CHAPTER 05

Endocrine, nutritional or metabolic diseases

This chapter has 148 four-character categories.

Code range starts with 5A00

This chapter includes endocrine diseases, nutritional diseases as well as metabolic diseases.

Exclusions: Transitory endocrine or metabolic disorders specific to fetus or newborn (BlockL1‑KB6)

Pregnancy, childbirth or the puerperium (Chapter 18)

Coded Elsewhere: Symptoms, signs or clinical findings of endocrine, nutritional or metabolic diseases (MA50-MA6Y)

Endocrine, nutritional or metabolic diseases complicating pregnancy, childbirth or the puerperium (JB64.2)

This chapter contains the following top level blocks:

* Endocrine diseases
* Nutritional disorders
* Metabolic disorders
* Postprocedural endocrine or metabolic disorders

Endocrine diseases (BlockL1‑5A0)

Coded Elsewhere: Neoplasms of the endocrine system

Endocrine tumours

Disorders of the thyroid gland or thyroid hormones system (BlockL2‑5A0)

Disorders due to dysfunction of thyroid gland and regulation systems of thyroid hormone actions including dysfunction of the pituitary, hypothalamus, or thyroid hormone receptors.

5A00 Hypothyroidism

5A00.0 Congenital hypothyroidism

Hypothyroidism is a condition where the thyroid gland produces too little or no thyroid hormone, and the condition arises at birth. Common clinical features include decreased activity and increased sleep, feeding difficulty and constipation, prolonged jaundice, myxedematous facies, large fontanels (especially posterior), macroglossia, a distended abdomen with umbilical hernia, and hypotonia.

Coded Elsewhere: Congenital central hypothyroidism (5A61.41)

5A00.00 Permanent congenital hypothyroidism with diffuse goitre

A condition caused by a partial or complete loss of thyroid function due to failure of the thyroid to correctly develop during the antenatal period. This condition is characterised by a swollen, smooth thyroid gland, and in infants by a dull look, puffy face, and thick tongue that sticks out. This condition may also present with choking episodes, constipation, dry brittle hair, jaundice, lack of muscle tone, low hairline, poor feeding, short height, sleepiness, or sluggishness.

Exclusions: transitory congenital goitre with normal function (KB62)

5A00.01 Permanent congenital hypothyroidism without goitre

This is a permanent congenital state in which the thyroid gland does not make enough thyroid hormone. This diagnosis is without swelling of the thyroid gland.

5A00.02 Pendred syndrome

Pendred syndrome is characterised by the association of congenital bilateral neurosensory deafness, thyroid goitre, cochleovestibular malformation and potential vestibular dysfunction.

5A00.03 Transient congenital hypothyroidism

Transient congenital hypothyroidism is defined as transient thyroid dysfunction with mildly elevated thyroid-stimulating hormone (TSH) and low thyroxine (FT4) levels which return to normal either very promptly and spontaneously, or after several months of thyroxine therapy. The disorder is due to a variety of causes including iodine deficiency or exposure to iodine-containing compounds, transplacental passage of blocking maternal antibodies, and dyshormonogenesis.

Exclusions: Transitory congenital goitre with normal function (KB62)

5A00.04 Congenital hypothyroidism due to iodine deficiency

Hypothyroidism is a condition which arises at birth where the thyroid gland produces too little or no thyroid hormone and it can be induced by iodine-deficiency.

Exclusions: Subclinical iodine-deficiency hypothyroidism (5A00.22)

5A00.0Y Other specified congenital hypothyroidism

5A00.0Z Congenital hypothyroidism, unspecified

5A00.1 Iodine-deficiency-related thyroid disorders or allied conditions

Any condition caused by aberrant thyroid function due to a deficiency of iodine. Confirmation is by blood test.

Exclusions: congenital iodine-deficiency syndrome (5A00.04)

Subclinical iodine-deficiency hypothyroidism (5A00.22)

5A00.10 Iodine-deficiency-related diffuse goitre

Diffuse enlargement of the thyroid gland due to Iodine-deficiency

5A00.11 Iodine-deficiency-related multinodular goitre

Multinodular enlargement of the thyroid gland due to Iodine-deficiency

Inclusions: Iodine-deficiency-related nodular goitre

5A00.1Z Iodine-deficiency-related thyroid disorders or allied conditions, unspecified

5A00.2 Acquired hypothyroidism

Acquired hypothyroidism is a condition where the thyroid gland produces too little or no thyroid hormone, and the condition arises only after birth.

Exclusions: Postprocedural hypothyroidism (5D40)

iodine-deficiency-related hypothyroidism (5A00.1)

Coded Elsewhere: Acquired central hypothyroidism (5A61.40)

Dementia due to acquired hypothyroidism (6D85.Y)

5A00.20 Hypothyroidism due to medicaments or other exogenous substances

A condition caused by an underactive thyroid due to a medicaments or other exogenous substances. This condition may present with fatigue, increased sensitivity to cold, constipation, dry skin, weight gain, muscle weakness, elevated blood cholesterol, muscle aches, joint pain or swelling, heavier or irregular menstrual periods, thinning hair, depression, or impaired memory.

5A00.21 Myxoedema coma

A life-threatening hypothyroid condition with long-standing severe untreated hypothyroidism in whom adaptive mechanisms fail to maintain homeostasis.

5A00.22 Subclinical iodine-deficiency hypothyroidism

A condition with elevated serum TSH level, but with normal thyroid hormone levels, which is induced by iodine-deficiency

5A00.2Y Other specified acquired hypothyroidism

5A00.2Z Acquired hypothyroidism, unspecified

5A00.Z Hypothyroidism, unspecified

5A01 Nontoxic goitre

Enlargement of the thyroid gland due to follicular multiplication, unaccompanied by hyperthyroidism or thyrotoxicosis

Exclusions: congenital goitre NOS (5A00.00)

congenital parenchymatous goitre (5A00.00)

iodine-deficiency-related goitre (5A00.1)

congenital diffuse goitre (5A00.00)

5A01.0 Nontoxic diffuse goitre

Diffuse enlargement of the thyroid gland due to follicular multiplication, unaccompanied by hyperthyroidism or thyrotoxicosis

5A01.1 Nontoxic single thyroid nodule

Single tumour of the thyroid gland due to follicular multiplication, unaccompanied by hyperthyroidism or thyrotoxicosis

5A01.2 Nontoxic multinodular goitre

Multiple nodules of the thyroid gland due to follicular multiplication, unaccompanied by hyperthyroidism or thyrotoxicosis

5A01.Z Nontoxic goitre, unspecified

5A02 Thyrotoxicosis

A hypermetabolic condition associated with elevated levels of free thyroxine and/or free triiodothyronine resulting in excess synthesis and secretion of thyroid hormone

Exclusions: neonatal thyrotoxicosis (KB62.0)

Coded Elsewhere: Dysthyroid exophthalmos (9A20.00)

5A02.0 Thyrotoxicosis with diffuse goitre

Thyrotoxicosis occurs by the ingestion of excessive amounts of exogenous thyroid hormone in the form of thyroid hormone supplements such as the most widely used supplement levothyroxine.

Inclusions: Toxic diffuse goitre

Graves disease

5A02.1 Thyrotoxicosis with toxic single thyroid nodule

Inclusions: Thyrotoxicosis with toxic uninodular goitre

5A02.2 Thyrotoxicosis with toxic multinodular goitre

Thyrotoxicosis caused by functioning thyroid multinodules

5A02.3 Thyrotoxicosis from ectopic thyroid tissue

5A02.4 Thyrotoxicosis factitia

A condition of thyrotoxicosis caused by the ingestion of exogenous thyroid hormone

5A02.5 Thyroid crisis

Thyrotoxic crisis (or thyroid storm) is a rare but severe complication of hyperthyroidism, which may occur when a thyrotoxic patient becomes very sick or physically stressed.

Inclusions: Thyroid storm

5A02.6 Secondary hyperthyroidism

Overproduction of thyroid hormone in the thyroid gland induced by dysfunction of the pituitary gland or hypothalamus.

Coding Note: Code aslo the casusing condition

5A02.Y Other specified thyrotoxicosis

5A02.Z Thyrotoxicosis, unspecified

5A03 Thyroiditis

Thyroiditis is the inflammation of the thyroid gland. It includes acute and chronic forms of thyroiditis. Thyroiditis is usually caused by autoimmune reaction to the thyroid, resulting in inflammation and damage to the thyroid cells. The symptoms include fatigue, weight gain, depression, dry skin, and constipation.

Exclusions: Acquired hypothyroidism (5A00.2)

Thyrotoxicosis (5A02)

Coded Elsewhere: Postpartum thyroiditis (JB44.5)

5A03.0 Acute thyroiditis

Acute thyroiditis is a rare form of thyroiditis directly caused by an infection, frequently bacterial.

5A03.1 Subacute thyroiditis

A self-limited thyroiditis associated with a triphasic clinical course of hyperthyroidism, hypothyroidism, and return to normal thyroid function. It is thought to be caused by a viral infection.

Inclusions: de Quervain thyroiditis

giant-cell thyroiditis

granulomatous thyroiditis

Exclusions: Autoimmune thyroiditis (5A03.2)

5A03.2 Autoimmune thyroiditis

A chronic inflammatory disorder of the thyroid gland associated with abnormal circulatory antibodies.

5A03.20 Hashimoto thyroiditis

5A03.21 Painless thyroiditis

A destructive thyroiditis which has an autoimmune basis in the non-postpartum period. An inflammation of the thyroid gland characterised by transient hyperthyroidism, followed by hypothyroidism and then recovery.

5A03.2Y Other specified autoimmune thyroiditis

5A03.Y Other specified thyroiditis

5A03.Z Thyroiditis, unspecified

5A04 Hypersecretion of calcitonin

This is the process of elaborating, releasing, and oozing a 32-amino acid linear polypeptide hormone that is produced in humans primarily by the parafollicular cells (also known as C-cells) of the thyroid, and in many other animals in the ultimobranchial body.

Inclusions: Hypersecretion of thyrocalcitonin

C-cell hyperplasia of thyroid

5A05 Generalised resistance to thyroid hormone

Decreased thyroid hormone action, generally induced by mutation of thyroid hormone receptors.

5A06 Sick-euthyroid syndrome

5A0Y Other specified disorders of the thyroid gland or thyroid hormones system

5A0Z Disorders of the thyroid gland or thyroid hormones system, unspecified

Diabetes mellitus (BlockL2‑5A1)

A metabolic disorder with heterogenous aetiologies which is characterised by chronic hyperglycaemia and disturbances of carbohydrate, fat and protein metabolism resulting from defects in insulin secretion, insulin action, or both.

Coded Elsewhere: Diabetes mellitus in pregnancy (JA63)

Neonatal diabetes mellitus (KB60.2)

5A10 Type 1 diabetes mellitus

Diabetes mellitus type 1 (type 1 diabetes, T1DM, formerly insulin dependent or juvenile diabetes) is a form of diabetes mellitus that results from destruction of insulin-producing beta cells, mostly by autoimmune mechanisms. The subsequent lack of insulin leads to increased blood and urine glucose.

Exclusions: Type 2 diabetes mellitus (5A11)

Diabetes mellitus, other specified type (5A13)

Diabetes mellitus in pregnancy (JA63)

Coded Elsewhere: Pre-existing type 1 diabetes mellitus in pregnancy (JA63.0)

5A11 Type 2 diabetes mellitus

Diabetes mellitus type 2 (formerly noninsulin-dependent diabetes mellitus (NIDDM) or adult-onset diabetes) is a metabolic disorder that is characterised by high blood glucose in the context of insulin resistance and relative insulin deficiency.

Inclusions: non-insulin-dependent diabetes of the young

Exclusions: Diabetes mellitus in pregnancy (JA63)

Diabetes mellitus, other specified type (5A13)

Idiopathic Type 1 diabetes mellitus (5A10)

Coded Elsewhere: Pre-existing type 2 diabetes mellitus in pregnancy (JA63.1)

5A12 Malnutrition-related diabetes mellitus

5A13 Diabetes mellitus, other specified type

Diabetes mellitus which cannot be classified as either Type 1 or Type 2 diabetes mellitus.

Exclusions: Diabetes mellitus in pregnancy (JA63)

Type 2 diabetes mellitus (5A11)

Idiopathic Type 1 diabetes mellitus (5A10)

5A13.0 Diabetes mellitus due to genetic defects of beta cell function

Other specified diabetes mellitus due to genetic defects of beta cell function is a form of diabetes, which is associated with monogenetic defects in beta-cell function.

5A13.1 Diabetes mellitus due to genetic defects in insulin action

Other specified diabetes mellitus due to genetic defects in insulin action is a form of diabetes, which results from genetically determined abnormalities of insulin action. The metabolic abnormalities associated with mutations of the insulin receptor may range from hyperinsulinemia and modest hyperglycaemia to severe diabetes.

5A13.2 Diabetes mellitus due to diseases of the exocrine pancreas

Other specified diabetes mellitus due to diseases of the exocrine pancreas is a form of diabetes, which caused by any process that diffusely injures the pancreas. Acquired processes include pancreatitis, trauma, infection, pancreatectomy, and pancreatic carcinoma. With the exception of that caused by cancer, damage to the pancreas must be extensive for diabetes to occur.

5A13.3 Diabetes mellitus due to endocrinopathies

Other specified diabetes mellitus due to endocrinopathies is a form of diabetes caused by several hormones (e.g., growth hormone, cortisol, glucagon, epinephrine), which antagonize insulin action. Excess amounts of these hormones (e.g., acromegaly, Cushing’s syndrome, glucagonoma, pheochromocytoma, respectively) can cause diabetes. This generally occurs in individuals with preexisting defects in insulin secretion, and hyperglycaemia typically resolves when the hormone excess is resolved.

5A13.4 Diabetes mellitus due to drug or chemical

Other specified diabetes mellitus due to drug or chemical is a form of diabetes, which is caused by drug or chemical substance that impairs insulin secretion and insulin action.

5A13.5 Diabetes mellitus due to uncommon forms of immune-mediated diabetes

Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes is a form of diabetes, which is caused by two known conditions. The stiff-man syndrome is an autoimmune disorder of the central nervous system characterised by stiffness of the axial muscles with painful spasms. Patients usually have high titres of the GAD autoantibodies, and approximately one-third will develop diabetes.

5A13.6 Diabetes mellitus due to other genetic syndromes

Other specified diabetes mellitus due to other genetic syndromes is a form of diabetes, which is associated with genetic syndromes.

Coding Note: Use additional code, if desired, to identify any associated genetic syndrome

Coded Elsewhere: Wolfram syndrome (5A61.5)

Maternally inherited diabetes and deafness (LD2H.Y)

Thiamine-responsive megaloblastic anaemia syndrome (5C63.Y)

Woodhouse-Sakati syndrome (5A61.0)

Mitochondrial myopathy with diabetes mellitus (8C73.Y)

5A13.7 Diabetes mellitus due to clinically defined subtypes or syndromes

Diabetes mellitus that has clinically defined subtypes or associated syndromes

5A13.Y Diabetes mellitus due to other specified cause

5A14 Diabetes mellitus, type unspecified

Exclusions: Idiopathic Type 1 diabetes mellitus (5A10)

Type 2 diabetes mellitus (5A11)

Diabetes mellitus, other specified type (5A13)

Diabetes mellitus in pregnancy (JA63)

Acute complications of diabetes mellitus (BlockL3‑5A2)

5A20 Diabetic hyperosmolar hyperglycaemic state

Coding Note: Code aslo the casusing condition

5A20.0 Hyperosmolar hyperglycaemic state without coma

Coding Note: Code aslo the casusing condition

5A20.1 Hyperosmolar hyperglycaemic state with coma

Coding Note: Code aslo the casusing condition

5A20.Z Diabetic hyperosmolar hyperglycaemic state, unspecified

Coding Note: Code aslo the casusing condition

5A21 Hypoglycaemia in the context of diabetes mellitus

Coding Note: Code aslo the casusing condition

5A21.0 Hypoglycaemia in the context of diabetes mellitus without coma

Coding Note: Code aslo the casusing condition

5A21.1 Hypoglycaemia in the context of diabetes mellitus with coma

Coding Note: Code aslo the casusing condition

5A21.Z Hypoglycaemia in the context of diabetes, unspecified

Coding Note: Code aslo the casusing condition

5A22 Diabetic acidosis

Coding Note: Code aslo the casusing condition

5A22.0 Diabetic ketoacidosis without coma

Coding Note: Code aslo the casusing condition

5A22.1 Diabetic lactic acidosis

Coding Note: Code aslo the casusing condition

5A22.2 Diabetic metabolic acidosis

Coding Note: Always assign an additional code for diabetes mellitus

5A22.3 Diabetic ketoacidosis with coma

Coding Note: Code aslo the casusing condition

5A22.Y Other specified diabetic acidosis

Coding Note: Code aslo the casusing condition

5A22.Z Diabetic acidosis, unspecified

Coding Note: Code aslo the casusing condition

5A23 Diabetic coma

Coding Note: Code aslo the casusing condition

5A24 Uncontrolled or unstable diabetes mellitus

Brittle diabetes mellitus is a term used to describe particularly hard-to-control Type 1 or Type 2 diabetes mellitus. It results in frequent, extreme swings in blood glucose levels, causing hyperglycaemia that could lead to ketoacidosis or hypoglycaemia.

Coding Note: Code aslo the casusing condition

5A2Y Other specified acute complications of diabetes mellitus

Coding Note: Code aslo the casusing condition

Other disorders of glucose regulation or pancreatic internal secretion (BlockL2‑5A4)

Exclusions: Benign neoplasm of endocrine pancreas (2E92.9)

Multiple endocrine neoplasia type 1 (2F7A.0)

Malignant neoplasm of pancreas (2C10)

5A40 Intermediate hyperglycaemia

A metabolic disorder characterised by glucose levels too high to be considered normal, though not high enough to meet the criteria for diabetes.

Inclusions: prediabetes

Impaired glucose regulation

Exclusions: Diabetes mellitus, other specified type (5A13)

Idiopathic Type 1 diabetes mellitus (5A10)

Type 2 diabetes mellitus (5A11)

Diabetes mellitus, type unspecified (5A14)

Elevated blood glucose level (MA18.0)

5A40.0 Impaired fasting glucose

Impaired glucose tolerance is a metabolic disorder with FPG 110–125 mg/dl (6.1–6.9 mmol/l).

5A40.1 Impaired glucose tolerance

Impaired glucose tolerance (IGT) is a metabolic disorder, which is characterised by 2-h postload glucose 140–199 mg/dl (7.8–11.1 mmol/l).

5A40.Y Other specified intermediate hyperglycaemia

5A40.Z Intermediate hyperglycaemia, unspecified

5A41 Hypoglycaemia without associated diabetes

Exclusions: Hypoglycaemia in the context of diabetes mellitus (5A21)

Coded Elsewhere: Neonatal hypoglycaemia (KB60.4)

5A42 Increased secretion of glucagon

Exclusions: Multiple endocrine neoplasia type 1 (2F7A.0)

Coded Elsewhere: Glucagonoma (2C10.1)

5A43 Abnormal secretion of gastrin

Coded Elsewhere: Gastrinoma (2C10.1)

5A43.0 Drug-induced hypergastrinaemia

A form of hypergastrinaemia that can be induced by drugs.

