

**Disease Overview**(<https://www.ncbi.nlm.nih.gov/medgen/?term=C0031117>)  
<https://www.disgenet.org/search/0/C0000786/>

**Name:**

**Count im Datensatz:**

**Beschreibung:**

**Ort wo es auftritt:**

**C0033578**

Name: Prostatic Neoplasms

Count im datensatz: 485

**Beschreibung:** A benign, borderline, or malignant neoplasm that affects the prostate gland. Representative examples include benign prostate phyllodes tumor, prostatic intraepithelial neoplasia, prostate carcinoma, and prostate sarcoma

→ Tumor or cancer of the Prostate

Ort wo es auftritt: PROSTATE

**C0017661**

**Name: IGA Glomerulonephritis**

**Count im Datensatz: 450**

**Beschreibung:**End-stage renal disease (ESRD) is a major public health problem, affecting 1 in 1,000 individuals and with an annual death rate of 20% despite dialysis treatment. IgA nephropathy (IgAN) is the most common form of glomerulonephritis, a principal cause of ESRD worldwide, affecting up to 1.3% of the population. Kidneys of patients with IgA nephropathy show deposits of IgA-containing immune complexes with proliferation of the glomerular mesangium. Typical clinical features include onset before age 40 with hematuria and proteinuria, and episodes of gross hematuria following mucosal infections are common; 30% of patients develop progressive renal failure. Although not generally considered a hereditary disease, striking ethnic variation in prevalence (Julian et al., 1985; D'Amico, 1987) and familial clustering (Scolari et al., 1999), along with subclinical renal abnormalities among relatives of cases, suggest a genetic component (Gharavi et al., 2000). Genetic Heterogeneity of IgA Nephropathy A locus for familial IgA nephropathy, called IGAN1, on chromosome 6q22-q23, was described by Gharavi et al. (2000). Another locus, IGAN2 (613944), was identified by Paterson et al. (2007) on chromosome 2q36. IGAN3 (616818) is caused by mutation in the SPRY2 gene (602466) on chromosome 13q31. Polymorphisms in the ACE (106180) and AGT (106150) genes have been associated with progression to chronic renal failure in patients with IgA nephropathy.

→ Berger disease

**Ort wo es auftritt: renal**

**C1458155**

**Name: Mammary Neoplasms/ Breast neoplasm**

**Count im Datensatz: 433**

**Beschreibung:** A tumor (abnormal growth of tissue) of the breast.

**Ort wo es auftritt:** breast

**C0028754**

**Name:** Obesity

**Count im Datensatz:** 298

**Beschreibung:** Accumulation of substantial excess body fat. Obesity is a complex disease involving an excessive amount of body fat. Obesity isn't just a cosmetic concern. It's a medical problem that increases the risk of other diseases and health problems, such as heart disease, diabetes, high blood pressure and certain cancers.(  
<https://www.mayoclinic.org/diseases-conditions/obesity/symptoms-causes/syc-20375742>)

**Ort wo es auftritt:** effect nearly every part of the body , like brain, blood vessels, heart, liver, bones etc(<https://www.healthline.com/health/obesity/how-obesity-affects-body>)

**C0031117**

**Name:** Peripheral Neuropathy/ Peripheral nerve damag

**Count im Datensatz:** 293

**Beschreibung:** Peripheral neuropathy is a general term for any disorder of the peripheral nervous system. The main clinical features used to classify peripheral neuropathy are distribution, type (mainly demyelinating versus mainly axonal), duration, and course.  
-> Peripheral neuropathy, a result of damage to the nerves located outside of the brain and spinal cord (peripheral nerves), often causes weakness, numbness and pain, usually in the hands and feet. It can also affect other areas and body functions including digestion, urination and circulation. Your peripheral nervous system sends information from your brain and spinal cord (central nervous system) to the rest of your body. The peripheral nerves also send sensory information to the central nervous system.  
Peripheral neuropathy can result from traumatic injuries, infections, metabolic problems, inherited causes and exposure to toxins. One of the most common causes is diabetes. People with peripheral neuropathy generally describe the pain as stabbing, burning or tingling. In many cases, symptoms improve, especially if caused by a treatable condition. Medications can reduce the pain of peripheral neuropathy.( <https://www.mayoclinic.org/diseases-conditions/peripheral-neuropathy/symptoms-causes/syc-20352061>)

