



Creating a data dictionary for pediatric autonomic disorders

Jeffrey R. Boris¹ · Hasan Abdallah² · Shelley Ahrens³ · Gisela Chelimsky⁴ · Thomas C. Chelimsky⁵ · Philip R. Fischer^{3,6,7} · John E. Fortunato⁸ · Raewyn Gavin⁹ · Janice L. Gilden¹⁰ · Renato Gonik¹¹ · Blair P. Grubb¹² · Kelsey M. Klaas³ · Erin Marriott¹³ · Lauren E. Marsillio^{14,15,16} · Marvin S. Medow¹⁷ · Lucy Norcliffe-Kaufmann¹⁸ · Mohammed T. Numan¹⁹ · Erin Olufs²⁰ · Laura A. Pace²¹ · Paul T. Pianosi²² · Pippa Simpson²³ · Julian M. Stewart¹⁷ · Sally Tarbell²⁴ · Natalie R. Van Waning²⁵ · Debra E. Weese-Mayer^{14,15,16}

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Abstract

Purpose Whether evaluating patients clinically, documenting care in the electronic health record, performing research, or communicating with administrative agencies, the use of a common set of terms and definitions is vital to ensure appropriate use of language. At a 2017 meeting of the Pediatric Section of the American Autonomic Society, it was determined that an autonomic data dictionary comprising aspects of evaluation and management of pediatric patients with autonomic disorders would be an important resource for multiple stakeholders.

Methods Our group created the list of terms for the dictionary. Definitions were prioritized to be obtained from established sources with which to harmonize. Some definitions needed mild modification from original sources. The next tier of sources included published consensus statements, followed by Internet sources. In the absence of appropriate sources, we created a definition.

Results A total of 589 terms were listed and defined in the dictionary. Terms were organized by Signs/Symptoms, Triggers, Comorbid Disorders, Family History, Medications, Medical Devices, Physical Examination Findings, Testing, and Diagnoses.

Conclusion Creation of this data dictionary becomes the foundation of future clinical care and investigative research in pediatric autonomic disorders, and can be used as a building block for a subsequent adult autonomic data dictionary.

Keywords Child · Adolescent · Autonomic nervous system · Medical informatics · Health information

Introduction

The autonomic nervous system (ANS) controls involuntary bodily functions, both at rest and during activity, affecting virtually all organ systems. Consequently, there is a wide range of conditions associated with dysregulation of the ANS. One of the earliest documented disorders of the ANS was reported in 1928, when carcinoid tumors of the appendix were described [1]. Over the next century, a panoply of diverse disorders, derangements, and dysfunctions of the ANS have been delineated, including diseases primary to the ANS as well as ANS disturbances secondary to other conditions [2]. A medical literature search for “autonomic nervous system” in 2022 identified nearly 170,000 entries, and a similar search for “autonomic nervous system disease” listed over 41,000 entries [3].

Although these represent substantially fewer literature entries compared with myocardial infarction (> 270,000 entries) or cerebrovascular accident (> 410,000 entries), it is noteworthy that research in myocardial infarction and cerebrovascular accident seem to have more relatively “recent” research, with inflection points around the 1960s and 1970s, respectively, as compared with the slow steady growth of ANS publications since the 1930s [3]. In other words, while disorders that affect single organs have not only had an explosion of relatively recent research, they have also compiled a significant body of study; the same cannot be said about the ANS. Despite the much wider range of ANS involvement and disorders across the human body as compared with numerous conditions that act on isolated organs, there is a relatively limited knowledge about the ANS, its function, and its pathophysiologic processes, especially as it pertains to pediatric patients, the progression into adulthood, prognosis, and effect on other disorders.

✉ Jeffrey R. Boris
jeffreyborismd@earthlink.net

Extended author information available on the last page of the article

Autonomic nervous system disorders

The majority of ANS-related conditions, as with the large majority of medical disorders, are predominantly described in adults, involving diabetes mellitus, Parkinson's disease, multiple system atrophy, and pure autonomic failure, to name a few [2]. This likely follows from a larger proportion of clinical and research personnel in the field of adult medicine. However, there are also manifestations of ANS disorders in pediatric and adolescent patients [4, 5]. The earliest description of the ANS in the context of pediatric patients was in 1927 [6], in which leukocyte levels were assessed in the setting of induced vasoconstriction and vasodilation. Since that time, the medical literature has evolved, growing with increasing documentation of awareness, recognition, description, and interventions for ANS disorders in childhood. However, the breadth of understanding of the ANS in the context of children remains limited as compared with adults, despite significant advances in the field as well as attempts to review and summarize them to encourage greater awareness among providers [7].

Most notable is the difference in the various ANS disorders observed and reported in the pediatric population as compared to adults. The aforementioned disorders listed above that are seen in adults are not described in children and adolescents. However, a subset of ANS disorders overlap between these populations, including orthostatic intolerance, various subtypes of syncope, initial orthostatic hypotension, orthostatic hypotension, and postural orthostatic tachycardia syndrome. There are also ANS-affecting conditions that tend to be uniquely identified in childhood. These are often the rare syndromic and genetic conditions that involve ANS disorders, including the various hereditary and sensory neuropathies (such as familial dysautonomia), Rett syndrome, rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD), and congenital central hypoventilation syndrome (CCHS), as well as certain tumors of neural crest origin, such as neuroblastoma, ganglioneuroblastoma, and ganglioneuroma [7]. Interestingly, breath-holding spells, seen in infants up through young children, may be manifestations of neurocardiogenic syncope, which also can be found in adults. However, the way that they present, with episodes specifically triggered with mild pain or anger, and typically occurring after prolonged crying at end-expiration, is unique to children. Breath-holding spells are actually relatively common, occurring in up to almost 5% of children [8].

Utility of the data dictionary

Though long considered to have been “discovered” in 1921, the initial descriptions of the ANS were in 1903 and 1908 by Langley [9] and Kappers [10], respectively. However,

as occurs when new findings are described in medicine and science, there is often discussion and dispute around the appropriate terminology for common use. This includes disagreement on the nomenclature utilized to describe the ANS [11]. Similar discourse continues today. For example, naming conventions around syncope have evolved in the past 20 years [12–14], although much of the change in today's literature attempts to be more evidence-based as opposed to opinion-based. To that end, the use of language and, specifically, nomenclature, with its appropriate and agreed-upon terms and definitions, is key to the ability of practitioners to care for their patients and for researchers to advance the discipline in coordinated fashion. Other groups within medicine have recognized this, such as neonatology, emergency medicine, ambulatory pediatric cardiology, and neurologists who manage stroke, and have taken steps to provide data dictionaries for general use [15–18], the benefits of which are numerous. At a minimum, a commonly agreed upon vocabulary allows clinicians both to ensure that they are caring for patients with appropriately defined conditions and to allow communication among providers in the joint care of these patients. Increasingly, though, common agreed-upon terminology is facilitating research. It allows multi-center studies to attempt to ensure that the data they gather are consistent, rigorous, and reporting on homogeneous cohorts. Furthermore, with today's increased use of complex computing and compilation, the ability to parse “big data” is more easily applied with a standardized set of terms and definitions. These data dictionaries can be incorporated by governmental agencies for billing and surveillance, as they are often assembled by committees composed of subject matter experts within the specific field, and therefore represent an ideal source for ensuring that common language is used within this oversight. Typically with adoption by the federal government, insurance payors will also implement the use of these terms, facilitating the ability to document, bill, and reimburse for these diagnoses and related procedures. The harmonization of these data dictionaries with existing clinical use dictionaries, either by direct juxtaposition and incorporation or by acknowledging overlapping terms and updating them to be “more correct,” allows for even larger data analyses across disciplines not previously considered. Lastly, having the availability of these lexicons for general use means that they can also be incorporated by the companies who manufacture electronic health records in a vendor-agnostic way. This then feeds back both into the above benefits as well as allowing clinicians to capture discrete data while generating documentation, and to build datasets of patient information that help to improve internal clinic function and to advance clinical and translational research.

Recognizing these numerous benefits, the Pediatric Section of the American Autonomic Society set out to

generate a data dictionary specific to pediatric disorders of the ANS. The end product data dictionary is designed to be used both for clinical documentation and for research purposes. Although disorders of orthostatic intolerance are the most frequently diagnosed ANS disorder in children and adolescents, inclusion of less commonly identified autonomic disorders, such as the hereditary sensory and autonomic neuropathies, Rett syndrome, ROHHAD, and CCHS, allow for practitioners and researchers to leverage the same advantages afforded by this dictionary. Within informatics, incorporation of existing terms and definitions from other established terminologies is the more desired approach. This allows for better harmonization with other dictionaries in the future, and it saves the informaticists from having to recreate an entire set of terms and definitions for each application.

Methods

The concept for creating the data dictionary came out of an initial research meeting of the Pediatric Section of the American Autonomic Society sponsored by the Children's Hospital of Philadelphia in 2017. At the time, it was felt that the data dictionary would be used as a foundational dictionary for internal research projects within our group. However, over time, it became apparent that the dictionary could be applicable in multiple other disciplines, and should be made available for other providers and researchers working in the emerging discipline of pediatric autonomic medicine. The dictionary needed to be harmonized with existing catalogs of common data elements, essentially integrating and unifying components from the various known resources into our dictionary. To that end, as best as possible, we utilized the National Cancer Institute Thesaurus (NCIt) [19] as the source for dictionary terms and definitions, as it is one of the largest compilations of biomedical lexicons. At times, some of the NCIt definitions were out of date, inadequate, or inconsistent across similar terms, so in our capacity as subject matter experts, we elected to modify them. In other instances, the specific required term did not exist in the NCIt, prompting acquisition of needed definitions for terms from other consensus-driven documents or single subject matter expert publications. If none of those options could be used, we generated our own definitions with support from existing literature. The work for this project encompassed numerous in-person meetings, virtual meetings, and email correspondences. Because there is so much overlap of ANS dysregulation with other, non-ANS disorders, our dictionary not surprisingly appears to include widely disparate terms across medical conditions and their care. But it is for this very reason, such as the overlap of postural orthostatic tachycardia syndrome with entities such as joint hypermobility

syndromes, functional gastrointestinal disorders, and mast cell activation syndrome, that this inclusive approach was taken. This strategy allows for meticulous documentation of the signs and symptoms across these disorders, patient and family histories, the spectrum of clinical testing necessary to correctly diagnose affected patients, and the monitoring of longitudinal treatments.

As in much of medicine, the understanding of clinical syndromes and their pathophysiology, genetic basis, and other aspects of their manifestations evolve over time. Research augments the ability to comprehend these various relationships as well as to better care for these patients. Because of this, it is understood that our data dictionary will need to be a dynamic instrument, updated over time as science and medicine advance and evolve. However, it will also be a signpost to be used by adult ANS providers, who can take advantage of the work done here to create an adult-patient-based ANS data dictionary as well as to be able to care for children with autonomic conditions who advance into adulthood. The data dictionary can be a tool useful for integration with other data vocabularies, such as in offering reciprocity of new terms and updated definitions introduced by our group to the NCIt or inclusion in larger multidisciplinary studies. Lastly, it can be a step forward in helping clinicians, researchers, and informaticists to find gaps in our definitions and data content with the aim to optimize the instrument we have prepared.

Data elements and definitions

Our data dictionary is divided into nine sections. The first section is "Signs/Symptoms," in which the parts of what would be considered the history of the present illness would be obtained. The second section is "Triggers," which delineates known or suspected events or other factors that initiated the onset of the signs or symptoms. The third section is "Co-morbid Disorders," denoting various co-occurring diseases, disorders, and syndromes, followed by the fourth section, "Family History," which lists the diseases and syndromes determined to occur in family members. The fifth and sixth sections are "Medications" and "Medical Devices," respectively, allowing for recording of pharmacologic and other therapeutic interventions. The seventh section is the specific "Physical Examination Findings" typically associated with these disorders, followed by the extensive eighth section on "Testing," including laboratory, imaging, and autonomic testing. The last section is on "Diagnoses." These nine sections are divided in the manner of the documentation of a classic history and physical examination. As the electronic health record advances, these categories may change, further supporting the notion of the data dictionary being a fluid, "living" document that will be prospectively updated every 3–5 years.

Pediatric autonomic disorders data dictionary

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Signs/symptoms	Objective evidence of disease perceptible to the examining practitioner (sign) and subjective evidence of disease perceived by the patient (symptom)			National Cancer Institute (NCI) Thesaurus	NCI Thesaurus Code C100104
Cardiovascular	Pertaining to the heart or blood vessels			NCI Thesaurus	NCI Thesaurus Code C25292
Dizziness	A nonspecific term indicating a sensation of lightheadedness, unsteadiness, turning, vertigo, or rocking			Modified from NCI Thesaurus AND Reilly BM (1990) Dizziness. In: Walker HK, et al. (eds.) Clinical Methods: The History, Physical, and Laboratory Examinations, 3 rd Ed. Butterworth, Boston, Ch. 212 [20]	NCI Thesaurus Code C37943
Lightheadedness	A sensation of faintness or wooziness that may be accompanied by visual or auditory changes, diaphoresis, nausea, or tachycardia			Modified from NCI Thesaurus	NCI Thesaurus Code C118255
Vertigo	A sensation of motion as if the external world is moving around the patient, or as if the patient is moving or spinning			Modified from NCI Thesaurus	NCI Thesaurus Code C38057
Blurred vision	The loss of visual acuity (sharpness of vision) resulting in a loss of ability to see small details			NCI Thesaurus	NCI Thesaurus Code C27123
Tunnel vision	Loss of peripheral vision with retention of central field of vision			www.medicinenet.com	
Blackout	A transient loss of vision characterized by perceived complete loss of light and color due to hypoxia and/or hypotension				
Grayout	A transient loss of vision characterized by a perceived dimming of light and color due to hypoxemia and/or hypotension				

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Whiteout	A transient loss of vision characterized by perceived brightening of light due to hypoxia and/or hypotension				
Presyncope	An episode of lightheadedness which may precede an episode of syncope			Modified from NCI Thesaurus	NCI Thesaurus Code C80100
Syncope	A transient loss of consciousness and postural tone due to cerebral hypoperfusion, characterized by a rapid onset, short duration, and spontaneous complete recovery			Brignole M, et al. Eur Heart J 2018;39:1883–1948 [21] 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Raynaud phenomenon	An episodic vasoconstriction followed by vasodilation resulting in discoloration of the skin and pain in the affected areas, often involving fingers or toes. Classically associated with triphasic color changes (white, blue, red) but may be biphasic. Often occurs in response to cold temperatures or emotional stress. May be primary or secondary to an underlying autoimmune disease			Modified from NCI Thesaurus	NCI Thesaurus Code C50724
Venous pooling	The accumulation of blood in veins of the extremities or splanchnic circulation due to gravitational pull when a person assumes an upright position				Also called dependent acrocyanosis
Excessive venous pooling	The disproportionate accumulation of blood in the veins when a person changes position from supine to seated position or standing, especially in the lower extremities, that can appear as acrocyanosis with or without edema			Stewart JM and Weldon A. J Appl Physiol (1985) 2000;89(4):1505–12 [22]	
Palpitations	Sensation of irregular, rapid, and/or forceful beating of the heart			Modified from NCI Thesaurus	NCI Thesaurus Code C37999
Tachycardia	An abnormally rapid heart rate exceeding the 95th percentile for age			Modified from NCI Thesaurus	NCI Thesaurus Code C38029
Exercise intolerance	A condition of inability or decreased ability to perform physical exercise at the normally expected level or duration for people of that age, size, sex, and muscle mass			Vissing J. International Neurology 2016 John Wiley & Sons p. 516. Hoboken. [23]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Neurologic	Pertaining to the nerves or nervous system			NCI Thesaurus	NCI Thesaurus Code C25262
Headache	Pain in various parts of the head, not necessarily confined to the area of distribution of any nerve			Modified from NCI Thesaurus	NCI Thesaurus Code C34661
Migraine	Recurrent headache disorder manifesting in attacks lasting 4–72 h. Typical characteristics of the headache are unilateral location, pulsating quality, moderate or severe intensity, aggravation by routine physical activity, and association with nausea and/or photophobia and phonophobia			Olesen J, et al. Cephalalgia 2018;38:1–211 [24]	
Tension-type headache	Episodes of headache, typically bilateral, pressing or tightening in quality and of mild to moderate intensity, lasting minutes to days. The pain does not worsen with routine physical activity and is not associated with nausea, although photophobia or phonophobia may be present			Olesen J, et al. Cephalalgia 2018;38:1–211 [24]	
Cluster headache	Attacks of severe, strictly unilateral pain which is orbital, supraorbital, temporal or in any combination of these sites, lasting 15–180 min, and occurring from once every other day to eight times a day. The pain is associated with ipsilateral conjunctival injection, lacrimation, nasal congestion, rhinorrhea, forehead and facial sweating, miosis, ptosis and/or eyelid edema, and/or with restlessness or agitation			Olesen J, et al. Cephalalgia 2018;38:1–211 [24]	
New daily persistent headache	Persistent headache, daily from its onset, which is clearly remembered. The pain lacks characteristic features, and may be migraine-like or tension-type-like, or have elements of both			Olesen J, et al. Cephalalgia 2018;38:1–211 [24]	
Fatigue	Overall tiredness and lack of energy that is not improved with rest			Modified from NCI Thesaurus	NCI Thesaurus Code C3036
Excessive fatigue	Overall tiredness and lack of energy disproportionate to the level of exertion				

