

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 Lidlle (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.

Health care specialties

Allergists are physicians specialized in the diagnosis and treatment of allergies. More information [HERE](#).

Cardiologists are physicians practicing cardiology, a branch of medicine that deals with the disorders of the heart and some parts of the circulatory system. More information [HERE](#).

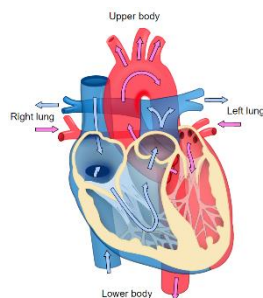


Figure source [HERE](#).

Endocrinologists are specialist in endocrinology, a branch of biology and medicine that deals with the endocrine system in health and diseases. The endocrine system is a system of glands which secretes hormones (e.g. thyroid hormone, growth hormone, and insulin), which are chemicals that affect the actions of different organ systems in the body. It involves positive feedback mechanisms, where one hormone stimulates the release of another secondary hormone, and negative feedback, where an excess of secondary hormone inhibits the release of the primary hormone, maintaining the balance (homeostasis). More information [HERE](#).

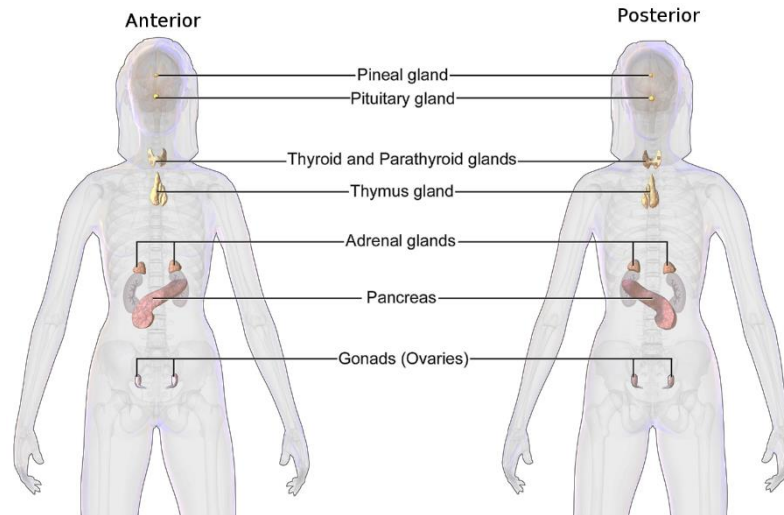


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Gastroenterologists are physicians practicing gastroenterology, a branch of medicine that deals with the digestive system and its disorders. More information [HERE](#).

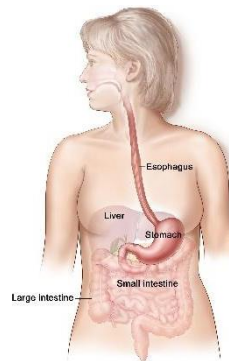


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Geneticists are biologists who study genetic (the science of genes), heredity, and variation of organisms, or physicians specialized in genetics. They can evaluate, diagnose, and manage patients with hereditary conditions or congenital malformations, genetic risk calculations, and mutation analysis, and refer patients to other medical specialists. They perform studies, test, and counsel patients with genetic disorders. More information [HERE](#).

Hematologists are physicians practicing hematology, a branch of medicine that focuses on the study of the cause, prognosis, treatment, and prevention of diseases related to blood. More information [HERE](#).

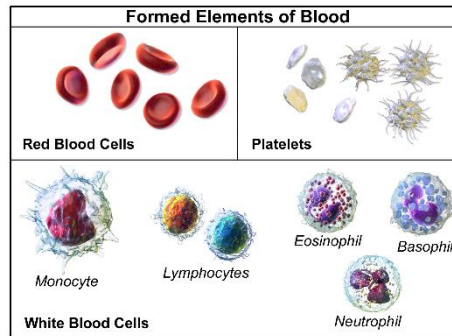


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Hepatologists are physicians practicing hepatology, a branch of medicine that focuses on the study of liver, gallbladder, biliary tree, and pancreas as well as management of their disorders. More information [HERE](#).

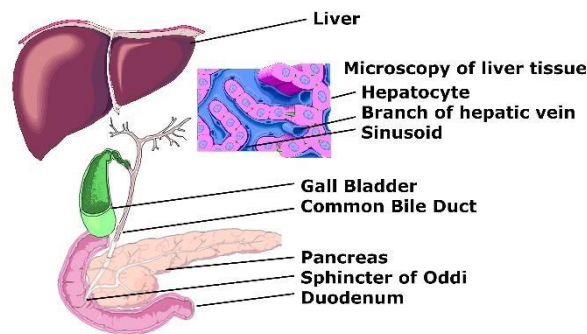


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Immunologists are specialists in immunology, a branch of biology that focuses on the study of immune system, also known as the body's defense system, in all organisms. It is essential to understand the immune system in health and diseases (e.g. autoimmune diseases, hypersensitivities, immune deficiency, and transplant rejection). Immunology has applications in many disciplines of medicine such as organ transplantation, oncology, rheumatology, virology, bacteriology, parasitology, psychiatry, and dermatology. More information [HERE](#).

Infectiologists are physicians practicing infectiology, a branch of medicine that covers the diagnosis and treatment of complex infections. They serve as consultants to other physicians in cases of complex infections and manage patients with immunodeficiency (e.g. HIV). More information [HERE](#).

Neonatologists are physicians practicing neonatology, a subspecialty of pediatrics that deals with newborns requiring special medical care. Examples of these special cases include

prematurity, low birth weight, intrauterine growth restriction, congenital malformations (birth defects), sepsis, pulmonary hypoplasia or birth asphyxia. More information [HERE](#).