5A43.1 Zollinger-Ellison syndrome

A syndrome characterised by the presence of a gastrin-secreting tumour, usually in the pancreas or duodenum, resulting in increased gastric acidity and formation of gastric ulcers. Signs and symptoms include abdominal pain and diarrhea. It may be sporadic or a manifestation of multiple endocrine neoplasia type 1.

Coded Elsewhere: Anastomotic ulcer due to Zollinger-Ellison syndrome (DA62.Y)

Gastric ulcer due to Zollinger-Ellison syndrome (DA60.Y)

Duodenal ulcer due to Zollinger-Ellison syndrome (DA63.Y)

5A43.Y Other specified abnormal secretion of gastrin

5A43.Z Abnormal secretion of gastrin, unspecified

5A44 Insulin-resistance syndromes

Coding Note: Code aslo the casusing condition

5A45 Persistent hyperinsulinaemic hypoglycaemia of infancy

Congenital isolated hyperinsulinism, or Persistent hyperinsulinaemic hypoglycaemia of infancy (PHHI) is defined by an inappropriate oversecretion of insulin by the endocrine pancreas that is responsible for profound hypoglycaemia, which requires aggressive medical and/or surgical treatment to prevent severe and irreversible brain damage. PHHI is a genetically heterogeneous disorder with two types of histological lesions: diffuse (DiPHHI) and focal (FoPHHI) which are clinically indistinguishable.

5A4Y Other specified disorders of glucose regulation and pancreatic internal secretion

5A4Z Disorders of glucose regulation and pancreatic internal secretion, unspecified

Disorders of the parathyroids or parathyroid hormone system (BlockL2‑5A5)

Disorders of the parathyroids and parathyroid hormone system generally refer to conditions with inappropriate secretion and/or actions of parathyroid hormone that cause dysregulation of calcium metabolism.

Exclusions: Hypocalcaemic vitamin D dependent rickets (5C63.20)

Hypovitaminosis D (5B57)

Hyperphosphataemic familial tumoural calcinosis (5C54.1)

Hypocalcaemic vitamin D resistant rickets (5C63.21)

5A50 Hypoparathyroidism

Hypoparathyroidism is a condition with insufficient biological actions of parathyroid hormone due to impaired secretion of parathyroid hormone or refractoriness of target tissues to parathyroid hormone.

Exclusions: Postprocedural hypoparathyroidism (5D42)

tetany NOS (MB47.D)

Coded Elsewhere: Transitory neonatal hypoparathyroidism (KB64)

5A50.0 Hypoparathyroidism due to impaired parathyroid hormone secretion

Hypoparathyroidism due to impaired PTH secretion is a condition with low circulating PTH level and hypocalcaemia caused by being unable to secrete PTH from parathyroids in response to hypocalcaemia with pathological or functional defects in parathyroids.

Coded Elsewhere: CATCH 22 phenotype (LD44.N0)

5A50.00 Idiopathic hypoparathyroidism

Exclusions: Autoimmune polyendocrinopathy type 1 (5B00)

5A50.01 Secondary hypoparathyroidism

Coding Note: Code aslo the casusing condition

5A50.02 Hypoparathyroidism due to destruction of the parathyroid glands

Dysfunction of parathyroid glands can be caused by several etiologies such as radiation, destruction of parathyroid glands by granulomatous disease or cancer infiltration, and deposition of iron or copper.

5A50.03 Autoimmune hypoparathyroidism

5A50.0Y Other specified hypoparathyroidism due to impaired parathyroid hormone secretion

5A50.0Z Hypoparathyroidism due to impaired parathyroid hormone secretion, unspecified

5A50.1 Pseudohypoparathyroidism

Pseudohypoparathyroidism is a condition with refractoriness to parathyroid hormone of its target tissues especially kidney that causes hypocalcaemia and hyperphosphataemia even in the presence of high circulating levels of biologically active parathyroid hormone.

5A50.Y Other specified hypoparathyroidism

5A50.Z Hypoparathyroidism, unspecified

5A51 Hyperparathyroidism

Hyperparathyroidism refers to overproduction of parathormone and s most frequently due to a tumour in one of the parathyroid glands. It may also occur in response to low calcium levels, as encountered in various situations such as vitamin D deficiency or chronic kidney disease.

Hyperparathyroidism results in weakening of the bones through loss of calcium.

Exclusions: Adult osteomalacia (FB83.2)

infantile and juvenile osteomalacia (5B57.0)

5A51.0 Primary hyperparathyroidism

Primary hyperparathyroidism is a condition with enhanced PTH secretion and high circulatory PTH level caused by abnormal parathyroid pathology such as adenoma, hyperplasia and cancer. Primary hyperparathyroidism usually causes hypercalcaemia by enhanced PTH actions.

5A51.1 Secondary hyperparathyroidism

Secondary hyperparathyroidism is a condition with enhanced PTH secretion and high circulatory PTH level caused by metabolic changes such and hypocalcaemia, hyperphosphatemia and low 1,25-dihydroxyvitamin D.

Coding Note: Code aslo the casusing condition

Exclusions: secondary hyperparathyroidism of renal origin (GB90.4)

5A51.2 Familial hypocalciuric hypercalcaemia

Familial Hypocalciuric Hypercalcaemia (FHH) or benign familial hypercalcaemia is an autosomal dominant disorder of calcium metabolism that is often asymptomatic and that is biologically characterised by a significant but moderate hypercalcaemia. Serum levels of parathyroid hormone are normal or slightly increased, and urinary calcium excretion is relatively low for hypercalcaemia. CASR, GNA11 and AP2S1 have been identified as causative genes.

5A51.Y Other specified hyperparathyroidism

5A51.Z Hyperparathyroidism, unspecified

5A5Y Other specified disorders of the parathyroids or parathyroid hormone system

5A5Z Disorders of the parathyroids or parathyroid hormone system, unspecified

Disorders of the pituitary hormone system (BlockL2‑5A6)

Clinical status with increased, decreased, or dysregulated secretion of pituitary hormones, which is caused by a variety of tumourous, non-tumourous, and genetic disorders.

5A60 Hyperfunction of pituitary gland

A disease characterised by hypersecretion of adenohypophyseal hormones such as growth hormone, pralactin, thyrotopin, luteinising hormone, follicle stimulating hormone or adrenocorticotropic hormone.

Clinical status with excessive production of one or more pituitary hormones, which is mostly caused by hormone-producing pituitary adenomas.

Exclusions: Nelson syndrome (5A70.3)

overproduction of pituitary ACTH (5A70.0)

overproduction of thyroid-stimulating hormone (5A02)

Cushing syndrome (5A70)

Multiple endocrine neoplasia type 1 (2F7A.0)

Multiple endocrine neoplasia type 4 (2F7A.0)

5A60.0 Acromegaly or pituitary gigantism

Acromegaly is an acquired disorder related to excessive production of growth hormone (GH) and characterised by progressive somatic disfigurement (mainly involving the face and extremities) and systemic manifestations. The main clinical features are broadened extremities (hands and feet), widened thickened and stubby fingers, and thickened soft tissue. The disease also has rheumatologic, cardiovascular, respiratory and metabolic consequences which determine its prognosis. In the majority of cases, acromegaly is related to a pituitary adenoma, either purely GH-secreting (60%) or mixed. Transsphenoidal surgery is often the first-line treatment. When surgery fails to correct GH/IGF-I hypersecretion, medical treatment with somatostatin analogs and/or radiotherapy can be used.

Inclusions: Overproduction of growth hormone

Exclusions: constitutional gigantism (5B12)

increased secretion from endocrine pancreas of growth hormone-releasing hormone (BlockL2‑5A4)

Constitutional tall stature (5B12)

5A60.1 Hyperprolactinaemia

Increased peripheral blood levels of prolactin often associated with galactorrhea, sometimes associated with normal ovarian function, but often resulting in a spectrum of ovulatory dysfunction varying between short luteal phase (inadequate preovulatory follicular development), anovulatory cycles, amenorrhea and hypogonadotropic hypogonadism

Coded Elsewhere: Prolactinoma of pituitary gland (2F37.Y)

5A60.2 Syndrome of inappropriate secretion of antidiuretic hormone

Syndrome of inappropriate antidiuretic hormone (ADH) secretion (SIADH) is characterised by continued ADH secretion, leading to hyponatremia, hypoosmolality and natriuresis. Exact prevalence is unknown. The disease has been described in all age groups. SIADH is often associated with tumours, pulmonary disease, central nervous system disorders or exposure to drugs. Occasionally, it is found in patients with adrenal, thyroid or pituitary insufficiency. The disorder is caused by gain-of-function mutations in the gene encoding the vasopressin V2 receptor. Fluid restriction is the most common treatment. The outcome is related to the underlying and associated disorders.

5A60.20 Nephrogenic syndrome of inappropriate antidiuresis

5A60.2Y Other specified syndrome of inappropriate secretion of antidiuretic hormone

5A60.2Z Syndrome of inappropriate secretion of antidiuretic hormone, unspecified

5A60.3 Central precocious puberty

Central precocious puberty is defined as the onset of pubertal changes before 8 years of age in girls and before 9.5 years of age in boys due to the overproduction of gonadotropin-releasing hormone (GnRH) by the hypothalamus. It may be idiopathic with no apparent cause (90% of cases in girls, 50% of cases in boys) or secondary to a lesion (tumour or malformation) in the hypothalamus. Other causes may include traumatic brain injury, or genetic disorders, affecting behavioural and psychological development, and final body height.

5A60.Y Other specified hyperfunction of pituitary gland

5A60.Z Hyperfunction of pituitary gland, unspecified

5A61 Hypofunction or certain specified disorders of pituitary gland

Clinical status with disordered function of the pituitary gland without excessive pituitary hormone production, which is caused by a variety of diseases

Exclusions: Postprocedural hypopituitarism (5D43)

Craniopharyngioma (2A00)

Coded Elsewhere: Non-secreting pituitary adenoma (2F37.0)

5A61.0 Hypopituitarism

A disorder manifesting a deficiency or decrease of one or more pituitary hormones, which is caused by a variety of diseases such as tumour, trauma/surgery, irradiation, inflammation and haemorrhage/infarction.

Inclusions: pituitary cachexia

pituitary short stature

Coded Elsewhere: Prader-Willi syndrome (LD90.3)

Argonz-del Castillo Syndrome (5A60.1)

5A61.1 Adrenocorticotropic hormone deficiency

Deficiency of adrenocorticotropic hormone (ACTH) resulting in functional hypocortisolism. Includes deficiency of corticotropin releasing hormone (CRH, CRF).

5A61.2 Gonadotropin deficiency

Deficiency of Luteinizing Hormone (LH), Follicle Stimulating Hormone (FSH) resulting in hypogonadism (male and female). Includes deficiency of Gonadotropin Releasing Hormone (GnRH, LHRH).

Exclusions: Ovarian dysfunction (5A80)

Testicular hypofunction (5A81.1)

5A61.3 Growth hormone deficiency

Deficiency of growth hormone in children, adolescents and adults. Includes deficiency of growth hormone releasing hormone (GHRH) and excess of central somatostatin, leading to growth hormone deficiency. Includes idiopathic, inborn and acquired forms of growth hormone deficiency.

Exclusions: Hypopituitarism (5A61.0)

5A61.4 Thyroid stimulating hormone deficiency

Deficiency of thyroid stimulating hormone (TSH), leading to secondary (pituitary) or tertiary (hypothalamic) hypothyroidism Includes deficiency of TSH releasing hormone (TRH).

5A61.40 Acquired central hypothyroidism

Central Hypothyroidism is a condition where the thyroid gland produces too little or no thyroid hormone, induced by dysfunction of either hypothalamus or pituitary.

5A61.41 Congenital central hypothyroidism

5A61.4Y Other specified thyroid stimulating hormone deficiency

5A61.4Z Thyroid stimulating hormone deficiency, unspecified

5A61.5 Central diabetes insipidus

Central diabetes insipidus (CDI) is a hypothalamus-pituitary disease characterised by polyuria and polydipsia due to a vasopressin (AVP) deficiency. The condition may be associated with deficient secretion of antidiuretic hormone (ADH) and is most frequently idiopathic (possibly due to autoimmune injury to the ADH-producing cells), or may be induced by trauma, pituitary surgery, or hypoxic or ischaemic encephalopathy.

Inclusions: ADH - [antidiuretic hormone secretion] deficiency

Exclusions: Nephrogenic diabetes insipidus (GB90.4A)

5A61.6 Oxytocin deficiency

Isolated oxytocin deficiency or oxytocin deficiency in combination with anterior and/or posterior pituitary deficiencies.

5A61.Y Other specified hypofunction or disorders of pituitary gland

5A6Z Disorders of the pituitary hormone system, unspecified

Disorders of the adrenal glands or adrenal hormone system (BlockL2‑5A7)

Coded Elsewhere: Gonadotropin deficiency (5A61.2)

Growth hormone deficiency (5A61.3)

Thyroid stimulating hormone deficiency (5A61.4)

Oxytocin deficiency (5A61.6)

Adrenal incidentaloma (2F37.Y)

5A70 Cushing syndrome

Cushing syndrome results from excess of corticosteroid hormones in the body due to overstimulation of the adrenal glands by excessive amounts of the hormone ACTH, secreted either by a tumuor of the pituitary gland (Cushing's disease) or by a malignant tumour in the lung or elsewhere. Symptoms include weight gain, reddening of the face and neck, excess growth of body and facial hair, raised blood pressure, loss of mineral from the bones (osteoporosis), raised blood glucose levels, and sometimes mental disturbances.

5A70.0 Pituitary-dependent Cushing disease

Pituitary-dependent Cushing disease is caused by a pituitary tumour, generally benign (adenoma) but rarely malignant (carcinoma), which secretes adrenocorticotropin (ACTH) autonomously, leading to hypercortisolism. The condition is associated with increased morbidity and mortality that can be mitigated by treatments that result in sustained endocrine remission. Transsphenoidal pituitary surgery (TSS) remains the mainstay of treatment for this disease but requires considerable neurosurgical expertise and experience in order to optimize patient outcomes.

5A70.1 Ectopic ACTH syndrome

5A70.2 Pseudo-Cushing syndrome

This is a condition in which patients display the signs, symptoms, and abnormal hormone levels seen in Cushing's syndrome. However, pseudo-Cushing's syndrome is not caused by a problem with the hypothalamic-pituitary-adrenal axis as Cushing's is; it is an idiopathic condition.

5A70.3 Nelson syndrome

5A70.Y Other specified Cushing syndrome

5A70.Z Cushing syndrome, unspecified

5A71 Adrenogenital disorders

Disorders of the reproductive system resulting from pathologic androgen production secondary to abnormalities in cortisol and/or aldosterone production

5A71.0 46,XX disorders of sex development induced by androgens of fetal origin

This refers to 46,XX disorders of sex development induced by any natural or synthetic compound, usually a steroid hormone, that stimulates or controls the development and maintenance of male characteristics in vertebrates by binding to androgen receptors, of fetal origin.

5A71.00 Glucocorticoid resistance

Glucocorticoid resistance is a rare genetic endocrine condition characterised by generalised, partial, target tissue resistance to glucocorticoids. The clinical spectrum of the condition is broad, ranging from asymptomatic to severe cases of hyperandrogenism, fatigue and/or mineralocorticoid excess.

5A71.01 Congenital adrenal hyperplasia

Congenital adrenal hyperplasia (CAH) refers to a group of conditions associated with either complete (classical form) or partial (non-classical) anomalies in the biosynthesis of adrenal hormones. The condition is characterised by insufficient production of cortisol, or of aldosterone (classical form with salt wasting), associated with overproduction of adrenal androgens. In the classical form, metabolic decompensation (dehydration with hyponatraemia, hyperkalaemia and acidosis associated with mineralocorticoid deficiency, and hypoglycaemia associated with glucocorticoid deficiency) may be life-threatening from the neonatal period onwards. Genital variations may be noted at birth in affected females. Chronic hyperandrogenism may lead to accelerated growth during childhood, but advanced bone maturation may lead to a deficit in final height. Adults tend to be overweight and metabolic disturbances, bone anomalies and fertility problems may also be present. Non-classical forms are associated with later onset, during the peri- or postpubertal period, and manifest with signs of hyperandrogenism (acne, hirsutism, menstrual problems and infertility).

5A71.0Y Other specified 46,XX disorders of sex development induced by androgens of fetal origin

5A71.0Z 46,XX disorders of sex development induced by androgens of fetal origin, unspecified

5A71.1 46,XX disorders of sex development induced by androgens of maternal origin

This refers to 46,XX disorders of sex development induced by any natural or synthetic compound, usually a steroid hormone, that stimulates or controls the development and maintenance of male characteristics in vertebrates by binding to androgen receptors, of maternal origin.

5A71.Y Other specified adrenogenital disorders

5A71.Z Adrenogenital disorders, unspecified

5A72 Hyperaldosteronism

5A72.0 Primary hyperaldosteronism

5A72.1 Secondary hyperaldosteronism

Coding Note: Code aslo the casusing condition

5A72.Z Hyperaldosteronism, unspecified

5A73 Hypoaldosteronism

Exclusions: Congenital adrenal hyperplasia (5A71.01)

5A74 Adrenocortical insufficiency

A condition in which the adrenal glands do not produce adequate amounts of steroid hormones, primarily cortisol. It may also include impaired production of aldosterone (a mineralocorticoid), which regulates sodium conservation, potassium secretion, and water retention and also accompanies impaired production of adrenal androgens.

Coded Elsewhere: X-linked adrenoleukodystrophy (5C57.1)

5A74.0 Acquired adrenocortical insufficiency

This is a acquired condition in which the adrenal glands do not produce adequate amounts of steroid hormones, primarily cortisol; but may also include impaired production of aldosterone (a mineralocorticoid), which regulates sodium conservation, potassium secretion, and water retention.

Exclusions: Amyloidosis (5D00)

Coded Elsewhere: Adrenocorticotropic hormone deficiency (5A61.1)

5A74.1 Adrenal crisis

Adrenal crisis is a life-threatening condition that indicates severe adrenal insufficiency caused by insufficient levels of cortisol.

Coded Elsewhere: Waterhouse-Friderichsen syndrome (1C1C.1)

5A74.Y Other specified adrenocortical insufficiency

5A74.Z Adrenocortical insufficiency, unspecified

5A75 Adrenomedullary hyperfunction

Idiopathic overstimulation of the adrenal medulla resulting in pathologic epinephrine/norepinephrine-mediated sympathetic output

5A76 Certain specified disorders of adrenal gland

5A76.0 Premature adrenarche

Premature development of pubic and/or axillary hair without central or peripheral precocious puberty. Children show premature clinical and/or laboratory signs of androgen action without estrogen action.

