Diagnoseliste for SMT sjeldne medisinske tilstander
Utarbeidet av Helsedirektoratet Sist endret 31.12.2019

KODE	BESKRIVELSE	LISTE
AAG	Aagenæs syndrom	A
AAR	Aarskog syndrom	A
AAT	Alfra-antitrypsinmangel	B
ABE	Abetalipoproteinemi	A
ACH	Achalasi	A
ACP	Achondroplasi	A
ACR	Acromegali	A
ADR	Adrenogenitalt syndrom	A
ADY	Angiodysplastisk syndrom	A
AFI	Alvorlig faktor I-mangel	A
AFM	Alvorlig faktor VII-mangel	A
AFV	Alvorlig faktor V-mangel	A
AIC	Aicardi syndrom	A
AIP	Akutt intermitterende porfyri	A
AKP	Akutte porfyrier	A
ALA	Alagilles syndrom	A
ALP	Alport syndrom	A
ALS	Amytrofisk lateral sklerose	A
AMC	Arthrogryposis multiplex congenita	A
AMY	Amyloidose, primær og sekundær	A
ANG	Angelman syndrom	A
APE	Apert syndrom	A
APL	Aplastisk anemi	B
APS	APS type 1 og type 2	A
ARG	Arvelig gingival fibromatose	A
ATD	Alvorlig trombocytt funksjonsdefekter	A
BAR	Bartters Syndrom	A
BBI	Blandet bindevevssykdom	A
BEH	Behçet syndrom	A
BET	Bethlem myopati	A
BFL	Børjeson-Forsman-Lehmann syndrom	A
BMD	Becker muskeldystrofi	A
BOD	Blepharosspasm-oromandibular dystonia syndrom	A
BOR	Brankiootorenalt syndrom	A
BPE	Blepharofimosis-ptose-epicanthus inversus(BPES)	A

KODE	BESKRIVELSE	LISTE
BRO	Bronchopulmonal dysplasi	A
BUL	Bulløs slimhinne- pemfigoid	A
BWI	Beckwith-Wiedemann syndrom	A
CAD	CADASIL (Cerebral Autosomal dominant arteriopati med subkortikal infarsering)	B
CAM	Camtomelic dysplasi	A
CAR	Carpenter syndrom	A
CAS	Castleman	B
CCH	Cri du Chat syndrom	A
CDY	Chondrodysplasi	A
CEA	Cerebellær ataxi	A
CED	Cranioectodermal dysplasi	A
CFI	Cystisk fibrose	A
CHA	CHARGE association	A
CHE	Cherubisme	A
CHG	Carbohydratdefekt-glycoproteinsyndrom	A
CHO	Choanalatresi	A
CHR	Chorea-acanthocytose	A
CIS	Kuldeindusert svettesyndrom	A
CLE	Cleidocranial dysplasi	A
CLS	Coffin Lowry syndrom	A
CMS	Craniofacial misdannelse, sammensatt	A
CMT	Chracot-Marie_Tooth syndrom	A
COH	Cohen syndrom	A
COL	Cornelia de Lange syndrom	A
CON	Congenitt muskeldystrofi	A
COW	Cowden syndrom(multiple hamartomer)	A
CPS	Komplekst regionalt smertesyndrom	B
CRA	Craniosynostose	A
CRO	Crouzons syndrom	A
CSI	Coffin-Siris syndrom	A
CST	Churg-Strauss syndrom	A
CUR	Currarinos syndrom	A
DBL	Døvblind	A
DCH	Diastrofisk chondrodysplasi	A
DEC	Dercums syndrom /Adiposis dolorosa	A
DER	Dermatomyositt	A