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Brain Fog	An informal designation used to refer to several different forms of cognitive dysfunction characterized by slowed mentation, memory loss or dysfunction, and/or difficulty concentrating				
Insomnia	A sleep disorder characterized by difficulty in falling asleep and/or remaining asleep			NCI Thesaurus	NCI Thesaurus Code C28286
Paresthesia	Altered sensation, as in tingling or “pins and needles”			NCI Thesaurus	NCI Thesaurus Code C50771
Pain	An unpleasant sensory and emotional experience associated with, or resembling that associated with, actual or potential tissue damage			Raja SN, et al. Pain 2020;161(9):1976–82 [25]	
Myalgia	Painful sensation originating from a muscle or group of muscles			NCI Thesaurus	NCI Thesaurus Code C27009
Arthralgia	Pain in a joint			NCI Thesaurus	NCI Thesaurus Code C50464
Abdominal pain	Painful sensation in the abdominal region			NCI Thesaurus	NCI Thesaurus Code C26682
Pelvic pain	Painful sensation in the pelvis			NCI Thesaurus	NCI Thesaurus Code C40122
Heat intolerance	A feeling of being overheated when the surrounding temperature rises			https://medlineplus.gov/ency/article/003094.htm	
Cold intolerance	An abnormal sensitivity to a cold environment or cold temperatures			https://medlineplus.gov/ency/article/003095.htm	
Diaphoresis	A clinical finding in which there is excessive or unpredictable sweating			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Hyperhidrosis	Excessive perspiration			NCI Thesaurus	NCI Thesaurus Code C78345
Hypohidrosis	Impaired ability (partial or complete) to produce or deliver sweat to the surface of the skin in the presence of an appropriate stimulus			Leung AKC J R Soc Promot Health 1999;119(2):101–7 [26]	
Anhidrosis	Lack of sweating or the ability to sweat when provoked by the appropriate stimulus			NCI Thesaurus	NCI Thesaurus Code C34385

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Alacrima	Lack of tears due to absence or hyosecretion of tears, or to agenesis or improper development of lacrimal gland ducts and associated structures			Adams J et al. Clin Genet 2018;94(1):54–60 [27]	
Hyperpyrexia	Extreme elevation of body temperature above normal			NCI Thesaurus	NCI Thesaurus Code C111817
Xerophthalmia	Dryness of the eye(s) due to inadequate production of tears			Modified from NCI Thesaurus	NCI Thesaurus Code C34503
Numbness	Decrease or loss of superficial sensation in an anatomic region of the body			Modified from NCI Thesaurus	NCI Thesaurus Code C34857
Photophobia	Increased sensitivity of the eyes to light, which can result in the avoidance of light exposure			NCI Thesaurus	NCI Thesaurus Code C28210
Hyperacusis	A disorder in which an individual has an abnormally low noise tolerance, and increased sensitivity to sounds			NCI Thesaurus	NCI Thesaurus Code C116366
Seizure	A transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain			Fisher RS, et al. Epilepsia 2005;46(4):470–2 [28]	
Gastrointestinal	Pertaining to the upper alimentary tract, stomach, or intestines			Modified from NCI Thesaurus	NCI Thesaurus Code C13359
Nausea	A difficult-to-describe sensation of sick or queasy feeling usually perceived as being in the stomach that can escalate in severity and may precede vomiting			Stern RM, Koch KL, Andrews PLR (2001) Nausea: Mechanisms and Management. Oxford University Press, New York [29]	
Vomiting/emesis	Forceful ejection of the contents of the stomach through the mouth			NCI Thesaurus	NCI Thesaurus Code C3442
Early satiety	The sensation of feeling full or no longer hungry earlier while consuming a non-oversized meal or after eating less than usual preventing completion of a meal			Modified from www.medlineplus.gov	
Constipation	Difficulties in defecation that include infrequent bowel movements, hard or lumpy stools, excessive straining, sensation of incomplete evacuation or blockage and, in some instances, the use of manual maneuvers to facilitate evacuation			Aziz I, et al. Expert Rev Gastroenterol Hepatol 2020;14(1):39–46 [30]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Diarrhea	A disorder characterized by passage of three or more loose or liquid stools per day (or more frequent passage than is normal for the individual)			www.who.int/news-room/fact-sheets/detail/diarrhoeal-disease	
Abdominal bloating	The feeling of increased pressure within the abdomen			Iovino P, et al. World J Gastroenterol 2014;20(39):14407–19 [31]	
Abdominal distension	Protrusion of the abdomen			NCI Thesaurus	NCI Thesaurus Code C78161
Visceral hypersensitivity	Heightened sensitivity or reduced thresholds for pain and discomfort associated with the gastrointestinal tract			Modified from Mertz H. Aliment Pharmacol Ther 2003;17(5):623–33 [32] AND Faure C, Di Lorenzo C, Thapar N. (2017) Pediatric Neurogastroenterology Gastrointestinal Motility and Functional Disorders in Children, 2 nd Ed. Springer International Publishing, Switzerland [33]	Also called visceral hyperalgesia
Xerostomia	Dryness of the oral mucosa secondary to a decrease in saliva production, or a change in saliva composition			NCI Thesaurus	NCI Thesaurus Code C26917
Dysphagia	A symptom referring to difficulty in swallowing			NCI Thesaurus	NCI Thesaurus Code C2980
Aerophagia	An episodic or chronic disorder in which patients swallow large quantities of air which accumulate in the gastrointestinal tract to cause abdominal distention and bloating			Bredenoord AJ Clin Gastroenterol Hepatol 2013;11(1):6–12 [34]	
Respiration/control of breathing	Pertaining to the organs involved in the exchange of carbon dioxide and oxygen, including the trachea, bronchi, and lung parenchyma			Modified from NCI Thesaurus	NCI Thesaurus Code C25656
Apnea	Transient cessation of respiration lasting longer than 6 s			Modified from NCI Thesaurus	NCI Thesaurus Code C26698
Obstructive sleep apnea	Cessation of air flow during sleep due to upper airway obstruction			NCI Thesaurus	NCI Thesaurus Code C116337
Dyspnea	A subjective experience of shortness of breath				
Exertional dyspnea	A subjective sense of shortness of breath with physical exertion			Pianos PT Pediatr Exerc Sci 2018;30(4):442–9 [35]	
Cyanosis	A bluish or purplish discoloration of the skin and/or mucous membranes resulting from an increased amount of deoxygenated hemoglobin in the blood			Modified from NCI Thesaurus	NCI Thesaurus Code C26737

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Central cyanosis	Cyanosis that is noted to occur over the entire body surface area and visible mucosa			www.ncbi.nlm.nih.gov	
Perioral cyanosis	Cyanosis that is noted to occur surrounding the mouth that may or may not include the lips				
Peripheral cyanosis	Cyanosis that is noted to occur in the distal upper and lower extremities			Modified from www.ncbi.nlm.nih.gov	Also called acrocyanosis
Breath-holding spell	A clinical condition in which there is a brief involuntary cessation of breathing at end expiration in response to emotional or painful stimulus seen in infants, toddlers, and children			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Simple breath-holding spell	A clinical condition in which there is a breath-holding spell without syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Complex breath-holding spell	A clinical condition in which there is a breath-holding spell associated with syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Pallid breath-holding spell	A clinical condition in which there is a breath-holding spell associated with pale appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Cyanotic breath-holding spell	A clinical condition in which there is a breath-holding spell associated with cyanotic appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Breath holding	Episodic apnea, which is more common in young children and usually resolves by age six			NCI Thesaurus	NCI Thesaurus Code C121558
Chest-related symptoms					
Chest pain	Discomfort felt in the upper abdomen, thorax, neck, or shoulders			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Chest pressure	Sense of weight or compression felt in the thorax that may radiate to contiguous body parts				

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Chest wall pain	Discomfort felt in the upper abdomen, thorax, neck, or shoulders whose source is musculoskeletal. Pain may be reproducible by palpation				
Other systems					
Joint hypermobility	The capability that a joint (or a group of joints) has to move, passively and/or actively, beyond normal limits along physiological axes			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	
Easy bleeding	Bleeding that is disproportionate to the offending trauma			NCI Thesaurus	NCI Thesaurus Code C95552
Easy bruising	Appearance of skin bruises following perceived minimal contact or injury			NCI Thesaurus	NCI Thesaurus Code C119027
Joint subluxation	A partial or brief and reversible abnormal separation where two or more bones meet				
Joint dislocation	An abnormal separation where two or more bones meet			NCI Thesaurus	NCI Thesaurus Code C26750
Hyperelastic skin	Skin that can be stretched beyond what is considered normal, and returns to normal after it is stretched			www.medlineplus.gov	
Dysmenorrhea	Abnormally painful abdominal cramps during menstruation			NCI Thesaurus	NCI Thesaurus Code C34559
Menorrhagia	Heavy bleeding during menstruation			Modified from NCI Thesaurus	NCI Thesaurus Code C26829
Metrorrhagia	Uterine bleeding that occurs during a time not associated with the normal menstrual period			NCI Thesaurus	NCI Thesaurus Code C34818
Pruritus	An intense itching sensation			NCI Thesaurus	NCI Thesaurus Code C3344
Urticaria	A transient, itchy skin eruption characterized by raised pale wheals with a surrounding red flare			Modified from NCI Thesaurus	NCI Thesaurus Code C3432
Flushing	Reddening of the skin from a transient increase in blood flow			NCI Thesaurus	NCI Thesaurus Code C26775
Dermatographism (Darier sign)	Urticaria due to physical allergy in which a pale, raised welt or wheal with a red flare on each side is elicited by stroking or scratching the skin			https://medical-dictionary.thefreedictionary.com/dermatographism	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Food intolerance	An adverse reaction to food(s) in which there is difficulty digesting or metabolizing a particular food not due to an immunologic etiology			Bruijnzeel-Koomen C, et al. Allergy 1995;50(8):623–35 [37]	
Food allergies	An adverse reaction to food(s) due to an immunologic mechanism			Sicherer SH and Sampson HA. J Allergy Clin Immunol 2014;133(2):291–307 [38]	
Triggers	Something that sets into motion some course of events; a device that activates or releases or causes something to happen			NCI Thesaurus	NCI Thesaurus Code C54190
Infection	The invasion and multiplication of microorganisms such as bacteria, <u>viruses</u> , and parasites that are not normally present within the body			www.medicinenet.com	
Epstein–Barr virus	A species of Herpetoviridae that is responsible for infectious mononucleosis (glandular fever)			NCI Thesaurus	NCI Thesaurus Code C14204
Mononucleosis	A clinical entity characterized by pharyngitis, cervical lymph node enlargement, fatigue and fever, which results most often from a primary Epstein–Barr virus infection			Balfour HH Jr, et al. Clin Transl Immunology 2015;4(2):e33 [39]	
Upper respiratory tract infection	An infectious process affecting the upper respiratory tract (nose, paranasal sinuses, pharynx, or larynx)			Modified from NCI Thesaurus	NCI Thesaurus Code C35650
Influenza	An acute viral infection of the respiratory tract, occurring in isolated cases, in epidemics, or in pandemics; it is caused by serologically different strains of viruses (influenza viruses) designated A, B, and C, has a 3-day incubation period, and usually lasts for 3–10 days			NCI Thesaurus	NCI Thesaurus Code C53482
Gastroenteritis	An inflammatory disorder that affects the upper and/or lower gastrointestinal tract			Modified from NCI Thesaurus	NCI Thesaurus Code C34632
Pneumonia	An acute, acute and chronic, or chronic inflammation focally or diffusely affecting the lung parenchyma, due to infections, treatment (e.g., radiation), or exposure (inhalation) to chemicals			NCI Thesaurus	NCI Thesaurus Code C3333

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Sinusitis	An acute or chronic inflammatory process affecting the mucous membranes of any sinus cavity			NCI Thesaurus	NCI Thesaurus Code C35024
Varicella	A highly contagious viral infection caused by the varicella zoster virus			NCI Thesaurus	NCI Thesaurus Code C96407
Severe acute respiratory syndrome coronavirus 2	A positive-sense single-stranded RNA virus in the genus <i>Betacoronavirus</i>			NCI Thesaurus	NCI Thesaurus Code C169076 Also called SARS-CoV-2, SARS coronavirus 2
COVID-19 infection	An acute infection of the respiratory tract that is caused by the severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2)			NCI Thesaurus	NCI Thesaurus Code C171133
Enterovirus	A (+) strand single-strand RNA virus of the picornavirus family			NCI Thesaurus	NCI Thesaurus Code C14203
Hand, foot, and mouth disease	A clinical syndrome that is usually caused by enterovirus infection, and that is characterized by fever, anorexia, and painful sores in the mouth, distal extremities, and/or other sites, including the buttocks			NCI Thesaurus	NCI Thesaurus Code C128439
Cellulitis	Inflammation of the dermis and subcutaneous tissues caused by a bacterial infection			NCI Thesaurus	NCI Thesaurus Code C26715
Appendicitis	Acute inflammation of the vermiform appendix			NCI Thesaurus	NCI Thesaurus Code C35145
Concussion	Concussion is a traumatic brain injury induced by biomechanical forces			McCrory P, et al. Br J Sports Med 2017;51(11):877–887 [40]	
Surgery	A diagnostic or treatment procedure performed by manual and/or instrumental means, often involving an incision and the removal or replacement of a diseased organ or tissue			NCI Thesaurus	NCI Thesaurus Code C15329
Menarche	Onset of menses			NCI Thesaurus	NCI Thesaurus Code C3228
Adolescent growth spurt	The fast and intense increase in the rate of growth in height and weight that occurs during the pubertal stage of the human life cycle			Modified from Soliman A, et al. Indian J Endocrinol Metab 2014;18(suppl 1):S53-S62 [41]	
Co-morbid disorders	The coexistence of two or more disease processes			NCI Thesaurus	NCI Thesaurus Code C16457

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Syncope	A transient loss of consciousness and postural tone due to cerebral hypoperfusion, characterized by a rapid onset, short duration, and spontaneous complete recovery			Brignole M, et al. Eur Heart J 2018;39:1883–1948 [21] 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Reflex syncope	A transient loss of consciousness and postural tone due to a reflex response that encompasses vasodilation and/or bradycardia leading to systemic hypotension and cerebral hypoperfusion			https://www.uptodate.com/contents/reflex-syncope-in-adults-and-adolescents-treatment 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17] Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Neurally mediated syncope	A type of reflex syncope typically triggered by orthostasis leading to vasodilation and/or bradycardia that includes vasodepressor cardioinhibitory, and mixed syncope			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	Also called vasovagal syncope, neurocardiogenic syncope
Vasodepressor syncope	A type of neurally mediated syncope characterized primarily by reflex vasodilation			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Cardioinhibitory syncope	A type of neurally mediated syncope characterized primarily by reflex bradycardia			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Mixed syncope	A type of neurally mediated syncope characterized by reflex vasodilation and reflex bradycardia			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Situational syncope	A type of reflex syncope typically triggered by specific events, such as micturition, gastrointestinal stimulation, cough or sneeze, or exercise			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Syncope due to orthostatic hypotension	A syncopal event due to an inadequate increase in peripheral resistance and heart rate or due to severe hypovolemia upon standing			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Orthostatic hypotension	A progressive and sustained drop in blood pressure upon standing from a sitting or supine position in which there is a decrease of systolic blood pressure by ≥ 20 mmHg and/or a decrease of diastolic blood pressure by ≥ 10 mmHg within the first 3 min of upright position			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Initial orthostatic hypotension	A sudden drop in blood pressure upon standing from a sitting or supine position in which there is a decrease of systolic blood pressure by ≥ 40 mmHg and/or a decrease in diastolic blood pressure by ≥ 20 mmHg within the first 15 s of standing that spontaneously resolves with a relatively rapid return to pre-challenge values			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Cardiac syncope	A syncopal event due to a primary cardiac etiology, such as an arrhythmia or structural lesion				
Postural orthostatic tachycardia syndrome	A type of chronic orthostatic intolerance lasting ≥ 3 months associated with excessive upright sinus tachycardia in the absence of orthostatic hypotension, plus a constellation of typically daily symptoms that may include lightheadedness, dizziness, nausea, dyspnea, diaphoresis, headache, fatigue, and other symptoms of autonomic dysfunction. Excessive tachycardia is defined by present consensus as a sustained heart rate increase of at least 30 bpm in adults (40 bpm for adolescents), or a heart rate > 120 bpm (in adults), within 10 min of upright tilt table testing			Modified from 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	Also called POTS

