

## Glossary dedicated to CDG Symptom Prioritization Questionnaire – (CDGSPQ)

### WHO DEVELOPED THIS PROJECT?

This project is developed under the scope of **CDG & Allies – Professionals and Patient Associations International Network (CDG & Allies – PPAIN)** and led by Dr. Vanessa Ferreira, Prof. Dr. Paula Videira and Prof. Luísa Barros. We had the support of experts like Pf. Dr. Jaak Jaeken, Pf. Eva Morava, Dr. Mercedes Serrano, Andrea Miller (CDG Care), Merell Liddle (CDG Australia), Claudia Vazquez (CDG USA) and CDG & Allies - PPAIN colleagues. CDG & Allies-PPAIN is a unique international patient-led network focused on increasing CDG knowledge and ultimately, contributing to future therapies and better management of CDG.

### HELP CONTACT:

If you need help, please do not hesitate to contact us asking for help related to this survey at <https://worldcdg.org/contact>. We will be available to schedule a SKYPE, WhatsApp CALL, or Zoom meeting.

### CDG Ophthalmologic

**Nystagmus** is a condition of involuntary eye movements, that can result in reduced or limited vision. More information [HERE](#). Watch a video [HERE](#).

**Strabismus (a.k.a crossed eyes)** is a condition of misalignment of the eyes when looking at an object. More information [HERE](#).



Figure source [HERE](#).

**Retinitis pigmentosa** involves a gradual loss of the vision by the following order: at night; peripheral vision; central vision and problems with color vision. More information [HERE](#).

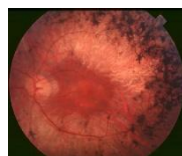


Figure legend: Fundus of patient with retinitis pigmentosa; Figure source [HERE](#).

## CDG Gastrointestinal

**(Gastrointestinal tract** is the tract from the mouth to the anus which includes all the organs of the digestive system. More information [HERE](#).)

**Dysphagia** is the difficulty in swallowing. More information [HERE](#).

**Anorexia** involves loss of appetite, no feeling of hunger and even an aversion to food.

**Abdominal bloating** occurs when substances such as gas or fluid accumulate in the abdomen. It causes a feeling of fullness. More information [HERE](#).

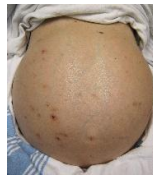


Figure source [HERE](#).

**Abdominal pain** is the pain that you feel anywhere between your chest and groin. This is often referred to as the stomach region or belly. More information [HERE](#).

**Nausea** is a diffuse sensation of unease and discomfort, often perceived as an urge to vomit. More information [HERE](#).

**Gastroesophageal reflux** happens when stomach acid flows back into the esophagus (tube connecting your mouth and stomach). This can irritate the esophagus and cause **heartburn** (a burning sensation in the chest). More information [HERE](#) and [HERE](#).

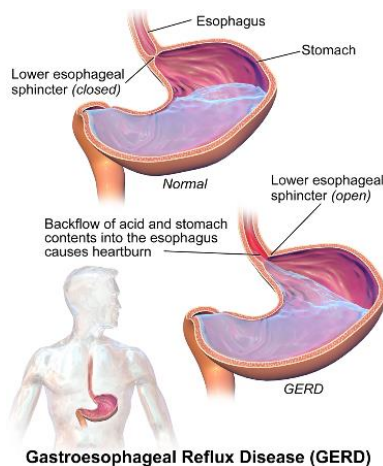


Figure source [HERE](#).

**Vomiting** is an uncontrollable reflex that expels the contents of the stomach through the mouth. More information [HERE](#).



Figure source [HERE](#).

**Gastrointestinal bleeding** reveals a disorder in the digestive tract. The blood often appears in vomit or stools but isn't always visible. However, it may cause the stools to look black or tarry. The bleeding can range from mild to severe and can be life-threatening. More information [HERE](#) and [HERE](#).

**Constipation** occurs when bowel movements become less frequent and stools become difficult to pass. More information [HERE](#).

**Diarrhea** is the condition of having at least three loose or liquid stools per day. It can last for a few days and result in dehydration due to fluid loss. More information [HERE](#).