Figure source [HERE](#):

Nephrologists are physicians practicing nephrology, a branch of medicine that focuses on the study of the kidneys in terms of normal function and disease, the health preservation, and the treatment, from diet and medication to renal replacement therapy (dialysis and kidney transplantation). It also studies systemic conditions affecting the kidneys (e.g. diabetes and autoimmune disease), and systemic diseases occurring because of kidney disease (e.g. renal osteodystrophy and hypertension). More information [HERE](#).

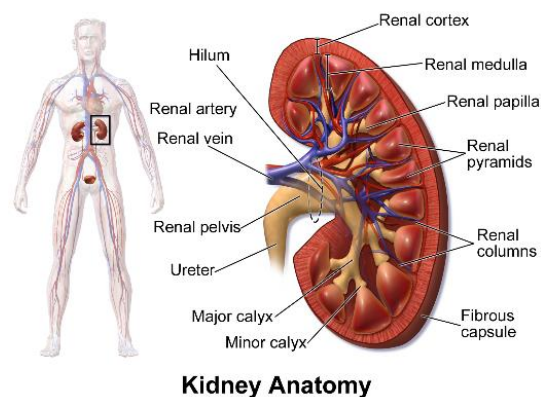


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Neurologists are physicians specializing in neurology and trained to investigate, or diagnose and treat disorders of the nervous system. More information [HERE](#).

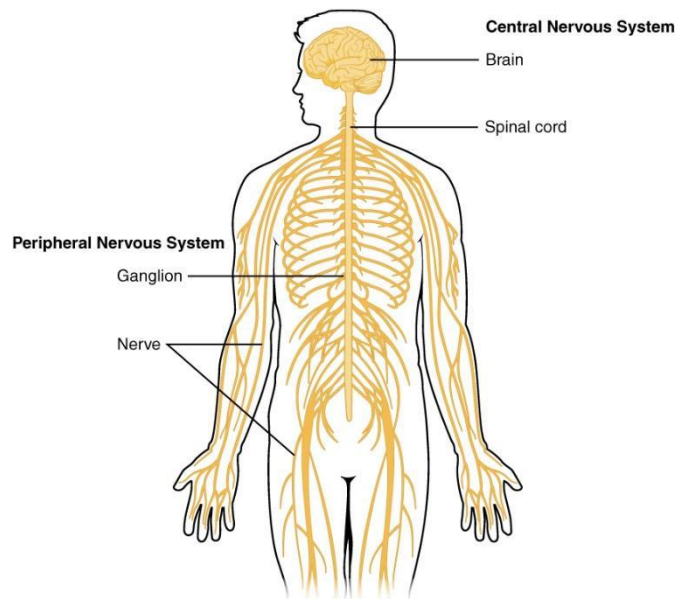


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Ophthalmologists are physicians practicing ophthalmology, a branch of medicine and surgery that diagnoses and treats of eye disorders. More information [HERE](#).



Figure source [HERE](#).

Pediatricians are physicians practicing pediatrics, which is the branch of medicine that deals with diseases in infants, children, and adolescents. More information [HERE](#).



Figure source [HERE](#).

Physiatrists are physicians practicing physiatry/physical medicine and rehabilitation, a branch of medicine that enhances and restores functional ability and quality of life to people with physical impairments or disabilities. More information [HERE](#).



Figure source [HERE](#).

Psychiatrists are physicians specialized in psychiatry, the branch of medicine focus on the diagnosis, prevention, study, and treatment of mental disorders. More information [HERE](#).



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Respiratory physicians/Pulmonologists are medical specialists that deal with diseases involving the respiratory tract (e.g. pneumonia, asthma, tuberculosis, emphysema, and complicated chest infections). More information [HERE](#).

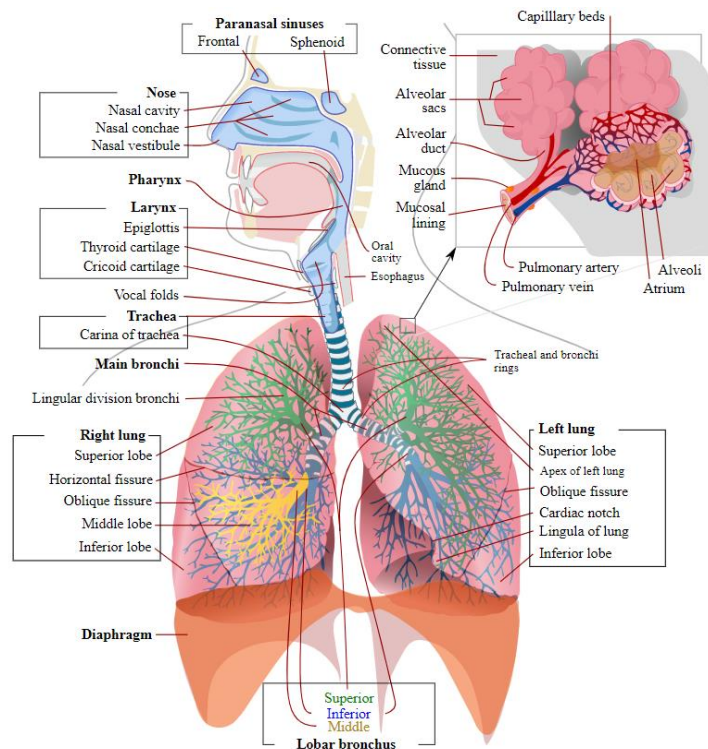


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Rheumatologists are physicians practicing rheumatology, a branch of medicine that deals with the diagnosis and therapy of rheumatic diseases. They treat immune-mediated disorders of the musculoskeletal system, soft tissues, autoimmune diseases, vasculitides, and inherited connective tissue disorders. More information [HERE](#).