KODE	BESKRIVELSE	LISTE
DMD	Duchenne muskeldystrofi	A
DMY	Dystrofia myotonica	A
DUB	Dubowitz syndrom	A
DYC	Dyskeratosis Congenita	A
DYS	Dysmeli	B
EBD	Epidermolysis bullosa dystrofisk junctional	A
EBS	Epidermolysis bullosa simplex	B
ECD	Erdheim-Chester disease	A
ECR	Ellis van Creveld syndrom	A
EDA	Ehlers-Danlos syndrom	A
EDM	Emery-Dreifuss muskeldystrofi	A
EKT	Ektodermal dysplasi	A
ENE	Epidermal nevus	B
EPI	Epifyseal dysplasi	A
EWJ	Ekman-Westborg-Julin	A
FAB	Fabry sykdom	A
FAC	Facialis parese, medfødt	A
FAM	Familiær Dysautonomi	A
FAN	Fanconi Anemi	A
FAP	Familiær adenomatøs polypose	A
FAT	Friedreich ataxi	A
FCD	Fibrøs cemento-ossøs dysplasi	A
FEN	Fenylketonuri (Føllings sykdom)	B
FHH	Familiær hypokalsiurisk hyperkalsemi	B
FMD	Frontometaphyseal dysplasi	A
FOP	Fibrodysplasia ossificans progressiva	A
FSH	Freeman Sheldon syndrom	A
FSM	Facioscapulohumeral muskeldystrofi	A
GAC	Godartet arvelig chorea	A
GAL	Gallegangsatresi	A
GAS	Gardner syndrom	A
GBU	Gjellebuesyndrom	A
GCY	Granulocytopeni	A
GLY	Glykogenose	A
GMS	Gietelmanns syndrom	B
GOL	Goldenhar syndrom	A
GOR	Gorlin syndrom	A

KODE	BESKRIVELSE	LISTE
GRO	Grouchy syndrom	A
GSY	Glutarsyreuri	A
GUI	Guillain-Barre	B
HAB	Hemofili A og Hemofili B	A
HAN	Hereditært angioneurotisk ødem	A
HCH	Hypochondroplasi	A
HCP	Hereditær koproporfyri	A
HEM	Hemihypertrofi	A
HEN	Hennenkam syndrom	A
HFF	Hypofosfatemi	A
HFR	Hypofosfatemisk rakitt	A
HGE	Hyper IgE mangel	A
HGG	Hypogammaglobulinemi	A
HGM	Hyper IgM syndrom	A
HMI	Hemifacial Microsomi	A
HNE	Hereditær nevropati, alle typer	A
HOL	Holoprosencephali	A
HOM	Homocystinuri	A
HOR	Holt Oram syndrom	A
HPA	Hypoparathyreoidisme	A
HPI	Hypopituitarisme	A
HSP	Hallervorden-Spatz syndrom (P-KAN)	A
HTE	Hunter syndrom	A
HTT	Huntingtons sykdom	A
HUR	Hurler syndrom	A
HYP	Hypofosfatasi	A
IAG	IgA, IgG mangel (begge samtidig)	A
ICM	Ichtyoser, sjeldne medfødte	A
IHS	Idiopatisk hypersomni	B
ILF	Idiopatisk lungefibrose	B
ILM	Inklusjonslegememyositt	A
IMM	Immunsvikt (medfødt)	A
INC	Incontinentia pigmenti	A
INT	Intestinal lymfangiectasi	A
JBL	Johanson-Blizzard syndrom	A
JOS	Juvenil osteoporose	A
JOU	Joubert syndrom	A

KODE	BESKRIVELSE	LISTE
KAB	Kabuki syndrom	A
KEP	Kongentital erytropoietisk porfyri	A
KFE	Klippel-Feil syndrom	B
KJD	Kjevedystoni	A
KLS	Kleine Levin syndrom	B
KPP	Keratoderma-Palmi-Plantaris	B
KRN	Kronisk neutropeni	A
KRO	Svært sjeldne kromosomavvik eller genfeil som gir sammensatte vansker	A
KTP	Kronisk trombotisk trombocytopenisk purpura	A
KTW	Klippel-Trènaunay-Weber syndrom	A
LAD	Lacrimo-auriculo-dento-digitalt syndrom	A
LAS	Laings distale myopati	A
LCD	Long Chain 3 Hydroxyasyl CoA Dehydrogenasemangel (LCHAD-mangel)	A
LCH	Langerhans celle histiocytose	A
LDS	Loeys-Dietz syndrom type	A
LDY	Leukodystrofi	A
LEM	Lambert-Eaton-Myasthenic syndrom LEMS	A
LEO	LEOPARD syndrom	A
LEU	Leucocyttadhesjonsdefekt	A
LEW	Leri-Weill dyschondrosteose	A
LGA	Lennox-Gastaut syndrom	A
LGI	Limb Girdle syndrom	A
LHO	LHON (Leber Hereditary Optic Neuropathy, Leber Optic Atrophy)	B
LID	Lineær IgA Dermatose	A
LIP	Lipodystrofi	A
LIS	Lissencephalii	A
LKS	Landau-Kleffner syndrom	B
LMB	Laurence-Moon-Bardet-Biedl syndrom	A
LNY	Lesch-Nyhan syndrom	A
LPG	Lichen planus sykdom, generalisert form	B
LPI	Lysinurisk proteinintoleranse	B
LYM	Lymfangiom	A
MAP	Maple syrup urine disease	A
MAR	Marfan syndrom	A