BRISTOL STOOL CHART			
	Type 1	Separate hard lumps	<b>SEVERE CONSTIPATION</b>
	Type 2	Lumpy and sausage like	<b>MILD CONSTIPATION</b>
	Type 3	A sausage shape with cracks in the surface	<b>NORMAL</b>
	Type 4	Like a smooth, soft sausage or snake	<b>NORMAL</b>
	Type 5	Soft blobs with clear-cut edges	<b>LACKING FIBRE</b>
	Type 6	Mushy consistency with ragged edges	<b>MILD DIARRHEA</b>
	Type 7	Liquid consistency with no solid pieces	<b>SEVERE DIARRHEA</b>

Figure source [HERE](#).

**Fecal incontinence** is the inability to control bowel movements, causing feces to leak unexpectedly from the rectum. It can range from an occasional leakage of feces while passing gas to a complete loss of bowel control. More information [HERE](#).

**Failure to thrive** consists of an insufficient weight gain or inappropriate weight loss. More information [HERE](#).

## Gastrointestinal problems associated with food

**Lactose intolerance** happens when the small intestine doesn't produce enough of an enzyme (lactase) to digest milk sugar (lactose). Lactose intolerance causes diarrhea, gas and bloating, nausea, and sometimes, vomiting after eating or drinking dairy products. More information [HERE](#)

**Fermentable Oligo-, Di-, Mono-saccharides And Polyols - FODMAP intolerance** is the sensitivity to a group of sugars poorly digested/absorbed in the small intestine, prone to absorb water and ferment in the colon (by gut bacteria) (e.g. fructans, galactooligosaccharides, lactose, fructose, sorbitol, mannitol, xylitol, and maltitol), producing gas and expanding the intestinal wall. Although this natural process promotes beneficial alterations in the gut flora, it can cause pain and discomfort in some people with a highly sensitive gut to 'stretching' the intestinal wall (e.g. irritable bowel syndrome (IBS)). More information [HERE](#).

**Celiac disease** is an autoimmune disorder in which gluten (a protein found in wheat, barley and rye) triggers an immune response in the small intestine. Over time, this can damage the small intestine's lining and prevents it from absorbing some nutrients (malabsorption). The intestinal damage can cause diarrhea, fatigue, weight loss, bloating and anemia, and can lead to serious complications. In children, malabsorption can affect growth and development. Although there's no cure, following a strict gluten-free diet can help manage symptoms and promote intestinal healing. It can be diagnosed by a blood test for certain types of antibodies, followed by a biopsy of the small intestine if the first test is positive. More information [HERE](#).

**Non-celiac gluten (a protein found in wheat, barley and rye) sensitivity** shows some signs and symptoms identical to Celiac disease (e.g. abdominal pain, bloating, diarrhea, constipation, "foggy brain," rash or headache) although there is no damage to the tissues of the small intestine. The immune system seems to play a role, but the process isn't well understood. More information [HERE](#) and [HERE](#).

**Inflammatory bowel disease (IBD)** is a group of disorders, e.g. **Crohn's disease** and **Ulcerative colitis**, that cause chronic inflammation of the digestive tract. Crohn's disease causes inflammation of the lining of your digestive tract, which often spreads deep into affected tissues, while Ulcerative colitis causes long-lasting inflammation and sores (ulcers) in the innermost lining of your large intestine (colon) and rectum. Both involve debilitating and sometimes life-threatening complications: diarrhea; fever; fatigue; abdominal pain; blood in

the stools; reduced appetite; weight loss. More information [HERE](#).

**Food allergy** occurs when the immune system misidentifies a protein (e.g. gluten) found on some food (e.g. wheat) as a disease-causing agent (e.g. bacteria), produces antibodies against this protein, prompting an immune system response. This can cause tingling or itching in the mouth; hives, itching or eczema; swelling of the lips, face, tongue and throat or other parts of the body; wheezing, nasal congestion or trouble breathing; abdominal pain, diarrhea, nausea or vomiting; dizziness, lightheadedness or fainting. Some people trigger anaphylaxis, a severe and life-threatening allergic reaction, causing constriction of airways that makes it difficult to breathe, shock with a severe drop in blood pressure, rapid pulse, dizziness, lightheadedness or loss of consciousness. More information [HERE](#).

Figure legend: Hives on back caused by food allergy and Skin allergy tests. Figure source [HERE](#), [HERE](#) and [HERE](#).

### CDG Bladder

**Urinary incontinence** is an involuntary and unintentional discharge of urine. It can range from a few drops to permanent leakage. More information [HERE](#).