Exclusions: Central precocious puberty (5A60.3)

Congenital adrenal hyperplasia (5A71.01)

Peripheral precocious puberty (5A92)

5A76.Y Other specified disorders of adrenal gland

5A7Z Disorders of the adrenal glands or adrenal hormone system, unspecified

Disorders of the gonadal hormone system (BlockL2‑5A8)

Gonad has a capability to produce androgen and estrogen under the control by hypothalamic- pituitary- gonadal axis. Gonadal dysfunction is caused by either insufficient actions of gonadotropin or resistance to gonadotropin.

5A80 Ovarian dysfunction

Pathological processes of the OVARY

Exclusions: isolated gonadotropin deficiency (5A61.0)

Postprocedural ovarian failure (5D44)

Coded Elsewhere: Premature ovarian failure (GA30.6)

Hirsutism associated with hyperandrogenaemia (ED72.1)

Ovarian hyperstimulation syndrome (GA32.0)

HAIR-AN syndrome (5A44)

5A80.0 Clinical hyperandrogenism

Presence of hirsutism, acne or androgenic alopecia (scalp hair loss in women)

5A80.1 Polycystic ovary syndrome

Condition defined by the presence of at least 2 of the following 3 criteria: oligo/anovulation; clinical or biochemical signs of hyperandrogenism; presence of polycystic ovaries as identified by ultrasound.

Inclusions: Sclerocystic ovary syndrome

Exclusions: Polycystic ovary NOS (5A80.2)

5A80.2 Polycystic ovary

Ovary with increased size (> 7 mL) and stromal volume, and with increased number of follicles (12 or more measuring 2-0 mm in diameter), that may be present in women with PCOS, but also in women with normal ovulatory function and normal fertility (unilaterally or bilaterally).

Exclusions: Polycystic ovary syndrome (5A80.1)

5A80.3 Anovulation

lack of ovulation in the last 12 months, leading to amenorrhea, irregular or infrequent cycles

5A80.4 Oligo-ovulation

Oligo-ovulation (less than 4 ovulations in the last 12 months) not related to described categories of endocrine dysfunction. Excludes anovulation related to PCOS, hyperprolactinaemia or amenorrhea.

5A80.5 Diminished ovarian reserve

Condition characterised by ovaries with lower number of oocytes than expected for female chronologic age, marked by biochemical abnormalities (increased serum FSH levels, decreased serum AMH levels) and/or ultrasound findings (low antral follicle count) associated with ovarian ageing, reduced response to ovarian stimulation, and female infertility

5A80.Y Other specified ovarian dysfunction

5A80.Z Ovarian dysfunction, unspecified

5A81 Testicular dysfunction or testosterone-related disorders

Exclusions: isolated gonadotropin deficiency (5A61.0)

Klinefelter syndrome (LD50.3)

Postprocedural testicular hypofunction (5D45)

Azoospermia (GB04.0)

Oligospermia (GB04)

Coded Elsewhere: 46,XY gonadal dysgenesis (LD2A.1)

Testicular agenesis (LD2A.2)

46,XY disorder of sex development due to a defect in testosterone metabolism (LD2A.3)

46,XY disorder of sex development due to androgen resistance (LD2A.4)

46, XY disorders of sex development (LD2A.Y)

5A81.0 Testicular hyperfunction

A hypersecretion of testicular hormones.

Exclusions: McCune-Albright syndrome (FB80.0)

5A81.1 Testicular hypofunction

In pre-puberty, a disorder characterised by atrophied testes and sterility, abnormal height and absence of secondary sex characteristics. In post-puberty, a disorder characterised by depressed sexual function, loss of sex drive and sterility, muscle weakness and osteoporosis (due to loss of the androgen anabolic effect).

5A81.Y Other specified testicular dysfunction or testosterone-related disorders

5A81.Z Testicular dysfunction or testosterone-related disorders, unspecified

5A8Z Disorders of the gonadal hormone system, unspecified

Certain disorders of puberty (BlockL2‑5A9)

Exclusions: Central precocious puberty (5A60.3)

5A90 Disorder of puberty due to oestrogen resistance

5A91 Delayed puberty

This is when an organism has passed the usual age of onset of puberty with no physical or hormonal signs that it is beginning. Puberty may be delayed for several years and still occur normally, in which case it is considered constitutional delay, a variation of healthy physical development. Delay of puberty may also occur due to malnutrition, many forms of systemic disease, or to defects of the reproductive system (hypogonadism) or the body's responsiveness to sex hormones.

Inclusions: Delayed sexual development

Constitutional delay of puberty

5A92 Peripheral precocious puberty

Precocious puberty without activation of the GnRH-/gonadotropin axis. It includes gonadal tumours with sex hormone production and it may be part of McCune-Albright's syndrome.

Inclusions: Precocious menstruation

Exclusions: female heterosexual precocious pseudopuberty (5A71)

male isosexual precocious pseudopuberty (5A71)

Central precocious puberty (5A60.3)

Congenital adrenal hyperplasia (5A71.01)

Coded Elsewhere: Testotoxicosis (5A81.0)

McCune-Albright syndrome (FB80.0)

5A9Y Other disorders of puberty

5A9Z Disorders of puberty, unspecified

Polyglandular dysfunction (BlockL2‑5B0)

Exclusions: Ataxia-telangiectasia (4A01.31)

Pseudohypoparathyroidism (5A50.1)

dystrophia myotonica [Steinert] (8C71.0)

Coded Elsewhere: Multiple polyglandular tumours (2F7A.0)

5B00 Autoimmune polyendocrinopathy

This a subtype of autoimmune polyendocrine syndrome, in which multiple endocrine glands dysfunction as a result of autoimmunity. It is a genetic disorder attributed to a defect in the AIRE gene that normally confers immune tolerance. It is inherited in a recessive fashion.

Coded Elsewhere: X-linked immune dysregulation – polyendocrinopathy – enteropathy (4A01.21)

5B01 Polyglandular hyperfunction

5B0Y Other specified polyglandular dysfunction

5B0Z Polyglandular dysfunction, unspecified

Endocrine disorders, not elsewhere classified (BlockL2‑5B1)

Exclusions: Pseudohypoparathyroidism (5A50.1)

5B10 Carcinoid syndrome

5B11 Short stature, not elsewhere classified

Exclusions: Progeria (LD2B)

Silver-Russell syndrome (LD2F.1)

short-limbed stature with immunodeficiency (4A01.10)

short stature hypochondroplastic (LD24.01)

short stature achondroplastic (LD24.00)

renal short stature (BlockL2‑GB6)

pituitary related short stature (5A61.0)

Coded Elsewhere: Short stature due to growth hormone resistance (5A61.0)

5B12 Constitutional tall stature

Constitutional (familial) tall stature, a variant of the normal pattern of childhood growth and development, is defined as a condition in which the height of an individual is more than 2 SD above the corresponding mean height for a normal subject of the same age and gender. Distinguishing features are a family history of tall stature and lack of dimorphism or other clinical features suggesting pathologic causes of abnormally rapid growth.

Inclusions: Constitutional gigantism

5B1Y Other specified endocrine disorders, not elsewhere classified

5B3Y Other specified endocrine diseases

5B3Z Endocrine diseases, unspecified

Nutritional disorders (BlockL1‑5B5)

Nutritional disorders in all their forms result from imbalances (excess or deficiency) in energy and/or specific macro and micronutrients. They occur when the intake of essential macronutrients and micronutrients does not meet or exceeds the metabolic demands for those nutrients. Metabolic demands vary with age and other physiological conditions, they are also affected by environmental circumstances, including poor hygiene and sanitation, which lead to diarrhea and other infections.

Coded Elsewhere: Nutritional or toxic disorders of the nervous system (8D40-8D4Z)

Undernutrition (BlockL2‑5B5)

Undernutrition is a condition in which the body’s requirements are unmet due to underconsumption or to impaired absorption and use of nutrients. It can be produced by lack of access to food, or as a consequence of illness. Undernutrition commonly refers to a deficit in energy intake, but can also refer to deficiencies of specific nutrients, and can be either acute or chronic.

Inclusions: Malnutrition NOS

Exclusions: slim disease (1C62.3)

starvation (NF07.0)

Intestinal malabsorption (DA96.0)

Anorexia Nervosa (6B80)

Coded Elsewhere: Malnutrition in pregnancy (JA64)

Undernutrition-dehydration cataract (9B10.2Y)

5B50 Underweight in infants, children or adolescents

5B51 Wasting in infants, children or adolescents

5B52 Acute malnutrition in infants, children or adolescents

5B53 Stunting in infants, children or adolescents

5B54 Underweight in adults

Body mass index (BMI) <18.5 kg/m²

5B55 Vitamin A deficiency

Vitamin A deficiency (VAD) is defined as a state in which tissue concentrations of vitamin A are low enough to have adverse health consequences even if there is no evidence of clinical xerophthalmia. The term xerophthalmia encompasses the clinical spectrum of ocular manifestations of vitamin A deficiency, from milder stages of night blindness and Bitot’s spots, to potentially blinding stages of corneal xerosis, ulceration and necrosis (keratomalacia). In addition to the specific signs and symptoms of xerophthalmia and the risk of irreversible blindness, nonspecific symptoms include increased morbidity and mortality, poor reproductive health, increased risk of anaemia, and contributions to slowed growth and development.

Inclusions: Hypovitaminosis A

Coded Elsewhere: Acquired vitamin A deficiency anaemia (3A03.5)

5B55.0 Vitamin A deficiency with night blindness

Night blindness (poor adaptation to darkness) is generally the earliest manifestation of vitamin A deficiency. In mild cases, night blindness is apparent only after photic stress. Affected children no longer move around after dusk and prefer to sit in a secure corner, often unable to find their food or toys. Night blindness of recent onset in a preschool child is practically pathognomonic of vitamin A deficiency. All patients respond rapidly to therapy with vitamin A, usually within 48 hours.

5B55.1 Vitamin A deficiency with conjunctival xerosis

In conjunctival xerosis the epithelium of the conjunctiva is transformed from the normal columnar to the stratified squamous type, with a resultant loss of goblet cells, formation of a granular cell layer, and keratinization of the surface. Clinically, these changes are expressed as marked dryness or unwettability, the affected area appearing roughened, with fine droplets or bubbles on the surface, rather than smooth and glistening. Conjunctival xerosis first appears in the temporal quadrant, as an isolated oval or triangular patch adjacent to the limbus in the interpalpebral fissure. It is almost always present in both eyes.

5B55.2 Vitamin A deficiency with conjunctival xerosis or Bitot's spots

generalised conjunctival xerosis suggests advanced vitamin A deficiency. The entire conjunctiva appears dry, roughened, and corrugated, sometimes skin-like. In some individuals keratin and saprophytic bacilli accumulate on the xerotic surface, giving it a foamy or cheesy appearance. Such lesions are known as Bitot's spot. With treatment active conjunctival xerosis and Bitot's spot begin to resolve within 2-5 days. Most will disappear within 2 weeks, though a significant proportion of temporal lesions may persist, in shrunken form, for months.

5B55.3 Vitamin A deficiency with corneal xerosis

Clinically, the cornea develops classical xerosis, a hazy, lustreless, dry appearance, first apparent near the inferior limbus. Many children have characteristic superficial punctate lesions of the inferior-nasal aspects of their cornea that stain brightly with fluorescein. Early in the disease they are visible only through a slit-lamp examination. With more severe disease the punctate lesions become more numerous and spread upwards over the central cornea, and the corneal stroma becomes oedematous. Thick, keratinized plaques resembling Bitot's spot may form on the corneal surface. These are often densest in the interpalpebral zone. With treatment, these corneal plaques peel off, sometimes leaving superficial erosion which quickly heals. Corneal xerosis responds within 2-5 days to vitamin A therapy, the cornea regaining its normal appearance in 1-2 weeks.

5B55.4 Vitamin A deficiency with corneal ulceration or keratomalacia

Ulceration/keratomalacia indicates permanent destruction of part or all of the corneal stroma, resulting in permanent structural alteration. Ulcers are classically round to oval "punched-out" defects, as if a trephine or cork-borer had been applied to the eye. The surrounding cornea is generally xerotic but otherwise clear, and typically lacks the grey, infiltrated appearance of ulcers of bacterial origin. There may be more than one ulcer. Small ulcers are almost invariably confined to the periphery of the cornea, especially its inferior and nasal aspects. The ulceration may be shallow, but is commonly deep. Perforations become plugged with iris, thereby preserving the anterior chamber. In more advanced disease the necrotic stroma sloughs, leaving a large ulcer or descemetocele. As with smaller ulcers, this is usually peripheral and heals as a dense, white, adherent leukoma. With therapy, superficial ulcers often heal with surprisingly little scarring; deeper ulcers, especially perforations, form dense peripheral adherent leukomas.

5B55.5 Vitamin A deficiency with xerophthalmic scars of cornea or blindness

Xerophthalmia or "dry eye" remains the most important cause of childhood blindness in many developing countries. Healed sequelae of prior corneal disease related to vitamin A deficiency include opacities or scars of varying density (nebula, macula, leukoma), weakening and outpouching of the remaining corneal layers (staphyloma and descemetocele) and, where loss of intraocular contents has occurred, phthisis bulbi, a scarred shrunken globe. Such end-stage lesions are not specific for xerophthalmia and may arise from numerous other conditions, notably trauma and infection.

5B55.Y Vitamin A deficiency with other specified manifestations

5B55.Z Vitamin A deficiency, unspecified

5B56 Vitamin C deficiency

This condition groups several clinical consequences secondary to vitamin C deficiency with scurvy being the most severe presentation. The populations at risk of vitamin C deficiency are those for whom the fruit and vegetable supply is minimal. Epidemics of scurvy are associated with famine and war, when food supply is small and irregular. Children fed predominantly heat-treated (ultra-high-temperature or pasteurized) milk or unfortified formulas and not receiving fruits and fruit juices are at significant risk for symptomatic disease.

5B56.0 Scurvy

Scurvy is a disease caused by a lack of vitamin C (ascorbic acid) in the diet. Vitamin C plays a central role in collagen and ground-substance formation, metabolism of aromatic amino acids (phenylalanine, tyrosine), reduction of folic acid to folinic acid and a broad range of biochemical redox reactions. Clinical features include perifollicular haemorrhages, ecchymoses, swollen bleeding gums, stomatitis and epistaxis.

Coded Elsewhere: Scorbutic anaemia (3A03.2)

5B56.Y Other specified vitamin C deficiency

5B56.Z Vitamin C deficiency, unspecified

5B57 Vitamin D deficiency

Vitamin D is a fat-soluble vitamin contained naturally in very few foods, added to milk, available as a supplement, and produced endogenously with exposure to sunlight. Vitamin D deficiency can be caused by inadequate intake due to dietary factors (e.g., special diets (veganism), lactose intolerance or allergies) and/or limited exposure to sunlight due to geographic location, sun avoidance, or shiftwork. Severe deficiency results in disordered bone modelling called rickets in childhood (open growth plates), and osteomalacia in adults (fused growth plates).

5B57.0 Vitamin D deficiency rickets

Rickets is a disease of growing bone that is due to unmineralized matrix at the growth plates and occurs in children only before fusion of the epiphyses. There are many causes of rickets, including vitamin D disorders, calcium deficiency, phosphorous deficiency, and distal renal tubular acidosis. With the increased survival rate of very low birthweight infants, rickets in this age group has become a significant problem.

5B57.1 Vitamin D deficiency osteomalacia

Osteomalacia is a disorder of defective mineralization of newly formed osteoid at sites of bone turnover. Several different disorders cause osteomalacia via mechanisms that result in hypocalcaemia, hypophosphatemia, or direct inhibition of the mineralization process. Severe vitamin D deficiency, secondary to inadequate dietary intake, lack of sun exposure, gastric bypass or malabsorption (celiac disease), is the most common cause of osteomalacia in adults.

5B57.Y Other specified vitamin D deficiency

5B57.Z Vitamin D deficiency, unspecified

5B58 Vitamin E deficiency

Vitamin E deficiency is a condition that causes haemolysis and/or neurologic manifestations. Red blood cell fragility occurs and can produce a haemolytic anaemia. Neuronal degeneration produces peripheral neuropathies, ophthalmoplegia, and destruction of posterior columns of spinal cord. Neurologic disease is frequently irreversible if deficiency is not corrected early enough. Vitamin E deficiency may also contribute to the haemolytic anaemia and retrolental fibroplasia seen in premature infants.

Coded Elsewhere: Acquired vitamin E deficiency anaemia (3A03.6)

Dementia due to vitamin E deficiency (6D85.Y)

5B59 Vitamin K deficiency

Vitamin K is necessary for the synthesis of clotting factors II, VII, IX, and X, and deficiency of vitamin K can result in clinically significant bleeding. Vitamin K deficiency typically affects infants, who experience a transient deficiency related to inadequate intake, or patients of any age who have decreased vitamin K absorption. Mild vitamin K deficiency can affect long-term bone and vascular health.

Coded Elsewhere: Neonatal vitamin K deficiency (KA8F)

5B5A Vitamin B1 deficiency

Vitamin B1 deficiency manifests itself principally with changes involving the nervous system (polyneuritis and paralysis of the peripheral nerves), the cardiovascular system (cardiac insufficiency and generalised oedema), and also the gastrointestinal tract (constipation, vomiting, and abdominal pain).

5B5A.0 Beriberi

The clinical picture of Beriberi is usually divided into a dry (neuritic) type and a wet (cardiac) type. The disease is wet or dry depending on the amount of fluid which accumulates in the body due to factors like cardiac function, kidney lesions and others; even though the exact cause for this oedema has never been successfully explained. Many cases of thiamine deficiency show a mixture of the two main features and are more properly termed thiamine deficiency with cardiopathy and peripheral neuropathy. The infant shows signs of cyanosis and an acute cardiac attack can follow with the infant usually dying within 2 to 4 hours. The common age for this form of the deficiency disease is one month up through the third month. This type of deficiency responds very dramatically to thiamine.

5B5A.00 Dry beriberi

Neuritic form of Beri Beri

5B5A.01 Wet beriberi

Cardiac form of Beri Beri.

5B5A.0Z Beriberi, unspecified

5B5A.1 Wernicke-Korsakoff Syndrome

A thiamine-deficiency syndrome characterised by symmetric hyperaemic lesions of the brainstem, hypothalamus, thalamus, and mammillary bodies with glial proliferation, capillary dilatation, and perivascular haemorrhage. The syndrome is manifested by a confusional state, disorientation, ophthalmoplegia, nystagmus, diplopia, and ataxia (Wernicke encephalopathy), with severe loss of memory for recent events and confabulation (the invention of accounts of events to cover the loss of memory) (Korsakov psychosis) occurring following recovery. Defective binding of thiamine diphosphate by transketolase has been found. It appears that the disorder is of autosomal recessive inheritance but is expressed as clinical disease only in the event of thiamine deficiency.