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hyperadrenergic postural orthostatic tachycardia syndrome	A phenotype of postural orthostatic tachycardia syndrome in which there is excessive sympathetic vasoconstriction, increased peripheral resistance, and typically excessively high serum catecholamine levels, with greatly elevated upright tachycardia and hypertension				
Neuropathic postural orthostatic tachycardia syndrome	A phenotype of postural orthostatic tachycardia syndrome in which there is impaired sympathetic activity in the lower extremities or splanchnic vasculature with arteriolar dysregulation				
Dysautonomia	A condition in which altered function of one or more components of the autonomic nervous system adversely affects health			Goldstein DS Principles of Autonomic Medicine v 4.0 [42]	Also called autonomic nervous system dysfunction
Post-COVID-19 syndrome	A condition in which there are multiorgan symptoms and complications beyond the initial period of acute infection and illness with SARS-CoV-2			Venkatesan P Lancet Respir Med 2021;9(2):129 [43]	Also called long COVID, long-haul COVID, post-acute sequelae of SARS-CoV-2 infection (PASC)
Myalgic encephalomyelitis-chronic fatigue syndrome	A syndrome of unknown etiology characterized by an unexplained persistent or relapsing chronic fatigue that is of at least 6 months' duration, is not the result of ongoing exertion, is not substantially alleviated by rest, and results in substantial reduction of previous levels of occupational, educational, social, or personal activities			NCI Thesaurus	NCI Thesaurus Code C3037
Inappropriate sinus tachycardia	A sinus heart rate > 100 bpm at rest (with a mean 24-h heart rate > 90 bpm not due to primary causes) and is associated with distressing symptoms of palpitations			Sheldon RS Heart Rhythm 2015;12(6):e41-e63 [44]	
Hypertension	The average clinic-measured systolic and/or diastolic blood pressure \geq 95th percentile for age, sex, and height percentiles			Flynn JT Pediatrics 2017;140(3):e20171904 [45]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hypotension	Low systolic blood pressure, as defined by age parameters: 0–1 year: < 5th percentile for gestational and/or postnatal age 1–10 years: < 70 mmHg plus 2 X (age in years) > 10 years: < 90/50 mmHg			Dionne J Hypertension 2021;77(3):781–787 [46] Kleinman ME Circulation 2010;122(18Suppl3):S876-S908 [47]	
Celiac disease	An autoimmune genetic disorder with an unknown pattern of inheritance that damages the small intestine due to intolerance to dietary gluten			Modified from NCI Thesaurus	NCI Thesaurus Code C26714
Thyroid disease	A disorder in which there is abnormal function of the thyroid gland due to causes such as autoimmune disease, pituitary disease, toxins, or malignancy				
Hashimoto thyroiditis	An autoimmune disorder caused by the production of autoantibodies against thyroid tissue. There is progressive destruction of the thyroid follicles leading to hypothyroidism			NCI Thesaurus	NCI Thesaurus Code C27191
Graves disease	Hyperthyroidism associated with diffuse hyperplasia of the thyroid gland (goiter), resulting from production of antibodies that are directed against the thyrotropin receptor complex of the follicular epithelial cells. As a result, the thyroid gland enlarges and secretes increased amounts of thyroid hormones			NCI Thesaurus	NCI Thesaurus Code C3071
Eosinophilic esophagitis	Allergic inflammation of the esophagus. Morphologically, it is characterized by the presence of eosinophils infiltrating the esophageal epithelium			NCI Thesaurus	NCI Thesaurus Code C27105
Diabetes mellitus	A metabolic disorder characterized by abnormally high blood sugar levels due to diminished production of insulin or insulin resistance/desensitization			NCI Thesaurus	NCI Thesaurus Code C2985
Type 1 diabetes mellitus	A chronic condition characterized by minimal or absent production of insulin by the pancreas			NCI Thesaurus	NCI Thesaurus Code C2986

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Type 2 diabetes mellitus	A type of diabetes mellitus that is characterized by insulin resistance or desensitization and increased blood glucose levels			NCI Thesaurus	NCI Thesaurus Code C26747
Inflammatory bowel disease	A spectrum of small and large bowel inflammatory diseases of unknown etiology. It includes Crohn's disease, ulcerative colitis, microscopic colitis, and colitis of indeterminate type			Modified from NCI Thesaurus	NCI Thesaurus Code C3138
Crohn's disease	A gastrointestinal disorder characterized by chronic inflammation involving all layers of the intestinal wall, noncaseating granulomas affecting the intestinal wall and regional lymph nodes, and transmural fibrosis			NCI Thesaurus	NCI Thesaurus Code C2965
Ulcerative colitis	An inflammatory bowel disease involving the mucosal surface of the large intestine and rectum			NCI Thesaurus	NCI Thesaurus Code C2952
Irritable bowel syndrome	Gastrointestinal symptoms characterized by chronic abdominal pain and altered bowel habits in the absence of any organic cause. One of the functional gastrointestinal disorders			Modified from NCI Thesaurus	NCI Thesaurus Code C82343
Visceral hypersensitivity	Heightened sensitivity or reduced thresholds for pain and discomfort associated with the gastrointestinal tract			Modified from Mertz H. Aliment Pharmacol Ther 2003;17(5):623–33 [32] AND Faure C, Di Lorenzo C, Thapar N. (2017) Pediatric Neurogastroenterology Gastrointestinal Motility and Functional Disorders in Children, 2 nd Ed. Springer International Publishing, Switzerland [33]	Also called visceral hyperalgesia
Joint hypermobility	The capability that a joint (or a group of joints) has to move, passively and/or actively, beyond normal limits along physiological axes			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	
Ehlers–Danlos syndrome	A group of inherited connective tissue disorders characterized by loose and fragile skin and joint hypermobility			NCI Thesaurus	NCI Thesaurus Code C34568 Also called EDS

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hypermobile EDS	A type of Ehlers–Danlos syndrome characterized by generalized joint hypermobility, plus systemic manifestations of a generalized connective tissue disorder, positive family history, and/or musculoskeletal complications, plus exclusion of other types of Ehlers–Danlos syndrome			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called hEDS
Classical EDS	A type of Ehlers–Danlos syndrome characterized by skin hyperextensibility, generalized joint hypermobility, plus other minor criteria associated with a mutation in collagen in the <i>COL5A1</i> , <i>COL5A2</i> , <i>COL1A1</i> , or <i>COL1A2</i> genes			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called cEDS
Vascular EDS	A type of Ehlers–Danlos syndrome characterized by arterial, uterine, or gastrointestinal rupture at a young age, atraumatic formation of carotid-cavernous fistula, plus other minor criteria associated with a mutation in collagen in the <i>COL3A1</i> gene			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called vEDS
Hypermobility spectrum disorder	A group of clinically relevant conditions related to joint hypermobility in which phenotypic domains are limited to the musculoskeletal system			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	Also called HSD
Mast cell activation syndrome	A mast cell activation disorder in which there are episodic multisystem symptoms consistent with mast cell activation, appropriate response to medications targeting mast cell activation, and documented increase in validated markers of mast cell activation			Akin C. J Allergy Clin Immunol 2017;140(2):349–55 [49]	Also called MCAS
Median arcuate ligament syndrome	A condition characterized by abdominal pain attributed to compression of the celiac artery and the celiac ganglia by the median arcuate ligament			www.wikipedia.org	Also called MALS

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Amplified musculoskeletal pain syndrome	A condition in which the nerves register normal stimuli (lack of tissue injury) as pain, resulting in feelings of intense pain with even minor sensory input across the entire body			Modified from NCI Thesaurus	NCI Thesaurus Code C119048
Chronic regional pain syndrome	A chronic (lasting longer than 6 months) pain condition that most often affects one limb usually after an injury characterized by prolonged or excessive pain and changes in skin color, temperature, and/or swelling in the affected area			www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Complex-Regional-Pain-Syndrome-Fact-Sheet	
Cyclic vomiting syndrome	A condition characterized by recurrent, self-limiting stereotypical episodes of vomiting associated with intense nausea, pallor, and lethargy. One of the functional gastrointestinal disorders, it can be a migraine precursor			Modified from NCI Thesaurus	NCI Thesaurus Code C117014
Anorexia nervosa	A disorder characterized by a refusal to maintain a minimally normal body weight, an intense fear of gaining weight, a disturbance in body image, and, in postmenarchal females, the development of amenorrhea			Modified from NCI Thesaurus	NCI Thesaurus Code C34387
Bulimia nervosa	A disorder characterized by recurrent episodes of binge-eating over which the individual feels a lack of control; these episodes of binge-eating are followed by recurrent compensatory behavior to prevent weight gain, usually self-induced vomiting			NCI Thesaurus	NCI Thesaurus Code C34440
Binge-eating disorder	A disorder characterized by recurrent episodes of binge-eating without the affected individual's effort to make up for the binge episodes by excessively exercising, purging, and/or inappropriately using medications, like diet pills or laxatives			www.medicinenet.com	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Avoidant/restrictive food intake disorder	An eating or feeding disturbance (e.g., apparent lack of interest in eating or food; avoidance based on the sensory characteristics of food; concern about aversive consequences of eating) as manifested by persistent failure to meet appropriate nutritional and/or energy needs associated with one (or more) of the following: 1. Significant weight loss (or failure to achieve expected weight gain or faltering growth in children) 2. Significant nutritional deficiency 3. Dependence on enteral feeding or oral nutritional supplements 4. Marked interference with psychosocial functioning			DSM-V	
Overweight	Suggested by having a high amount of body fat in which the body mass index is between 25 and 29.9 kg/m ² (adult criteria), or at/above the 85th up to 95th percentile for age and gender. This is confirmed by other more precise methods			Modified from NCI Thesaurus PLUS Centers for Disease Control	NCI Thesaurus Code C94250
Obesity	Suggested by having a high amount of body fat in which the body mass index (BMI) is 30 kg/m ² or more (adult criteria), or at/above the 95th percentile for age and gender. This is confirmed by other more precise methods			Modified from NCI Thesaurus PLUS Centers for Disease Control	NCI Thesaurus Code C3283
Anxiety disorder	A category of psychiatric disorders which are characterized by anxious feelings or fear often accompanied by physical symptoms associated with anxiety			NCI Thesaurus	NCI Thesaurus Code C2878
Panic disorder	An anxiety disorder characterized by multiple unexpected panic attacks with persistent concern of recurring attacks			NCI Thesaurus	NCI Thesaurus Code C34890
Depression	A mental condition marked by ongoing feelings of sadness, despair, loss of energy, and difficulty dealing with normal daily life			NCI Thesaurus	NCI Thesaurus Code C2982

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Obsessive–compulsive disorder	A disorder characterized by the presence of persistent and recurrent irrational thoughts (obsessions), resulting in marked anxiety and repetitive excessive behaviors (compulsions) as a way to try to decrease that anxiety			NCI Thesaurus	NCI Thesaurus Code C88411
Attention deficit/hyperactivity disorder	A neurodevelopmental disorder defined by impairing levels of inattention, disorganization, and/or hyperactivity–impulsivity			DSM-V	Also called ADHD
Attention deficit/hyperactivity disorder, combined presentation	ADHD in which both inattention and hyperactivity–impulsivity are met for the past 6 months			DSM-V	
Attention deficit/hyperactivity disorder, predominantly inattentive presentation	ADHD in which inattention is met but hyperactivity–impulsivity is not met for the past 6 months			DSM-V	
Attention deficit/hyperactivity disorder, predominantly hyperactive–impulsive presentation	ADHD in which hyperactivity–impulsivity is met but inattention is not met for the past 6 months			DSM-V	
Oppositional defiant disorder	A behavior disorder characterized by a persistent pattern of defiant, disobedient, and hostile behavior towards authority figures, manifested by a frequent loss of temper, arguing, becoming angry or vindictive, or other negativistic behaviors			NCI Thesaurus	NCI Thesaurus Code C92565
Somatic symptom disorder	A. One or more somatic symptoms that are distressing or result in significant disruption of daily life B. Excessive thoughts, feelings, or behaviors related to the somatic symptoms or associated health concerns C. Although any one somatic symptom may not be continuously present, the state of being symptomatic is persistent (typically more than 6 months)			DSM-V	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Conversion disorder (functional neurological symptom disorder)	<p>A. One or more symptoms of altered voluntary motor or sensory function</p> <p>B. Clinical findings provide evidence of incompatibility between the symptom and recognized neurological or medical conditions</p> <p>C. The symptom or deficit is not better explained by another medical or mental disorder</p> <p>D. The symptom or deficit causes clinically significant distress or impairment in social, occupational, or other important areas of functioning or warrants medical evaluation</p>			DSM-V	
Functional gastrointestinal disorders	A group of gastrointestinal disorders that occur in which, after appropriate medical evaluation, the symptoms cannot be attributed to another medical condition. This includes functional nausea and vomiting disorders, functional abdominal pain disorders, and functional defecation disorders			Adapted from Hyams JS, et al. Gastroenterology 2016; S0016-5085(16)00181-5 [50]	
Sjögren syndrome	An autoimmune disorder affecting the salivary and lacrimal glands. Morphologically, it is characterized by the presence of lymphocytic and plasmacytic infiltrates that cause destruction of these glands			NCI Thesaurus	NCI Thesaurus Code C26883
Systemic lupus erythematosus	An autoimmune multiorgan disease typically associated with vasculopathy and autoantibody production			NCI Thesaurus	NCI Thesaurus Code C3201
Juvenile idiopathic arthritis	A group of chronic, inflammatory childhood disorders of unknown etiology that primarily involve joints			NCI Thesaurus	NCI Thesaurus Code C114357
Polycystic ovary syndrome	A disorder that manifests as multiple cysts on the ovaries. It results in hormonal imbalances and leads to irregular and abnormal menstrual periods, excess growth of hair, acne eruptions, and obesity			NCI Thesaurus	NCI Thesaurus Code C26862

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Endometriosis	The growth of functional endometrial tissue in anatomic sites outside the uterine body			NCI Thesaurus	NCI Thesaurus Code C3014
Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation	A neurocristopathy characterized by hypothalamic dysfunction with inability to maintain normal salt–water balance, growth hormone deficiency, hypothyroidism, early or late puberty, increased prolactin concentration, hypoventilation, attenuated/absent peripheral and central chemoreceptor responsiveness, and autonomic dysfunction			Modified from NCI Thesaurus	NCI Thesaurus Code C121944 Also called ROHHAD
Congenital central hypoventilation syndrome	A neurocristopathy associated with heterozygous <i>PHOX2B</i> gene mutations characterized by hypoventilation and hypoxemia typically during sleep with attenuated/absent peripheral and central chemoreceptor responsiveness, and autonomic dysregulation; symptoms typically appear in the newborn period and can co-occur with Hirschsprung disease and neural crest tumors			Modified from NCI Thesaurus	NCI Thesaurus Code C98889 Also called CCHS
Tracheostomy	A surgically created external opening into the trachea			NCI Thesaurus	NCI Thesaurus Code C122643
Hereditary and sensory neuropathy, type I	An autosomal dominant condition caused by mutation(s) in the <i>SPTLC1</i> gene characterized by distal sensory impairment with variable autonomic and motor involvement			NCI Thesaurus	NCI Thesaurus Code C170433
Hereditary and sensory neuropathy, type IIA (HSAN2A)	A rare congenital disorder caused by various mutations in the <i>WNK1</i> gene characterized by sensory loss, self-mutilation, and mild or absent autonomic involvement			Modified from https://www.ncbi.nlm.nih.gov/books/NBK49247/ and https://www.omim.org/entry/201300	
Hereditary and sensory neuropathy, type IIB (HSAN2B, Morvan's disease; congenital sensory neuropathy)	A rare autosomal recessive disorder caused by various mutations in the <i>FAM134B</i> gene characterized by decreased distal sensation and variable autonomic involvement			Modified from https://www.ncbi.nlm.nih.gov/books/NBK49247/ and https://www.omim.org/entry/613115	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hereditary and sensory neuropathy, type III (HSAN3, familial dysautonomia; Riley–Day syndrome)	A rare congenital disorder caused by a founder mutation in the <i>ELP-1</i> gene characterized by initial hypotonia and feeding difficulties with subsequent blood pressure instability and hypertensive vomiting attacks alternating with hypotension due to loss of sensory inputs from cranial nerves IX and X resulting in afferent baroreflex failure			Modified from https://www.omim.org/entry/223900	
Hereditary and sensory neuropathy, type IV (HSAN4, congenital insensitivity to pain with anhidrosis; CIPA)	A rare autosomal recessive disorder caused by different mutations in the <i>NTRK1</i> gene affecting peripheral sensory and autonomic fibers, and is characterized by inability to feel pain and temperature that leads to repeated unintentional self-injuries, severe cognitive impairment, and anhidrosis that leads to hyperpyrexia and febrile seizures			Modified from NCI Thesaurus and https://www.omim.org/entry/256800	NCI Thesaurus Code C118633
Hereditary and sensory neuropathy, type V (HSAN5)	An autosomal recessive condition caused by mutations in the <i>NGF</i> or <i>NTRK1</i> genes affecting peripheral small fibers characterized by loss of pain sensation, particularly in the extremities, and variable anhidrosis, with normal cognitive function as compared with HSAN4			Modified from NCI Thesaurus and https://www.omim.org/entry/608654	NCI Thesaurus Code C156360
Hereditary and sensory neuropathy, type VI (HSAN6)	An extremely rare inherited disorder caused by mutations in the <i>DST</i> gene characterized by neonatal expression of diminished pain and labile blood pressure			https://www.ncbi.nlm.nih.gov/medgen/761278 and https://www.omim.org/entry/614653	
Hereditary and sensory neuropathy, type VII (HSAN7)	A rare disorder caused by mutations in the <i>SCN11A</i> gene, characterized by insensitivity to pain as well as gastrointestinal dysfunction			Modified from NCI Thesaurus and https://www.omim.org/entry/615548	NCI Thesaurus Code C125388
Hereditary and sensory neuropathy, type VIII (HSAN8)	An autosomal recessive disorder caused by mutations in the <i>PRDM12</i> gene characterized by congenital insensitivity to pain; anhidrosis and alacrima may occur			Modified from https://www.ncbi.nlm.nih.gov/medgen/894363 and https://www.omim.org/entry/616488	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Rett syndrome	A progressive neurologic disorder caused by mutations in the <i>MECP2</i> gene on chromosome X. It almost exclusively affects girls. It is characterized by breath-holding spells, flushing, repetitive hand motions, lack of advancement in language, and regression in developmental milestones. Other signs and symptoms include slowed head growth, scoliosis, breathing abnormalities (including sleep-disordered breathing), and sleep disturbances			Modified from NCI Thesaurus	NCI Thesaurus Code C75488
Hirschsprung disease	A congenital disorder characterized by the absence of myenteric ganglion cells in the distal colon resulting in a functional stenosis of the distal colon and a massive distention of the proximal colon			NCI Thesaurus	NCI Thesaurus Code C34700
Neuroblastoma	A neuroblastic tumor characterized by the presence of neuroblastic cells, the absence of ganglion cells, and the absence of a prominent Schwannian stroma formation			NCI Thesaurus	NCI Thesaurus Code C3270
Ganglioneuroma	A benign neuroblastic tumor of the sympathetic nervous system that occurs in childhood. Ganglioneuroma typically arises from the sympathetic chain in the mediastinum or adrenal glands			Modified from NCI Thesaurus	NCI Thesaurus Code C3049
Ganglioneuroblastoma	A neuroblastic tumor characterized by the presence of neuroblastic cells, ganglion cells, and a stroma with Schwannian differentiation constituting more than 50% of the tumor volume			NCI Thesaurus	NCI Thesaurus Code C3790
Paraganglioma	A benign or malignant neoplasm arising from paraganglia located along the sympathetic or parasympathetic nerves, including carotid body tumors			Modified from NCI Thesaurus	NCI Thesaurus Code C3308