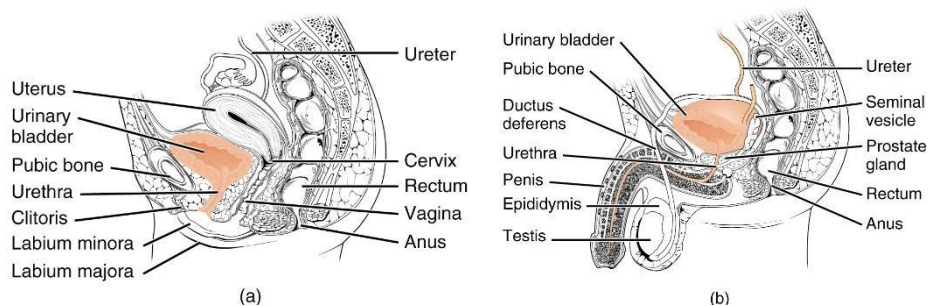


Figure legend: Female (a) and male (b) urinary system. Figure source [HERE](#).

### CDG Reproductive

**(Reproductive or genital system** comprehends all the anatomical organs involved in sexual reproduction).

**Infertility** is the inability of a person to reproduce by natural means. More information [HERE](#).

Menstrual flow usually occurs every 21 to 35 days and lasts 2 to 7 days. **Abnormal types of the menstrual period** include prolonged, heavy bleeding; excess pain during the period; absent periods and extra light periods. More information [HERE](#).

## CDG Skeletal

**Osteopenia/ Osteoporosis** are conditions showing low bone density and a higher risk of fractures. Osteoporosis is more severe than osteopenia. More information [HERE](#) and [HERE](#).

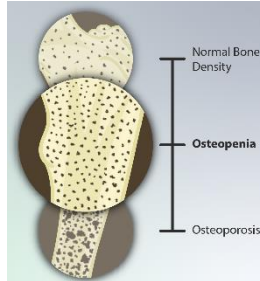


Figure source [HERE](#).

**Kyphosis** is an exaggerated, forward rounding of the back. More information [HERE](#).

**Scoliosis** is a sideways curvature of the spine. More information [HERE](#).

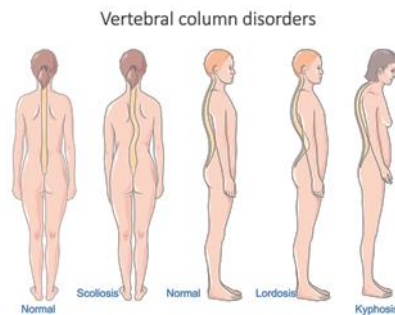


Figure source [HERE](#).

**Bone fracture** is a medical condition with a partial or complete break in the continuity of the bone.

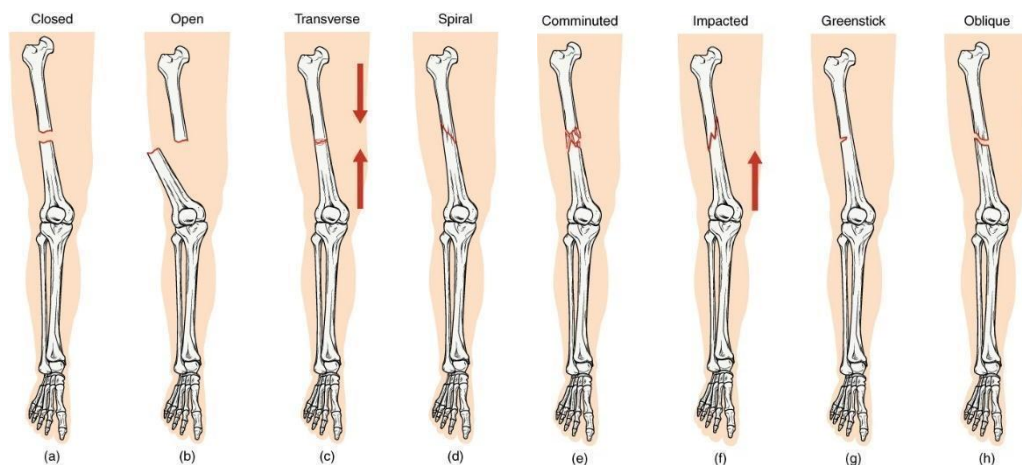


Figure source [HERE](#).