Coding Note: Chronic alcohol use may be associated with thiamine deficiency, but alcohol may also have effects on the brain via other mechanisms. This category should be used to describe cognitive symptoms due to chronic alcohol use if there is evidence of thiamine deficiency.

Exclusions: Amnestic disorder due to use of alcohol (6D72.10)

5B5A.10 Wernicke encephalopathy

Wernicke's encephalopathy is an acute neuropsychiatric syndrome characterised by nystagmus, ophthalmoplegia, changes in the mental status, an uncoordinated gait and truncal ataxia. Wernicke's encephalopathy is usually accompanied or followed by Korsakoff's syndrome/Korsakoff's dementia (a continuum of Wernicke's encephalopathy characterised by severe memory defects, ataxia, apathy, disorientation, confabulations, hallucinations, paralysis of muscles controlling the eye and coma). The disorder results from a deficiency in vitamin B1, and mostly occurs in adults with a history of alcohol abuse or in patients with AIDS.

5B5A.11 Korsakoff syndrome

A disease of the nervous system, caused by deficiency of vitamin B1 in the brain. This disease commonly follows Wernicke encephalopathy, and may present with inability to form new memories, amnesia, confabulation, or hallucinations.

Coding Note: Chronic alcohol use may be associated with thiamine deficiency, but alcohol may also have effects on the brain via other mechanisms. This category should be used to describe cognitive symptoms due to chronic alcohol use if there is evidence of thiamine deficiency.

Exclusions: Amnestic disorder due to use of alcohol (6D72.10)

5B5A.1Y Other specified Wernicke-Korsakoff Syndrome

Coding Note: Chronic alcohol use may be associated with thiamine deficiency, but alcohol may also have effects on the brain via other mechanisms. This category should be used to describe cognitive symptoms due to chronic alcohol use if there is evidence of thiamine deficiency.

5B5A.1Z Wernicke-Korsakoff Syndrome, unspecified

Coding Note: Chronic alcohol use may be associated with thiamine deficiency, but alcohol may also have effects on the brain via other mechanisms. This category should be used to describe cognitive symptoms due to chronic alcohol use if there is evidence of thiamine deficiency.

5B5A.Y Other specified vitamin B1 deficiency

5B5A.Z Vitamin B1 deficiency, unspecified

5B5B Vitamin B2 deficiency

The signs of riboflavin deficiency are sore throat, hyperaemia, oedema of the pharyngeal and oral mucous membranes, cheilosis, angular stomatitis, glossitis, seborrheic dermatitis, and normochromic normocytic anaemia associated with pure red cell cytoplasia of the bone marrow. The major cause of hyporiboflavinosis is inadequate dietary intake as a result of limited food supply, which is sometimes exacerbated by poor food storage or processing. Children in developing countries will commonly demonstrate clinical signs of riboflavin deficiency during periods of the year when gastrointestinal infections are prevalent. Decreased assimilation of riboflavin also results from abnormal digestion, such as that which occurs with lactose intolerance.

Inclusions: Riboflavin deficiency

Coded Elsewhere: Acquired riboflavin deficiency anaemia (3A03.41)

5B5C Vitamin B3 deficiency

Niacin deficiency classically results in pellagra, which is a chronic wasting disease associated with a characteristic erythematous dermatitis that is bilateral and symmetrical, a dementia after mental changes including insomnia and apathy preceding an overt encephalopathy, and diarrhoea resulting from inflammation of the intestinal mucous surfaces. Pellagra occurs endemically in poorer areas of Africa, China and India.

Inclusions: Niacin deficiency NOS

5B5C.0 Pellagra

Pellagra is a potentially life-threatening disorder due to niacin deficiency and is observed in malnourished individuals, especially alcoholics, and as a complication of isoniazid therapy. The diagnosis is often overlooked or delayed. Pellagra manifests as diarrhoea, dermatitis, dementia, which usually appear in this order. Gastrointestinal tract symptoms always precede skin involvement, which presents initially with a sunburn-like blistering erythema, typically affecting the dorsal surfaces of the hands, face, neck, arms, and feet. With time the skin becomes thickened, scaly and pigmented.

5B5C.Y Other specified vitamin B3 deficiency

5B5D Vitamin B6 deficiency

A deficiency of vitamin B6 alone is uncommon because it usually occurs in association with a deficit in other B-complex vitamins. Hypovitaminosis B6 may often occur with riboflavin (vitamin B2) deficiency. The classical clinical symptoms of vitamin B6 deficiency are a seborrheic dermatitis, microcytic anaemia, epileptiform convulsions, and depression and confusion. Infants are especially susceptible to insufficient intakes, which can lead to epileptiform convulsions. Moreover, there is usually a decrease in circulating lymphocytes and sometimes a normocytic, microcytic, or sideroblastic anaemia as well. As is the case with other micronutrient deficiencies, vitamin B6 deficiency results in an impairment of the immune system. Several medical conditions can also affect vitamin B6 metabolism and thus lead to deficiency symptoms.

Exclusions: Pyridoxine-responsive sideroblastic anaemia, not elsewhere classified (3A72.1)

Coded Elsewhere: Acquired pyridoxine deficiency anaemia (3A03.40)

Pyridoxine dependent epilepsy with antiquitin mutations (8A61.0Y)

5B5E Folate deficiency

Nutritional deficiency of folate is common in people consuming a limited diet. This can be exacerbated by malabsorption conditions, including coeliac disease and tropical sprue. Pregnant women are at risk for folate deficiency because pregnancy significantly increases the folate requirement, especially during periods of rapid fetal growth (i.e. in the second and third trimester). During lactation, losses of folate in milk also increase the folate requirement. During pregnancy, there is an increased risk of fetal neural tube defects (NTDs), with risk increasing 10-fold as folate status goes from adequate to poor. Between days 21 and 27 post-conception, the neural plate closes to form what will eventually be the spinal cord and cranium. Spina bifida, anencephaly, and other similar conditions are collectively called NTDs. They result from improper closure of the spinal cord and cranium, respectively, and are the most common congenital abnormalities associated with folate deficiency.

5B5F Vitamin B12 deficiency

Vegetarianism and poverty-imposed near-vegetarianism are the most common causes of nutritional cobalamin insufficiency worldwide in all age groups. In such populations, low maternal cobalamin status is associated with adverse pregnancy outcomes (preterm birth, intrauterine growth retardation, early recurrent miscarriage), neural tube defects, reduced neurocognitive performance in children, accelerated bone turnover, and low bone mineral density with fractures. Insufficient cobalamin intake is also seen in breast-fed infants of mothers with pernicious anaemia.

Inclusions: cobalamin deficiency

cyanocobalamin deficiency

Coded Elsewhere: Vitamin B12 deficiency anaemia due to low intake (3A01.2)

Vitamin B12 deficiency anaemia due to intrinsic factor deficiency (3A01.3)

Vitamin B12 deficiency anaemia due to intestinal disease (3A01.4)

Drug-induced vitamin B12 deficiency anaemia (3A01.5)

Acquired vitamin B12 deficiency anaemia (3A01.Y)

Dementia due to vitamin B12 deficiency (6D85.Y)

5B5G Biotin deficiency

Isolated biotin deficiency is rare. Signs of biotin deficiency in humans have been demonstrated in individuals who consume raw egg white over long periods and in total parenteral nutrition (TPN) before biotin supplementation in patients with malabsorption. The clinical findings of biotin deficiency include dermatitis, conjunctivitis, alopecia, and central nervous system abnormalities. In adults fed raw egg white (which contains avidin, a protein that binds biotin with such high affinity that it renders it biounavailable) or receiving biotin-free TPN for months to years, thinning of hair, frequently with loss of hair colour, has been reported. Most adults with the deficiency demonstrate a red, scaly, skin rash, frequently around the eyes, nose, and mouth. Most of the adults have neurological symptoms, including depression, lethargy, hallucinations, and paraesthesia of the extremities.

5B5H Pantothenic acid deficiency

Pantothenic deficiency is rare: only reported as a result of feeding semisynthetic diets or an antagonist to the vitamin. Experimental, isolated deficiency in humans produces fatigue, abdominal pain, vomiting, insomnia, and paraesthesias of the extremities.

5B5J Choline deficiency

Choline deficiency is rare. Individuals fed with total parenteral nutrition (TPN) solutions lacking choline develop fatty liver and liver damage.

5B5K Mineral deficiencies

Exclusions: Disorders of mineral absorption or transport (5C64)

Coded Elsewhere: Hypokalaemia (5C77)

Hypomagnesaemia (5C64.41)

5B5K.0 Iron deficiency

Iron deficiency is a state in which there is insufficient iron to maintain the normal physiological function of blood, brain and muscles. It can exist in the absence of anaemia if it has not lasted long enough or if it has not been severe enough to cause the haemoglobin concentration to fall below the threshold for the specific sex and age group. Iron deficiency is the most common nutritional deficiency.

Exclusions: Iron deficiency anaemia (3A00)

Coded Elsewhere: Acquired iron deficiency anaemia due to blood loss (3A00.0)

Acquired iron deficiency anaemia due to low intake (3A00.1)

Acquired iron deficiency anaemia due to decreased absorption (3A00.2)

Acquired iron deficiency anaemia due to increased requirement (3A00.3)

Acquired iron deficiency anaemia (3A00.Y)

Dementia due to iron deficiency (6D85.Y)

5B5K.1 Calcium deficiency

Hypocalcaemia is defined as a total serum calcium concentration of less than 8.4 mg/dl (2.1 mmol/litre) or an ionized calcium concentration of less than 4.48 mg/dl (1.12 mmol/litre). There are numerous causes of hypocalcaemia, being chronic kidney disease the most common cause. Other causes are: vitamin D deficiency, disorders associated with acquired or genetic hypoparathyroridism, including intravenous bisphosphonate therapy, post-thyroidectomy and post-parathyroidectomy, and acute pancreatitis. Hypocalcaemia may be associated with a spectrum of clinical manifestations, ranging from few symptoms if the hypocalcaemia is mild, to life-threatening seizures, refractory heart failure, or laryngospasm if it is severe. In addition to severity, the rate of development of hypocalcaemia and chronicity determine the clinical manifestations.

Exclusions: Disorders of calcium metabolism (5C64.5)

Coded Elsewhere: Neonatal hypocalcaemia (KB61.2)

Neonatal osteopenia (KB61.3)

Myopathy due to calcium deficiency (8D40.2)

5B5K.10 Tetany due to acute calcium deficiency

The hallmark of acute hypocalcaemia is tetany, which is characterised by neuromuscular irritability. The symptoms of tetany may be mild (peri-oral numbness, paresthesias of the hands and feet, muscle cramps) or severe (carpopedal spasm, laryngospasm, and focal or generalised seizures, which must be distinguished from the generalised tonic muscle contractions that occur in severe tetany). Other patients have less specific symptoms such as fatigue, hyperirritability, anxiety, and depression, and some patients, even with severe hypocalcaemia, have no neuromuscular symptoms. Factors that determine the variation in frequency and severity of symptoms include acid-base status (hypocalcaemia and alkalosis act synergistically to cause tetany), hypomagnesaemia, and potassium balance.

5B5K.1Y Other specified calcium deficiency

5B5K.1Z Calcium deficiency, unspecified

5B5K.2 Zinc deficiency

The clinical features of severe zinc deficiency in humans are growth retardation, delayed sexual and bone maturation, skin lesions, diarrhoea, alopecia, impaired appetite, increased susceptibility to infections mediated via defects in the immune system, and the appearance of behavioural changes. The effects of marginal or mild zinc deficiency are less clear. A reduced growth rate and impairments of immune defence are so far the only clearly demonstrated signs of mild zinc deficiency in humans. Other effects, such as impaired taste and wound healing, which have been claimed to result from a low zinc intake, are less consistently observed.

Coded Elsewhere: Neonatal nutritional zinc deficiency (5C64.21)

5B5K.3 Iodine deficiency

Iodine deficiency disorders (IDD), caused mainly by a low dietary supply of iodine, refer to all of the consequences of iodine deficiency in a population that can be prevented by ensuring that the population has an adequate intake of iodine. Iodine deficiency is the most frequent cause of preventable brain damage in childhood.

Coded Elsewhere: Iodine-deficiency-related thyroid disorders or allied conditions (5A00.1)

Acquired hypothyroidism (5A00.2)

Congenital hypothyroidism due to iodine deficiency (5A00.04)

5B5K.4 Fluorine deficiency

A condition caused by a deficiency of fluoride. Low Fluorine concentrations in an individual’s dental plaque and enamel may result in an increased risk for dental caries at any age. Fluorine deficiencies might also show negative effects on human’s bone health.

5B5K.5 Sodium chloride deficiency

Sodium and chloride are usually found together in most foods as sodium chloride, also termed salt. For that reason, the effects of sodium and chloride deficiency are considered together. Deficiency can be caused by poor intake or increased losses (e.g., diuretics increase the urinary excretion of water, sodium, and chloride; in cystic fibrosis the sodium and chloride content of sweat is very high; gastrointestinal losses are associated with diarrhoeal diseases, emesis, ostomy output and other causes).

5B5K.6 Copper deficiency

Dietary deficiency is rare; it has been observed in premature and low birthweight infants fed exclusively a cow’s milk diet and in individuals on long-term total parenteral nutrition without copper. Clinical manifestations include depigmentation of skin and hair, neurologic disturbances, leukopenia, hypochromic microcytic anaemia, and skeletal abnormalities.

Coded Elsewhere: Copper deficiency anaemia (3A03.3)

5B5K.7 Selenium deficiency

Selenium deficiency is rare but has been observed in individuals on long-term total parenteral nutrition lacking selenium. Clinical manifestations of deficiency arising from such situations are uncommon and poorly defined. They include muscular weakness and myalgia with, in several instances, the development of congestive heart failure. The importance of selenium for thyroid hormone metabolism is evident from changes in the T3–T4 ratio which develop after relatively mild selenium depletion in infants and elderly subjects.

5B5K.8 Chromium deficiency

Deficiency in humans is only described in long-term total parenteral nutrition patients receiving insufficient chromium. Hyperglycaemia or impaired glucose tolerance occurs. Elevated plasma free fatty acid concentrations, neuropathy, encephalopathy, and abnormalities in nitrogen metabolism are also reported.

5B5K.9 Manganese deficiency

5B5K.A Molybdenum deficiency

Molybdenum functions as a cofactor for a limited number of enzymes in humans: sulphite oxidase, xanthine oxidase and aldehyde oxidase. A rare severe metabolic defect causing molybdenum cofactor deficiency and preventing these enzymes from being synthesized has been described. Few infants with such defects survive the first days of life, and those who survive have severe neurological abnormalities. Although molybdenum deficiency related to a dietary deficiency is extremely rare in humans, it has been described in long-term total parenteral nutrition as being secondary to the administration of sulphite. Symptoms include: tachycardia, headache, night blindness, irritability and coma. Biochemical changes can consist of elevated plasma and methionine concentration, low serum uric acid concentration, high urinary thiosulfate and low urinary uric acid and sulphate levels.

5B5K.B Vanadium deficiency

A biological role of vanadium in humans has not yet been identified.

5B5K.Y Other specified mineral deficiency

5B5K.Z Mineral deficiency, unspecified

Sequelae of malnutrition and certain specified nutritional deficiencies (BlockL3‑5B6)

This refers to sequelae of malnutrition and certain specified nutritional deficiencies.

5B60 Sequelae of protein-energy malnutrition

This refers to a pathological condition resulting from protein-energy malnutrition.

5B61 Sequelae of vitamin A deficiency

This refers to a pathological condition resulting from vitamin A deficiency.

5B62 Sequelae of vitamin C deficiency

This refers to a pathological condition resulting from vitamin C deficiency.

5B63 Sequelae of rickets

Bowed legs and/or arms, knock-knees, deformities of the thoracic cage and/or spine and/or skeletal dysplasia secondary to chronic or advanced rickets

5B6Y Other specified sequelae of malnutrition and certain specified nutritional deficiencies

5B6Z Sequelae of malnutrition and certain specified nutritional deficiencies, unspecified

5B70 Essential fatty acid deficiency

Deficiency of EFA (linoleic acid, linolenic acid, arachidonic acid, docosapentaenoic acid, docosahexaenoic acid and eicosapentaenoic acid) can be caused by deficient intake, particularly, in rapidly growing infants (as preterm infants), in patients receiving parenteral nutrition without an adequate source of EFA, and in diseases with fat malabsorption. Clinical findings are: dermatitis, alopecia, and thrombocytopenia. The role of EFA during pregnancy and lactation has been highlighted, and the role of long-chain n-3 fatty acids as structural components for the development of the retinal function and central nervous system is now accepted. The prenatal period is a time of increased risk for omega-3 deficiency, as maternal tissue stores tend to decline as they are used for the developing fetus. Deficiency of n-3 EFA can affect growth, and cognitive and visual function in infants. The characteristic signs of deficiency attributed to the n-6 fatty acids are scaly skin rash, increased transepidermal water loss, reduced growth, and elevation of the plasma ratio of eicosatrienoic acid: arachidonic acid. EFA deficiency in special populations has been linked to hematologic disturbances and diminished immune response. Long-chain n-3 and n-6 fatty acids are essential nutrients and also, as part of the overall fat supply may affect the prevalence and severity of cardiovascular disease, diabetes, cancer and age-related functional decline.

5B71 Protein deficiency

5B7Y Other specified undernutrition

5B7Z Unspecified undernutrition

Overweight, obesity or specific nutrient excesses (BlockL2‑5B8)

Overweight or obesity (BlockL3‑5B8)

5B80 Overweight or localised adiposity

Overweight is a condition characterized by excessive adiposity. Overweight is assessed by the body mass index (BMI), which is a surrogate marker of adiposity calculated as weight (kg)/height² (m²). The BMI categories for defining overweight vary by age and gender in infants, children and adolescents. For adults, overweight (or pre-obesity) is defined by a BMI ranging from 25.00 to 29.99 kg/m². Localized adiposity is a condition characterized by accumulation of adipose tissue in specific regions of the body independently of BMI.

5B80.0 Overweight

Overweight is a condition characterized by excessive adiposity. Overweight is assessed by the body mass index (BMI), which is a surrogate marker of adiposity calculated as weight (kg)/height² (m²). The BMI categories for defining overweight vary by age and gender in infants, children and adolescents. For adults, overweight (or pre-obesity) is defined by a BMI ranging from 25.00 to 29.99 kg/m².

5B80.00 Overweight in infants, children or adolescents

Overweight is a condition characterized by excessive adiposity. Overweight is assessed by the body mass index (BMI), which is a surrogate marker of adiposity calculated as weight (kg)/height² (m²). In infants, children and adolescents, BMI categories for defining overweight vary by age and gender based on WHO growth charts. Children 0 to 5 years are overweight if weight-for-length/height or BMI-for-age is above 2 and less than or equal to 3 standard deviations of the median of the WHO Child Growth Standards. Children 5 to 19 years are overweight if BMI-for-age is above 1 and less than or equal to 2 standard deviations of the median of WHO Growth Reference for School-aged Children and Adolescents .