KODE	BESKRIVELSE	LISTE
MBD	Mb. Darier	A
MBI	Møbius sykdom	A
MBO	Mb. Osler	A
MCA	Mucocutan candidas (kronisk)	A
MCC	McCune Albright	A
MCM	M-CMTC (macrocephalicutis Marmorata)	A
MCN	Medfødt cyklisk neutropeni	A
MCS	Mastocytose	A
MDI	Miller Dieker syndrom	A
MEL	MELAS syndrom(mitokondrie-encefalopati)	A
MEN	Menke's syndrom	A
MER	MERRF Myoklon- epelepsi (ragged-red-fiber)	A
MFT	Multippel familiær trichoepiteliom	A
MFX	Mangel på faktor XIII	A
MGR	Myastena Gravis	A
MHD	Mandibulær hypoplasi, døvhet og progroid syndrom	A
MHS	May-Hegglins sykdom	B
MIA	Multiple idiopatiske apikale rotresposjoner	B
MIG	Migrasjonsforstyrrelse	A
MIR	Multiple idiopatiske cervikale rotresorpsjoner	B
MIT	Mitokondriemyopati	A
MND	Maxillo-nasal dysplasi (Binder syndrom)	A
MNE	Melnick Needles syndrom	A
MOR	Morquio syndrom	A
MPA	Mikroskopisk polyangitt	A
MPS	Mucopolysaccaridose	A
MRK	Mayer-Rokitansky-Küster-Hauser syndrom (MRKH-syndrom) - type 2	A
MUC	Mucolipidose	A
MUL	Multiple endocrine neoplasia type 1 (MEN-1)	A
MUN	Multiple endocrine neoplasia type 2B (MEN-2B)	A
MUS	Muskeldystrofi	B
NAI	Nail-Patella syndrom	A
NAR	Narkolepsi	B
NCL	Nevronal ceroid lipofuscinose	A
NET	Netherton syndrome	B
NEU	Neutropeni	A

KODE	BESKRIVELSE	LISTE
NEV	Nekrotiserende vaskulitt	A
NF1	Nevrofibromatose 1 med orale manifestasjoner	B
NF2	Nevrofibromatose 2	A
NHS	Nance Horan syndrom	A
NOO	Noonan syndrom	A
NPI	Nieman-Pick syndrom	A
OAV	Oculo-auriculo-vertebralt spectrum	A
OBL	Odho blefarofimose syndrom	A
OCF	Oculofaciacardiodentalt syndrom	A
OCR	Oculo-cerebro-renalt syndrom (Lowe syndrom)	A
OCU	Oculopharyngeal muskeldystrofi	A
ODD	Oculodentodigital dysplasi	A
OES	Øsofagusatresi (tidl Oesophagusatresi)	A
OFD	Orofacialt digitalt syndrom (OFD)	A
OFG	Orofacial granulomatose, inkluderer oral manifestasjoner av tilstandene Chrons, ulcerøs colitt og sarkoidose	A
OLI	Olivopontocerebellar atrofi (I-V)	A
OPD	Oto- pallato-digitalt syndrom	A
OPI	Opitz syndrom	A
OPT	Osteopetrose	A
ORM	Ormonds sykdom	A
OSC	Osteopathia striata med cranial stenose	A
OST	Osteogenesis imperfecta	A
PAC	Pachydermoperiostose	A
PCD	Primær ciliedyskinesi	A
PDU	Pseudupseudohypoparathyreoidisme	A
PEL	Pseudoxanthoma elasticum	B
PFE	Pfeiffer syndrom	A
PHP	Paramyotonia/Hyperkalemisk periodisk paralyse	A
PHY	Pseudohypoparathyreoidisme	A
PL1	Plasminogen mangel type I	A
PLE	Pappilon-Lefevre syndrom	A
PLS	Primær lateral sklerose	A
POL	Polymyositt	A
PON	Polyarteritis nodosa	B
POS	Poland syndrom	A
POX	Primær Oxalose	A

