PIDcare
Nationellt kvalitetsregister
för primär immunbrist

# Variabellista

Detta är en lista över variabler som kan registreras i PIDcare. Alla variabler är inte relevanta för alla diagnoser.

| Persondata | Registringsalternativ |
|---|---|
| Ansvarig läkare | |
| Ansvarig sjuksköterska | |
| Vårdenhet | |
| Kön | Man, Kvinna |
| Personnummer | |
| Reservnummer | |
| Förnamn | |
| Efternamn | |
| Landsting / regioner | Blekinge, Dalarna, Gotland, Gävleborg, Halland, Jämtland, Jönköping, Kalmar, Kronoberg, Norrbotten, Skåne, Stockholm, Södermanland, Uppsala, Värmland, Västerbotten, Västernorrland, Västmanland, Västra Götaland, Örebro, Östergötland |
| 1:a vårdkontakt | Datum |
| Symtomdebut | Datum |
| Släktskap mellan föräldrar | Ingen (0%), Kusiner (25%), Far- och morföräldrar (25%), Föräldrars syskon (25%), Halvsyskon (25%), Föräldrar (50%), Syskon (50%) |
| Hereditet för immunbrist | Ja, Nej, Okänt |
| Inga symtom | Ja (eller blank) |
| Avliden datum | Datum |
| Avlidenorsak | Hjärtsjukdom, Infektion, Lunginsufficiens, Malignitet, Okänd, Övrigt |

| Diagnos Immunbrist | Registringsalternativ |
|---|---|
| Datum | Startdatum, slutdatum |
| Diagnos | Major category, subcategory, disease enligt IUIS:s klassifikation för primära immunbrister[1]<br>537 diagnoser, se bilaga 1 |
| Kommentar | Kommentar i fritext finns för enstaka diagnoser |
| ICD-10 | C86.3, D46.9, D61.89, D70.0, D71, D72, D72.818, D76.1, D80.0, D80.2, D80.3, D80.4, D80.5, D80.6, D80.7, D80.9, D81.0, D81.1, D81.2, D81.3, D81.6, D81.7, D81.8, D81.9, D82.0, D82.1, D82.2, D82.3, D82.4, D82.8, D82.9, D83.9, D84.0, D84.1, D89.82, D89.9, K52.3, L98.2, M04.1, M04.2, M04.8 |

[1] IUIS: International Union of Immunological Societies

| Associerade diagnoser/komplikationer | Registringsalternativ |
|---|---|
| Datum | Startdatum |
| Associerade tillstånd | Astma, Celiaki, Diabetes mellitus typ 1, Diabetes mellitus typ 2, Giardia, Granulom, Hemolytisk anemi, förvärvad, Hypotyreos, IBD, ITP, KLL, KOL, Lymfadenopati, Lymfom, Myelom, Neutropeni, Perniciös anemi, Respiratorisk insufficiens, Salmonella, Splenomegali, Struma, Ventrikelcancer, Vitiligo, Övrig autoimmunitet, Övrig cancer |
| Organmanifestationer vid CVID | Inga organmanifestationer, Lung - Bronkiektasier, Lung - GLILD - Granulomatous-lymphocytic interstitial lung disease, Lung - Follikulär bronkiolit, Lung - Systemisk granulomatos/lymfoid infiltration, Tarm - Villusatrofi – celiakiliknande, Tarm - Kroniska tarminfektioner, Tarm - Lymfoid hyperplasi/granulom, Tarm - IBD, Tarm - Gastrit, Lever - Kronisk hepatopati med portahypertension, Lever - Nodulär regenerativ hyperplasi (NRH), Lever - Lymfoid infiltration och granulom, Autoimmunitet -Autoimmun cytopeni, Autoimmunitet - Vitiligo, Autoimmunitet - Tyreoidit, Autoimmunitet - Perniciös anemi, Autoimmunitet - Sjögrens syndrom, Malignitet - MALT-lymfom, Malignitet - Non Hodgkins lymfom, Malignitet - Hodgkins lymfom, Malignitet - Ventrikelcancer, Övrigt |
| Infektion | Pneumoni, Sinuit, Bronkit, Otit, PCR-verifierad virusinfektion, Sepsis, Meningit, Lungabscess, Ej luftvägsrelaterad infektion, Covid-19, verifierad |