5B80.01 Overweight in adults

5B80.0Z Overweight, unspecified

5B80.1 Localised adiposity

A condition characterised by accumulation of adipose tissue in specific regions of the body.

Coded Elsewhere: Benign symmetrical lipomatosis (EF02.1)

5B81 Obesity

Obesity is a chronic complex disease defined by excessive adiposity that can impair health. It is in most cases a multifactorial disease due to obesogenic environments, psycho-social factors and genetic variants. In a subgroup of patients, single major etiological factors can be identified (medications, diseases, immobilization, iatrogenic procedures, monogenic disease/genetic syndrome).

Body mass index (BMI) is a surrogate marker of adiposity calculated as weight (kg)/height² (m²). The BMI categories for defining overweight vary by age and gender in infants, children and adolescents. For adults, overweight is defined by a BMI greater than or equal to 30.00 kg/m². There are three levels of severity in recognition of different management options.

Coded Elsewhere: Obesity hypoventilation syndrome (7A42.0)

Syndromes with obesity as a major feature (LD29)

5B81.0 Obesity due to energy imbalance

Obesity is a chronic complex disease defined by excessive adiposity that can impair health. It is in most cases a multifactorial disease due to obesogenic environments, psycho-social factors and genetic variants. In a subgroup of patients, single major etiological factors can be identified (diseases, immobilization, iatrogenic procedures, monogenic disease/genetic syndrome).

5B81.00 Obesity in children or adolescents

In infants, children and adolescents, BMI categories for defining obesity vary by age and gender based on WHO growth charts. Children 0 to 5 years have obesity if weight-for-length/height or BMI-for-age is above 3 standard deviations of the median of the WHO Child Growth Standards.

Children aged 5 to 19 years have obesity if BMI-for-age is above 2 standard deviations of the median of WHO Growth Reference for School-aged Children and Adolescents.

5B81.01 Obesity in adults

Obesity is defined as a body mass index (BMI) greater than or equal to 30.00 kg/m². There are three levels of severity in recognition of different management options.

5B81.1 Drug-induced obesity

5B81.Y Other specified obesity

5B81.Z Obesity, unspecified

Certain specified nutrient excesses (BlockL3‑5B9)

Any disease caused by an excess of specific nutrients. Confirmation is by blood test.

5B90 Vitamin excesses

5B90.0 Hypervitaminosis A

Because vitamin A is fat soluble and can be stored, primarily in the liver, routine consumption of large amounts of vitamin A over a period of time can result in toxic symptoms, including liver damage, bone abnormalities and joint pain, alopecia, headaches, vomiting, and skin desquamation. Hypervitaminosis A appears to be due to abnormal transport and distribution of vitamin A and retinoids caused by overloading of the plasma transport mechanisms. Very high single doses can cause transient acute toxic symptoms that may include bulging fontanelles in infants; headaches in older children and adults; and vomiting, diarrhoea, loss of appetite, and irritability in all age groups. Rarely does toxicity occur from ingestion of food sources of preformed vitamin A. When this occurs, it usually results from very frequent consumption of liver products.

Coded Elsewhere: Pseudotumour Cerebri related to Hypervitaminosis A (8D41.2)

5B90.1 Hypercarotenaemia

Excessive intake of carotenoids is not associated with toxicity but can cause yellow coloration of the skin that disappears when intake is reduced. This disorder is especially likely to occur in children with liver disease, diabetes mellitus or hypothyroidism, and in those who do not have enzymes that metabolize carotenoids.

5B90.2 Hypervitaminosis D

Hypervitaminosis D is secondary to excessive intake of vitamin D. It can occur with long-term high intake or with a substantial, acute ingestion. Excess amounts result in abnormally high concentrations of calcium and phosphate in the serum. The signs and symptoms of vitamin D intoxication are secondary to hypercalcaemia. Gastrointestinal manifestations include nausea, vomiting, constipation, abdominal pain and pancreatitis. Possible cardiac findings are hypertension, decreased Q-T interval and arrhythmias. The central nervous system effects of hypercalcaemia include lethargy, hypotonia, confusion, disorientation, depression, psychosis, hallucinations and coma. Hypercalcaemia impairs renal concentrating mechanisms, which can lead to polyuria, dehydration and hypernatremia. Hypercalcaemia can also lead to acute renal failure, nephrolithiasis and nephrocalcinosis, which can result in chronic renal insufficiency. Deaths are usually associated with arrhythmias or dehydration.

5B90.3 Megavitamin-B6 syndrome

A disease caused by an excess of vitamin B6. This disease is characterised by progressive sensory ataxia, diminished or absent tendon reflexes, and impaired sense of touch, temperature and pain. Confirmation is by blood test.

Coded Elsewhere: Peripheral neuropathy due to vitamin B6 hyperalimentation (8D41.0)

5B90.Y Other specified vitamin excess

5B90.Z Unspecified vitamin excesses

5B91 Mineral excesses

Coded Elsewhere: Hyperkalaemia (5C76)

Iron overload diseases (5C64.10)

5B91.0 Hypercalcaemia

Hypercalcaemia is a condition caused by increased calcium levels. The higher the calcium levels and the faster its level rises, the more severe will be the symptoms. When present, symptoms are caused by dehydration secondary to urinary losses of calcium, water and other electrolytes, and to an increase in membrane potential caused by the elevation in extracellular fluid ionized calcium concentration. Patients with moderate to severe hypercalcaemia often complain of nausea and vomiting, symptoms likely related to dehydration as well as to the effects of the hypercalcaemia on central nervous system function. Because hypercalcaemia tends to hyperpolarize membranes, a range of neurologic and neuromuscular signs and symptoms can occur. Patients with mild hypercalcaemia often complain of fatigue, depressed mood and asthenia. Gastrointestinal motility is impaired; this commonly results in constipation.

Coded Elsewhere: Myopathy due to hypercalcaemia (8D41.1)

5B91.1 Zinc excess

Adverse effects associated with chronic intake of supplemental zinc include suppression of immune response, decrease in high-density lipoprotein (HDL) cholesterol and reduced copper status. Acute adverse effects of excess zinc include epigastric pain, nausea, vomiting, loss of appetite, abdominal cramps, diarrhoea, headaches and gastrointestinal distress.

Coded Elsewhere: Myelopathy due to excess of zinc (8D41.Y)

5B91.2 Sodium chloride excess

The main adverse effect of increased sodium chloride in the diet is increased blood pressure, which is a major risk factor for cardiovascular-renal diseases. However, evidence from a variety of studies, including observational studies and clinical trials, has demonstrated heterogeneity in the blood pressure responses to sodium intake. Those individuals with the greatest reductions in blood pressure in response to decreased sodium intake are termed “salt sensitive”.

5B91.3 Fluorine excess

The primary adverse effects associated with chronic, excess fluoride intake are enamel and skeletal fluorosis. Enamel fluorosis is a dose-response effect caused by fluoride intake during the pre-eruptive development of teeth. The development of skeletal fluorosis and its severity is directly related to the level and duration of exposure. The clinical signs in advanced stages may include dose-related calcification of ligaments, osteosclerosis, exostoses, possibly osteoporosis of long bones, muscle wasting and neurological defects due to hypercalcification of vertebrae.

Coded Elsewhere: Dental enamel fluorosis (DA07.0)

5B91.4 Aluminium excess

Patients receiving long-term parenteral nutrition are at increased risk of aluminium toxicity because of bypass of the gastrointestinal tract during parenteral nutrition infusion. Complications of aluminium toxicity include metabolic bone disease, aluminium-associated encephalopathy in adults and impaired neurological development in preterm infants.

5B91.5 Manganese excess

Manganese toxicity in humans is a well-recognised occupational hazard for people who inhale manganese dust. High concentrations of circulating manganese as a result of total parenteral nutrition have also been associated with manganese toxicity. People with chronic liver disease have neurological pathology and behavioural signs of manganese neurotoxicity, probably because elimination of manganese in bile is impaired. The most prominent effect is central nervous system pathology, especially in the extra-pyramidal motor system. The lesions and symptoms are similar to those of Parkinson’s disease.

Coded Elsewhere: Dementia or parkinsonism due to manganese toxicity (6D84.Y)

5B91.Y Other specified mineral excess

5B91.Z Unspecified mineral excess

5B9Y Other specified nutrient excesses

5B9Z Certain specified nutrient excesses, unspecified

5C1Y Other specified overweight, obesity or specific nutrient excesses

5C1Z Overweight, obesity or specific nutrient excesses, unspecified

5C3Y Other specified nutritional disorders

5C3Z Nutritional disorders, unspecified

Metabolic disorders (BlockL1‑5C5)

Exclusions: androgen resistance syndrome (LD2A.4)

Congenital adrenal hyperplasia (5A71.01)

Ehlers-Danlos syndrome (LD28.1)

Hereditary haemolytic anaemia due to enzyme deficiency (3A10)

Marfan syndrome (LD28.01)

5-alpha-reductase deficiency (5A81.1)

Coded Elsewhere: Cystic fibrosis (CA25)

Metabolic disorders following abortion, ectopic or molar pregnancy (JA05.5)

Inborn errors of metabolism (BlockL2‑5C5)

Inborn errors of metabolism comprise a large class of genetic diseases involving disorders of metabolism. The majority are due to defects of single genes that code for enzymes that facilitate conversion of various substances (substrates) into others (products).

Exclusions: Disorders of lipoprotein metabolism or certain specified lipidaemias (BlockL2‑5C8)

5C50 Inborn errors of amino acid or other organic acid metabolism

5C50.0 Phenylketonuria

Phenylketonuria is a hereditary metabolic disease, characterised by deficiency of phenylalanine hydroxylase, an enzyme necessary for the transformation of phenylalanine into tyrosine. Untreated, phenylketonuria leads to mental retardation, sometimes profound, as well as hypopigmentation. Dietary phenylalanine restriction allows patients to lead almost normal lives.

5C50.00 Classical phenylketonuria

Classical phenylketonuria is a severe form of phenylketonuria (PKU, ) an inborn error of amino acid metabolism characterised in untreated patients by severe intellectual deficit and neuropsychiatric complications.

5C50.01 Nonclassical phenylketonuria

Mild phenylketonuria is a rare form of phenylketouria (PKU, ), an inborn error of amino acid metabolism, characterised by symptoms of PKU of mild to moderate severity.

5C50.02 Embryofetopathy due to maternal phenylketonuria

Maternal phenylalaninaemia refers to developmental anomalies that may occur in offspring of women affected by phenylketonuria (PKU), and include fetal development disorders, including microcephaly, intrauterine growth retardation, and subsequent intellectual deficit, and embryo development disorders such as heart defects (usually conotruncal), corpus callosus agenesis, neuronal migration disorders, facial dysmorphism and more rarely cleft palate, tracheo-oesophageal abnormalities.

5C50.0Y Other specified phenylketonuria

5C50.0Z Phenylketonuria, unspecified

5C50.1 Disorders of tyrosine metabolism

Coded Elsewhere: Transitory tyrosinaemia of newborn (KB63.4)

Autosomal recessive dopa-responsive dystonia (8A02.11)

Oculocutaneous albinism type 1A (EC23.20)

Oculocutaneous albinism type 1B (EC23.20)

5C50.10 Alkaptonuria

Alkaptonuria is characterised by the accumulation of homogentisic acid (HGA) and its oxidised product benzoquinone acetic acid (BQA), leading to a darkening of the urine when it is left exposed to air, grey-blue colouration of the eye sclerae and the ear helix (ochronosis), and a disabling joint disease involving both the axial and peripheral joints (ochronotic arthropathy).

5C50.11 Tyrosinaemia type 1

Tyrosinemia type 1 is an inborn error of amino acid metabolism characterised by hepatorenal manifestations. The early-onset acute form of the disorder manifests between 15 days and 3 months after birth with hepatocellular necrosis. Septicaemia is a frequent complication. Renal tubular dysfunction occurs and is associated with phosphate loss and hypophosphatemic rickets. A later onset form has also been described and manifests with vitamin-resistant rickets caused by renal tubular dysfunction.

5C50.12 Tyrosinaemia type 2

Tyrosinemia type 2 is an inborn error of tyrosine metabolism characterised by hypertyrosinemia with oculocutaneous manifestations (eye redness, photophobia, excessive tearing and pain, palmoplantar hyperkeratosis) and, in some cases, intellectual deficit.

5C50.1Y Other specified disorders of tyrosine metabolism

5C50.1Z Disorders of tyrosine metabolism, unspecified

5C50.2 Disorders of histidine metabolism

Coded Elsewhere: Formiminoglutamic aciduria (3A02.Y)

5C50.20 Histidinaemia

Histidinemia is a disorder of histidine metabolism caused by a defect in histidase, and seems to be benign in most affected individuals.

5C50.21 Urocanic aciduria

This is an autosomal recessive metabolic disorder caused by a deficiency of the enzyme urocanase. It is a secondary disorder of histidine metabolism.

5C50.2Y Other specified disorders of histidine metabolism

5C50.2Z Disorders of histidine metabolism, unspecified

5C50.3 Disorders of tryptophan metabolism

Exclusions: Hartnup disease (5C60)

5C50.4 Disorders of lysine or hydroxylysine metabolism

Exclusions: Refsum disease (5C57.1)

Zellweger syndrome (5C57.0)

Glutaryl-CoA dehydrogenase deficiency (5C50.E1)

5C50.5 Disorders of the gamma-glutamyl cycle

Coded Elsewhere: Haemolytic anaemia due to glutathione synthetase deficiency (3A10.0Y)

Haemolytic anaemia due to gamma-glutamylcysteine synthetase deficiency (3A10.0Y)

5C50.6 Disorders of serine metabolism

5C50.7 Disorders of glycine metabolism

5C50.70 Glycine encephalopathy

Isolated nonketotic hyperglycinemia is an inborn disorder of glycine metabolism which onset is generally neonatal with coma, severe hypotonia, myoclonic seizures, and microcephaly, usually progressing to severe intellectual deficit and tetrapyramidal syndrome.

5C50.71 Sarcosinaemia

Sarcosinaemia is a metabolic disorder characterised by an increased concentration of sarcosine in plasma and urine due to sarcosine dehydrogenase deficiency. Prevalence has been estimated at 1:28,000 to 1:350,000 in newborn screening programs. Sarcosinaemia is most probably a benign condition without significant clinical problems. It is transmitted in an autosomal recessive manner. Mutations in the gene for sarcosine dehydrogenase, located on chromosome 9q34, have been associated with this deficiency.

5C50.7Y Other specified disorders of glycine metabolism

5C50.7Z Disorders of glycine metabolism, unspecified

5C50.8 Disorders of proline or hydroxyproline metabolism

5C50.9 Disorders of ornithine metabolism

Coded Elsewhere: Hyperornithinaemia-hyperammonaemia-homocitrullinuria (5C50.AY)

Ornithine carbamoyltransferase deficiency (5C50.AY)

5C50.A Disorders of urea cycle metabolism

Exclusions: Disorders of ornithine metabolism (5C50.9)

Lysinuric protein intolerance (5C60)

5C50.A0 Argininosuccinic aciduria

Arginosuccinicaciduria is an autosomal recessive inherited deficiency of arginosuccinate lyase, an enzyme involved in the urea cycle that leads to severe hyperammonemic coma in neonates or, in childhood, to hypotonia, growth failure, anorexia and chronic vomiting or behavioural disorders. Onset can also occur later with hyperammonemic coma or behavioural disorders that simulate psychiatric disorders.

5C50.A1 Carbamoylphosphate synthetase deficiency

Carbamyl phosphate synthetase deficiency is an urea cycle disorder strictly limited to the liver and intestine that results in congenital hyperammonemia and defective citrulline synthesis.

5C50.A2 Argininaemia

Arginase deficiency is a rare autosomal recessive amino acid metabolism disorder characterised clinically by variable degrees of hyperammonemia, developing from about 3 years of age, and leading to progressive loss of developmental milestones and spasticity in the absence of treatment.

5C50.A3 Citrullinaemia

5C50.AY Other specified disorders of urea cycle metabolism

5C50.AZ Disorders of urea cycle metabolism, unspecified

5C50.B Disorders of methionine cycle or sulphur amino acid metabolism

Coded Elsewhere: Hereditary megaloblastic anaemia due to transcobalamin deficiency (3A01.0)

5C50.C Disorders of beta or omega amino acid metabolism

Exclusions: 4-hydroxybutyric aciduria (5C50.E1)

Coded Elsewhere: Gamma aminobutyric acid transaminase deficiency (5C59.1)

5C50.D Disorders of branched-chain amino acid metabolism

Exclusions: Methylmalonic acidaemia (5C50.E0)

Propionic acidaemia (5C50.E0)

Isovaleric acidaemia (5C50.E0)

3-methylglutaconic aciduria (5C50.E0)

Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency (5C50.E0)

3-hydroxyisobutyric aciduria (5C50.E0)

5C50.D0 Maple-syrup-urine disease

Maple syrup urine disease (MSUD) is a disorder of branched-chain amino acids metabolism. Four forms are described. The early onset classic form manifests after birth by lethargy, poor feeding and neurological signs of intoxication. Clinical course without treatment is characterised by deepening coma with maple syrup odour of urine. Subacute MSUD manifests later with encephalopathy, mental disability, major hypotonia, opisthotonus and cerebral atrophy with severe outcome. The intermittent form of MSUD may manifest at any age and presents with repeated ketoacidotic coma. Thiamine-responsive MSUD is a very rare form characterised by improvement of the biochemical profile with thiamine therapy.

5C50.DY Other specified disorders of branched-chain amino acid metabolism

5C50.DZ Disorders of branched-chain amino acid metabolism, unspecified

5C50.E Organic aciduria

An inborn error of metabolism disrupting normal amino acid metabolism, particularly branched-chain amino acids, causing a buildup of acids, which are usually not present

5C50.E0 Classical organic aciduria

This a term used to classify a group of metabolic disorders which disrupt normal amino acid metabolism, particularly branched-chain amino acids, causing a buildup of acids which are usually not present.

Coded Elsewhere: Ketoacidosis due to beta-ketothiolase deficiency (5C50.DY)

5C50.E1 Cerebral organic aciduria

This is a term used to classify a group of metabolic disorders which disrupt normal amino acid metabolism, particularly branched-chain amino acids, causing a buildup of acids which are usually not present.

5C50.EY Other specified organic aciduria

5C50.EZ Organic aciduria, unspecified

5C50.F Disorders of peptide metabolism

A condition which refers to inborn errors in peptide metabolism.

Exclusions: Disorders of gamma aminobutyric acid metabolism (5C59.1)

5C50.F0 Prolidase deficiency

Prolidase deficiency is a very rare inborn error of metabolism characterised by mild to severe skin lesions particularly on the face, palms, lower legs and soles, together with other variable features.

5C50.F1 Carnosinaemia

Carnosinaemia is a very rare inherited disorder of the metabolism of peptides that presents with serum carnosinase deficiency, variable degrees of intellectual deficit, sometimes with seizures, while a few patients are asymptomatic.