### CDG Neuro and muscular

(**The neurological system** includes the network of neurons and cells that carry messages to and from the brain and spinal cord to various parts of the body. **The muscular system** is responsible for the movement of the human body).

**Intellectual development delay** comprehends an impaired understanding, learning, and adaptive functioning. More information [HERE](#).

**Social problems or disability** involves difficulties in verbal, non-verbal or written communication with others.

**Fine motor disability** comprises the poor coordination of small muscles during the movement. Examples of fine motor skills, usually requiring the synchronization of hands, fingers and the eyes are eating, writing, and buttoning a shirt. More information [HERE](#).

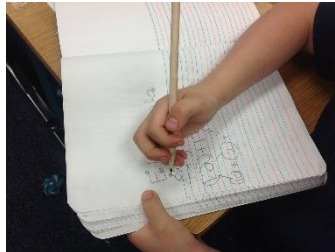


Figure source [HERE](#).

**Gross motor disability** comprises the poor coordination gross movements come from large muscle groups and whole-body movement. Examples of gross motor skills are standing up, walking and running, walking upstairs. More information [HERE](#).



**Figure legend:** Gross motor skills such as climbing and balancing. **Figure source** [HERE](#).

**Joint contracture** is a full limitation of motion of a joint. It results from shortening of muscles, tendons, ligaments, and joint capsules or heterotopic ossification (abnormal growth of bone in the non-skeletal tissues including muscle, tendons, or other soft tissues). More information [HERE](#).



Figure source [HERE](#).

**Speech impairments** embrace difficulties in the articulation of the words, fluency, and/or voice.

- Articulation problems involve errors in the production of speech sounds (e.g. omissions: (bo for boat); substitutions: (wabbit for rabbit); distortions: (shlip for sip)).
- Fluency problems comprehend difficulties with the rhythm and timing of speech (e.g. hesitations, repetitions, or prolongations of sounds, syllables, words, or phrases).
- Voice problems refer to the characteristics of the sounds produced, specifically the pitch, quality, and intensity of the sound. More information [HERE](#).

**Seizures** are a sudden, uncontrolled electrical disturbance in the brain. They can cause changes in the behavior, movements or feelings, and in levels of consciousness. **Epilepsy** is characterized by recurrent epileptic seizures. There are many types of seizures, which range in severity. Seizure types differ by where and how they begin in the brain. More information [HERE](#), [HERE](#) and [HERE](#).

**Stroke-like episodes (SLE) in PMM2-CDG** patients are acute neurological complications characterized by confusional status, the loss of movement from one leg or arm (monoparesis), or one side of the body (hemiparesis), and sometimes, epileptic seizures. They involve focal deficits that mimic clinically, but not neuroradiologically, an ischemic injury. Infections or fevers can trigger SLE. However, the underlying pathomechanisms are not understood, and clinical guidelines helping their diagnosis, and prevention and treatment are missing. More information [HERE](#).



## Emotional/ behavioral development disturbance

**Mood swings** are an extreme or rapid change in mood. It can happen any time at any place, and the duration and severity can also vary. More information [HERE](#).



Figure source [HERE](#).

**Repetitive behaviors** are categorized into two groups:

- Lower-order which involve movements such as hand-flapping, fidgeting with objects or body rocking, and vocalizations such as grunting or repeating certain phrases.
- Higher-order which include routines and rituals, insistence on sameness and intense interests. More information [HERE](#).

**Autistic features** include problems with social interaction with others (e.g. playing with others); unusual interest in objects; the need for sameness; great variation in abilities; under or overreaction to one or more of the five senses (sight, touch, taste, smell, or hearing); repeated actions or movements; unusual emotional reactions and expressions. More information [HERE](#).



Figure source [HERE](#).

**Aggressive behavior** can cause physical or emotional harm to others. It may range from verbal abuse to physical abuse or harming personal property. More information [HERE](#).

**Self-abusive behavior** involves the occurrence of behavior that results in physical injury to one's own body. More information [HERE](#).

**Tantrums** are an emotional outburst, typically related to those in emotional distress. It can involve stubbornness, crying, screaming, violence, defiance, angry ranting, and resistance. More information [HERE](#).



Figure source [HERE](#).

**Anxiety** is an intense, excessive feeling of fear, worry or apprehension about what's to come. It can cause an increased heart rate; breathing rapidly (hyperventilation); sweating; trembling; feeling tired; gastrointestinal problems; the urge to avoid things that trigger anxiety; trouble concentrating and sleeping. More information [HERE](#) and [HERE](#).