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Breath-holding spell	A clinical condition in which there is a brief involuntary cessation of breathing at end expiration in response to emotional or painful stimulus seen in infants, toddlers, and children			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Simple breath-holding spell	A clinical condition in which there is a breath-holding spell without syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Complex breath-holding spell	A clinical condition in which there is a breath-holding spell associated with syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Pallid breath-holding spell	A clinical condition in which there is a breath-holding spell associated with pale appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Cyanotic breath-holding spell	A clinical condition in which there is a breath-holding spell associated with cyanotic appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Seizure disorder	A brain disorder characterized by episodes of abnormally increased neuronal discharge resulting in transient episodes of sensory or motor neurological dysfunction, or psychic dysfunction that may or may not be associated with loss of consciousness or convulsions			NCI Thesaurus	NCI Thesaurus Code C3020 Also called epilepsy
Spinal cord injury	Traumatic damage of the spinal cord			NCI Thesaurus	NCI Thesaurus Code C50750
Autonomic dysreflexia	A form of paroxysmal autonomic adrenergic overactivity due to disinhibited spinal reflexes in spinal cord injury above the T6 level			Novak P. Am J Med 2019;132(4):420–436 [51]	
Family history	A record of a patient's background regarding health and disease events of blood relatives			NCI Thesaurus	NCI Thesaurus Code C17726

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Syncope	A transient loss of consciousness and postural tone due to cerebral hypoperfusion, characterized by a rapid onset, short duration, and spontaneous complete recovery			Brignole M, et al. Eur Heart J 2018;39:1883–1948 [21] 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Reflex syncope	A transient loss of consciousness and postural tone due to a reflex response that encompasses vasodilation and/or bradycardia leading to systemic hypotension and cerebral hypoperfusion			https://www.uptodate.com/contents/reflex-syncope-in-adults-and-adolescents-treatment 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17] Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Neurally mediated syncope	A type of reflex syncope typically triggered by orthostasis leading to vasodilation and/or bradycardia that includes vasodepressor cardioinhibitory, and mixed syncope			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	Also called vasovagal syncope, neurocardiogenic syncope
Vasodepressor syncope	A type of neurally mediated syncope characterized primarily by reflex vasodilation			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Cardioinhibitory syncope	A type of neurally mediated syncope characterized primarily by reflex bradycardia			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Mixed syncope	A type of neurally mediated syncope characterized by reflex vasodilation and reflex bradycardia			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Situational syncope	A type of reflex syncope typically triggered by specific events, such as micturition, gastrointestinal stimulation, cough or sneeze, or exercise			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Syncope due to orthostatic hypotension	A syncopal event due to an inadequate increase in peripheral resistance and heart rate or due to severe hypovolemia upon standing			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Orthostatic hypotension	A progressive and sustained drop in blood pressure upon standing from a sitting or supine position in which there is a decrease of systolic blood pressure by ≥ 20 mmHg and/or a decrease of diastolic blood pressure by ≥ 10 mmHg within the first 3 min of upright position			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Initial orthostatic hypotension	A sudden drop in blood pressure upon standing from a sitting or supine position in which there is a decrease of systolic blood pressure by ≥ 40 mmHg and/or a decrease in diastolic blood pressure by ≥ 20 mmHg within the first 15 s of standing that spontaneously resolves with a relatively rapid return to pre-challenge values			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Cardiac syncope	A syncopal event due to a primary cardiac etiology, such as an arrhythmia or structural lesion				
Postural orthostatic tachycardia syndrome	A type of chronic orthostatic intolerance lasting ≥ 3 months associated with excessive upright sinus tachycardia in the absence of orthostatic hypotension, plus a constellation of typically daily symptoms that may include lightheadedness, dizziness, nausea, dyspnea, diaphoresis, headache, fatigue, and other symptoms of autonomic dysfunction. Excessive tachycardia is defined by present consensus as a sustained heart rate increase of at least 30 bpm in adults (40 bpm for adolescents), or a heart rate > 120 bpm (in adults), within 10 min of upright tilt table testing			Modified from 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	Also called POTS

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hyperadrenergic postural orthostatic tachycardia syndrome	A phenotype of postural orthostatic tachycardia syndrome in which there is excessive sympathetic vasoconstriction, increased peripheral resistance, and typically excessively high serum catecholamine levels, with greatly elevated upright tachycardia and hypertension				
Neuropathic postural orthostatic tachycardia syndrome	A phenotype of postural orthostatic tachycardia syndrome in which there is impaired sympathetic activity in the lower extremities or splanchnic vasculature with arteriolar dysregulation				
Dysautonomia	A condition in which altered function of one or more components of the autonomic nervous system adversely affects health			Goldstein DS Principles of Autonomic Medicine v 4.0 [42]	Also called autonomic nervous system dysfunction
Post COVID-19 Syndrome	A condition in which there are multiorgan symptoms and complications beyond the initial period of acute infection and illness with SARS-CoV-2			Venkatesan P Lancet Respir Med 2021;9(2):129 [43]	Also called long COVID, long-haul COVID, post-acute sequelae of SARS-CoV-2 infection (PASC)
Inappropriate sinus tachycardia	A sinus heart rate > 100 bpm at rest (with a mean 24-h heart rate > 90 bpm not due to primary causes) and is associated with distressing symptoms of palpitations			Sheldon RS Heart Rhythm 2015;12(6):e41-e63 [44]	
Hypertension	The average clinic-measured systolic and/or diastolic blood pressure \geq 95th percentile for age, sex, and height percentiles			Flynn JT Pediatrics 2017;140(3):e20171904 [45]	
Myalgic encephalomyelitis-chronic fatigue syndrome	A syndrome of unknown etiology characterized by an unexplained persistent or relapsing chronic fatigue that is of at least 6 months' duration, is not the result of ongoing exertion, is not substantially alleviated by rest, and results in substantial reduction of previous levels of occupational, educational, social, or personal activities			NCI Thesaurus	NCI Thesaurus Code C3037

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Joint hypermobility	The capability that a joint (or a group of joints) has to move, passively and/or actively, beyond normal limits along physiological axes			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	
Hypermobility spectrum disorders	A group of clinically relevant conditions related to joint hypermobility in which phenotypic domains are limited to the musculo-skeletal system			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	Also called HSD
Ehlers–Danlos syndrome	A group of inherited connective tissue disorders characterized by loose and fragile skin and joint hypermobility			NCI Thesaurus	NCI Thesaurus Code C34568 Also called EDS
Hypermobile EDS	A type of Ehlers–Danlos syndrome characterized by generalized joint hypermobility, plus systemic manifestations of a generalized connective tissue disorder, positive family history, and/or musculoskeletal complications, plus exclusion of other types of Ehlers–Danlos syndrome			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called hEDS
Classical EDS	A type of Ehlers–Danlos syndrome characterized by skin hyperextensibility, generalized joint hypermobility, plus other minor criteria associated with a mutation in collagen in the <i>COL5A1</i> , <i>COL5A2</i> , <i>COL1A1</i> , or <i>COL1A2</i> genes			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called cEDS
Vascular EDS	A type of Ehlers–Danlos syndrome characterized by arterial, uterine, or gastrointestinal rupture at a young age, atraumatic formation of carotid-cavernous fistula, plus other minor criteria associated with a mutation in collagen in the <i>COL3A1</i> gene			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called vEDS
Dizziness	A nonspecific term indicating a sensation of lightheadedness, unsteadiness, turning, vertigo, or rocking			Modified from NCI Thesaurus AND Reilly BM (1990) Dizziness. In: Walker HK, et al. (eds.) Clinical Methods: The History, Physical, and Laboratory Examinations, 3 rd Ed. Butterworth, Boston, Ch. 212 [20]	NCI Thesaurus Code C37943

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Lightheadedness	A feeling of faintness or wooziness, or a sensation of motion within the head that may be accompanied by visual or auditory changes, diaphoresis, nausea, or tachycardia			Modified from NCI Thesaurus	NCI Thesaurus Code C118255
Vertigo	A feeling of movement, a sensation as if the external world was revolving around the patient			Modified from NCI Thesaurus	NCI Thesaurus Code C38057
Syncope	A transient loss of consciousness and postural tone due to cerebral hypoperfusion, characterized by a rapid onset, short duration, and spontaneous complete recovery			Brignole M, et al. Eur Heart J 2018;39:1883–1948 [14] 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Migraine	A common, severe type of vascular headache often associated with increased sympathetic activity, resulting in nausea, vomiting, and light sensitivity			NCI Thesaurus	NCI Thesaurus Code C89715
Mast cell activation syndrome	A mast cell activation disorder in which there are episodic multisystem symptoms consistent with mast cell activation, appropriate response to medications targeting mast cell activation, and documented increase in validated markers of mast cell activation			Akin C. J Allergy Clin Immunol 2017;140(2):349–55 [49]	Also called MCAS
Sjögren syndrome	An autoimmune disorder affecting the salivary and lacrimal glands. Morphologically, it is characterized by the presence of lymphocytic and plasmacytic infiltrates which cause destruction of these glands			NCI Thesaurus	NCI Thesaurus Code C26883
Systemic lupus erythematosus	An autoimmune multiorgan disease typically associated with vasculopathy and autoantibody production			NCI Thesaurus	NCI Thesaurus Code C3201
Rheumatoid arthritis	A chronic, systemic autoimmune disorder characterized by inflammation in the synovial membranes and articular surfaces			NCI Thesaurus	NCI Thesaurus Code C2884
Juvenile idiopathic arthritis	A group of chronic, inflammatory childhood disorders of unknown etiology that primarily involve joints			NCI Thesaurus	NCI Thesaurus Code C114357

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Thyroid disease	The classification of autoimmune thyroid disease (AITD) includes Hashimoto thyroiditis (HT) or chronic autoimmune thyroiditis and its variants, Graves disease (GD) and autoimmune atrophic thyroiditis or primary myxedema			NCI Thesaurus	NCI Thesaurus Code C91510
Hashimoto thyroiditis	An autoimmune disorder caused by the production of autoantibodies against thyroid tissue. There is progressive destruction of the thyroid follicles leading to hypothyroidism			NCI Thesaurus	NCI Thesaurus Code C27191
Graves disease	Hyperthyroidism associated with diffuse hyperplasia of the thyroid gland (goiter), resulting from production of antibodies that are directed against the thyrotropin receptor complex of the follicular epithelial cells. As a result, the thyroid gland enlarges and secretes increased amounts of thyroid hormones			NCI Thesaurus	NCI Thesaurus Code C3071
Pheochromocytoma	A benign or malignant neuroendocrine neoplasm of the sympathetic nervous system that secretes catecholamines. It arises from the chromaffin cells of the adrenal medulla			NCI Thesaurus	NCI Thesaurus Code C3326
Carcinoid syndrome	A syndrome characterized by signs and symptoms caused by advanced carcinoid tumors. They include skin flushing, diarrhea, wheezing, and tachycardia			NCI Thesaurus	NCI Thesaurus Code C3215
Paraganglioma	A benign or malignant neoplasm arising from paraganglia located along the sympathetic or parasympathetic nerves			NCI Thesaurus	NCI Thesaurus Code C3308
Hirschsprung disease	A congenital disorder characterized by the absence of myenteric ganglion cells in the distal colon			NCI Thesaurus	NCI Thesaurus Code C34700

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation	A neurocristopathy characterized by hypothalamic dysfunction with inability to maintain normal salt–water balance, growth hormone deficiency, hypothyroidism, early or late puberty, increased prolactin concentration, hypoventilation, attenuated/absent peripheral and central chemoreceptor responsiveness, and autonomic dysfunction			Modified from NCI Thesaurus	NCI Thesaurus Code C121944 Also called ROHHAD
Congenital central hypoventilation syndrome	A neurocristopathy associated with heterozygous <i>PHOX2B</i> gene mutations characterized by hypoventilation and hypoxemia typically during sleep with attenuated/absent peripheral and central chemoreceptor responsiveness, and autonomic dysregulation; symptoms typically appear in the newborn period and can co-occur with Hirschsprung disease and neural crest tumors			Modified from NCI Thesaurus	NCI Thesaurus Code C98889 Also called CCHS
Tracheostomy	A surgically created external opening into the trachea			NCI Thesaurus	NCI Thesaurus Code C122643
Neuroblastoma	A neuroblastic tumor characterized by the presence of neuroblastic cells, the absence of ganglion cells, and the absence of a prominent Schwannian stroma formation			NCI Thesaurus	NCI Thesaurus Code C3270
Ganglioneuroma	A benign neuroblastic tumor of the sympathetic nervous system that occurs in childhood. Ganglioneuroma typically arises from the sympathetic chain in the mediastinum or adrenal glands			Modified from NCI Thesaurus	NCI Thesaurus Code C3049
Ganglioneuroblastoma	A neuroblastic tumor characterized by the presence of neuroblastic cells, ganglion cells, and a stroma with Schwannian differentiation constituting more than 50% of the tumor volume			NCI Thesaurus	NCI Thesaurus Code C3790

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Neurofibromatosis type 1	An autosomal dominant form of neurofibromatosis caused by mutation(s) in the <i>NF1</i> gene, encoding neurofibromin. The clinical features of this condition may include smooth bordered areas of hyperpigmentation, known as cafe-au-lait spots, axillary freckling, Lisch nodules, short stature, cutaneous and subcutaneous tumors (neurofibromas), optic nerve gliomas, precocious puberty, and growth hormone excess			NCI Thesaurus	NCI Thesaurus Code C3273
Neurofibromatosis type 2	An autosomal dominant disorder caused by mutations in the <i>NF2</i> tumor suppressor gene. It is characterized by the development of peripheral and central nervous system tumors including acoustic schwannomas, neurofibromas, gliomas, and meningiomas			NCI Thesaurus	NCI Thesaurus Code C3274
Rett syndrome	A progressive neurologic disorder caused by mutations in the <i>MECP2</i> gene on chromosome X. It almost exclusively affects girls. It is characterized by breath-holding spells, flushing, repetitive hand motions, lack of advancement in language, and regression in developmental milestones. Other signs and symptoms include slowed head growth, scoliosis, breathing abnormalities (including sleep-disordered breathing), and sleep disturbances			Modified from NCI Thesaurus	NCI Thesaurus Code C75488
Waardenburg syndrome	A rare, autosomal dominant inherited syndrome caused by mutations in the <i>PAX3</i> , <i>MITF</i> , and <i>SNAI2</i> genes. Signs and symptoms include hearing loss, dystopia canthorum (widely spaced inner corners of the eyes), and changes in the color of the skin, hair (white forelock), and eyes			Modified from NCI Thesaurus	NCI Thesaurus Code C85222

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Visceral hypersensitivity	Heightened sensitivity or reduced thresholds for pain and discomfort associated with the gastrointestinal tract			Modified from Mertz H. Aliment Pharmacol Ther 2003;17(5):623–33 [32] AND Faure C, Di Lorenzo C, Thapar N. (2017) Pediatric Neurogastroenterology Gastrointestinal Motility and Functional Disorders in Children, 2 nd Ed. Springer International Publishing, Switzerland [33]	Also called visceral hyperalgesia
Multiple endocrine neoplasia	An autosomal dominant inherited neoplastic syndrome characterized by the development of various endocrine neoplasms and abnormalities in various anatomic sites			NCI Thesaurus	NCI Thesaurus Code C6432
Simple breath-holding spell	A clinical condition in which there is a breath-holding spell without syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Complex breath-holding spell	A clinical condition in which there is a breath-holding spell associated with syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Pallid breath-holding spell	A clinical condition in which there is a breath-holding spell associated with pale appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Cyanotic breath-holding spell	A clinical condition in which there is a breath-holding spell associated with cyanotic appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Seizure disorder	A brain disorder characterized by episodes of abnormally increased neuronal discharge resulting in transient episodes of sensory or motor neurological dysfunction, or psychic dysfunction that may or may not be associated with loss of consciousness or convulsions			NCI Thesaurus	NCI Thesaurus Code C3020 Also called epilepsy
Sudden death	An unexpected death without warning			NCI Thesaurus	NCI Thesaurus Code C107103
Sudden cardiac death	An unexpected death from a cardiac cause within a short time period from the onset of symptoms			NCI Thesaurus	NCI Thesaurus Code C50911