5C50.F2 Homocarnosinosis

Homocarnosinosis is a metabolic defect characterised by progressive spastic diplegia, intellectual deficit and retinitis pigmentosa. This extremely rare disorder has been reported in only one family, namely a woman and three of her children. The latter showed but their mother was symptom free. It is therefore uncertain whether there is a relationship between the biochemical defect and the clinical symptoms. Inheritance in the reported family seems to be autosomal dominant.

5C50.FY Other specified disorders of peptide metabolism

5C50.FZ Disorders of peptide metabolism, unspecified

5C50.G Trimethylaminuria

Trimethylaminuria is a metabolic disorder characterised by a body malodour similar to that of decaying fish.

Inclusions: Fish odour syndrome

5C50.Y Other specified inborn errors of amino acid or other organic acid metabolism

5C50.Z Inborn errors of amino acid or other organic acid metabolism, unspecified

5C51 Inborn errors of carbohydrate metabolism

Exclusions: Increased secretion of glucagon (5A42)

Diabetes mellitus (BlockL2‑5A1)

hypoglycaemia NOS (5A41)

Mucopolysaccharidosis (5C56.3)

5C51.0 Disorders of the pentose phosphate pathway

Coded Elsewhere: Haemolytic anaemia due to glucose-6-phosphate dehydrogenase deficiency (3A10.00)

5C51.1 Disorders of glycerol metabolism

5C51.2 Disorders of glyoxylate metabolism

Primary hyperoxaluria, or oxalosis, is a rare metabolic disorder transmitted as an autosomal recessive disease, including both type 1, the most frequent, and type 2, extremely rare. Hyperoxaluria type 1 is due to a defect of the peroxysomal hepatic enzyme L-alanine: glyoxylate aminotransferase (AGT). Hyperoxaluria type 2 is extremely rare and is due to glycerate dehydrogenase deficiency.

5C51.20 Primary hyperoxaluria type 1

Primary hyperoxaluria type 1 is a rare metabolic disorder due to a defect of the peroxysomal hepatic enzyme L-alanine: glyoxylate aminotransferase (AGT). The infantile form is characterised by chronic renal failure due to massive oxalate deposition. In other patients, urolithiasis develops with infections, haematuria, renal colic or acute renal failure due to complete obstruction. End-stage renal failure occurs before 15 years of age in half the cases and the resulting increase of circulating oxalate leads to its deposition in tissues causing cardiac conduction defects, hypertension, distal gangrene, and reduced joint mobility and pain.

5C51.2Y Other specified disorders of glyoxylate metabolism

5C51.2Z Disorders of glyoxylate metabolism, unspecified

5C51.3 Glycogen storage disease

The term Glycogen storage disease characterises a group of heterogeneous diseases resulting from defects in the process of glycogen synthesis or breakdown within muscles, liver, and other cell types.

5C51.4 Disorders of galactose metabolism

5C51.40 Galactose-1-phosphate uridyltransferase deficiency

Classic galactosemia is a life-threatening metabolic disease with onset in the neonatal period. Infants usually develop feeding difficulties, lethargy, and severe liver disease.

5C51.41 Galactokinase deficiency

Galactokinase deficiency is a rare mild form of galactosemia characterised by early onset of cataract and an absence of the usual signs of classic galactosemia, i.e. feeding difficulties, poor weight gain and growth, lethargy, and jaundice.

5C51.42 Glucose or galactose intolerance of newborn

5C51.4Y Other specified disorders of galactose metabolism

5C51.4Z Disorders of galactose metabolism, unspecified

5C51.5 Disorders of fructose metabolism

This refers to disorders of the metabolism of fructose in the phosphorylation of fructose to fructose 1-phosphate by fructokinase, thus trapping fructose for metabolism in the liver.

Coded Elsewhere: Fructose malabsorption (5C61.40)

5C51.50 Hereditary fructose intolerance

Hereditary fructose intolerance is an autosomal recessive disorder due to a deficiency of fructose-1-phosphate aldolase activity, which results in an accumulation of fructose-1-phosphate in the liver, kidney, and small intestine, and is characterised by severe abdominal pain, vomiting, and hypoglycaemia following ingestion of fructose or other sugars metabolised through fructose-1-phosphate.

Exclusions: Fructose malabsorption (5C61.40)

5C51.5Y Other specified disorders of fructose metabolism

5C51.5Z Disorders of fructose metabolism, unspecified

5C51.Y Other specified inborn errors of carbohydrate metabolism

5C51.Z Inborn errors of carbohydrate metabolism, unspecified

5C52 Inborn errors of lipid metabolism

Coded Elsewhere: Retinal dystrophy in lipid storage disorders (9B71.Y)

5C52.0 Inborn errors of fatty acid oxidation or ketone body metabolism

Coded Elsewhere: Adrenoleukodystrophy (8A44.1)

5C52.00 Disorders of carnitine transport or the carnitine cycle

5C52.01 Disorders of mitochondrial fatty acid oxidation

5C52.02 Disorders of ketone body metabolism

Coded Elsewhere: Cytosolic acetoacetyl-CoA thiolase deficiency (5C50.DY)

5C52.03 Sjögren-Larsson syndrome

Sjögren-Larsson syndrome is a neurocutaneous disorder caused by an inborn error of lipid metabolism and characterised by congenital ichthyosis, intellectual deficit, and spasticity.

5C52.0Y Other specified inborn errors of fatty acid oxidation or ketone body metabolism

5C52.0Z Inborn errors of fatty acid oxidation or ketone body metabolism, unspecified

5C52.1 Inborn errors of sterol metabolism

Coded Elsewhere: X-linked ichthyosis (EC20.01)

5C52.10 Disorders of cholesterol synthesis

Coded Elsewhere: Chondrodysplasia punctata, X-linked dominant (LD24.04)

Greenberg dysplasia (LD24.04)

Congenital hemidysplasia with ichthyosiform erythroderma and limbs defects (LD24.04)

Hyperalphalipoproteinaemia due to cholesteryl ester transfer protein deficiency (5C80.3)

5C52.11 Bile acid synthesis defect with cholestasis

Anomalies of bile acid synthesis are a group of sterol metabolism disorders due to enzyme deficiencies of bile acid synthesis in infants, children and adults, with variable manifestations that include cholestasis, neurological disease, and fat malabsorption. Eight inborn errors have been clearly identified, 7 of which lead to liver cholestasis and include: 3-beta-hydroxy-C27-steroid oxidoreductase deficiency (type 1), delta4-3-oxosteriod-5-beta reductase deficiency (type 2), oxysterol 7alpha-hydroxylase deficiency (type 3), 2-methylacyl-CoA racemase deficiency (type 4), bile acid CoA ligase deficiency, and cerebrotendinous xanthomatosis. Cholesterol 7alpha-hydroxylase deficiency leads to hypercholesterolaemia without liver cholestasis.

5C52.1Y Other specified inborn errors of sterol metabolism

5C52.1Z Inborn errors of sterol metabolism, unspecified

5C52.2 Neutral lipid storage disease

Neutral lipid storage disease (NLSD) refers to a group of diseases characterised by a deficit in the degradation of cytoplasmic triglycerides and their accumulation in cytoplasmic lipid vacuoles in most tissues of the body. The group is heterogeneous: NLSD with icthyosis (NLSDI/Dorfman-Chanarin disease) and NLSD with myopathy (NLSDM/neutral lipid storage myopathy) can be distinguished.

5C52.Y Other specified inborn errors of lipid metabolism

5C52.Z Inborn errors of lipid metabolism, unspecified

5C53 Inborn errors of energy metabolism

5C53.0 Disorders of pyruvate metabolism

5C53.00 Pyruvate kinase deficiency

This refers an enzyme involved in glycolysis. It catalyzes the transfer of a phosphate group from phosphoenolpyruvate (PEP) to ADP, yielding one molecule of pyruvate and one molecule of ATP.

Coded Elsewhere: Glycogen storage disease due to muscle pyruvate kinase deficiency (5C51.3)

Haemolytic anaemia due to red cell pyruvate kinase deficiency (3A10.Y)

5C53.01 Lactate dehydrogenase deficiency

This refers to a deficiency in the enzyme present in a wide variety of organisms, including plants and animals. This exist in four distinct enzyme classes. Two of them are cytochrome c-dependent enzymes, each acting on either D-lactate (EC 1.1.2.4) or L-lactate (EC 1.1.2.3). The other two are NAD(P)-dependent enzymes, each acting on either D-lactate (EC 1.1.1.28) or L-lactate (EC 1.1.1.27). This article is about the NAD(P)-dependent L-lactate dehydrogenase.

5C53.02 Pyruvate dehydrogenase complex deficiency

Pyruvate dehydrogenase deficiency (PDHD) is a rare neurometabolic disorder characterised by a wide range of clinical signs with metabolic and neurological components of varying severity. Manifestations range from often fatal, severe, neonatal to later-onset neurological disorders.

5C53.03 Pyruvate carboxylase deficiency

This is a deficiency in the enzyme of the ligase class that catalyzes the (depending on the species) irreversible carboxylation of pyruvate to form oxaloacetate (OAA).

5C53.0Y Other specified disorders of pyruvate metabolism

5C53.0Z Disorders of pyruvate metabolism, unspecified

5C53.1 Disorders of the citric acid cycle

5C53.2 Disorders of mitochondrial oxidative phosphorylation

An inborn error of metabolism in cellular respiration (oxidative phosphorylation) in the mitochondria, where a series of enzymes catalyze the transfer of electrons to molecular oxygen and the generation of energy-storing ATP

Coded Elsewhere: Neuropathy, ataxia, and retinitis pigmentosa (8C73.1)

5C53.20 Mitochondrial DNA depletion syndromes

The mitochondrial DNA (mtDNA) depletion syndrome (MDS) is a clinically heterogeneous group of mitochondrial disorders characterised by a reduction of the mtDNA copy number in affected tissues without mutations or rearrangements in the mtDNA. MDS is phenotypically heterogeneous, manifesting either as a hepatocerebral form, a myopathic form, a benign 'later-onset' myopathic form or a cardiomyopathic form.

Coded Elsewhere: Childhood-onset autosomal dominant optic atrophy (9C40.8)

5C53.21 Multiple mitochondrial DNA deletion syndromes

This is the multiple DNA located in organelles called mitochondria, structures within eukaryotic cells that convert the chemical energy from food into a form that cells can use, adenosine triphosphate (ATP).

Coded Elsewhere: Progressive external ophthalmoplegia, autosomal dominant (9C82.0)

Progressive external ophthalmoplegia, autosomal recessive (9C82.0)

Autosomal dominant optic atrophy plus syndrome (9C40.8)

Deafness - optic atrophy syndrome (LD2H.Y)

Autosomal dominant optic atrophy or cataract (9C40.8)

5C53.22 Coenzyme Q10 deficiency

This is a deficiency in a 1,4-benzoquinone, where Q refers to the quinone chemical group, and 10 refers to the number of isoprenyl chemical subunits in its tail. This oil-soluble, vitamin-like substance is present in most eukaryotic cells, primarily in the mitochondria. It is a component of the electron transport chain and participates in aerobic cellular respiration, generating energy in the form of ATP.

Coded Elsewhere: Cerebellar atrophy - ataxia - seizures (LD90.Y)

5C53.23 Mitochondrial protein translation defects

This refers to defects in the enzyme that belongs to the family of hydrolases, specifically those acting on acid anhydrides to catalyse transmembrane movement of substances.

Coded Elsewhere: Pontocerebellar hypoplasia type 6 (LD20.01)

Mitochondrial myopathy with sideroblastic anaemia (3A72.0Y)

5C53.24 Leigh syndrome

Leigh syndrome or subacute necrotizing encephalomyelopathy is a progressive neurological disease defined by specific neuropathological features associating brainstem and basal ganglia lesions. Loss of motor milestones, hypotonia with poor head control, recurrent vomiting, and a movement disorder are common initial symptoms. Pyramidal and extrapyramidal signs, nystagmus, breathing disorders, ophthalmoplegia and peripheral neuropathy are often noted later. Epilepsy is relatively uncommon. Leigh syndrome has multiple causes, all of which imply a defect in aerobic energy production, ranging from the pyruvate dehydrogenase complex to the oxidative phosphorylation pathway.

Coded Elsewhere: Maternally inherited Leigh syndrome (8C73.Y)

5C53.25 Isolated ATP synthase deficiency

5C53.2Y Other specified disorders of mitochondrial oxidative phosphorylation

5C53.2Z Disorders of mitochondrial oxidative phosphorylation, unspecified

5C53.3 Disorders of mitochondrial membrane transport

An inborn error of metabolism in proteins in the membranes of mitochondria, which serve to transport molecules and other factors such as ions into or out of the organelles

5C53.30 Mitochondrial substrate carrier disorders

Coded Elsewhere: Autosomal recessive sideroblastic anaemia, pyridoxine-refractory (3A72.00)

5C53.31 Mitochondrial protein import disorders

This refers to disorders in the enzyme belongs to the family of hydrolases, specifically those acting on acid anhydrides to catalyse transmembrane movement of substances.

Coded Elsewhere: Deafness-dystonia optic atrophy syndrome (8A02.12)

5C53.3Y Other specified disorders of mitochondrial membrane transport

5C53.3Z Disorders of mitochondrial membrane transport, unspecified

5C53.4 Disorders of creatine metabolism

An inborn error of metabolism in creatine which serves as an energy shuttle between the mitochondrial sites of ATP production and the cytosol where ATP is utilized

5C53.Y Other specified inborn errors of energy metabolism

5C53.Z Inborn errors of energy metabolism, unspecified

5C54 Inborn errors of glycosylation or other specified protein modification

Congenital Disorders of Glycosylation (CDG) syndromes are a group of glycoprotein synthesis disorders characterised by neurological manifestations that can be associated with multivisceral involvement. The CDG syndromes are associated with different enzymatic deficits.

5C54.0 Disorders of protein N-glycosylation

Congenital disorders involving defective N-glycosylation of proteins (the addition of glycans linked to the polypeptide chain by a beta-linkage between the anomeric carbon of N-acetylglucosamine and the amido group of L-asparagine).

5C54.1 Disorders of protein O-glycosylation

Congenital disorders involving defective O-linked glycosylation, which typically occurs via an alpha linkage of the glycan to the hydroxyl group of a serine or threonine residue on a protein

Coded Elsewhere: Multiple osteochondromas (LD24.20)

5C54.2 Disorders of multiple glycosylation or other pathways

Coded Elsewhere: Hereditary inclusion body myositis (4A41.20)

5C54.Y Other specified congenital disorders of glycosylation and protein modification

5C54.Z Congenital disorders of glycosylation and protein modification, unspecified

5C55 Inborn errors of purine, pyrimidine or nucleotide metabolism

Exclusions: Xeroderma pigmentosum (LD27.1)

Calculus of kidney (GB70.0)

5C55.0 Disorders of purine metabolism

Coded Elsewhere: Primary gout (FA25.0)

Haemolytic anaemia due to adenosine deaminase excess (3A10.1)

Immunodeficiency due to purine nucleoside phosphorylase deficiency (4A01.1Y)

Severe combined immunodeficiency T- B- due to adenosine deaminase deficiency (4A01.10)

5C55.00 Xanthinuria

5C55.01 Lesch-Nyhan syndrome

Lesch-Nyhan syndrome (LNS) is the most severe form of hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency, a hereditary disorder of purine metabolism, and is associated with uric acid overproduction (UAO), neurological troubles, and behavioural problems. Patients are normal at birth. Psychomotor delay becomes evident within 3 to 6 months with a delay in head support and sitting, hypotonia and athetoid movements. Sandy urine in diapers or crystalluria with urinary tract obstruction are common forms of presentation. Patients usually show mild to moderate intellectual deficit. Diagnosis is suspected when psychomotor delay occurs in a patient with elevated UA in blood and urine. Undetectable HPRT enzyme activity in peripheral blood or in intact cells (erythrocyte, fibroblast) and molecular genetic testing confirm the diagnosis. Inheritance is X-linked recessive.

5C55.0Y Other specified disorders of purine metabolism

5C55.0Z Disorders of purine metabolism, unspecified

5C55.1 Disorders of pyrimidine metabolism

Coded Elsewhere: Hereditary orotic aciduria (3A03.0)

Haemolytic anaemia due to pyrimidine 5' nucleotidase deficiency (3A10.Y)

5C55.2 Disorders of nucleotide metabolism

Coded Elsewhere: Haemolytic anaemia due to adenosine triphosphatase deficiency (3A10.Y)

5C55.Y Other specified inborn errors of purine, pyrimidine or nucleotide metabolism

5C55.Z Inborn errors of purine, pyrimidine or nucleotide metabolism, unspecified

5C56 Lysosomal diseases

Exclusions: Glycogen storage disease due to LAMP-2 deficiency (5C51.3)

5C56.0 Sphingolipidosis

Coded Elsewhere: Krabbe disease (8A44.4)

Niemann-Pick disease type C (5C56.0Y)

Niemann-Pick disease type A (5C56.0Y)

Niemann-Pick disease type B (5C56.0Y)

5C56.00 Gangliosidosis

5C56.01 Fabry disease

Fabry disease (FD) is a progressive, inherited, multisystemic lysosomal storage disease characterised by specific neurological, cutaneous, renal, cardiovascular, cochleo-vestibular and cerebrovascular manifestations.

Coded Elsewhere: Glomerular disease associated with Fabry disease (GB4Z)

5C56.02 Metachromatic leukodystrophy

Metachromatic leukodystrophy is a neurodegenerative disease characterised by an accumulation of sulfatides (sulphated glycosphingolipids, especially sulfogalactosylceramides or sulfogalactocerebrosides) in the nervous system and kidneys. Three forms of the disease exist: late infantile, juvenile and adult.

5C56.0Y Other specified sphingolipidosis

5C56.0Z Sphingolipidosis, unspecified

5C56.1 Neuronal ceroid lipofuscinosis

Neuronal ceroid lipofuscinoses (NCLs) are a group of inherited progressive degenerative brain diseases characterised clinically by a decline of mental and other capacities, epilepsy, and vision loss through retinal degeneration, and histopathologically by intracellular accumulation of an autofluorescent material, ceroid lipofuscin, in the neuronal cells in the brain and in the retina.

5C56.2 Glycoproteinosis

These are lysosomal storage diseases affecting glycoproteins, resulting from defects in lysosomal function. The term is sometimes reserved for conditions involving degradation of glycoproteins.

5C56.20 Mucolipidosis

Exclusions: Sialidosis (mucolipidosis type 1) (5C56.21)

Coded Elsewhere: Mucolipidosis type 4 (5C56.0Y)

Wolman disease (5C56.0Y)

5C56.21 Oligosaccharidosis

5C56.2Y Other specified glycoproteinosis

5C56.2Z Glycoproteinosis, unspecified

5C56.3 Mucopolysaccharidosis

Inclusions: Disorders of glycosaminoglycan metabolism

5C56.30 Mucopolysaccharidosis type 1

Mucopolysaccharidosis type 1 (MPS 1) is a rare lysosomal storage disease belonging to the group of mucopolysaccharidoses. There are three variants, differing widely in their severity, with Hurler syndrome (57% of cases) being the most severe, Scheie syndrome (20% of cases) the mildest and Hurler-Scheie syndrome (23% of cases) giving an intermediate phenotype.

