turned inward ('crossed') to a relatively severe degree, usually

defined as 10 diopters or more.

Heterogeneous

Developmental regression
Loss of developmental skills, as manifested by loss of developmental milestones.

EEG with focal sharp slow waves
EEG with focal sharp transient waves of a duration between 80 and 200 msec followed by a slow wave.

Cerebellar vermis atrophy
Wasting (atrophy) of the vermis of cerebellum.

Dystonia
An abnormally increased muscular tone that causes fixed abnormal postures. There is a slow, intermittent twisting motion that leads to exaggerated turning and posture of the extremities and trunk.

Unsteady gait

Hyperreflexia
Hyperreflexia is the presence of hyperactive stretch reflexes of the muscles.

Saccadic smooth pursuit
An abnormality of tracking eye movements in which smooth pursuit is interrupted by an abnormally high number of saccadic movements.

Falls

Autism
Autism is a neurodevelopmental disorder characterized by impaired social interaction and communication, and by restricted and repetitive behavior. Autism begins in childhood. It is marked by the presence of markedly abnormal or impaired development in social interaction and communication and a markedly restricted repertoire of activity and interest. Manifestations of the disorder vary greatly depending on the developmental level and chronological age of the individual (DSM-IV).

Visual hallucinations
Visual perceptions that are not elicited by a corresponding stimulus from the outside world.

Flexion contracture
A flexion contracture is a bent (flexed) joint that cannot be straightened actively or passively. It is thus a chronic loss of joint motion due to structural changes in muscle, tendons, ligaments, or skin that prevents normal movement of joints.

Tetraparesis
Weakness of all four limbs.

Abnormality of vision
Abnormality of eyesight (visual perception).

Impulsivity
Acting on the spur of the moment in response to immediate stimuli; acting on a momentary basis without a plan or consideration of outcomes; difficulty establishing or following plans; a sense of urgency and self-harming behavior under emotional distress.

Dysphagia
Difficulty in swallowing.

Aggressive behavior
Aggressive behavior can denote verbal aggression, physical aggression against objects, physical aggression against people, and may also include aggression towards oneself.

Atonic seizure
Atonic seizure is a type of motor seizure characterized by a sudden loss or diminution of muscle tone without apparent preceding myoclonic or tonic event lasting about 1 to 2 seconds, involving head, trunk, jaw, or limb musculature.

Skewfoot
A type of flat-foot characterized by hindfoot abductovalgus, metatarsus adductus, and Achilles tendon shortening. The predominant radiographic findings include forefoot adduction with lateral subluxation of the navicular on the talus and heel valgus. Very abnormal shoe wear is noted on the medial side. Calluses occurunder the metatarsal heads and thehead of the plantar-flexed talus.

Increased CSF protein
Increased concentration of protein in the cerebrospinal fluid.

Ptosis
The upper eyelid margin is positioned 3 mm or more lower than usual and covers the superior portion of the iris (objective); or, the upper lid margin obscures at least part of the pupil (subjective).

Facial tics
Sudden, repetitive, nonrhythmic motor movements (spasms), involving the eyes and muscles of the face.

Microcephaly
Head circumference below 2 standard deviations below the mean for age and gender.

Neurological speech impairment

Cerebral edema
Abnormal accumulation of fluid in the brain.

Oral-pharyngeal dysphagia

Failure to thrive

Failure to thrive (FTT) refers to a child whose physical growth is substantially below the norm.

Abnormal head movements

Paroxysmal dyskinesia
Episodic bouts of involuntary movements with dystonic, choreic, ballistic movements, or a combination thereof. There is no loss of consciousness during the attacks.

Tongue fasciculations
Fasciculations or fibrillation affecting the tongue muscle.

Headache
Cephalgia, or pain sensed in various parts of the head, not confined to the area of distribution of any nerve.

Agitation
A state of exceeding restlessness and excessive motor activity associated with mental distress or a feeling of inner tension.