**Ort wo es auftritt:** peripheral nervous system

**C0038356**

**Name:** Stomach Neoplasms/ Tumor of stomach

**Count im Datensatz:** 284

**Beschreibung:** A tumor (abnormal growth of tissue) of the stomach.

**Ort wo es auftritt:** stomach

**C0004352**

**Name:** Autistic Disorder

**Count im Datensatz:** 112

**Beschreibung:** Mental or Behavioral Dysfunction

- **Autism, the prototypic pervasive developmental disorder**

**Ort wo es auftritt: Brain and nervous system**

**C2239176**

**Name: Liver carcinoma/** Hepatocellular cancer (liver cancer)

**Count im Datensatz: 222**

**Beschreibung:** Hepatocellular carcinoma is the major histologic type of malignant primary liver neoplasm. It is the fifth most common cancer and the third most common cause of death from cancer worldwide. The major risk factors for HCC are chronic hepatitis B virus (HBV) infection, chronic hepatitis C virus (HCV) infection, prolonged dietary aflatoxin exposure, alcoholic cirrhosis, and cirrhosis due to other causes. Hepatoblastomas comprise 1 to 2% of all malignant neoplasms of childhood, most often occurring in children under 3 years of age. Hepatoblastomas are thought to be derived from undifferentiated hepatocytes (Taniguchi et al., 2002)

**Ort wo es auftritt: Liver**

**C0003873**

**Name: Rheumatoid Arthritis**

**Count im Datensatz: 219**

**Beschreibung:** - Rheumatoid arthritis is an inflammatory disease, primarily of the joints, with autoimmune features and a complex genetic component

Abnormality of limbs

- Abnormality of metabolism/homeostasis
- Abnormality of the cardiovascular system
- Abnormality of the immune system
- Abnormality of the musculoskeletal system
- Constitutional symptom
- Growth abnormality

**Ort wo es auftritt: They mainly affect the bones, joints and spine, as well as the associated muscles, tendons and ligaments. In some forms, organs such as the skin, heart, lungs, kidneys or the eyes, as well as the nervous system, may also be affected.**

**C0025202**

**Name: melanoma/** Malignant Melanoma/skin cancer

**Count im Datensatz: 204**

**Beschreibung:** The presence of a melanoma, a malignant cancer originating from pigment producing melanocytes. Melanoma can originate from the skin or the pigmented layers of the eye (the uvea)

**Ort wo es auftritt: skin**

**C0024121**

**Name: Lung Neoplasms/** Tumor of lung

**Count im Datensatz: 178**

**Beschreibung:** Tumor of the lung

**Ort wo es auftritt: Lung**

**C0027627**

**Name: Neoplasm Metastasis**

**Count im Datensatz: 174**

**Beschreibung:** The spread or migration of cancer cells from one part of the body (the organ in which it first appeared) to another. The secondary tumor contains cells that are like those in the original (primary) tumor.

**Ort wo es auftritt:** Brain, lymph nodes, bone, liver and lungs(<https://www.futuremedicine.com/doi/10.2217/fon-2020-1103>)

**C0011860**

**Name:** Diabetes Mellitus, Non-Insulin-Dependent/ Type 2 diabetes

**Count im Datensatz: 173**

**Beschreibung:** Type 2 diabetes is a disorder characterized by abnormally high blood sugar levels. In this form of diabetes, the body stops using and making insulin properly. Insulin is a hormone produced in the pancreas that helps regulate blood sugar levels. Specifically, insulin controls how much glucose (a type of sugar) is passed from the blood into cells, where it is used as an energy source. When blood sugar levels are high (such as after a meal), the pancreas releases insulin to move the excess glucose into cells, which reduces the amount of glucose in the blood.\n\nMost people who develop type 2 diabetes first have insulin resistance, a condition in which the body's cells use insulin less efficiently than normal.