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Medications	A drug product that contains one or more active and/or inactive ingredients; it is intended to treat, prevent or alleviate the symptoms of disease			NCI Thesaurus	NCI Thesaurus Code C459
Fludrocortisone	A synthetic adrenocortical steroid possessing very potent mineralocorticoid properties and high glucocorticoid activity; it is used only for its mineralocorticoid effects			www.rxlist.com	
Midodrine	A direct-acting prodrug and sympathomimetic agent with antihypertensive properties			NCI Thesaurus	NCI Thesaurus Code C61846
Droxidopa	A synthetic amino acid analog that is directly metabolized to norepinephrine by dopa-decarboxylase			https://www.accessdata.fda.gov/drugsatfda_docs/label/2017/203202s0071bl.pdf	
Beta blocker	Natural or synthetic beta adrenergic antagonists selectively or non-selectively blocking or diminishing physiologic beta-adrenergic agonist actions on the sympathetic system			NCI Thesaurus	NCI Thesaurus Code C29576
Propranolol	A synthetic, nonselective beta-adrenergic receptor blocker with antianginal, antiarrhythmic, antihypertensive properties			NCI Thesaurus	NCI Thesaurus Code C62073
Atenolol	A synthetic isopropylamino-propanol derivative used as an antihypertensive, hypotensive and antiarrhythmic, atenolol acts as a peripheral, cardioselective beta blocker specific for beta-1 adrenergic receptors, without intrinsic sympathomimetic effects			NCI Thesaurus	NCI Thesaurus Code C28836
Metoprolol	A cardioselective competitive beta-1 adrenergic receptor antagonist with antihypertensive properties and devoid of intrinsic sympathomimetic activity			NCI Thesaurus	NCI Thesaurus Code C61845
Nadolol	A nonselective beta-adrenergic antagonist with antihypertensive and antiarrhythmic activities			NCI Thesaurus	NCI Thesaurus Code C29280
Nebivolol	A beta-1 adrenergic receptor antagonist with antihypertensive and vasodilatory activity			NCI Thesaurus	NCI Thesaurus Code C66221

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Ivabradine	An orally bioavailable, hyperpolarization-activated, cyclic nucleotide-gated (HCN) channel blocker, with negative chronotropic activity that selectively binds to the intracellular portion of the HCN channel pore and blocks HCN channels in the pacemaker cells within the sinoatrial (SA) node			NCI Thesaurus	NCI Thesaurus Code C65995
Pyridostigmine	The bromide salt form of pyridostigmine, a quaternary ammonium carbamate derivative and an acetylcholinesterase inhibitor			NCI Thesaurus	NCI Thesaurus Code C47697
Desmopressin	A synthetic octapeptide, and an analog of human hormone arginine vasopressin with antidiuretic and coagulant activities			NCI Thesaurus	NCI Thesaurus Code C61701
Stimulant	Any agent capable of having a stimulating effect on the central nervous system (CNS)			NCI Thesaurus	NCI Thesaurus Code C47795
Methylphenidate	A synthetic central nervous system stimulant			NCI Thesaurus	NCI Thesaurus Code C62045
Dexmethylphenidate	The D-threo-enantiomer of methylphenidate, a central nervous system stimulant, appearing to block the reuptake of norepinephrine and dopamine into the presynaptic neuron, increasing their availability in the extracellular space			NCI Thesaurus	NCI Thesaurus Code C26642
Mixed amphetamine salts	A combination of two synthetic agents with central nervous system stimulant activity			NCI Thesaurus	NCI Thesaurus Code C26643
Lisdexamfetamine	The dimesylate form and prodrug of the D-isomer of amphetamine, a non-catecholamine sympathomimetic amine with central nervous system (CNS)-stimulating activity			NCI Thesaurus	NCI Thesaurus Code C76628
Atomoxetine	The hydrochloride salt of atomoxetine, a phenoxy-3-propylamine derivative and selective non-stimulant, norepinephrine reuptake inhibitor with cognitive-enhancing activity			NCI Thesaurus	NCI Thesaurus Code C47405

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Modafinil	A synthetic central nervous system stimulant with wakefulness-promoting activity			NCI Thesaurus	NCI Thesaurus Code C26661
Armodafinil	The R-enantiomer of the racemic synthetic agent modafinil with central nervous system (CNS) stimulant and wakefulness-promoting activities			NCI Thesaurus	NCI Thesaurus Code C65241
Psychotropic medication	A classification of medication that acts on the central nervous system, affecting brain function, level of consciousness, mood, emotions, and behavior			NCI Thesaurus	NCI Thesaurus Code C82395
Sertraline	The hydrochloride salt of sertraline, a synthetic derivative of naphthalenamine with anti-serotonergic and anti-depressant properties			NCI Thesaurus	NCI Thesaurus Code C29451
Citalopram	A bicyclic phthalene derivative and selective serotonin reuptake inhibitor (SSRI), with antidepressant and anxiolytic activities			NCI Thesaurus	NCI Thesaurus Code C61680
Escitalopram	The active S-stereoisomer of the selective serotonin reuptake inhibitor (SSRI) citalopram with antidepressant, anti-obsessive–compulsive and anti-bulimic properties			NCI Thesaurus	NCI Thesaurus Code C61754
Fluoxetine	The hydrochloride salt form of fluoxetine, a diphenhydramine derivative and selective serotonin reuptake inhibitor with antidepressant, antiobsessional, and anti-bulimic effects			NCI Thesaurus	NCI Thesaurus Code C2829
Bupropion	An aminoketone with antidepressant activity			NCI Thesaurus	NCI Thesaurus Code C62012
Venlafaxine	A synthetic phenethylamine bicyclic derivative with antidepressant activity			NCI Thesaurus	NCI Thesaurus Code C1278
Paroxetine	The hydrochloride salt form of paroxetine, a phenylpiperidine derivative and a selective serotonin reuptake inhibitor (SSRI) with antidepressant and anxiolytic properties			NCI Thesaurus	NCI Thesaurus Code C1837

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Duloxetine	The hydrochloride salt of duloxetine, a fluoxetine derivative belonging to the class of selective serotonin (5-HT) and norepinephrine (NE) reuptake inhibitors (SSNRIs) and exhibiting antidepressant activity			NCI Thesaurus	NCI Thesaurus Code C65496
Aripiprazole	A quinoline derivative and atypical anti-psychotic agent			NCI Thesaurus	NCI Thesaurus Code C47403
Olanzapine	A synthetic derivative of thienobenzodiazepine with antipsychotic, anti-nausea, and antiemetic activities			NCI Thesaurus	NCI Thesaurus Code C47639
Quetiapine	A dibenzothiazepine derivative with antipsychotic property			NCI Thesaurus	NCI Thesaurus Code C61917
Risperidone	A benzisoxazole derivative with antipsychotic property			NCI Thesaurus	NCI Thesaurus Code C29416
Ziprasidone	A benzothiazolylpiperazine derivative and an atypical antipsychotic agent with an antischizophrenic property			NCI Thesaurus	NCI Thesaurus Code C47788
Mirtazapine	A synthetic tetracyclic derivative of the piperazino-azepines with antidepressant activity			NCI Thesaurus	NCI Thesaurus Code C29265
Buspirone	An anxiolytic agent chemically and pharmacologically unrelated to benzodiazepines, barbiturates, or other sedative/hypnotic drugs			NCI Thesaurus	NCI Thesaurus Code C62013
Trazodone	A synthetic triazolopyridine derivative, antidepressant and sedative, trazodone acts as a serotonin uptake inhibitor, chemically unrelated to tricyclic, tetracyclic, or other antidepressants			NCI Thesaurus	NCI Thesaurus Code C29510
Antihistamine	Any agent that binds to the histamine H1 receptor, thereby antagonizing histamine mediated allergic reactions, such as bronchoconstriction, vasodilation, and upregulated capillary permeability			NCI Thesaurus	NCI Thesaurus Code C29578
Cetirizine	Cetirizine is a metabolite of hydroxyzine and a selective peripheral histamine H1-receptor antagonist			NCI Thesaurus	NCI Thesaurus Code C1042

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Loratadine	A piperidine histamine H1-receptor antagonist with anti-allergic properties and without sedative effects			NCI Thesaurus	NCI Thesaurus Code C29162
Fexofenadine	A second generation, long-lasting selective histamine H1 receptor antagonist with antiinflammatory property			NCI Thesaurus	NCI Thesaurus Code C61764
Diphenhydramine	A first generation antihistamine and ethanolamine with sedative and anti-allergic properties			NCI Thesaurus	NCI Thesaurus Code C61728
Cyproheptadine	The hydrochloride salt of a synthetic methyl-piperidine derivative with antihistaminic and anti-serotonergic properties			NCI Thesaurus	NCI Thesaurus Code C28949
Acid suppression					
Ranitidine	A member of the class of histamine H2-receptor antagonists with antacid activity			NCI Thesaurus	NCI Thesaurus Code C29412
Cimetidine	A histamine H(2)-receptor antagonist			NCI Thesaurus	NCI Thesaurus Code C374
Famotidine	A propanimidamide and histamine H2-receptor antagonist with antacid activity			NCI Thesaurus	NCI Thesaurus Code C29045
Omeprazole	A benzimidazole with selective and irreversible proton pump inhibition activity			NCI Thesaurus	NCI Thesaurus Code C716
Lansoprazole	A substituted benzimidazole prodrug with selective and irreversible proton pump inhibitor activity			NCI Thesaurus	NCI Thesaurus Code C29150
Esomeprazole	The S-isomer of omeprazole, with gastric proton pump inhibitor activity			NCI Thesaurus	NCI Thesaurus Code C65538
Pantoprazole	A substituted benzimidazole and proton pump inhibitor with antacid activity			NCI Thesaurus	NCI Thesaurus Code C29346
Rabeprazole	An alpha-pyridylmethyl-sulfinyl benzimidazole and a selective and irreversible proton pump inhibitor with antisecretory property			NCI Thesaurus	NCI Thesaurus Code C29410
Clonidine	A centrally-acting alpha-adrenergic agonist that also acts on imidazoline receptors that inhibits central sympathetic activity with antihypertensive activity			Modified from NCI Thesaurus	NCI Thesaurus Code C380

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Carbidopa	A hydrazine derivative of dopa and a peripheral dopa decarboxylase inhibitor that is used as an adjunct to levodopa administration to prevent peripheral biosynthesis of levodopa to dopamine			NCI Thesaurus	NCI Thesaurus Code C47431
Dexmedetomidine	A short-acting imidazole derivate and active D-isomer of medetomidine with antihypertensive, analgesic, anxiolytic, and sedative properties			Modified from NCI Thesaurus	NCI Thesaurus Code C47479
Erythromycin	A broad-spectrum, macrolide antibiotic with antibacterial activity			NCI Thesaurus	NCI Thesaurus Code C476
Pregabalin	A 3-isobutyl derivative of gamma-amino butyric acid (GABA) with anti-convulsant, anti-epileptic, anxiolytic, and analgesic activities			NCI Thesaurus	NCI Thesaurus Code C64625
Naltrexone	A noroxymorphone derivative with competitive opioid antagonistic property			NCI Thesaurus	NCI Thesaurus Code C62055
Nonsteroidal anti-inflammatory drug	A pharmacological agent that is not a steroid and has potential anti-inflammatory, analgesic, antipyretic, and antiplatelet activities			NCI Thesaurus	NCI Thesaurus Code C257
Ibuprofen (note similar to naproxen)	A propionic acid derivative and nonsteroidal anti-inflammatory drug (NSAID) with anti-inflammatory, analgesic, and antipyretic effects			NCI Thesaurus	NCI Thesaurus Code C561
Naproxen	A propionic acid derivative and a nonsteroidal anti-inflammatory drug (NSAID) with anti-inflammatory, antipyretic, and analgesic activities			NCI Thesaurus	NCI Thesaurus Code C680
Ketorolac	A synthetic pyrrolizine carboxylic acid derivative with anti-inflammatory, analgesic, and antipyretic activities			NCI Thesaurus	NCI Thesaurus Code C1219
Celecoxib	A nonsteroidal anti-inflammatory drug (NSAID) with a diaryl-substituted pyrazole structure			NCI Thesaurus	NCI Thesaurus Code C1728
Ondansetron	A carbazole derivative with antiemetic activity			NCI Thesaurus	NCI Thesaurus Code C1119
Granisetron	An indazole derivative with antiemetic properties			NCI Thesaurus	NCI Thesaurus Code C62031

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Aprepitant	A small molecule, high-affinity substance P antagonist with antiemetic activity			NCI Thesaurus	NCI Thesaurus Code C49173
Scopolamine	A tropane alkaloid derived from plants of the nightshade family (Solanaceae), specifically <i>Hyoscyamus niger</i> and <i>Atropa belladonna</i> , with anticholinergic, antiemetic, and antivertigo properties			NCI Thesaurus	NCI Thesaurus Code C47712
Meclizine	A synthetic piperazine with anti-emetic, sedative and histamine H1 antagonistic properties			NCI Thesaurus	NCI Thesaurus Code C61610
Melatonin	A hormone involved in sleep regulatory activity, and a tryptophan-derived neurotransmitter, which inhibits the synthesis and secretion of other neurotransmitters such as dopamine and GABA			NCI Thesaurus	NCI Thesaurus Code C2267
Zolpidem	The tartrate salt of an imidazopyridine with non-benzodiazepine benzodiazepine-receptor agonist and sedative-hypnotic activities			NCI Thesaurus	NCI Thesaurus Code C29553
Eszopiclone	A nonbenzodiazepine, cyclopyrrolone, and active stereoisomer of zopiclone with hypnotic and sedative activity and without significant anxiolytic activity			NCI Thesaurus	NCI Thesaurus Code C65545
Laxatives	Synthetic compounds or mixtures administered orally or rectally with laxative activity. A laxative preparation is used to promote bowel movement to treat constipation or to clean the colon for rectal and bowel examinations			NCI Thesaurus	NCI Thesaurus Code C29697
Lubiprostone	A bicyclic fatty acid derived from prostaglandin E1 and a chloride channel activator with laxative activity			NCI Thesaurus	NCI Thesaurus Code C66040
Psyllium	The husk of the dried ripe seeds of <i>Plantago psyllium</i> , <i>P. indica</i> , <i>P. ovata</i> , and <i>P. arenaria</i> plants with laxative and cholesterol-lowering activities			NCI Thesaurus	NCI Thesaurus Code C29250
Docusate sodium	The sodium salt of docusate, a dioctyl salt and an emollient laxative with stool-softening activity			NCI Thesaurus	NCI Thesaurus Code C29000

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Senna	An extract made from the dried leaflets on the pods of <i>Cassia angustifolia</i> or <i>Cassia acutifolia</i> with cathartic activity			NCI Thesaurus	NCI Thesaurus Code C61324
Mineral oil	A mixture of liquid paraffinic, naphthenic, and polycyclic aromatic hydrocarbons obtained from petroleum in which the amount and types of aromatic hydrocarbons are dependent on the treatment process			NCI Thesaurus	NCI Thesaurus Code C29264
Linaclotide	A synthetic, fourteen amino acid peptide and agonist of intestinal guanylate cyclase type C (GC-C), which is structurally related to the guanylin peptide family, with secretagogue, analgesic, and laxative activities			NCI Thesaurus	NCI Thesaurus Code C81001
Prucalopride	A highly selective serotonin 5-HT ₄ receptor agonist that has been shown to stimulate gut motility			Quigley EMM Therap Adv Gastroenterol 2012;5(1):23–30 [52]	
Lactulose	A disaccharide (sugar) synthesized from fructose and galactose; lactulose is used to treat constipation and some liver diseases			NCI Thesaurus	NCI Thesaurus Code C29148
Polyethylene glycol 3350	A polymer made by joining molecules of ethylene oxide and water together in a repeating pattern used as an osmotic laxative			Modified from NCI Thesaurus	NCI Thesaurus Code C762
Bisacodyl	A synthetic pyridinyl-methylene-diacetate ester derivative stimulant laxative, Bisacodyl acts with a parasympathetic effect directly on mucosal sensory nerves, increasing peristaltic contractions			NCI Thesaurus	NCI Thesaurus Code C28870
Tegaserod	A 5-HT ₄ agonist that functions as a motility agent, by stimulating the peristaltic reflex and intestinal secretion, as an analgesic by inhibition of visceral sensitivity			www.drugs.com	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Cisapride	A substituted piperidiny benzamide prokinetic agent. Cisapride facilitates release of acetylcholine from the myenteric plexus, resulting in increased gastrointestinal motility. In addition, cisapride has been found to act as a serotonin agonist, stimulating type 4 receptors, and a serotonin 5-HT ₃ receptor antagonist			NCI Thesaurus	NCI Thesaurus Code C1210
Metoclopramide	A substituted benzamide and a derivative of para-aminobenzoic acid (PABA) that is structurally related to procainamide, with gastropromkinetic and antiemetic effects			NCI Thesaurus	NCI Thesaurus Code C62046
Divalproex	A stable coordination compound composed of sodium valproate and valproic acid with anticonvulsant and antiepileptic activities			NCI Thesaurus	NCI Thesaurus Code C28996
Amitriptyline	A derivative of dibenzocycloheptadiene and a tricyclic antidepressant. Amitriptyline inhibits the re-uptake of norepinephrine and serotonin by the presynaptic neuronal membrane in the central nervous system (CNS), thereby increasing the synaptic concentration of norepinephrine and serotonin. Due to constant stimulation to these receptors, amitriptyline may produce a downregulation of adrenergic and serotonin receptors, which may contribute to the antidepressant activity			NCI Thesaurus	NCI Thesaurus Code C62005