KODE	BESKRIVELSE	LISTE
PPH	Pemphigus (alle varianter)	A
PRC	Pure Red Cell Anemia	A
PRO	Proteus syndrom	A
PRS	Pierre Robin sekvens	A
PSE	Pseudoachondrodysplasi	A
PVA	Porfyria variegata	A
PWI	Prader-Willi syndrom	A
PYG	Pyoderma gangrenosym	A
RET	Rett syndrom	A
RIE	Rieger syndrom	A
ROB	Robin sekvens	A
ROM	Romberg syndrom	A
RPK	Residiverende polykondritt	B
RTA	Rubinstein-Taybi syndrom	A
RTS	Rothmund-Thomson syndrom	A
SAN	Sanfilippo syndrom	A
SAP	SAPHO syndrom	A
SAT	SATB assosiert syndrom	A
SCH	Schizencefali	A
SCS	Sætre-Chotzen syndrom	A
SCT	Spondylocarpotarsal stenosis syndrom	A
SDI	Schwacman-Diamond syndrom	A
SEM	Servelle Martorell syndrom	A
SEP	Spondyloepifyseal/-metafyseal dysplasi	A
SGI	Schinzel-Giedeon syndrom	A
SHU	Scapulo-humoral muskeldystrofi	A
SIO	Schimke-immuno-ossøs dysplasi	A
SJS	Scwarz-Jampel syndom	A
SKL	Systemisk sklerose (Sklerodermi)	A
SLA	Sjøgren-Larsson syndrom	A
SMA	Smith-Magenis syndrom	A
SMU	Spinal muskelatrofi	A
SOL	Solitary median maxillary sentral incisor syndrome	A
SOT	Sotos syndrom/Cerebral gigantisme	A
SPA	Kompleks hereditær spastisk paraparese	A
SPS	Stiff person syndrom	A
SPY	Spyttkjertelaplasi	A

KODE	BESKRIVELSE	LISTE
SQU	Spastisk quadriplegi	A
SRA	Seropositiv juvenil reumatoid artritt	A
SRU	Silver Russel syndrom	A
STI	Sticklers syndrom	A
SWE	Sturge-Webers syndrom	A
SWS	Stüve Weidemanns syndrom	A
TAB	TAB2 assosiert syndrom	A
TAK	Takayasus arteritt	B
TDO	Tricho-dento-ossøst-syndrom	A
THO	Thomsens myotoni	A
TRE	Treacher Collins syndrom	A
TRI	Trigonocephali	A
TRP	Tricho-rhino-phalangealt syndrom	A
TUB	Tuberøs sklerose	A
TUM	Tumoral calcinose	A
TVM	Transvers myelitt	A
USH	Usher syndrom	A
VCF	Velocardiofacialt syndrom	A
VDW	Van der Woude syndrom	A
VHL	Von Hippel Lindau syndrom	A
VWI	Von Willebrands syndrom B-liste	B
WAD	Weyers acrofaciale dysostose	A
WDD	Worster Drought Disease	A
WEA	Weaver syndrom	A
WEG	Wegeners granulomatose	A
WES	West syndrom	A
WHI	Wolff-Hirschorns syndrom	A
WIL	Williams syndrom	A
WIS	Wiedmann-Steiner syndrom	A
WLS	Wilsons sykdom	A