5C56.31 Mucopolysaccharidosis type 2

Mucopolysaccharidosis type 2 (MPS 2) is a lysosomal storage disease belonging to the group of mucopolysaccharidoses. The clinical picture ranges from severe (the most frequent form) with early psychomotor regression, facial dysmorphism (macroglossia, constantly opened mouth, coarse features), hepatosplenomegaly, limited joint motion, carpal tunnel syndrome, dysostosis multiplex, small size, behavioural disorders and psychomotor regression leading to intellectual deficit, deafness, cardiac and respiratory disorders, and cutaneous signs, to mild (normal intelligence, milder dysmorphism and dysostoses).

Inclusions: Hunter syndrome

5C56.32 Mucopolysaccharidosis type 4

Mucopolysaccharidosis type IV (MPS IV) is a lysosomal storage disease belonging to the group of mucopolysaccharidoses, and characterised by spondylo-epiphyso-metaphyseal dysplasia. It exists in two clinically indistinguishable forms, A and B. A deficiency in one of the two enzymes required for the degradation of keratan sulfate (KS) is responsible for the MPS IV subtypes: N-acetylgalactosamine-6-sulfate sulfatase in MPS IVA, and beta-D-galactosidase in MPS IVB.

5C56.33 Mucopolysaccharidosis type 6

Mucopolysaccharidosis type 6 (MPS VI) is a lysosomal storage disease with progressive multisystem involvement, associated with a deficiency of arylsulfatase B (ASB) leading to the accumulation of dermatan sulfate. The disorder shows a wide spectrum of symptoms from slowly to rapidly progressing forms.

5C56.3Y Other specified mucopolysaccharidosis

5C56.3Z Mucopolysaccharidosis, unspecified

5C56.4 Disorders of sialic acid metabolism

This refers to any disorders of the N- or O-substituted derivatives of neuraminic acid, a monosaccharide with a nine-carbon backbone.

5C56.Y Other specified lysosomal diseases

5C56.Z Lysosomal diseases, unspecified

5C57 Peroxisomal diseases

Peroxisomal disorders represent a class of medical conditions caused by defects in peroxisome functions. This may be due to defects in single enzymes important for peroxisome function or in peroxins, proteins encoded by PEX genes that are critical for normal peroxisome assembly and biogenesis.

Coded Elsewhere: Primary hyperoxaluria type 1 (5C51.20)

Adrenoleukodystrophy (8A44.1)

Rhizomelic chondrodysplasia punctata (LD24.04)

Glutaric aciduria type 3 (5C50.E0)

5C57.0 Disorders of peroxisome biogenesis

Peroxisome biogenesis disorders (PBDs) include the Zellweger syndrome spectrum (PBD-ZSD) and rhizomelic chondrodysplasia punctata type 1 (RCDP1). PBD-ZSD represents a continuum of disorders including infantile Refsum disease, neonatal adrenoleukodystrophy, and Zellweger syndrome. Collectively, PBDs are autosomal recessive developmental brain disorders that also result in skeletal and craniofacial dysmorphism, liver dysfunction, progressive sensorineural hearing loss, and retinopathy.

5C57.1 Disorders of peroxisomal alpha-, beta- or omega-oxidation

Coded Elsewhere: Congenital bile acid synthesis defect type 4 (5C52.11)

5C57.Y Other specified peroxisomal diseases

5C57.Z Peroxisomal diseases, unspecified

5C58 Inborn errors of porphyrin or heme metabolism

Inclusions: defects of catalase and peroxidase

Coded Elsewhere: X-linked sideroblastic anaemia, pyridoxine-responsive (3A72.00)

5C58.0 Disorders of bilirubin metabolism or excretion

Coded Elsewhere: Neonatal hyperbilirubinaemia (KA87)

5C58.00 Crigler-Najjar syndrome

Crigler-Najjar syndrome is an autosomal recessive disorder of bilirubin metabolism characterised by unconjugated hyperbilirubinemia due to a hepatic deficit of bilirubin glucuronosyltransferase activity. Two types have been described, CNS types 1 and 2, depending on whether the enzymatic deficit is complete or partial: clinical manifestations vary accordingly. Patients present with isolated jaundice that appears early in life. Biological analyses detect severe unconjugated hyperbilirubinemia with normal liver function tests. Abdominal imaging studies (plain X-rays, CT scans or ultrasonograms) and liver histology findings are normal. Diagnosis is generally confirmed by genomic DNA analysis.

5C58.01 Gilbert syndrome

Gilbert's syndrome is an inherited liver disorder characterised by jaundice due to unconjugated hyperbilirubinemia, resulting from a partial deficiency in hepatic bilirubin glucuronosyltransferase activity.

5C58.02 Dubin-Johnson syndrome

Dubin-Johnson syndrome (DJS) is a benign, inherited liver disorder characterised clinically by chronic, predominantly conjugated, hyperbilirubinemia and histopathologically by black-brown pigment deposition in parenchymal liver cells.

5C58.03 Progressive familial intrahepatic cholestasis

Progressive familial intrahepatic cholestasis (PFIC) refers to a heterogeneous group of autosomal recessive disorders of childhood that disrupt bile formation and present with cholestasis of hepatocellular origin. Three types of PFIC have been identified and are related to mutations in hepatocellular transport system genes involved in bile formation. PFIC1 and PFIC2 usually appear in the first months of life, whereas onset of PFIC3 may also occur later in infancy, in childhood or even during young adulthood.

5C58.04 Benign recurrent intrahepatic cholestasis

5C58.0Y Other specified disorders of bilirubin metabolism or excretion

5C58.0Z Disorders of bilirubin metabolism or excretion, unspecified

5C58.1 Porphyrias

Porphyrias constitute a group of diseases characterised by intermittent neuro-visceral manifestations, cutaneous lesions or by the combination of both. All porphyrias are caused by a deficiency in one of the enzymes of the heme biosynthesis pathway resulting in an accumulation of porphyrins and/or their precursors in the liver or bone marrow. Clinical signs of the disease usually appear in adulthood, but some porphyrias affect children. Porphyrias can be classified according to the main location of the metabolic anomaly. Direct or indirect neurotoxicity may cause neurological manifestations. Transmission of hereditary porphyrias is autosomal and either dominant with weak penetrance or recessive with complete penetrance. Diagnosis is mainly based on the measurement of porphyrins and their precursors in biological samples.

5C58.10 Porphyria cutanea tarda

Porphyria cutanea tarda (PCT) is due to an accumulation of uroporphyrins in plasma from blockage of the normal haem synthetic pathway in the liver at the level of uroporphyrinogen decarboxylase (URO-D). The majority of cases are sporadic and frequently associated with iron overload. PCT manifests as skin fragility and blistering in light-exposed skin, particularly on the dorsa of the hands, together with hypertrichosis.

5C58.11 Liver diseases due to porphyria

The porphyrias are a group of rare inherited or acquired disorders of certain enzymes that normally participate in the production of porphyrins and heme. Pathogenesis is storage of porphyrins, porphyrinogens, and their precursors. They manifest with either neurological complications or skin problems, or occasionally both. Porphyrias are classified as hepatic or erythropoietic based on the sites of accumulation of heme precursors, either in the liver or bone marrow and red blood cells.

Coded Elsewhere: Porphyria cutanea tarda (5C58.10)

Variegate porphyria (5C58.13)

Hepatoerythropoietic porphyria (5C58.1Y)

Erythropoietic protoporphyria (5C58.12)

Congenital erythropoietic porphyria (5C58.12)

Acute porphyrias (5C58.1Y)

5C58.12 Erythropoietic porphyrias

Erythropoietic porphyrias are associated clinically with photosensitivity and biochemically with abnormal accumulation of porphyrins in erythrocytes. They include erythropoietic protoporphyria and the very rare congenital erythropoietic porphyria.

5C58.13 Variegate porphyria

Variegate porphyria is a form of acute hepatic porphyria characterised by the occurrence of neuro-visceral attacks with or without the presence of cutaneous lesions (bullous photodermatitis).

5C58.1Y Other specified porphyrias

5C58.1Z Porphyrias, unspecified

5C58.Y Other specified inborn errors of porphyrin or heme metabolism

5C58.Z Inborn errors of porphyrin or heme metabolism, unspecified

5C59 Inborn errors of neurotransmitter metabolism

5C59.0 Disorders of biogenic amine metabolism

5C59.00 Disorders of catecholamine synthesis

Any condition caused by failure to correctly synthesize catecholamines. Confirmation is by blood test.

5C59.01 Disorders of pterin metabolism

Any condition caused by failure to correctly metabolize pterin.

Coded Elsewhere: Dopa-responsive dystonia (8A02.11)

5C59.0Y Other specified disorders of biogenic amine metabolism

5C59.0Z Disorders of biogenic amine metabolism, unspecified

5C59.1 Disorders of gamma aminobutyric acid metabolism

Coded Elsewhere: 4-hydroxybutyric aciduria (5C50.E1)

5C59.2 Disorders of pyridoxine metabolism

Coded Elsewhere: Pyridoxal dependent epilepsy (8A61.00)

Pyridoxine dependent epilepsy with antiquitin mutations (8A61.0Y)

5C59.Y Other specified inborn errors of neurotransmitter metabolism

5C59.Z Inborn errors of neurotransmitter metabolism, unspecified

5C5A Alpha-1-antitrypsin deficiency

Alpha-1-antitrypsin deficiency (AATD) is a genetic disorder that manifests as pulmonary emphysema, liver cirrhosis and, rarely, as the skin disease panniculitis, and is characterised by low serum levels of AAT, the main protease inhibitor (PI) in human serum.

5C5Y Other specified inborn errors of metabolism

5C5Z Inborn errors of metabolism, unspecified

Disorders of metabolite absorption or transport (BlockL2‑5C6)

5C60 Disorders of amino acid absorption or transport

Any condition caused by deficiencies in amino acid absorption and transport.

Exclusions: Disorders of tryptophan metabolism (5C50.3)

Coded Elsewhere: Fanconi syndrome (GB90.42)

5C60.0 Oculocerebrorenal syndrome

Oculocerebrorenal syndrome of Lowe (OCRL) is a multisystem disorder characterised by congenital cataracts, glaucoma, intellectual disabilities, postnatal growth retardation and renal tubular dysfunction with chronic renal failure.

5C60.1 Cystinosis

Cystinosis is a metabolic disease characterised by an accumulation of cystine inside the lysosomes of different tissues due to a defect in cystine transport out of lysosomes. There are three clinical forms : infantile, juvenile and ocular. The infantile form is severe, multisystem disease, with impaired proximal tubular reabsorptive capacity, with severe fluid-electrolyte balance alterations, cystine deposits in various organs and progression towards renal failure after 6 years of age. Juvenile cystinosis appear around 8 years of age and has an intermediate clinical picture with end-stage renal disease occurring after the age of 15.The ocular, adult form presents with photophobia.

5C60.2 Cystinuria

Cystinuria is a renal tubular aminoacid transport disorder characterised by recurrent formation of kidneys cystine stones.

5C60.Y Other specified disorders of amino acid absorption or transport

5C60.Z Disorders of amino acid absorption or transport, unspecified

5C61 Disorders of carbohydrate absorption or transport

5C61.0 Glucose-galactose malabsorption

Glucose-galactose malabsorption is characterised by diarrhoea and severe neonatal dehydration. Around 300 cases have been described to date. Moderate glucosuria has also been reported, but fructose absorption is normal. Glucose-galactose malabsorption is caused by a mutation in the SLC5A1 gene, encoding the glucose-sodium cotransporter, SGTL1. The mode of transmission is autosomal recessive. The fatal consequences of this syndrome can be avoided by following a glucose and galactose restricted diet.

Exclusions: Glucose or galactose intolerance of newborn (5C51.42)

5C61.1 Maltase-glucoamylase deficiency

Chronic diarrhea due to glucoamylase deficiency is characterised by chronic diarrhoea in infancy or childhood in association with intestinal glucoamylase deficiency.

5C61.2 Congenital sucrase-isomaltase deficiency

Congenital sucrase-isomaltase deficiency (CSID) is a carbohydrate intolerance disorder characterised by malabsorption of oligosaccharides and disaccharides. CSID is transmitted as an autosomal recessive trait and is caused by mutations in the brush-border membrane complex sucrase-isomaltase (SI), which is required for the breakdown of sucrose and starch into monosaccharides. The SI deficiency results in an accumulation of disaccharides in the lumen, causing osmotic diarrhoea. The prognosis for patients is good as the starch intolerance usually resolves during the first few years of life and sucrose intolerance usually improves with age.

5C61.3 Alpha, alpha trehalase deficiency

Alpha, alpha trehalase deficiency is characterised by diarrhoea and vomiting after ingestion of trehalose, a disaccharide found mainly in mushrooms. The disease is very rare in most populations but the incidence has been estimated at least 1 in 13 in Greenland. Isolated trehalose intolerance is due to a deficiency of trehalase (TREH; 11q23.3), a brush-border membrane glycoprotein.

5C61.4 Acquired monosaccharide malabsorption

This is an acquired condition in which the cells lining the intestine cannot take in one or all of the sugars glucose, galactose or fructose, which prevents proper digestion of these molecules and larger molecules made from them.

It may cause osmotic diarrhoea.

5C61.40 Fructose malabsorption

5C61.4Y Other specified acquired monosaccharide malabsorption

5C61.4Z Acquired monosaccharide malabsorption, unspecified

5C61.5 Disorders of facilitated glucose transport

Coded Elsewhere: Glycogen storage disease due to GLUT2 deficiency (5C51.3)

5C61.6 Lactose intolerance

Lactose intolerance is the inability to digest lactose, a sugar found in milk and some dairy products, due to a deficiency of lactase, the enzyme that metabolizes lactose. Lactose intolerance occurs when lactose is not completely broken down and consequently the sugar cannot be absorbed into the blood.

5C61.60 Primary lactase deficiency

5C61.61 Congenital lactase deficiency

This is a congenital deficiency of lactase (EC 3.2.1.108), inherited as an autosomal recessive trait, presenting in infancy and manifested by profuse watery diarrhoea in response to dietary milk, due to inability to digest lactose, a sugar found in milk and to a lesser extent milk-derived dairy products. The condition may lead to marasmus and death if lactose is not eliminated from the diet.

5C61.62 Secondary lactase deficiency

This form of lactase deficiency results from some sort of damage to the intestines either due to a disease or surgery.

Coding Note: Code aslo the casusing condition

5C61.6Z Lactose intolerance, unspecified

5C61.Y Other specified disorders of carbohydrate absorption or transport

5C61.Z Disorders of carbohydrate absorption or transport, unspecified

5C62 Disorders of lipid absorption or transport

5C63 Disorders of vitamin or non-protein cofactor absorption or transport

Coded Elsewhere: Hereditary factor X deficiency (3B14.1)

Combined deficiency of vitamin K-dependent clotting factors (3B14.2)

5C63.0 Disorders of cobalamin metabolism or transport

Coded Elsewhere: Hereditary Vitamin B12 deficiency anaemia (3A01.0)

Neonatal vitamin B12 deficiency anaemia (3A01.1)

Methylmalonic aciduria, vitamin B12 responsive (5C50.E0)

Congenital or neonatal vitamin B12 deficiency anaemia (3A01.Y)

5C63.1 Disorders of folate metabolism or transport

Coded Elsewhere: Formiminoglutamic aciduria (3A02.Y)

5C63.2 Disorders of vitamin D metabolism or transport

5C63.20 Hypocalcaemic vitamin D dependent rickets

Hypocalcaemic vitamin D-dependent rickets (VDDR-I) is an early-onset hereditary vitamin D metabolism disorder characterised by severe hypocalcaemia leading to osteomalacia and rachitic bone deformations, and moderate hypophosphatemia.

5C63.21 Hypocalcaemic vitamin D resistant rickets

Hypocalcaemic vitamin D-resistant rickets is a hereditary disorder of vitamin D action characterised by hypocalcaemia, severe rickets and in many cases alopecia.

5C63.22 Hypophosphataemic rickets

Hypophosphatemic rickets is a group of genetic diseases characterised by hypophosphatemia, rickets, and normal serum levels of calcium.

5C63.2Y Other specified disorders of vitamin D metabolism or transport

5C63.2Z Disorders of vitamin D metabolism or transport, unspecified

5C63.Y Other specified disorders of vitamin or non-protein cofactor absorption or transport

5C63.Z Disorders of vitamin or non-protein cofactor absorption or transport, unspecified

5C64 Disorders of mineral absorption or transport

A condition in which there is a deviation or interruption in the processing of a specific mineral in the body: its absorption, transport, storage, and utilization

Exclusions: Disorders of the parathyroids or parathyroid hormone system (BlockL2‑5A5)

Vitamin D deficiency (5B57)

dietary mineral deficiency (5B5K)

5C64.0 Disorders of copper metabolism

Any condition caused by failure to correctly metabolize copper.

Coded Elsewhere: X-linked cutis laxa (LD28.2)

5C64.00 Wilson disease

Wilson disease is an autosomal recessive disorder of copper metabolism characterised by the toxic accumulation of copper, mainly in the liver and central nervous system that may present as hepatic, neurologic or psychiatric forms.

5C64.0Y Other specified disorders of copper metabolism

5C64.0Z Disorders of copper metabolism, unspecified

5C64.1 Disorders of iron metabolism

This refers to any disorders of the set of chemical reactions maintaining human homeostasis of iron. The control of this necessary but potentially toxic substance is an important part of many aspects of human health and disease.

Exclusions: Sideroblastic anaemia (3A72)

Iron deficiency anaemia (3A00)

5C64.10 Iron overload diseases

Iron overload is the accumulation of excess iron in body tissues. Iron overload usually occurs as a result of a genetic predisposition to absorb and store iron in excess amounts, the most common form of which is hereditary hemochromatosis. Iron overload can also occur as a complication of other hematologic disorders that require chronic transfusion therapy, repeated injections of parenteral iron, or excessive iron ingestion. Excessive iron stores usually accumulate in the reticuloendothelial tissues and cause little damage (“hemosiderosis”). If overload continues, iron eventually begins to accumulate in tissues such as hepatic parenchyma, pancreas, heart and synovium, causing hemochromatosis.

Coded Elsewhere: Friedreich ataxia (8A03.10)

Atransferrinaemia (3A00.Y)

Microcytic anaemia with liver iron overload (3A00.Y)

5C64.1Y Other specified disorders of iron metabolism

5C64.1Z Disorders of iron metabolism, unspecified

5C64.2 Disorders of zinc metabolism

Any condition caused by failure to correctly metabolize zinc. These conditions may present with dermatitis, diarrhoea, alopecia, loss of appetite, growth impairment, neuropsychological changes, or immune deficiency syndromes.