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Nortriptyline	A tricyclic antidepressant agent used for short-term treatment of various forms of depression. Nortriptyline blocks the norepinephrine presynaptic receptors, thereby blocking the reuptake of this neurotransmitter and raising the concentration in the synaptic cleft in the CNS. Nortriptyline also binds to alpha-adrenergic, histaminergic and cholinergic receptors. Long-term treatment with nortriptyline produces a downregulation of adrenergic receptors due to the increased stimulation of these receptors			NCI Thesaurus	NCI Thesaurus Code C62060
Gabapentin	A synthetic analog of the neurotransmitter gamma-aminobutyric acid with anticonvulsant activity			NCI Thesaurus	NCI Thesaurus Code C1108
Triptans	A family of tryptamine-based drugs used as abortive medication in the treatment of migraine and cluster headaches			www.wikipedia.org	
Sumatriptan	A sulfonamide triptan that selectively binds to and activates serotonin 5-HT1D receptors in the central nervous system, thereby constricting cerebral blood vessels			NCI Thesaurus	NCI Thesaurus Code C1240
Rizatriptan	A triptan that selectively binds to and activates serotonin 5-HT1B receptors expressed in intracranial arteries, and to 5-HT1D receptors located on peripheral trigeminal sensory nerve terminals in the meninges and central terminals in brain stem sensory nuclei			NCI Thesaurus	NCI Thesaurus Code C61930
Eletriptan	A triptan that binds to and acts at serotonin 5-HT1B receptors located on intracranial blood vessels which leads to vasoconstriction. This drug may also exert its effects by binding to and activating 5-HT 1D receptors on sensory nerve endings in the trigeminal system, which results in the inhibition of pro-inflammatory neuropeptide release			NCI Thesaurus	NCI Thesaurus Code C47508

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Zolmitriptan	A triptan that selectively binds to and activates serotonin (5-HT) 1B receptors expressed in intracranial arteries and 5-HT 1D receptors located on peripheral trigeminal sensory nerve terminals in the meninges and central terminals in brainstem sensory nuclei, resulting in constriction of cranial vessels, reduction of vessel pulsation and inhibition of nociceptive transmission			NCI Thesaurus	NCI Thesaurus Code C47789
Dihydroergotamine mesylate	An ergot derivative with agonistic activity for alpha-adrenergic, serotonergic, and dopaminergic receptors			NCI Thesaurus	NCI Thesaurus Code C47491
Topiramate	A sulfamate-substituted monosaccharide with anticonvulsant property			NCI Thesaurus	NCI Thesaurus Code C47764
Onabotulinumtoxin A	An injectable formulation of a neurotoxin derived through the fermentation of the Hall strain of Clostridium botulinum type A with neuromuscular transmission inhibitory and analgesic activities			NCI Thesaurus	NCI Thesaurus Code C82623
Cromolyn sodium	A synthetic mast cell stabilizer with anti-inflammatory activity			NCI Thesaurus	NCI Thesaurus Code C61691
Montelukast	It is a type of leukotriene receptor antagonist and a type of antiasthmatic agent			NCI Thesaurus	NCI Thesaurus Code C66189
Quercetin	A polyphenolic flavonoid with potential chemopreventive activity			NCI Thesaurus	NCI Thesaurus Code C792
Aspirin	An orally administered non-steroidal antiinflammatory agent; acetylsalicylic acid			NCI Thesaurus	NCI Thesaurus Code C287
Epinephrine	A hormone and neurotransmitter			NCI Thesaurus	NCI Thesaurus Code C2292
Medical device	Any physical object that is useful for prevention, diagnosis, monitoring, or treatment of disease or other conditions			NCI Thesaurus	NCI Thesaurus Code C16830
Compression stockings	Specialized hosiery worn on the lower extremities available in varying degrees of pressure that creates increased peripheral venous return				
Abdominal binder	A wide band fastened snugly about the abdomen for support			https://medical-dictionary.thefreedictionary.com/abdominal+binder	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Gastric pacemaker	A device that is implanted under the skin and has electrodes that attach to the stomach that is programmed to stimulate the stomach to empty			NCI Thesaurus	NCI Thesaurus Code C157863
Cefaly	An external trigeminal nerve stimulation device used for the treatment of migraine			www.cefaly.us	
Transcutaneous electrical nerve stimulation	A non-invasive form of electroanalgesia that uses conductive gel pads to transmit electrical energy from an external stimulator to peripheral nerves			NCI Thesaurus	NCI Thesaurus Code C21032
Vagus nerve stimulator	A medical device that delivers electrical impulses to the vagus nerve, which can be implanted or external				
IB-Stim	A medical device that delivers electrical impulses to cranial nerve branches as a peripheral electrical nerve field stimulator to reduce abdominal pain				
Physical examination findings	A systemic evaluation of the body and its functions using visual inspection, palpation, percussion, and auscultation			NCI Thesaurus	NCI Thesaurus Code C20989
Vital signs	Vital signs (such as height, weight, and blood pressure) directly measure an individual's current state of attributes			NCI Thesaurus	NCI Thesaurus Code C154628
Heart rate	A measurement that describes the frequency of rate of contractions of the systemic (often left) ventricle measured within a unit time	Numeric, in bpm	0–300	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Supine heart rate	Heart rate measured in a posterior recumbent body position whereby the person lies on its back and faces upward	Numeric, in bpm	0–300	NCI Thesaurus	NCI Thesaurus Code C62167
Sitting heart rate	Heart rate measured in the posture of one who occupies a seat	Numeric, in bpm	0–300	NCI Thesaurus	NCI Thesaurus Code C62122
Standing heart rate	Heart rate measured during the act of assuming or maintaining an erect upright position	Numeric, in bpm	0–300	NCI Thesaurus	NCI Thesaurus Code C62126

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Blood pressure	A measurement that describes the pressure of the circulating blood against the walls of the blood vessels. Measurement is taken from the right arm	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Supine blood pressure	Blood pressure measured in a posterior recumbent body position whereby the person lies on its back and faces upward	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	NCI Thesaurus	NCI Thesaurus Code C62167
Seated blood pressure	Blood pressure measured in the posture of one who occupies a seat	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	NCI Thesaurus	NCI Thesaurus Code C62122
Standing blood pressure	Blood pressure measured during the act of assuming or maintaining an erect upright position	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	NCI Thesaurus	NCI Thesaurus Code C62126

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Blood pressure, other extremity	A measurement that describes the pressure of the circulating blood against the walls of the blood vessels. Measurement is taken from an extremity other than the right arm	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		Note: denote which extremity (LUE, RLE, LLE)
Pulse pressure	The force of a heart contraction measured by the difference between the diastolic and systolic blood pressure measurements	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	NCI Thesaurus	NCI Thesaurus Code C100945
Height	A measurement that describes the vertical measurement or distance from the base, or bottom, of the patient, to the top of the patient; this can be taken as the dimension of extension of a patient who cannot stand	Numeric, in cm	10–230	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Weight	A measurement that describes the vertical force exerted by a mass of the patient as a result of gravity	Numeric, in kg	0.2–500	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Oxygen saturation	A measurement that describes the determination of oxygen-hemoglobin saturation of blood	Numeric, in %	20–100	Modified from 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Respiratory rate	A measurement that describes the rate of breathing (inhalation and exhalation) measured within a unit time	Numeric, in breaths per minute	0–100	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Tidal volume	The volume of air moved into and out of the lungs per breath during breathing at rest			Modified from NCI Thesaurus	NCI Thesaurus Code C111324
Nystagmus	A condition where the eyes move rapidly and uncontrollably			www.aaopt.org/eye-health/diseases/what-is-nystagmus	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Joint hypermobility	The capability that a joint (or a group of joints) has to move, passively and/or actively, beyond normal limits along physiological axes			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	
Finger	Any of the digits of the hand			NCI Thesaurus	NCI Thesaurus Code C32608
Wrist	A joint between the distal end of the radius and the proximal row of carpal bones			NCI Thesaurus	NCI Thesaurus Code C33894
Elbow	A type of hinge joint located between the forearm and upper arm			NCI Thesaurus	NCI Thesaurus Code C32497
Shoulder	A ball-and-socket joint at the upper end of the humerus, located at the junction of humerus and scapula			NCI Thesaurus	NCI Thesaurus Code C33548
Knee	A joint connecting the lower part of the femur with the upper part of the tibia			NCI Thesaurus	NCI Thesaurus Code C32898
Waist	The part of the body in humans between the ribs and the hips, usually the narrowest part of the torso			www.dictionary.com	
Ankle	A gliding joint between the distal ends of the tibia and fibula and the proximal end of the talus			NCI Thesaurus	NCI Thesaurus Code C32078
Beighton score	A clinical assessment method for classifying generalized joint hypermobility consisting of nine separate tests. A score of 5/9, or greater, in adults, and 6/9, or greater, in children indicates a positive test	Numeric, in integers	0–9	Juul-Kristensen B Am J Med Genet C Semin Med Genet 175(1):116–147 [53]	
Venous pooling	The accumulation of blood in veins of the extremities or splanchnic circulation due to gravitational pull when a person assumes an upright position				Also called dependent acrocyanosis
Pallor	A clinical finding in which there appears to be a loss of color and/or perfusion, or a white appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Diaphoresis	A clinical finding in which there is excessive or unpredictable sweating			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Mydriasis	Dilation of the pupil			www.medicinenet.com	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Miosis	Constriction of the pupil			www.medicinenet.com	
Alacrima	Lack of tears due to absence or hyposecretion of tears, or to agenesis or improper development of lacrimal gland ducts and associated structures			Adams J Clin Genet 2018;94(1):54–60 [26]	
Cyanosis	A bluish or purplish discoloration of the skin and mucous membranes resulting from an increased amount of deoxygenated hemoglobin in the blood			Modified from NCI Thesaurus	NCI Thesaurus Code C26737
Central cyanosis	Cyanosis that is noted to occur over the entire body surface area and visible mucosa			www.ncbi.nlm.nih.gov	
Perioral cyanosis	Cyanosis that is noted to occur surrounding the mouth that may or may not include the lips				
Peripheral cyanosis	Cyanosis that is noted to occur in the upper and lower extremities			www.ncbi.nlm.nih.gov	Also called acrocyanosis
Dermatographism (Darier sign)	Urticaria due to physical allergy in which a pale, raised welt or wheal with a red flare on each side is elicited by stroking or scratching the skin			https://medical-dictionary.thefreedictionary.com/dermatographism	
Allodynia	Pain due to a stimulus that does not normally provoke pain			www.ncbi.nlm.nih.gov	
Urticaria	A transient, itchy skin eruption characterized by hives			Modified from NCI Thesaurus	NCI Thesaurus Code C3432
Areflexia	A finding indicating the complete absence of neurological reflexes			NCI Thesaurus	NCI Thesaurus Code C115420
Hyperreflexia	Autonomic nervous system overreaction to stimuli, most commonly after spinal cord injury at a T-5 level and above			NCI Thesaurus	NCI Thesaurus Code C43248
Anhidrosis	Lack of sweating or the ability to sweat when provoked by the appropriate stimulus			NCI Thesaurus	NCI Thesaurus Code C34385
Scoliosis	A congenital or acquired spinal deformity characterized by lateral curvature of the spine			NCI Thesaurus	NCI Thesaurus Code C78603
Hypotonia	A condition of decreased tone of the skeletal muscles and diminished resistance to passive stretching			NCI Thesaurus	NCI Thesaurus Code C87070
Testing	A procedure for critical evaluation			NCI Thesaurus	NCI Thesaurus Code C15336

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Laboratory	Any procedure that involves testing or manipulating a sample of blood, urine, or other body substance in a laboratory setting			NCI Thesaurus	NCI Thesaurus Code C25294
Morning (AM) cortisol	A hormone made by the adrenal cortex. Measurement is made between 6 and 8 AM	Numeric, in mcg/dl	0–100	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C2290
Thyroid-stimulating hormone	A peptide hormone secreted by the anterior pituitary that promotes the growth of the thyroid gland and stimulates the synthesis of thyroid hormones and the release of thyroxine by the thyroid gland	Numeric, in $\mu\text{U}/\text{ml}$	0–50	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C2280
T4/thyroxine	A hormone synthesized and secreted by the thyroid gland containing four iodine atoms and is converted to triiodothyronine (T3) in the body, influencing metabolism and organ function	Numeric, in $\mu\text{g}/\text{dl}$	0–50	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C2302
Ferritin	An iron protein complex, containing up to 23% iron, formed by the union of ferric iron with apoferritin; it is found in the intestinal mucosa, spleen, bone marrow, reticulocytes, and liver, and regulates iron storage and transport from the intestinal lumen to plasma	Numeric, in ng/ml	0–500	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C16577
Hemoglobin	The red respiratory protein of erythrocytes, consisting of approximately 3.8% heme and 96.2% globin (64.5 KD), which as oxyhemoglobin (HbO_2) transports oxygen from the lungs to the tissues where the oxygen is readily released and HbO_2 becomes Hb	Numeric, in g/dl	0–30	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C16676
Hematocrit	A measure of the volume of red blood cells expressed as a percentage of the total blood volume	Numeric, in %	0–90	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C64796
Blood urea nitrogen	A quantitative measurement of the amount of urea nitrogen present in a serum sample	Numeric, in mg/dl	0–40	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C61019

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Creatinine	The breakdown product of creatine, a constituent of muscle tissue, that is excreted by the kidney and whose serum level is used to evaluate kidney function	Numeric, in mg/dl	0–5	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C399
Tryptase	A family of serine endoproteases usually expressed in mast cell secretory granules	Numeric, in ng/ml	0–50	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C120030
Baseline	A starting point to which things may be compared			NCI Thesaurus	NCI Thesaurus Code C25213
Post-flush	A point in time after systemic flushing has occurred				
Urinary <i>n</i> -methylhistamine	The major metabolite of histamine			Afrin LB and Molderings GJ. World J Hematol 2014;3(1):1–17 [54] www.wikipedia.org	
Urinary prostaglandin-D2	A prostaglandin that binds to the receptor PTGDR as well as CRTH2, and is a major prostaglandin produced by mast cells				
Urinary 11 β -prostaglandin F2 α	The most abundant metabolic product of prostaglandins released by activated mast cells			www.mayocliniclabs.com	
IgA anti-tissue transglutaminase	An autoantibody against the transglutaminase protein found in celiac disease	Numeric, in U/ml	0–50	www.wikipedia.org , values based on ABIM Laboratory Test Reference Ranges Jan 2019	
IgA anti-endomysial reactivity	An autoantibody against tissue transglutaminase found in endomysium, seen in celiac disease			www.wikipedia.org	
Calprotectin	A heterodimeric or heterotrimeric protein complex comprised of protein S100A8 and protein S100A9 expressed in the cytosol of neutrophils and macrophages and plays a role in multiple host defense related processes including immunomodulation, neutrophil chemotaxis and both antibacterial and antifungal responses			NCI Thesaurus	NCI Thesaurus Code C105971
Antinuclear antibody	An autoimmune antibody that is directed against structures within the nucleus of the cell	Numeric, as ratio	1:1 to 1:16,000	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C70547

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
C-reactive protein	An annular pentameric protein of hepatic origin found in blood plasma cleaved during biological activation and is associated with host defense mechanisms and inflammatory responses	Numeric, in ng/ml	0–20	www.wikipedia.org , NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C60651
Erythrocyte sedimentation rate	A quantitative measurement of the distance that red blood cells travel in one hour in a sample of unclotted blood	Numeric, in mm/h	0–120	NCI Thesaurus, values based on ABIM Laboratory Test Reference Ranges Jan 2019	NCI Thesaurus Code C74611
Tilt-table testing	A medical procedure, also called head upright tilt testing, which assesses orthostatic tolerance utilizing passive upright posture on a mechanical tilt table				Also called head up tilt test
Standing test	A medical procedure which assesses orthostatic tolerance utilizing active upright posture while standing motionless				
Heart rate	A measurement that describes the frequency of rate of contractions of the systemic (often left) ventricle measured within a unit time	Numeric, in bpm	0–300	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Supine heart rate	Heart rate measured in a posterior recumbent body position whereby the person lies on its back and faces upward	Numeric, in bpm	0–300	NCI Thesaurus	NCI Thesaurus Code C62167
Upright heart rate	Heart rate measured while in the upright posture during tilt table testing or standing test	Numeric, in bpm	0–300		
Blood pressure	A measurement that describes the pressure of the circulating blood against the walls of the blood vessels. Measurement is taken from the right arm	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Supine blood pressure	Blood pressure measured while lying down during tilt table testing or standing test	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	NCI Thesaurus	NCI Thesaurus Code C62167
Seated blood pressure	Blood pressure measured in the posture of one who occupies a seat	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200	NCI Thesaurus	NCI Thesaurus Code C62122
Upright blood pressure	Blood pressure that is measured while in the upright posture during tilt table testing or standing test	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		
End-tidal CO ₂	A noninvasive measurement of exhaled carbon dioxide at the end of exhalation	Numeric, in mmHg	20–70	Modified from www.aastweb.org Technical Guideline End-Tidal CO ₂ May 2018	
Near infrared regional spectroscopy/NIRS	A spectroscopic method to measure regional blood flow/oxygenation to the forehead, often used in comparison to skeletal and splanchnic light-based technology, used to non-invasively monitor tissue oxygen status			Pelicer A and del Carmen Bravo M. Semin Fetal Neonatal Med 2011;16(1):42–9 [55]	
Oxygen saturation	A measurement that describes the determination of oxygen-hemoglobin saturation of blood	Numeric, in %	20–100	2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Duration of supine position	Amount of time spent while lying down in the face-up position prior to tilt table testing	Numeric, in min	0–120		
Duration of upright tilt	Amount of time spent while on the tilt table in the upright position during testing	Numeric, in min	0–120		