| Undersökningar | Registringsalternativ |
|---|---|
| Datum | Datum för vikt/längd |
| BMI | Räknas ut automatiskt |
| Längd | cm |
| Vikt | kg |
| Rökning | Rökare, Icke-rökare, Ex-rökare |

| Mikrobiologisk undersökning | Registringsalternativ |
|---|---|
| Datum | Datum för provtagning |
| Typ av diagnostik Bakterier | Nph, Sputum, BAL, Sterilt punktat, Svalg, Serologi, PCR, Övrigt, Kvantifierad sputumodling, Urin, Blod |
| Resultat Bakterier | Negativ odling, Chlamydia pneumoniae, Haemophilus influenzae, Moraxella, Mycoplasma, Pneumokocker, Pseudomonas, Stafylokocker, Streptokocker, Annan bakterie, Ej representativt prov |
| Typ av diagnostik Svamp | NPH, Sputum, BAL, Sterilt punktat, valg, Serologi, PCR, Mikroskopi, Övrigt |
| Resultat Svamp | Negativt, Aspergillus, Candida, Pneumocystis, Övrigt |
| Resultat | RSV positivt, RSV negativt, Influensa positivt, Influensa negativt, Luftvägsblock positivt, Luftvägsblock negativt, Övrigt, SARS-CoV-2-ak positivt, SARS-CoV-2-ak negativt, SARS-CoV-2-RNA positivt, SARS-CoV-2-RNA negativt |

| Fyslab/Röntgen/PatCyt/Genetik | Registringsalternativ |
|---|---|
| Datum | Datum för undersökning |
| Spirometri | Värde för: FVC (%), FEV1 (%), Datum, FVC (L), VC (%), VC (L), FEV1 (L) |
| Röntgen: Typ av undersökning | Rtg pulm, CT pulm, Rtg sinus, CT sinus, PETCT |
| Röntgen: Resultat | Inget avvikande, Bronkiektasi, Infiltrat, Kronisk sinuit, Akut sinuit, Annat avvikande |
| Röntgen: Annat avvikande | Kommentar |
| Benmärgsundersökning | Kommentar |
| Genetisk undersökning | Påvisad mutation: Ja, Nej |
| Genetisk undersökning: Resultat | Gendefekt anges |
| Genetisk undersökning: Kommentar | Fritext |

| Labvärden | Registringsalternativ |
|---|---|
| S-IgG | g/L |
| S-IgA | g/L |
| S-IgM | g/L |
| S-IgG1 | g/L |
| S-IgG2 | g/L |
| S-IgG3 | g/L |
| S-IgG4 | g/L |
| S-anti-IgA | Negativ, Svag positiv, Positiv, Stark positiv, |
| Sekretoriskt IgA | Påvisad, Ej påvisad, |
| B-Leukocyter | x10^9/L |
| B-Hb | g/L |
| B-TPK | x10^9/L |
| B-Neutrofila granulo | x10^9/L |
| B-Eosinofila granulo | x10^9/L |
| B-Basofila | x10^9/L |
| B-Lymfocyter | x10^9/L |
| B-Monocyter | x10^9/L |
| P-Natrium | mmol/L |
| P-Kalium | mmol/L |
| P-Calcium | mmol/L |
| S-kreatinin | mikromol/L |
| P-Bilirubin | mikromol/L |
| P-ASAT | mikrokat/L |
| P-ALAT | mikrokat/L |
| P-ALP | mikrokat/L |
| P-GT | mikrokat/L |
| LD | mikrokat/L |
| P-Albumin | g/L |
| Alfa-1-antitrypsin | g/L |
| P-Orosomucoid | g/L |
| P-Haptoglobin | g/L |
| CRP | mg/L |
| SR | mm |
| S-Calciumjon, fri | mmol/L |
| S-TSH | mE/L |
| S-T4 | pmol/L |
| S-B12/Kobalamin | pmol/L |
| B-Folat | nmol/L |
| 25-OH-VitaminD | nmol/L |
| Beta-2-mikroglobulin | mg/L |