5C64.20 Acrodermatitis enteropathica

Acrodermatitis enteropathica is an uncommon autosomal recessive disorder of intestinal zinc absorption. Signs usually appear within the first months of life with an exudative and crusted erythema located predominantly around body orifices (mouth, anogenital) and on the scalp and distal extremities. The signs are often misdiagnosed as being due to infection. The condition responds rapidly to zinc supplementation which must be continued throughout life.

5C64.21 Zinc deficiency syndromes

Coded Elsewhere: Acrodermatitis enteropathica (5C64.20)

5C64.2Y Other specified disorders of zinc metabolism

5C64.2Z Disorders of zinc metabolism, unspecified

5C64.3 Disorders of phosphorus metabolism or phosphatases

Any condition caused by errors in phosphorus metabolism, or in phosphatase activity.

Exclusions: Adult osteomalacia (FB83.2)

Osteoporosis (FB83.1)

Coded Elsewhere: Hypophosphataemic rickets (5C63.22)

Phosphate losing hypophosphataemia (GB90.48)

5C64.4 Disorders of magnesium metabolism

Any condition caused by failure to correctly metabolize magnesium.

5C64.40 Hypermagnesaemia

This is an electrolyte disturbance in which there is an abnormally elevated level of magnesium in the blood. Usually this results in excess of magnesium in the body.

5C64.41 Hypomagnesaemia

This is an electrolyte disturbance in which there is an abnormally low level of magnesium in the blood. Normal magnesium levels in humans fall between 1.5 - 2.5 mg/dL. Usually a serum level less than 0.7 mmol/L is used as reference for hypomagnesemia (not hypomagnesia which refers to low magnesium content in food/supplement sources).

Coded Elsewhere: Neonatal hypomagnesaemia (KB61.0)

5C64.4Z Disorders of magnesium metabolism, unspecified

5C64.5 Disorders of calcium metabolism

This refers to and disorders in the mechanism by which the body maintains adequate calcium levels. Derangements of this mechanism lead to hypercalcaemia or hypocalcaemia, both of which can have important consequences for health.

Exclusions: Hyperparathyroidism (5A51)

Chondrocalcinosis (FA26.2)

Coded Elsewhere: Familial hypocalciuric hypercalcaemia (5A51.2)

Hypercalciuria (MF98.0)

Nephrocalcinosis (GB57)

Hypercalcaemia (5B91.0)

5C64.6 Disorders of sodium metabolism

Coded Elsewhere: Tubular disorders of sodium or potassium transport (GB90.46)

Congenital sodium diarrhoea (DA90.1)

5C64.7 Disorders of chloride metabolism

Coded Elsewhere: Congenital chloride diarrhoea (DA90.1)

5C64.Y Disorders of other specified mineral absorption and transport

5C64.Z Disorders of mineral absorption or transport, unspecified

5C6Y Other specified disorders of metabolite absorption or transport

5C6Z Disorders of metabolite absorption or transport, unspecified

Disorders of fluid, electrolyte or acid-base balance (BlockL2‑5C7)

5C70 Volume depletion

Exclusions: Hypovolaemic shock (MG40.1)

5C70.0 Dehydration

Dehydration occurs when there is an insufficient amount or excessive loss of water in the body. This can be caused by vomiting, diarrhoea, fever, use of diuretics, profuse sweating, or decreased water intake.

Coded Elsewhere: Dehydration of newborn (KB63.1)

5C70.1 Hypovolaemia

This is a state of decreased blood volume; more specifically, decrease in volume of blood plasma. It is thus the intravascular component of volume contraction (or loss of blood volume due to things such as haemorrhaging or dehydration), but, as it also is the most essential one, hypovolemia and volume contraction are sometimes used synonymously.

Exclusions: Traumatic shock, not elsewhere classified (NF0A.4)

Hypovolaemic shock (MG40.1)

5C70.Y Other specified volume depletion

5C70.Z Volume depletion, unspecified

5C71 Hyperosmolality or hypernatraemia

Serum sodium concentrations in excess of 145 mmol/L; increased serum concentration of osmotically active particles

Coded Elsewhere: Hypernatremia of newborn (KB63.21)

5C72 Hypo-osmolality or hyponatraemia

Serum sodium concentrations of less than 135 mEq/L; decreased serum concentration of osmotically active particles

Inclusions: sodium [na] deficiency

Exclusions: Syndrome of inappropriate secretion of antidiuretic hormone (5A60.2)

Coded Elsewhere: Hyponatremia of newborn (KB63.20)

5C73 Acidosis

Acidosis is an abnormally acidic state of the blood and tissues.

Exclusions: diabetic acidosis (BlockL2‑5A1)

Coded Elsewhere: Kussmaul respiration (5A22.Y)

5C73.0 Acute respiratory acidosis

This is an acute condition in which decreased ventilation (hypoventilation) causes increased blood carbon dioxide concentration and decreased pH (a condition generally called acidosis). Carbon dioxide is produced continuously as the body's cells respire, and this CO2 will accumulate rapidly if the lungs do not adequately expel it through alveolar ventilation. Alveolar hypoventilation thus leads to an increased PaCO2 (called hypercapnia). The increase in PaCO2 in turn decreases the HCO3-/PaCO2 ratio and decreases pH.

5C73.1 Chronic respiratory acidosis

This is a chronic condition in which decreased ventilation (hypoventilation) causes increased blood carbon dioxide concentration and decreased pH (a condition generally called acidosis). Carbon dioxide is produced continuously as the body's cells respire, and this CO2 will accumulate rapidly if the lungs do not adequately expel it through alveolar ventilation. Alveolar hypoventilation thus leads to an increased PaCO2 (called hypercapnia). The increase in PaCO2 in turn decreases the HCO3?/PaCO2 ratio and decreases pH.

5C73.2 Anion gap metabolic acidosis

This is a form of metabolic acidosis characterised by a high anion gap. The list of agents that cause high anion gap metabolic acidosis is similar to but broader than the list of agents that cause a serum osmolal gap.

5C73.Y Other specified acidosis

5C73.Z Acidosis, unspecified

5C74 Alkalosis

Alkalosis is an abnormally basic state of the blood and tissues.

5C75 Mixed disorder of acid-base balance

This is a condition where more than one of the normal mechanisms that regulate the amount of acid or base content in the body are dysfunctional.

5C76 Hyperkalaemia

Inclusions: Potassium [K] excess

Potassium [K] overload

Coded Elsewhere: Hyperkalaemia of newborn (KB63.31)

5C77 Hypokalaemia

Coded Elsewhere: Hypokalaemia of newborn (KB63.30)

5C78 Fluid overload

This is the condition where there is too much fluid in the blood. The opposite condition is hypovolemia, which is too little fluid volume in the blood. Fluid volume excess in the intravascular compartment occurs due to an increase in total body sodium content and a consequent increase in extracellular body water. The mechanism usually stems from compromised regulatory mechanisms for sodium handling as seen in congestive heart failure (CHF), kidney failure, and liver failure. It may also be caused by excessive intake of sodium from foods, intravenous (IV) solutions and blood transfusions, medications, or diagnostic contrast dyes.

5C7Y Other specified disorders of fluid, electrolyte or acid-base balance

5C7Z Disorders of fluid, electrolyte or acid-base balance, unspecified

Disorders of lipoprotein metabolism or certain specified lipidaemias (BlockL2‑5C8)

Exclusions: Sphingolipidosis (5C56.0)

Coded Elsewhere: Lipoid dermatoarthritis (FA38.Y)

Multicentric reticulohistiocytosis (EE8Y)

Lipoid proteinosis (LD27.Y)

5C80 Hyperlipoproteinaemia

Disorders of lipoprotein metabolism that result in high levels of lipoproteins in the circulating blood

5C80.0 Hypercholesterolaemia

5C80.00 Primary hypercholesterolaemia

This is a genetic disorder characterised by high cholesterol levels, specifically very high levels of low-density lipoprotein (LDL, "bad cholesterol"), in the blood and early cardiovascular disease.

Coded Elsewhere: Sitosterolaemia (5C52.1Y)

5C80.01 Secondary hypercholesterolaemia

Coding Note: Code aslo the casusing condition

5C80.0Z Hypercholesterolaemia, unspecified

5C80.1 Hypertriglyceridaemia

A form of hyperlipidaemia characterised by abnormally elevated levels of triglyceride-rich lipoproteins in the blood. It is associated with an elevated risk of cardiovascular morbidity.

Inclusions: Hyperlipidaemia, group B

Endogenous hyperglyceridaemia

5C80.2 Mixed hyperlipidaemia

Elevated levels of both LDL cholesterol and triglycerides in the blood

Inclusions: Hyperbetalipoproteinaemia with prebetalipoproteinaemia

Hypercholesterolaemia with endogenous hyperglyceridaemia

Hyperlipidaemia, group C

Exclusions: cerebrotendinous cholesterosis [van Bogaert-Scherer-Epstein] (5C52.11)

5C80.3 Hyperalphalipoproteinaemia

A condition in which high-density lipoprotein is elevated in the blood.

5C80.Y Other specified hyperlipoproteinaemia

5C80.Z Hyperlipoproteinaemia, unspecified

5C81 Hypolipoproteinaemia

Disorders characterised by low level of lipoproteins of any type in the blood

Inclusions: High-density lipoprotein deficiency

5C81.0 Hypoalphalipoproteinaemia

A disorder characterised by low levels of high-density lipoprotein in the blood.

5C81.1 Hypobetalipoproteinaemia

Hypobetalipoproteinemia (HBL) constitutes a group of lipoprotein metabolism disorders that are characterised by permanently low levels (below the 5th percentile) of apolipoprotein B and LDL cholesterol. There are two types of HBL: familial hypobetalipoproteinemia and chylomicron retention disease (CMRD; see these terms). The familial form can be severe with early onset (abetalipoproteinemia/homozygous familial hypobetalipoproteinemia; see this term) or benign (benign familial hypobetalipoproteinemia; see this term). (Please add the sentence). Severe familial HBL and CMRD appear in infancy or childhood. As a result they are often associated with growth delay, diarrhoea with steatorrhoea, and fat malabsorption. Benign familial hypobetalipoproteinemia is generally asymptomatic, but in adults is occasionally associated with dietary intolerance to fat. HBL disorders are caused by mutations in proteins involved in the synthesis, secretion and catabolism of lipoproteins containing apolipoprotein B (LDL, VLDL and chylomicrons).

5C81.Y Other specified hypolipoproteinaemia

5C81.Z Hypolipoproteinaemia, unspecified

5C8Y Other specified disorders of lipoprotein metabolism or lipidaemias

5C8Z Unspecified disorders of lipoprotein metabolism or lipidaemias

5C90 Metabolic or transporter liver disease

Exclusions: Alcoholic liver disease (DB94)

Non-alcoholic fatty liver disease (DB92)

Drug-induced or toxic liver disease (DB95)

Acute fatty liver of pregnancy (JA65.0)

Coded Elsewhere: Bile acid synthesis defect with cholestasis (5C52.11)

Progressive familial intrahepatic cholestasis (5C58.03)

Benign recurrent intrahepatic cholestasis (5C58.04)

Glycogen storage disease (5C51.3)

Disorders of galactose metabolism (5C51.4)

Disorders of fructose metabolism (5C51.5)

Alpha-1-antitrypsin deficiency (5C5A)

Reye syndrome (8E46)

5C90.0 Liver diseases due to urea cycle defects

This is a group of liver diseases due to defects in the urea cycle, which is a metabolic cycle of nitrogen-containing compounds that produces the waste product urea.

Coded Elsewhere: Argininosuccinic aciduria (5C50.A0)

Carbamoylphosphate synthetase deficiency (5C50.A1)

Argininaemia (5C50.A2)

Ornithine carbamoyltransferase deficiency (5C50.AY)

5C90.1 Liver diseases due to disorders of porphyrin or bilirubin metabolism or transport

These are liver diseases due to disorders of porphyrin and bilirubin metabolism and transport

Exclusions: Defects of catalase and peroxidase (5C58)

Coded Elsewhere: Liver diseases due to porphyria (5C58.11)

Crigler-Najjar syndrome (5C58.00)

Gilbert syndrome (5C58.01)

Dubin-Johnson syndrome (5C58.02)

Rotor syndrome (5C58.0Y)

5C90.2 Liver diseases due to disorders of amino acid metabolism

This is liver disease due to the disorder of the various biochemical processes responsible for the synthesis of proteins and amino acids, and the breakdown of proteins (and other large molecules, too) by catabolism.

Coded Elsewhere: Disorders of tyrosine metabolism (5C50.1)

Citrullinaemia (5C50.A3)

5C90.3 Liver disease due to disorders of lysosomal storage

This is liver disease due to a group of approximately 50 rare inherited metabolic disorders that result from defects in lysosomal function.

Coded Elsewhere: Gaucher disease (5C56.0Y)

Niemann-Pick disease (5C56.0Y)

Wolman disease (5C56.0Y)

Cholesteryl ester storage disease (5C56.0Y)

5C90.4 Liver diseases due to mitochondrial disorders

This is liver disease due to a group of disorders caused by dysfunctional mitochondria, the organelles that generate energy for the cell.

5C90.5 Liver diseases due to disorders of mineral metabolism

This is a liver disease due to a disorder of the organic compound required by an organism as a vital nutrient in limited amounts.

Coding Note: Code aslo the casusing condition

5C90.Y Other specified metabolic or transporter liver disease

5C90.Z Metabolic or transporter liver disease, unspecified

Other metabolic disorders (BlockL2‑5D0)

Exclusions: histiocytosis X (chronic) (2B31.2)

Coded Elsewhere: Tophaceous gout (FA25.20)

5D00 Amyloidosis

Amyloidosis is a vast group of diseases defined by the presence of insoluble protein deposits in tissues. Its diagnosis is based on histological findings. Amyloidoses are classified according to clinical signs and biochemical type of amyloid protein involved. Most amyloidoses are multisystemic, 'generalised' or 'diffuse'. There are a few forms of localised amylosis. The most frequent forms are AL amyloidosis (immunoglobulins), AA (inflammatory), and ATTR (transthyretin accumulation).

Exclusions: Dementia due to Alzheimer disease (6D80)

Coded Elsewhere: Monoclonal immunoglobulin deposition disease (2A83.5)

5D00.0 AL amyloidosis

AL amyloid is due to the deposition of immunoglobulin light chains in glomeruli where they are seen as Congo red binding fibrils and immuno-stain specifically for kappa or lambda light chains. By light microscopy there is amorphous hyaline material in the mesangium and capillary walls. A light chain producing plasma cell or B-cell dysplasia is responsible. Other organs are also involved in this systemic disease.

Coded Elsewhere: Isolated cerebral amyloid angiopathy (8B22.3)

5D00.1 AA amyloidosis

AA amyloid is due to the deposition of the acute phase reactant serum amyloid A protein (SAA) in glomeruli where they are seen as Congo red binding fibrils which immuno- stain specifically for SAA. Chronic inflammation is responsible. Other organs are also involved in this systemic disease.

5D00.2 Hereditary amyloidosis

Hereditary amyloidosis (familial amyloidosis) is an inherited disorder that often affects the liver, nerves, heart and kidneys. Many different types of gene abnormalities present at birth are associated with an increased risk of amyloid disease. The type and location of an amyloid gene abnormality can affect the risk of certain complications, the age at which symptoms first appear, and the way the disease progresses over time.

5D00.20 Hereditary ATTR amyloidosis

5D00.21 Non-neuropathic heredofamilial amyloidosis

This is an amyloidosis (the formation of insoluble proteins, or amyloids) of inherited origin that does not affect the peripheral nerves. The most common sites of deposits are associated with the kidney and heart.

Coded Elsewhere: Familial Mediterranean fever with amyloidosis (4A60.0)

5D00.2Y Other specified hereditary amyloidosis

5D00.2Z Hereditary amyloidosis, unspecified

5D00.3 Dialysis-associated amyloidosis

Dialysis-related amyloidosis develops when proteins in blood are deposited in joints and tendons — causing pain, stiffness and fluid in the joints, as well as carpal tunnel syndrome. This type generally affects people on long-term dialysis.

5D00.Y Other specified amyloidosis

5D00.Z Amyloidosis, unspecified

5D01 Tumour lysis syndrome

This is a group of metabolic complications that can occur after treatment of cancer, usually lymphomas and leukaemias, and sometimes even without treatment. These complications are caused by the breakdown products of dying cancer cells and include hyperkalaemia, hyperphosphataemia, hyperuricaemia and hyperuricosuria, hypocalcaemia, and consequent acute uric acid nephropathy and acute renal failure.

5D0Y Other specified metabolic disorders

5D2Z Metabolic disorders, unspecified

Postprocedural endocrine or metabolic disorders (BlockL1‑5D4)

Any endocrine or metabolic disorder caused by or subsequent to any medical procedure.

Coded Elsewhere: Injury or harm arising from surgical or medical care, not elsewhere classified (NE80-NE8Z)

5D40 Postprocedural hypothyroidism

5D40.0 Postirridation hypothyroidism

5D40.00 Hypothyroidism postradioactive iodine ablation

5D40.0Y Other specified postirridation hypothyroidism

5D40.0Z Postirridation hypothyroidism, unspecified

5D40.Y Other specified postprocedural hypothyroidism

5D40.Z Postprocedural hypothyroidism, unspecified

5D41 Postprocedural hypoinsulinaemia

This is a low level of insulin that can result after medical procedures, including radiation, and it carries a risk of developing diabetes mellitus.

Inclusions: Postpancreatectomy hyperglycaemia

Postsurgical hypoinsulinaemia

5D42 Postprocedural hypoparathyroidism

This refers to a postprocedural decreased function of the parathyroid glands with underproduction of parathyroid hormone. This can lead to low levels of calcium in the blood, often causing cramping and twitching of muscles or tetany (involuntary muscle contraction), and several other symptoms.

Inclusions: Parathyroprival tetany

5D43 Postprocedural hypopituitarism

This is the postprocedural decreased (hypo) secretion of one or more of the eight hormones normally produced by the pituitary gland at the base of the brain. If there is decreased secretion of most pituitary hormones, the term panhypopituitarism (pan meaning "all") is used.

5D44 Postprocedural ovarian failure

A condition in women characterised by amenorrhea, caused by or subsequent to any intervention. This condition may also present with hot flashes, night sweats, irritability, poor concentration, decreased sex drive, pain during sex, vaginal dryness.

5D45 Postprocedural testicular hypofunction

A condition in men characterised by testosterone deficiency, caused by or subsequent to any intervention. This condition may present with fatigue, decreased libido, erectile dysfunction, negative mood states, decreased lean body mass, increased fat mass, or decreased bone mineral density.

5D46 Postprocedural adrenocortical hypofunction

A condition caused by or subsequent to any medical procedure. This condition is characterised by adrenocortical hormone deficiency. This condition may present with chronic fatigue, muscle weakness, loss of appetite, weight loss or abdominal pain.