### **CDG Hematology and immunology**

(**Hematology** is the study of blood and related conditions. **Immunology** is the study of the immune system, responsible for the body's defense mechanisms).

**Coagulopathy** is a condition with impaired blood's ability to coagulate (form clots). It increases the **tendency toward prolonged or excessive bleeding and bruising**, which may occur spontaneously, following an injury or during medical procedures.

Coagulopathy can result from reduced levels or absence of blood-clotting proteins such as clotting factors or coagulation factors. It can also result from dysfunction of platelets (small disk-shaped cells in the bloodstream that help in the clotting process) or from **reduced levels of platelets (Thrombocytopenia)**. More information [HERE](#), [HERE](#) and [HERE](#).



Figure source [HERE](#).

**Thrombosis** is the **formation of a blood clot inside an uninjured blood vessel** (by platelets and fibrin), obstructing the flow of blood through the circulatory system. A piece of the clot can break off as an embolus which can travel through the circulation and lodge somewhere else as a thromboembolism. More information [HERE](#).

**Infections** are the invasion of an organism's body tissues by disease-causing agents (e.g. viruses, fungi and parasites), their multiplication, and the reaction of specific host tissues/cells (immune system) to the infectious agents and their toxins. Infections can affect a single organ/ system (localized infection - e.g. gastrointestinal infections) or the entire body (systemic infection - e.g. sepsis), and the symptoms depend on the type of disease.

- **Gastrointestinal infections** can cause abdominal cramps and discomfort, diarrhea, nausea, vomiting, fever, loss of appetite, muscle aches, dehydration, headache, mucus or blood in the stools and weight loss.
- **Urinary tract infections** can affect a part of the urinary tract (e.g. bladder or kidney). **Bladder infection** cause pain while urinating, frequent urination, and feeling the need to urinate even with the bladder empty. In addition to these symptoms, **kidney infection** includes fever and flank pain.
- **Sepsis** is a life-threatening condition that occurs when the body's response to infection is out of balance, causing injury to its own tissues and organs. It involves fever, increased heart rate, increased breathing rate, and confusion. More information [HERE](#), [HERE](#), and [HERE](#). Watch a video [HERE](#).

### Other CDG signs and symptoms

**Unexplained fever episodes** are episodes of fever that happen without any visible or known reason.

**Insomnia** is a sleep disorder that can make it hard to fall asleep or to stay asleep. More information [HERE](#).

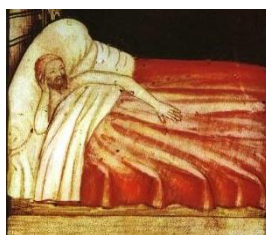


Figure source [HERE](#).

**Shortness of breath** (dyspnea) is described as an intense tightening in the chest or difficulty breathing. Its severity depends on when occurring (at rest or being associated with activity, exertion, or exercise). More information [HERE](#).



Figure source [HERE](#).

**Fluid retention** is the buildup of fluid in the body's tissue, resulting in swelling of this tissue (e.g. around your eyes, legs, ankles, feet, abdomen) and/ or weight gain. More information [HERE](#).



Figure source [HERE](#).

**Fatigue** is a feeling of tiredness, and it can be physical or mental. Physical fatigue is the temporary physical inability of muscles to perform optimally, while mental fatigue is a temporary inability to maintain optimal cognitive performance. **Fatigue** usually can be improved by periods of rest. More information [HERE](#).

**Pain** is "an unpleasant sensory and emotional experience associated with, or resembling that associated with, actual or potential tissue damage." according to the [International Association for the Study of Pain](#).

**Elevated serum transaminases (liver enzymes):** Alanine aminotransferase (ALT) and/ or Aspartate aminotransferase (AST), may be an indicator of liver damage. More information [HERE](#).

**Impact of signs and symptoms on the patient's everyday life** means that they prevent the patient from carrying out 1) daily activities and tasks (e.g. playing, preparing meals, household chores, shopping); 2) motor and sensorial functioning (e.g. difficulties in maintaining body positions, eye problems); 3) personal care (e.g. personal hygiene, dressing/undressing, eating/ drinking; take medication); 4) understanding and learning 5) social life, relation and communication with others; 6) controlling general behavior; 7) taking care of finances and handling administrative tasks.