| Labvärden (forts.) | Registringsalternativ |
|---|---|
| CD3 | x10^9/ |
| CD4 | x10^9/ |
| CD8 | x10^9/ |
| CD19 | x10^9/L |
| CD56 | x10^9/L |
| Switchade minnes B-celler, IgD- CD27+ (%) | % |
| Aktiverade B-celler, CD21-CD38-(CD21 low) (%) | % |
| Transitionella B-celler, IgM++ CD38++ (%) | % |
| Naiva CD4+ T-celler, CD4+naiva (CD45RA+ CCR7+) (%) | % |
| Regulatoriska CD4+ T-celler, CD4+CD25+CD127- (%) | % |
| S-IgE | g/L |
| S-IgD | mg/L |
| Serum Amyloid A (SAA) | mg/L |
| HBV | HBsAg positiv, HBsAg negativ, Pos PCR, Neg PCR |
| HCV | anti HCV positiv, anti HCV negativ, Pos PCR, Neg PCR |
| HIV | Positiv, Negativ, Pos PCR, Neg PCR |
| Tetanustiter | Avsaknad, Sänkt, Normalt |
| Difterititer | Avsaknad, Sänkt, Normalt |
| Pneumokocktiter | Avsaknad, Sänkt, Normalt |
| Haemophilustiter | Avsaknad, Sänkt, Normalt |

| Behandlingar: Vaccinationer/SCT/Genterapi | Registringsalternativ |
|---|---|
| Vaccinationer | Vac. Hemophilus influenzae B, Vac. Meningokock, Vac. Mässling, rubella, Parotit, Vac. Pneumokock, poly., Vac. Pneumokock. konj., Vac. Varicella, HPV-vaccin, Influensavaccin, Kombinationsvaccin, Covid-19 Dos 1 – Pfizer-BioNTech, Covid-19 Dos 2 – Pfizer-BioNTech, Covid-19 Dos 1 – Moderna, Covid-19 Dos 2 – Moderna, Covid-19 Dos 1 – Astra Zeneca, Covid-19 Dos 2 – Astra Zeneca, Covid-19 Dos 1 – Annat vaccin, Covid-19 Dos 2 – Annat vaccin |
| Hematopoetisk stamscellstransplantation | Hematopoetisk stamscellstransplantation |
| Genterapi | Kommentar |

| Behandling, Immunglobulin | Registreringsalternativ |
|---|---|
| Datum | Startdatum, ev slutdatum |
| Preparat | Beriglobin, Cutaqiug, Cuvitru, Gammanorm, Hizentra, HyQvia, Subcuvia, Xembify<br>Gammagard 50 mg/ml, Gammagard 100 mg/ml, Octagam, Privigen, Kiovig, Gamunex, Vivaglobin |
| Enhet | ml |
| Beredningsform | Subkutant, fSCIG, Rapid-push, Intravenöst, Intramuskulärt |
| Intervall | per dygn, per vecka, var 10:e dag, per två veckor, per tre veckor, per fyra veckor |
| Orsak till utsättning | Doshöjning, Dossänkning, Utsättningsförsök, Enligt program, Terapisvikt, Patientens önskemål, Biverkning, Annan orsak, Byte pga upphandling, Tillfälligt uppehåll |