**Ort wo es auftritt:** pancreas/bauchspeicheldrüse

**C0151744**

**Name:** Myocardial Ischemia/ Heart Disease,

**Count im Datensatz: 171**

**Beschreibung:** A disorder of cardiac function caused by insufficient blood flow to the muscle tissue of the heart. The decreased blood flow may be due to narrowing of the coronary arteries, to obstruction by a thrombus, or less commonly, to diffuse narrowing of arterioles and other small vessels within the heart. Severe interruption of the blood supply to the myocardial tissue may result in necrosis of cardiac muscle (myocardial infarction)

**Ort wo es auftritt:** heart

**C0002395**

**Name:** Alzheimer's Disease

**Count im Datensatz: 167**

**Beschreibung:** Alzheimer disease is a degenerative disease of the brain that causes dementia, which is a gradual loss of memory, judgment, and ability to function. This disorder usually appears in people older than age 65, but less common forms of the disease appear earlier in adulthood

**Ort wo es auftritt:** Brain

**C0014175**

**Name: Endometriosis**

**Count im Datensatz: 161**

**Beschreibung: The growth of endometrial tissue outside the uterus**

**Ort wo es auftritt:**

The most common sites of endometriosis include:

- The ovaries
- The fallopian tubes
- Ligaments that support the uterus (uterosacral ligaments)
- The posterior cul-de-sac, i.e., the space between the uterus and rectum
- The anterior cul-de-sac, i.e., the space between the uterus and bladder
- The outer surface of the uterus
- The lining of the pelvic cavity

Occasionally, endometrial tissue is found in other places, such as:

- The intestines
- The rectum
- The bladder
- The vagina
- The cervix
- The vulva
- Abdominal surgery scars

**C0024668**

**Name: Mammary Neoplasms, Experimental**

**Count im Datensatz: 152**

**Beschreibung: Experimentally induced mammary neoplasms in animals to provide a model for studying human BREAST NEOPLASMS**

**Ort wo es auftritt: breast**

**C0376634**

**Name: Craniofacial Abnormalities**

**Count im Datensatz: 149**

**Beschreibung: Congenital structural deformities, malformations, or other abnormalities of the cranium and facial bones**

**Ort wo es auftritt: cranium and facial bones**

**C0024667**

**Name: Animal Mammary Neoplasms/(breast cancer)animal**

**Count im Datensatz: 138**

**Beschreibung: Tumors or cancer of the MAMMARY GLAND in animals (MAMMARY GLANDS, ANIMAL)**

**Ort wo es auftritt: [Brustdrüse](#) /breast**

C0009375

**Name: Colonic Neoplasms**

**Count im Datensatz: 127**

**Beschreibung:** Tumor of colon/ A benign or malignant neoplasm that affects the colon. Representative examples of benign neoplasms include lipoma and leiomyoma. Representative examples of malignant neoplasms include carcinoma, lymphoma, and sarcoma. Colonic adenomas always exhibit epithelial dysplasia and are considered premalignant neoplasms.

**Ort wo es auftritt:**colon

C0007134

**Name: Renal Cell Carcinoma/**

**Count im Datensatz: 127**

**Beschreibung:** A type of carcinoma of the kidney with origin in the epithelium of the proximal convoluted renal tubule

**Ort wo es auftritt:** kidney cancer

C0242656

**Name: Disease Progression/** Course of illness

**Count im Datensatz: 127**

**Beschreibung:** A process that manifests as the worsening of a disease over time

**Ort wo es auftritt:** Every body

C0007131

**Name: Non-Small Cell Lung Carcinoma/** Non-small cell lung cancer

**Count im Datensatz: 117**

**Beschreibung:** Eine Gruppe von mindestens drei verschiedenen histologischen Typen von Lungenkrebs, darunter nicht-kleinzellige Plattenepithelkarzinome, Adenokarzinome und großzellige Karzinome. Nicht-kleinzellige Lungenkarzinome sprechen schlecht auf die herkömmliche Chemotherapie an.