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Angle of incline	The angle at which a tilt table is elevated during testing	Numeric, in degrees	0–85	Illigens BMW and Gibbons CH Clin Auton Res 2009;19(2):79–87 [56]	
Thermoregulatory sweat testing	A test that evaluates the integrity of central and peripheral sympathetic sudomotor pathways from the central nervous system to cutaneous sweat glands by raising core body temperature and visualizing degree and extend of sweat production with an indicator dye				
Quantitative sudomotor axon reflex testing (QSART)	A test that evaluates post-ganglionic sympathetic cholinergic sudomotor function by measuring the axon-reflex mediated sweat response over time and has achieved widespread clinical use. Sweat glands are stimulated via iontophoresis of a cholinergic agent and the sweat production is measured as an increase of humidity through a hygrometer			Illigens BMW and Gibbons CH Clin Auton Res 2009;19(2):79–87 [56]	
Sweat output	The amount of perspiration generated during QSART testing	Numeric, in $\mu\text{l}/\text{cm}^2$	0.5–10	Illigens BMW and Gibbons CH Clin Auton Res 2009;19(2):79–87 [56]	
Valsalva	A test that evaluates (1) sympathetic adrenergic functions using the blood pressure responses; and (2) cardiovagal (parasympathetic) functions using the heart rate responses. Valsalva maneuver consists of forced expiration against the resistance of a semi closed glottis with the expiratory pressure during strain at 40 mmHg for 15–20 s			Modified from Novak P J Vis Exp 2011;53:2502 [57]	
Valsalva phase 1 heart rate	Measurement of the minimum heart rate in the first phase of Valsalva testing	Numeric, in bpm	0–300		
Valsalva phase 1 blood pressure	Measurement of the maximum blood pressure in the first phase of Valsalva testing	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Valsalva phase 2 heart rate	Measurement of the maximum heart rate in the second phase of Valsalva testing	Numeric, in bpm	0–300		
Valsalva phase 2 blood pressure	Measurement of the minimum blood pressure in the second phase of Valsalva testing	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		
Valsalva phase 3 heart rate	Measurement of the maximum heart rate in the third phase of Valsalva testing	Numeric, in bpm	0–300		
Valsalva phase 3 blood pressure	Measurement of the minimum blood pressure in the third phase of Valsalva testing	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		
Valsalva phase 4 heart rate	Measurement of the minimum heart rate in the fourth phase of Valsalva testing	Numeric, in bpm	0–300		
Valsalva phase 4 blood pressure	Measurement of the maximum blood pressure in the fourth phase of Valsalva testing	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		
Valsalva ratio	The ratio of maximal tachycardia to the maximal bradycardia induced by the Valsalva maneuver	Numeric, no units	1.00–2.00	Levin AB Am J Cardiol 1966;18(1):90–9 [58] Novak P J Vis Exp 2011;53:2502 [57]	
Maximal decrease in mean blood pressure in phase 2	The difference between the highest and lowest average blood pressure measurements during phase 2	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Maximal pulse pressure decrease in phase 2	The difference in pulse pressure between the maximal and the lowest blood pressure measurements in phase 2	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		
Overshoot in phase 4	The difference between the maximal and the baseline blood pressure measurement in phase 4	Numeric, in mmHg; displayed as systolic pressure over diastolic pressure	0–200		
Pressure recovery time	The amount of time for blood pressure to return to baseline after phase 4	Numeric, in seconds	0–10		
Pupillometry	A technique used to measure the diameter of the pupil in response to stimuli			NCI Thesaurus	NCI Thesaurus Code C120540
Pupil diameter	A measurement of the widest part of the opening of the iris through which light passes	Numeric, in mm	2–8		
Initial pupil diameter	A measurement of the pupil diameter at baseline prior to light stimulus	Numeric, in mm	2–8	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
End pupil diameter	A measurement of the pupil diameter after light stimulus	Numeric, in mm	2–8	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
Percent pupillary constriction (delta)	The percent decrease in pupil diameter after light stimulus	Numeric, in %	-50–0	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
Pupillary latency	The time delay from light stimulus to onset of pupillary constriction	Numeric, in seconds	0–5	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
Average constriction velocity	The mean speed of pupillary constriction after light stimulus	Numeric, in mm/s	-5–0	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
Maximum constriction velocity	The peak speed of pupillary constriction after light stimulus	Numeric, in mm/s	-8–0	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
Average re-dilation velocity	The mean speed of pupillary relaxation after removal of light stimulus	Numeric, in mm/s	0–5	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	
T75	Time from light stimulus until pupil diameter returns to 75% of initial pupil diameter after re-dilation	Numeric, in seconds	0.5–5.0	Winston M, et al. Clin Auton Res 2020;30(1):43–51 [59]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Gastric emptying	A test that evaluates the process of emptying food from the stomach, including wireless motility capsule, scintigraphy, and breath test			Modified from www.medicinenet.com	
Sitz marker	A test that evaluates the transit of food and/or stool through the intestinal tract using radiopaque markers			Kim ER and Rhee PL J Neurogastroenterol Motil 2012;18(1):94–99 [60]	
SmartPill (wireless motility capsule)	A proprietary ingestible capsule for gastrointestinal tract functional assessment that measures pressure, pH, temperature, and transit time as it travels through the entire gastrointestinal tract			NCI Thesaurus	NCI Thesaurus Code C132255
Endoscopy	A diagnostic or therapeutic procedure in which an endoscope is inserted in a tubular organ to examine the structural architecture and/or remove abnormal tissues			NCI Thesaurus	NCI Thesaurus Code C16546
Esophagogastroduodenoscopy	An endoscopic procedure that visualizes the upper part of the gastrointestinal tract up to the duodenum			NCI Thesaurus	NCI Thesaurus Code C78144
Colonoscopy	Endoscopic examination of the luminal surface of the colon			NCI Thesaurus	NCI Thesaurus Code C16540
Abdominal ultrasonography	A method of ultrasound imaging in which the ultrasound probe is pressed against the skin of the abdomen to create an image of the abdominal organs			NCI Thesaurus	NCI Thesaurus Code C71375
Celiac artery Doppler ultrasonography	Diagnostic imaging that uses sound waves (ultrasound) applying the Doppler effect, with frequency-shifted ultrasound reflections produced by moving targets (usually red blood cells) in the bloodstream along the ultrasound axis in direct proportion to the velocity of movement of the targets, to determine both direction and velocity of blood flow in the celiac artery			Modified from NCI Thesaurus	NCI Thesaurus Code C62781
CT angiography of celiac axis	Angiography that uses multi-detector computed tomography to assess the anatomy of the celiac artery			NCI Thesaurus	NCI Thesaurus Code C157338

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Percutaneous celiac ganglion block	The injection of a local anesthetic with the aim of inhibiting neurotransmission without the ablation of neurons			Chak A Gastrointest Endosc 2009;69(2 Suppl):S172-3 [61]	
Electrogastrogram	A test in which the electrical current generated by the muscle of the stomach is sensed and recorded			www.medicinenet.com	
Electrocardiogram	The record produced by the variations in electrical potential caused by electrical activity of the heart muscle and detected at the body surface, as a method for studying the action of the heart muscle			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Echocardiogram	The recording of the position and motion of the heart walls or internal structures of the heart by the echo obtained from beams of the ultrasonic waves directed through the chest wall			NCI Thesaurus	NCI Thesaurus Code C37945
Holter monitor	An ambulatory electrocardiography technique that records a continuous electrocardiographic rhythm pattern for 24 h or more to detect heart arrhythmias and assess heart rate variability			Modified from NCI Thesaurus	NCI Thesaurus Code C38064
Cardiac event monitor	A medical device used to record the electrical activity of the heart. Often, the wearer will input a signal that is used as an event marker when they feel symptoms such as tachycardia, as the device continuously records cardiac activity			NCI Thesaurus	NCI Thesaurus Code C80401
Implantable loop recorder	A subcutaneous device used for diagnosing heart rhythm disorders			Bisignani A, et al. J Arrhythm 2019;35(1):25–32 [62]	
Diagnoses	The investigation, analysis and recognition of the presence and nature of disease, condition, or injury from expressed signs and symptoms; also, the scientific determination of any kind; the concise results of such an investigation			NCI Thesaurus	NCI Thesaurus Code C15220

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Dizziness	A nonspecific term indicating a sensation of lightheadedness, unsteadiness, turning, vertigo, or rocking			Modified from NCI Thesaurus AND Reilly BM (1990) Dizziness. In: Walker HK, et al. (eds.) Clinical Methods: The History, Physical, and Laboratory Examinations, 3 rd Ed. Butterworth, Boston, Ch. 212 [20]	NCI Thesaurus Code C37943
Lightheadedness	A feeling of faintness or wooziness, or a sensation of motion within the head that may be accompanied by visual or auditory changes, diaphoresis, nausea, or tachycardia			Modified from NCI Thesaurus	NCI Thesaurus Code C118255
Vertigo	A feeling of movement, a sensation as if the external world was revolving around the patient			Modified from NCI Thesaurus	NCI Thesaurus Code C38057
Orthostatic intolerance	A condition in which there is the presence of ≥ 1 symptoms (e.g., dizziness, lightheadedness, nausea, dyspnea, vision change) occurring specifically when assuming or maintaining upright position and resolving in the seated or supine position			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Chronic orthostatic intolerance	A condition in which there is orthostatic intolerance lasting for at least 3 months with associated functional impairment			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Syncope	A transient loss of consciousness and postural tone due to cerebral hypoperfusion, characterized by a rapid onset, short duration, and spontaneous complete recovery			Brignole M, et al. Eur Heart J 2018;39:1883–1948 [21] 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Reflex syncope	A transient loss of consciousness and postural tone due to a reflex response that encompasses vasodilation and/or bradycardia leading to systemic hypotension and cerebral hypoperfusion			https://www.uptodate.com/contents/reflex-syncope-in-adults-and-adolescents-treatment 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17] Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Neurally mediated syncope	A type of reflex syncope typically triggered by orthostasis leading to vasodilation and/or bradycardia that includes vasodepressor cardioinhibitory, and mixed syncope			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	Also called vasovagal syncope, neurocardiogenic syncope
Vasodepressor syncope	A type of neurally mediated syncope characterized primarily by reflex vasodilation			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Cardioinhibitory syncope	A type of neurally mediated syncope characterized primarily by reflex bradycardia			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Mixed syncope	A type of neurally mediated syncope characterized by reflex vasodilation and reflex bradycardia			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Situational syncope	A type of reflex syncope typically triggered by specific events, such as micturition, gastrointestinal stimulation, cough or sneeze, or exercise			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Syncope due to orthostatic hypotension	A syncopal event due to an inadequate increase in peripheral resistance and heart rate or due to severe hypovolemia upon standing			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Orthostatic hypotension	A progressive and sustained drop in blood pressure upon standing from a sitting or supine position in which there is a decrease of systolic blood pressure by ≥ 20 mmHg and/or a decrease of diastolic blood pressure by ≥ 10 mmHg within the first 3 min of upright position			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Initial orthostatic hypotension	A sudden drop in blood pressure upon standing from a sitting or supine position in which there is a decrease of systolic blood pressure by ≥ 40 mmHg and/or a decrease in diastolic blood pressure by ≥ 20 mmHg within the first 15 s of standing that spontaneously resolves with a relatively rapid return to pre-challenge values			Modified from Brignole M, et al. Eur Heart J 2018;39:e43-e80 [21]	
Cardiac syncope	A syncopal event due to a primary cardiac etiology, such as an arrhythmia or structural lesion				

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Postural orthostatic tachycardia syndrome	A type of chronic orthostatic intolerance lasting ≥ 3 months associated with excessive upright sinus tachycardia in the absence of orthostatic hypotension, plus a constellation of typically daily symptoms that may include lightheadedness, dizziness, nausea, dyspnea, diaphoresis, headache, fatigue, and other symptoms of autonomic dysfunction. Excessive tachycardia is defined by present consensus as a sustained heart rate increase of at least 30 bpm in adults (40 bpm for adolescents), or a heart rate > 120 bpm (in adults), within 10 min of upright tilt-table testing			Modified from 2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	Also called POTS
Hyperadrenergic postural orthostatic tachycardia syndrome	A phenotype of postural orthostatic tachycardia syndrome in which there is excessive sympathetic vasoconstriction, increased peripheral resistance, and typically excessively high serum catecholamine levels, with greatly elevated upright tachycardia and hypertension				
Neuropathic postural orthostatic tachycardia syndrome	A phenotype of postural orthostatic tachycardia syndrome in which there is impaired sympathetic activity in the lower extremities or splanchnic vasculature with arteriolar dysregulation				
Dysautonomia	A condition in which altered function of one or more components of the autonomic nervous system adversely affects health			Goldstein DS Principles of Autonomic Medicine v 4.0 [42]	Also called autonomic nervous system dysfunction
Inappropriate sinus tachycardia	A sinus heart rate > 100 bpm at rest (with a mean 24-h heart rate > 90 bpm not due to primary causes) and is associated with distressing symptoms of palpitations			Sheldon RS Heart Rhythm 2015;12(6):e41-e63 [44]	
Hypertension	The average clinic-measured systolic and/or diastolic blood pressure ≥ 95 th percentile for age, sex, and height percentiles			Flynn JT Pediatrics 2017;140(3):e20171904 [45]	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hypotension	Low systolic blood pressure, as defined by age parameters: 0–1 year: < 5th percentile for gestational and/or postnatal age 1–10 years: < 70 mmHg plus 2 X (age in years) > 10 years: < 90/50 mmHg			Dionne J Hypertension 2021;77(3):781–787 [46] Kleinman ME Circulation 2010;122(18Suppl3):S876–S908 [47]	
Post COVID-19 syndrome	A condition in which there are multiorgan symptoms and complications beyond the initial period of acute infection and illness with SARS-CoV-2			Venkatesan P Lancet Respir Med 2021;9(2):129 [43]	Also called long COVID, long-haul COVID, post-acute sequelae of SARS-CoV-2 infection (PASC)
Myalgic encephalomyelitis-chronic fatigue syndrome	A syndrome of unknown etiology characterized by an unexplained persistent or relapsing chronic fatigue that is of at least 6 months' duration, is not the result of ongoing exertion, is not substantially alleviated by rest, and results in substantial reduction of previous levels of occupational, educational, social, or personal activities			NCI Thesaurus	NCI Thesaurus Code C3037
Psychogenic pseudosyncope	The appearance of transient loss of consciousness in the absence of true loss of consciousness			Raj V Auton Neurosci 2014;184:66–72 [63]	
Sinus tachycardia	A disorder characterized by an electrocardiographic finding of abnormally rapid heart rate with its origin in the sinus node			NCI Thesaurus	NCI Thesaurus Code C26889
Tachycardia secondary to arrhythmia	A disorder characterized by an electrocardiographic finding of an abnormally rapid heart rate due to an abnormal heart rhythm				
Mast cell activation syndrome	A mast cell activation disorder in which there are episodic multisystem symptoms consistent with mast cell activation, appropriate response to medications targeting mast cell activation, and documented increase in validated markers of mast cell activation			Akin C. J Allergy Clin Immunol 2017;140(2):349–55 [49]	Also called MCAS
Ehlers–Danlos syndrome	A group of inherited connective tissue disorders characterized by loose and fragile skin and joint hypermobility			NCI Thesaurus	NCI Thesaurus Code C34568 Also called EDS

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hypermobile EDS	A type of Ehlers–Danlos syndrome characterized by generalized joint hypermobility, plus systemic manifestations of a generalized connective tissue disorder, positive family history, and/or musculoskeletal complications, plus exclusion of other types of Ehlers–Danlos syndrome			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called hEDS
Classical EDS	A type of Ehlers–Danlos syndrome characterized by skin hyperextensibility, generalized joint hypermobility, plus other minor criteria associated with a mutation in collagen in the <i>COL5A1</i> , <i>COL5A2</i> , <i>COL1A1</i> , or <i>COL1A2</i> genes			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called cEDS
Vascular EDS	A type of Ehlers–Danlos syndrome characterized by arterial, uterine, or gastrointestinal rupture at a young age, atraumatic formation of carotid-cavernous fistula, plus other minor criteria associated with a mutation in collagen in the <i>COL3A1</i> gene			Malfait F, et al. Am J Med Genet C Semin Med Genet 2017;175(1):8–26 [48]	Also called vEDS
Hypermobility spectrum disorder	A group of clinically relevant conditions related to joint hypermobility in which phenotypic domains are limited to the musculoskeletal system			Castori M, et al. Am J Med Genet C Semin Med Genet 2017;175(1):148–57 [36]	Also called HSD
Median arcuate ligament syndrome	A condition characterized by abdominal pain attributed to compression of the celiac artery and the celiac ganglia by the median arcuate ligament			www.wikipedia.org	Also called MALS
Small fiber neuropathy	Neuropathy caused by damage to the small myelinated (A-delta) fibers or unmyelinated C fibers in the peripheral nerves			NCI Thesaurus	NCI Thesaurus Code C125389
Sjögren syndrome	An autoimmune disorder affecting the salivary and lacrimal glands. Morphologically, it is characterized by the presence of lymphocytic and plasmacytic infiltrates which cause destruction of these glands			NCI Thesaurus	NCI Thesaurus Code C26883