Dissociated sensory loss
A pattern of sensory loss with selective loss of touch sensation and proprioception without loss of pain and temperature, or vice-versa.

Areflexia
Absence of neurologic reflexes such as the knee-jerk reaction.

CSF pleocytosis
An increased white blood cell count in the cerebrospinal fluid.

Poor head control
Difficulty to maintain correct position of the head while standing or sitting.

Aspiration
Inspiration of a foreign object into the airway.

Atypical absence seizure
An atypical absence seizure is a type of generalised non-motor (absence) seizure characterised by interruption of ongoing activities and reduced responsiveness. In comparison to a typical absence seizure, changes in tone may be more pronounced, onset and/or cessation may be less abrupt, and the duration of the ictus and post-ictal recovery may be longer. Although not always available, an EEG often demonstrates slow (<3 Hz), irregular, generalized spike-wave activity.

Vomiting
Forceful ejection of the contents of the stomach through the mouth by means of a series of involuntary spasmic contractions.

Dysarthria
Dysarthric speech is a general description referring to a neurological speech disorder characterized by poor articulation. Depending on the involved neurological structures, dysarthria may be further classified as spastic,

flaccid, ataxic, hyperkinetic and hypokinetic, or mixed.

Anorexia
A lack or loss of appetite for food (as a medical condition).

Photopsia
Perceived flashes of light.

Focal motor seizure
A type of focal-onset seizure characterized by a motor sign as its initial semiological manifestation.

Paresthesia
Abnormal sensations such as tingling, pricking, or numbness of the skin with no apparent physical cause.

Genetic anticipation
A mode of inheritance in which the severity of a disorder increases or the age of onset decreases as the disorder is passed from one generation to the next.

Tremor
An unintentional, oscillating to-and-fro muscle movement about a joint axis.

Intention tremor
A type of kinetic tremor that occurs during target directed movement is called intention tremor. That is, an oscillatory cerebellar ataxia that tends to be absent when the limbs are inactive and during the first part of voluntary movement but worsening as the movement continues and greater precision is required (e.g., in touching a target such as the patient's nose or a physician's finger).

Alien limb phenomenon
Alien limb phenomenon refers to involuntary motor activity of a limb in conjunction with the feeling of estrangement from that limb.

Attention deficit hyperactivity disorder
Attention deficit hyperactivity disorder (ADHD) manifests at age 2-3 years or by first grade at the latest. The main symptoms are distractibility, impulsivity, hyperactivity, and often trouble organizing tasks and projects, difficulty going to sleep, and social problems from being aggressive, loud, or impatient.

Muscle weakness
Reduced strength of muscles.

Generalized myoclonic seizure
A generalized myoclonic seizure is a type of generalized motor seizure characterised by bilateral, sudden, brief (<100 ms) involuntary single or multiple contraction of muscles or muscle groups of variable topography (axial, proximal limb, distal). Myoclonus is less regularly repetitive and less sustained than is clonus.

First dorsal interossei muscle weakness

Behavioral abnormality
An abnormality of mental functioning including various affective, behavioural, cognitive and perceptual

abnormalities.

Mental deterioration
Loss of previously present mental abilities, generally in adults.

Abnormal brainstem morphology
An anomaly of the brainstem.

Spontaneous pain sensation
Spontaneous pain is a kind of neuropathic pain which occurs without an identifiable trigger.

Sensory neuropathy
Peripheral neuropathy affecting the sensory nerves.

Vertical nystagmus
Vertical nystagmus may present with either up-beating or down-beating eye movements or both. When present in the straight-ahead position of gaze it is referred to as upbeat nystagmus or downbeat nystagmus.

Retinal degeneration
A nonspecific term denoting degeneration of the retinal pigment epithelium and/or retinal photoreceptor cells.

Migraine
Migraine is a chronic neurological disorder characterized by episodic attacks of headache and associated symptoms.

Impaired temperature sensation
A reduced ability to discriminate between different temperatures.

Intellectual disability
Subnormal intellectual functioning which originates during the developmental period. Intellectual disability, previously referred to as mental retardation, has been defined as an IQ score below 70.