**Ort wo es auftritt:**lung

C0023467

**Name: Leukemia, Myelocytic, Acute/** Leukemia

**Count im Datensatz: 115**

**Beschreibung:** Cytogenetically normal acute myeloid leukemia (CN-AML) is one form of a cancer of the blood-forming tissue (bone marrow) called acute myeloid leukemia. In normal bone marrow, early blood cells called hematopoietic stem cells develop into several types of blood cells: white blood cells (leukocytes) that protect the body from infection, red blood cells (erythrocytes) that carry oxygen, and platelets (thrombocytes) that are involved in blood clotting. In acute myeloid leukemia, the bone marrow makes large numbers of abnormal, immature white blood cells called myeloid blasts. Instead of developing into normal white blood cells, the myeloid blasts develop into cancerous

**leukemia cells. The large number of abnormal cells in the bone marrow interferes with the production of functional white blood cells, red blood cells, and platelets.**

**Ort wo es auftritt:** bone marrow/ liver, spleen, central nervous system (brain and spinal cord), and testicles.

**C0004096**

**Name:** Astma

**Count im Datensatz:** 113

**Beschreibung:** Asthma is characterized by increased responsiveness of the tracheobronchial tree to multiple stimuli, leading to narrowing of the air passages with resultant dyspnea, cough, and wheezing

**Ort wo es auftritt:** Lung

**C0005695**

**Name:** Bladder Neoplasm/ Tumors, Bladder

**Count im Datensatz:** 111

**Beschreibung:** The presence of a neoplasm of the urinary bladder.

**Ort wo es auftritt:** urinary bladder.

**C0023893**

**Name:** Liver Cirrhosis, Experimental /liver disease

**Count im Datensatz:** 109

**Beschreibung:** Cirrhosis of the liver is a serious liver disease. It develops gradually due to persistent stresses such as alcohol abuse or chronic liver inflammation (hepatitis).

**Ort wo es auftritt:** liver

**C0000786**

**Name:** Spontaneous abortion

**Count im Datensatz:** 108

**Beschreibung:** Female Urogenital Diseases and Pregnancy Complications

**Ort wo es auftritt:** Urogenital

**C0001418**

**Name:** Adenocarcinoma

**Count im Datensatz:** 108

**Beschreibung:** A common cancer characterized by the presence of malignant glandular cells. Morphologically, adenocarcinomas are classified according to the growth pattern (e.g., papillary, alveolar) or according to the secreting product (e.g., mucinous, serous).

**Representative examples of adenocarcinoma are ductal and lobular breast carcinoma, lung adenocarcinoma, renal cell carcinoma, hepatocellular carcinoma (hepatoma), colon adenocarcinoma, and prostate adenocarcinoma.**

**Ort wo es auftritt:** that can occur in several parts of the body.

->breast, colon, lung or prostate

**C0007621**

**Name: Neoplastic Cell Transformation/ Tumorigenesis**

**Count im Datensatz: 106**

**Beschreibung: A pathologic process that involves the transformation of normal cells to a neoplastic state and resulting in polyclonal or monoclonal neoplastic cell proliferation.**

**Ort wo es auftritt: several organ**

**C0011853**

**Name: Diabetes Mellitus, Experimental**

**Count im Datensatz: 106**

**Beschreibung: Nutritional and Metabolic Diseases; Endocrine System Diseases/ Experimental Model of Disease**

**Ort wo es auftritt: Pancrease**

**C0919267**

**Name: ovarian neoplasm/ Ovarian tumor**

**Count im Datensatz: 101**

**Beschreibung: Ovarian cancer, the leading cause of death from gynecologic malignancy, is characterized by advanced presentation with loco-regional dissemination in the peritoneal cavity and the rare incidence of visceral metastases (Chi et al., 2001). These typical features relate to the biology of the disease, which is a principal determinant of outcome (Auersperg et al., 2001). Epithelial ovarian cancer is the most common form and encompasses 5 major histologic subtypes: papillary serous, endometrioid, mucinous, clear cell, and transitional cell. Epithelial ovarian cancer arises as a result of genetic alterations sustained by the ovarian surface epithelium (Stany et al., 2008; Soslow, 2008).**

**Ort wo es auftritt: Ovarian**

**C0021390**

**Name: Inflammatory Bowel Diseases**

**Count im Datensatz: 100**

**Beschreibung: Inflammatory bowel disease is a systemic disorder comprised of two major disorders: ulcerative colitis and Crohn disease. Crohn disease can affect any part of the digestive system while ulcerative colitis is confined to the colon. Both disorders may affect sites outside of the digestive system [from SNOMEDCT\_US]**

**Ort wo es auftritt: bowel**