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Irritable bowel syndrome	Gastrointestinal symptoms characterized by chronic abdominal pain and altered bowel habits in the absence of any organic cause. One of the functional gastrointestinal disorders			Modified from NCI Thesaurus	NCI Thesaurus Code C82343
Rumination	An eating disorder most commonly observed in infants and adolescents characterized by the repeated regurgitation and rechewing of food for a period of at least 1 month; this behavior is not associated with a gastrointestinal or other medical reason. One of the functional gastrointestinal disorders				
Nausea	A difficult-to-describe sensation of sick or queasy feeling usually perceived as being in the stomach that can escalate in severity and may precede vomiting			Stern RM, Koch KL, Andrews PLR (2001) Nausea: Mechanisms and Management. Oxford University Press, New York [29]	
Constipation	Difficulties in defecation that include infrequent bowel movements, hard or lumpy stools, excessive straining, sensation of incomplete evacuation or blockage and, in some instances, the use of manual maneuvers to facilitate evacuation			Aziz I, et al. Expert Rev Gastroenterol Hepatol 2020;14:39–46 [30]	
Gastroparesis	Paralysis of the muscles of the stomach wall resulting in delayed emptying of the gastric contents into the small intestine			NCI Thesaurus	NCI Thesaurus Code C80512
Dysmotility	A descriptive term used to describe diseases of the gastrointestinal tract in which the muscles and/or nerves do not work normally			Modified from www.medicinenet.com	
Migraine	A common, severe type of vascular headache often associated with increased sympathetic activity, resulting in nausea, vomiting, and light sensitivity			NCI Thesaurus	NCI Thesaurus Code C89715
Tension headache	A headache associated with muscle tightness that may radiate to other parts of the body			NCI Thesaurus	NCI Thesaurus Code C117070

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Cluster headache	A headache disorder that is characterized by periodic severe, unilateral orbital, supraorbital, and/or temporal pain, and is associated with ipsilateral cranial autonomic symptoms			NCI Thesaurus	NCI Thesaurus Code C117077
New persistent daily headache	A bilateral, unremitting headache of mild to moderate intensity that occurs daily and may be associated with photophobia, phonophobia, or mild nausea			NCI Thesaurus	NCI Thesaurus Code C117082
Craniocervical instability	A pathological condition in which ligamentous connections from the skull to the spine are incompetent			Henderson FC Am J Med Genet C Semin Med Genet 2017;175(1):195–211 [64]	
Cerebrospinal fluid leak	The loss of cerebrospinal fluid into the surrounding tissues			NCI Thesaurus	NCI Thesaurus Code C50487
Thyroid disease	The classification of autoimmune thyroid disease (AITD) includes Hashimoto thyroiditis (HT) or chronic autoimmune thyroiditis and its variants, Graves disease (GD) and autoimmune atrophic thyroiditis or primary myxedema			NCI Thesaurus	NCI Thesaurus Code C91510
Graves disease	Hyperthyroidism associated with diffuse hyperplasia of the thyroid gland (goiter), resulting from production of antibodies that are directed against the thyrotropin receptor complex of the follicular epithelial cells. As a result, the thyroid gland enlarges and secretes increased amounts of thyroid hormones			NCI Thesaurus	NCI Thesaurus Code C3071
Hashimoto thyroiditis	An autoimmune disorder caused by the production of autoantibodies against thyroid tissue. There is progressive destruction of the thyroid follicles leading to hypothyroidism			NCI Thesaurus	NCI Thesaurus Code C27191
Adrenal failure, hypoadrenalism, Addison disease	A long-standing and persistent diminished production of adrenocortical hormones due to a disorder originating within the adrenal glands			NCI Thesaurus	NCI Thesaurus Code C26689

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Pheochromocytoma	A benign or malignant neuroendocrine neoplasm of the sympathetic nervous system that secretes catecholamines. It arises from the chromaffin cells of the adrenal medulla			NCI Thesaurus	NCI Thesaurus Code C3326
Carcinoid syndrome	A syndrome characterized by signs and symptoms caused by advanced carcinoid tumors. They include skin flushing, diarrhea, wheezing, and tachycardia			NCI Thesaurus	NCI Thesaurus Code C3215
Paraganglioma	A benign or malignant neoplasm arising from paraganglia located along the sympathetic or parasympathetic nerves			NCI Thesaurus	NCI Thesaurus Code C3308
Neuroblastoma	A neuroblastic tumor characterized by the presence of neuroblastic cells, the absence of ganglion cells, and the absence of a prominent Schwannian stroma formation			NCI Thesaurus	NCI Thesaurus Code C3270
Ganglioneuroma	A benign neuroblastic tumor of the sympathetic nervous system that occurs in childhood. Ganglioneuroma typically arises from the sympathetic chain in the mediastinum or adrenal glands			Modified from NCI Thesaurus	NCI Thesaurus Code C3049
Ganglioneuroblastoma	A neuroblastic tumor characterized by the presence of neuroblastic cells, ganglion cells, and a stroma with Schwannian differentiation constituting more than 50% of the tumor volume			NCI Thesaurus	NCI Thesaurus Code C3790
Eosinophilic esophagitis	Allergic inflammation of the esophagus. Morphologically, it is characterized by the presence of eosinophils infiltrating the esophageal epithelium			NCI Thesaurus	NCI Thesaurus Code C27105
Visceral hypersensitivity	Heightened sensitivity or reduced thresholds for pain and discomfort associated with the gastrointestinal tract			Modified from Mertz H. Aliment Pharmacol Ther 2003;17(5):623–33 [31] AND Faure C, Di Lorenzo C, Thapar N. (2017) Pediatric Neurogastroenterology Gastrointestinal Motility and Functional Disorders in Children, 2nd Ed. Springer International Publishing, Switzerland [33]	Also called visceral hyperalgesia

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Gastroesophageal reflux	The backward flow of stomach acid contents into the esophagus (the tube that connects the mouth to the stomach)			NCI Thesaurus	NCI Thesaurus Code C92560
Breath-holding spell	A clinical condition in which there is a brief involuntary cessation of breathing at end expiration in response to emotional or painful stimulus seen in infants, toddlers, and children			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Simple breath-holding spell	A clinical condition in which there is a breath-holding spell without syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Complex breath-holding spell	A clinical condition in which there is a breath-holding spell associated with syncope			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Pallid breath-holding spell	A clinical condition in which there is a breath-holding spell associated with pale appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Cyanotic breath-holding spell	A clinical condition in which there is a breath-holding spell associated with cyanotic appearance			2017 AHA/ACC Key Data Elements and Definitions for Ambulatory Electronic Health Records in Pediatric and Congenital Cardiology [17]	
Seizure disorder	A brain disorder characterized by episodes of abnormally increased neuronal discharge resulting in transient episodes of sensory or motor neurological dysfunction, or psychic dysfunction that may or may not be associated with loss of consciousness or convulsions			NCI Thesaurus	NCI Thesaurus Code C3020 Also called epilepsy
Spinal cord injury	Traumatic damage of the spinal cord			NCI Thesaurus	NCI Thesaurus Code C50750
Autonomic dysreflexia	A form of paroxysmal autonomic adrenergic overactivity due to disinhibited spinal reflexes in spinal cord injury above the T6 level			Novak P. Am J Med 2019;132(4):420–436 [51]	

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Anxiety disorder	A category of psychiatric disorders which are characterized by anxious feelings or fear often accompanied by physical symptoms associated with anxiety			NCI Thesaurus	NCI Thesaurus Code C2878
Panic disorder	An anxiety disorder characterized by multiple unexpected panic attacks with persistent concern of recurring attacks			NCI Thesaurus	NCI Thesaurus Code C34890
Depression	A mental condition marked by ongoing feelings of sadness, despair, loss of energy, and difficulty dealing with normal daily life			NCI Thesaurus	NCI Thesaurus Code C2982
Obsessive–compulsive disorder	A disorder characterized by the presence of persistent and recurrent irrational thoughts (obsessions), resulting in marked anxiety and repetitive excessive behaviors (compulsions) as a way to try to decrease that anxiety			NCI Thesaurus	NCI Thesaurus Code C88411
Attention deficit/hyperactivity disorder	A neurodevelopmental disorder defined by impairing levels of inattention, disorganization, and/or hyperactivity–impulsivity			DSM-V	Also called ADHD
Somatic symptom disorder	A. One or more somatic symptoms that are distressing or result in significant disruption of daily life B. Excessive thoughts, feelings, or behaviors related to the somatic symptoms or associated health concerns C. Although any one somatic symptom may not be continuously present, the state of being symptomatic is persistent (typically more than 6 months)			DSM-V	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Conversion disorder (functional neurological symptom disorder)	<p>A. One or more symptoms of altered voluntary motor or sensory function</p> <p>B. Clinical findings provide evidence of incompatibility between the symptom and recognized neurological or medical conditions</p> <p>C. The symptom or deficit is not better explained by another medical or mental disorder</p> <p>D. The symptom or deficit causes clinically significant distress or impairment in social, occupational, or other important areas of functioning or warrants medical evaluation</p>			DSM-V	
Hereditary and sensory neuropathy, type I	An autosomal dominant condition caused by mutation(s) in the <i>SPTLC1</i> gene characterized by distal sensory impairment with variable autonomic and motor involvement			NCI Thesaurus	NCI Thesaurus Code C170433
Hereditary and sensory neuropathy, type IIA (HSAN2A)	A rare congenital disorder caused by various mutations in the <i>WNK1</i> gene characterized by sensory loss, self-mutilation, and mild or absent autonomic involvement			Modified from https://www.ncbi.nlm.nih.gov/books/NBK49247/ and https://www.omim.org/entry/201300	
Hereditary and sensory neuropathy, type IIB (HSAN2B, Morvan's disease; congenital sensory neuropathy)	A rare autosomal recessive disorder caused by various mutations in the <i>FAM134B</i> gene characterized by decreased distal sensation and variable autonomic involvement			Modified from https://www.ncbi.nlm.nih.gov/books/NBK49247/ and https://www.omim.org/entry/613115	
Hereditary and sensory neuropathy, type III (HSAN3, familial dysautonomia; Riley-Day syndrome)	A rare congenital disorder caused by a founder mutation in the <i>ELP-1</i> gene characterized by initial hypotonia and feeding difficulties with subsequent blood pressure instability and hypertensive vomiting attacks alternating with hypotension due to loss of sensory inputs from cranial nerves IX and X resulting in afferent baroreflex failure			Modified from https://www.omim.org/entry/223900	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Hereditary and sensory neuropathy, type IV (HSAN4, congenital insensitivity to pain with anhidrosis; CIPA)	A rare autosomal recessive disorder caused by different mutations in the <i>NTRK1</i> gene affecting peripheral sensory and autonomic fibers, and is characterized by inability to feel pain and temperature that leads to repeated unintentional self-injuries, severe cognitive impairment, and anhidrosis that leads to hyperpyrexia and febrile seizures			Modified from NCI Thesaurus and https://www.omim.org/entry/256800	NCI Thesaurus Code C118633
Hereditary and sensory neuropathy, type V (HSAN5)	An autosomal recessive condition caused by mutations in the <i>NGF</i> or <i>NTRK1</i> genes affecting peripheral small fibers characterized by loss of pain sensation, particularly in the extremities, and variable anhidrosis, with normal cognitive function as compared with HSAN4			Modified from NCI Thesaurus and https://www.omim.org/entry/608654	NCI Thesaurus Code C156360
Hereditary and sensory neuropathy, type VI (HSAN6)	An extremely rare inherited disorder caused by mutations in the <i>DST</i> gene characterized by neonatal expression of diminished pain and labile blood pressure			https://www.ncbi.nlm.nih.gov/medgen/761278 and https://www.omim.org/entry/614653	
Hereditary and sensory neuropathy, type VII (HSAN7)	A rare disorder caused by mutations in the <i>SCN11A</i> gene, characterized by insensitivity to pain as well as gastrointestinal dysfunction			Modified from NCI Thesaurus and https://www.omim.org/entry/615548	NCI Thesaurus Code C125388
Hereditary and sensory neuropathy, type VIII (HSAN8)	An autosomal recessive disorder caused by mutations in the <i>PRDM12</i> gene characterized by congenital insensitivity to pain; anhidrosis and alacrima may occur			Modified from https://www.ncbi.nlm.nih.gov/medgen/894363 and https://www.omim.org/entry/616488	

Terminology concept	Definition	Data element	Permissible values	Source of definition	Comments
Rett syndrome	A progressive neurologic disorder caused by mutations in the <i>MECP2</i> gene on chromosome X. It almost exclusively affects girls. It is characterized by breath-holding spells, flushing, repetitive hand motions, lack of advancement in language, and regression in developmental milestones. Other signs and symptoms include slowed head growth, scoliosis, breathing abnormalities (including sleep-disordered breathing), and sleep disturbances			Modified from NCI Thesaurus	NCI Thesaurus Code C75488
Hirschsprung disease	A congenital disorder characterized by the absence of myenteric ganglion cells in the colon that occurs distally but can involve the proximal colon resulting in a functional stenosis of the distal colon and a massive distention of the proximal colon			Modified from NCI Thesaurus	NCI Thesaurus Code C34700
Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation	A neurocristopathy characterized by hypothalamic dysfunction with inability to maintain normal salt-water balance, growth hormone deficiency, hypothyroidism, early or late puberty, increased prolactin concentration, hypoventilation, attenuated/absent peripheral and central chemoreceptor responsiveness, and autonomic dysfunction			Modified from NCI Thesaurus	NCI Thesaurus Code C121944 Also called ROHHAD
Congenital central hypoventilation syndrome	A neurocristopathy associated with heterozygous <i>PHOX2B</i> gene mutations characterized by hypoventilation and hypoxemia typically during sleep with attenuated/absent peripheral and central chemoreceptor responsiveness, and autonomic dysregulation; symptoms typically appear in the newborn period and can co-occur with Hirschsprung disease and neural crest tumors			Modified from NCI Thesaurus	NCI Thesaurus Code C98889 Also called CCHS
Tracheostomy	A surgically created external opening into the trachea			NCI Thesaurus	NCI Thesaurus Code C122643

HR heart rate, BP blood pressure, bpm beats per minute

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Declarations

Conflict of interest This manuscript does not contain clinical studies or patient data. The authors declare that they have no conflict of interest.


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Authors and Affiliations

Jeffrey R. Boris¹  · Hasan Abdallah² · Shelley Ahrens³ · Gisela Chelimsky⁴ · Thomas C. Chelimsky⁵ · Philip R. Fischer^{3,6,7} · John E. Fortunato⁸ · Raewyn Gavin⁹ · Janice L. Gilden¹⁰ · Renato Gonik¹¹ · Blair P. Grubb¹² · Kelsey M. Klaas³ · Erin Marriott¹³ · Lauren E. Marsillio^{14,15,16} · Marvin S. Medow¹⁷ · Lucy Norcliffe-Kaufmann¹⁸ · Mohammed T. Numan¹⁹ · Erin Olufs²⁰ · Laura A. Pace²¹ · Paul T. Pianosi²² · Pippa Simpson²³ · Julian M. Stewart¹⁷ · Sally Tarbell²⁴ · Natalie R. Van Waning²⁵ · Debra E. Weese-Mayer^{14,15,16}

- ¹ Jeffrey R. Boris, MD LLC, P.O. Box 16, Moylan, PA 19065, USA
- ² Children's Heart Institute, Herndon, VA, USA
- ³ Mayo Clinic, Rochester, MN, USA
- ⁴ Children's Hospital of Richmond, Virginia Commonwealth University Health, Richmond, VA, USA
- ⁵ Virginia Commonwealth University Health, Richmond, VA, USA
- ⁶ Sheikh Shakhboub Medical City, Abu Dhabi, UAE
- ⁷ Khalifa University College of Medicine and Health Sciences, Abu Dhabi, UAE
- ⁸ Lurie Children's Hospital, Chicago, IL, USA
- ⁹ Starship Children's Hospital, Auckland, NZ, USA
- ¹⁰ Rosalind Franklin University of Medicine and Science, North Chicago, IL, USA
- ¹¹ University of Florida College of Medicine, Gainesville, FL, USA
- ¹² University of Toledo, Toledo, OH, USA
- ¹³ American Family Children's Hospital, Madison, WI, USA
- ¹⁴ Ann & Robert H Lurie Children's Hospital of Chicago, Chicago, IL, USA
- ¹⁵ Stanley Manne Children's Research Institute, Chicago, IL, USA
- ¹⁶ Northwestern University Feinberg School of Medicine, Chicago, IL, USA
- ¹⁷ New York Medical College, Valhalla, NY, USA
- ¹⁸ New York University Grossman School of Medicine, New York, NY, USA
- ¹⁹ University of Texas Houston McGovern Medical School, Houston, TX, USA
- ²⁰ University of Iowa Hospitals and Clinics, Iowa City, IA, USA
- ²¹ Metrodora Institute, Salt Lake City, UT, USA
- ²² University of Minnesota Medical School, Minneapolis, MN, USA
- ²³ Medical College of Wisconsin, Milwaukee, WI, USA
- ²⁴ Northwestern Feinberg School of Medicine, Chicago, IL, USA
- ²⁵ University of Iowa Children's Hospital, Iowa City, IA, USA