Dyscalculia
A specific learning disability involving mathematics and arithmetic.

Gait ataxia
A type of ataxia characterized by the impairment of the ability to coordinate the movements required for normal walking. Gait ataxia is characteirzed by a wide-based staggering gait with a tendency to fall.

Coma
Complete absence of wakefulness and content of conscience, which manifests itself as a lack of response to any kind of external stimuli.

Gaze-evoked horizontal nystagmus
Horizontal nystagmus made apparent by looking to the right or to the left.

Hearing impairment
A decreased magnitude of the sensory perception of sound.

Hyperhidrosis
Abnormal excessive perspiration (sweating) despite the lack of appropriate stimuli like hot and humid weather.

Anxiety
Intense feelings of nervousness, tenseness, or panic, often in reaction to interpersonal stresses; worry about the negative effects of past unpleasant experiences and future negative possibilities; feeling fearful, apprehensive, or threatened by uncertainty; fears of falling apart or losing control.

Choreoathetosis
Involuntary movements characterized by both athetosis (inability to sustain muscles in a fixed position) and chorea (widespread jerky arrhythmic movements).

Transient unilateral blurring of vision
Transient blurring of vision associated with the aura phase of migraine.

Cerebral atrophy
Atrophy (wasting, decrease in size of cells or tissue) affecting the cerebrum.

Choking episodes
Incidents in which a piece of food or other objects get stuck in the upper airway and provoke coughing, gagging, inability to talk, and difficulty breathing.

Short stature
A height below that which is expected according to age and gender norms. Although there is no universally accepted definition of short stature, many refer to "short stature" as height more than 2 standard deviations below the mean for age and gender (or below the 3rd percentile for age and gender dependent norms).

Vestibular dysfunction
An abnormality of the functioning of the vestibular apparatus.

Exotropia
A form of strabismus with one or both eyes deviated outward.

Cerebellar atrophy
Cerebellar atrophy is defined as a cerebellum with initially normal structures, in a posterior fossa with normal size, which displays enlarged fissures (interfolial spaces) in comparison to the foliae secondary to loss of tissue. Cerebellar atrophy implies irreversible loss of tissue and result from an ongoing progressive disease until a final stage is reached or a single injury, e.g. an intoxication or infectious event.

Migraine with aura
A type of migraine in which there is an aura characterized by focal neurological phenomena that usually proceed, but may accompany or occur in the absence of, the headache. The symptoms of an aura may include fully reversible visual, sensory, and speech symptoms but not motor weakness. Visual symptoms may include flickering lights, spots and lines and/or loss of vision and/or unilateral sensory symptoms such as paresthesias or numbness. At least one of the symptoms of an aura develops gradually over 5 or more minutes and/or different symptoms occur in succession.

Postural instability

A tendency to fall or the inability to keep oneself from falling; imbalance. The retropulsion test is widely regarded as the gold standard to evaluate postural instability, Use of the retropulsion test includes a rapid balance perturbation in the backward direction, and the number of balance correcting steps (or total absence thereof) is used to rate the degree of postural instability. Healthy subjects correct such perturbations with either one or two large steps, or without taking any steps, hinging rapidly at the hips while swinging the arms forward as a counterweight. In patients with balance impairment, balance correcting steps are often too small, forcing patients to take more than two steps. Taking three or more steps is generally considered to be abnormal, and taking more than five steps is regarded as being clearly abnormal. Markedly affected patients continue to step backward without ever regaining their balance and must be caught by the examiner (this would be called true retropulsion). Even more severely affected patients fail to correct entirely, and fall backward like a pushed toy soldier, without taking any corrective steps.

EEG with multifocal slow activity
Multifocal slowing of cerebral electrical activity recorded along the scalp by electroencephalography (EEG).

Hypotonia
Hypotonia is an abnormally low muscle tone (the amount of tension or resistance to movement in a muscle). Even when relaxed, muscles have a continuous and passive partial contraction which provides some resistance to passive stretching. Hypotonia thus manifests as diminished resistance to passive stretching. Hypotonia is not the same as muscle weakness, although the two conditions can co-exist.

Limb hypertonia

Abnormal T-wave
An abnormality of the T wave on the electrocardiogram, which mainly represents the repolarization of the ventricles.

Dyskinesia
A movement disorder which consists of effects including diminished voluntary movements and the presence of involuntary movements.

Neurodevelopmental delay

Focal-onset seizure
A focal-onset seizure is a type of seizure originating within networks limited to one hemisphere. They may be discretely localized or more widely distributed, and may originate in subcortical structures.

Pallor
Abnormally pale skin.

Generalized hypotonia
Generalized muscular hypotonia (abnormally low muscle tone).

Bulbar signs

Abnormal pyramidal sign

Functional neurological abnormalities related to dysfunction of the pyramidal tract.

Athetosis
A slow, continuous, involuntary writhing movement that prevents maintenance of a stable posture. Athetosis involves continuous smooth movements that appear random and are not composed of recognizable sub-movements or movement fragments. In contrast to chorea, in athetosis, the same regions of the body are repeatedly involved. Athetosis may worsen with attempts at movement of posture, but athetosis can also occur at rest.

Arrhythmia
Any cardiac rhythm other than the normal sinus rhythm. Such a rhythm may be either of sinus or ectopic origin and either regular or irregular. An arrhythmia may be due to a disturbance in impulse formation or conduction or both.

Impaired smooth pursuit
An impairment of the ability to track objects with the ocular smooth pursuit system, a class of rather slow eye movements that minimizes retinal target motion.

Delayed speech and language development
A degree of language development that is significantly below the norm for a child of a specified age.

Difficulty walking
Reduced ability to walk (ambulate).

Complex febrile seizure
A febrile seizure that has any of the following features: focal semiology (or associated with post-ictal neurologic abnormalities beyond drowsiness, such as a Todd's paresis), prolonged seizure beyond 15 minutes, or recurring (occurring more than once) in a 24 hour period.

Hemiplegia
Paralysis (complete loss of muscle function) in the arm, leg, and in some cases the face on one side of the body.

Global developmental delay
A delay in the achievement of motor or mental milestones in the domains of development of a child, including motor skills, speech and language, cognitive skills, and social and emotional skills. This term should only be used to describe children younger than five years of age.

Abnormal myelination
Any anomaly in the process by which myelin sheaths are formed and maintained around neurons.

Respiratory distress
Respiratory distress is objectively observable as the physical or emotional consequences from the experience of dyspnea. The physical presentation of respiratory distress is generally referred to as labored breathing, while the sensation of respiratory distress is called shortness of breath or dyspnea.

Diarrhea
Abnormally increased frequency of loose or watery bowel movements.

Facial hypotonia
Reduced muscle tone of a muscle that is innervated by the facial nerve (the seventh cranial nerve).

Rigidity
Continuous involuntary sustained muscle contraction. When an affected muscle is passively stretched, the degree of resistance remains constant regardless of the rate at which the muscle is stretched. This feature helps to distinguish rigidity from muscle spasticity.

EEG with generalized sharp slow waves
EEG with generalized sharp transient waves of a duration between 80 and 200 msec followed by a slow wave.

Gastrointestinal dysmotility
Abnormal intestinal contractions, such as spasms and intestinal paralysis, related to the loss of the ability of the gut to coordinate muscular activity because of endogenous or exogenous causes.

Cardiac conduction abnormality
Any anomaly of the progression of electrical impulses through the heart.

Dehydration

Abdominal distention
Distention of the abdomen.

Ocular flutter
Ocular flutter is an abnormal eye movement consisting of repetitive, irregular, involuntary bursts of horizontal saccades without an intersaccadic interval. It is generally superimposed on normal oculomotor behaviour and its occurrence may be favoured by various events, such as blinks, the triggering of normal saccades or optokinetic stimulation.