| Behandling, Antibiotika | Registreringsalternativ |
|---|---|
| Datum | Startdatum, slutdatum |
| Preparat | Tetracykliner, Penicilliner med utvidgat spektrum, Trimetoprim och/eller sulfa, Makrolider, Fluorokinoloner, Antibakteriella i komb. (J01RA), Betalaktamaskänsliga penicilliner, Betalaktamasresistenta penicilliner, Betalaktamashämmare, Komb. av penicil., inkl komb. m. betalaktamashäm., Monobaktamer, Karbapenemer, Cefalosporiner, Medellångverkande sulfonamider, Långverkande sulfonamider, Sulfonamider och trimetoprim, kombinationer, Kortverkande sulfonamider, Linkosamider, Streptograminer, Kinoloner, Rimactan, Antibakteriella aminoglykosider, Antibiotika mot Tuberkulos, Glykopeptider, Polymyxiner, Antibakteriella steroider, Imidazolderivat, Nitrofuranderivat, Övriga antibakteriella medel |
| Indikation | Bihåleinflammation (Sinuit), Lunginflammation (Pneumoni), Luftrörskatarr (Bronkit), Halsfluss (Tonsillit), Öroninflammation (Otit), Annan luftvägsinfektion, Annat (Ej luftvägsinfektion), Profylax/långtidsbehandling, Inflammationsdämpande behandling |

| Övrig behandling | Registringsalternativ |
|---|---|
| Datum | Startdatum, slutdatum |
| Preparat | Abatecept, Adalimumab, Anakinra, Azatioprin, Baricitinib, Budesonid, Dupilumab, Etanercept, Ibrutinib, Infliximab, Kolkicin, Mycofenolsyra, Palivizumab, Prednisolon, Rituximab, Ruoxitinib, Sirolimus, Tocilizumab, Tofacitinib, Ustekinumab, Vedulizumab, Ciklosporin, Dexametason, Everolimus, Filgrastim, Kolekalciferol, Omalizumab, Takrolimus |
| Indikation | Atopisk dermatit, Autoimmun cytopeni, Autoinflammation, GLILD, Hudabscesser, Inflammation, Kolit, Lymfoproliferation, PTLD, RSV profylax, Splenomegali, Vaskulit, Kronisk mucocutan candidiasis, Övrigt |

| Behandling, Immunglobulin hjälpmedel | Registringsalternativ |
|---|---|
| Datum | Ordinationsdatum, utsättningsdatum |
| Antal pumpar | |
| Administrationsset | Freedom60 admset 8 ml/h, Freedom60 admset 10 ml/h, Freedom60 admset 30 ml/h, Freedom60 admset 45 ml/h, Freedom60 admset 60 ml/h, Freedom60 admset 120 ml/h, Freedom60 admset 180 ml/h, Freedom60 admset 275 ml/h, Freedom60 admset 600 ml/h, Freedom60 admset 900 ml/h, Freedom60 admset 1200 ml/h, Freedom60 admset 2400 ml/h, VersaRate 6 steg, VersaRate Plus 12 steg, Reglerbar flödeskontroll |
| Infusionsset/nålar | Comfort short 1 nål, Cleo 29G 1 nål, Ecoflo 25G 1 nål, EMED SAFQ 27G 1 nål, EMED SAFQ 27G 2 nålar, EMED SAFQ 27G 3 nålar, EMED SAFQ 27G 4 nålar, EMED SAFQ 27G 5 nålar, High-Flo 26G 1 nål, High-Flo 26G 2 nålar, High-Flo 26G 3 nålar, High-Flo 26G 4 nålar, High-Flo 24G 1 nål, High-Flo 24G 2 nålar, High-Flo 24G 3 nålar, High-Flo 24G 4 nålar, Neria 27G 1 nål, Neria Multi 27G 2 nålar, Neria Multi 27G 3 nålar, Neria Multi 27G 4 nålar, Neria Soft 1 nål, Neria Soft45 27 G 1 nål, OrbitSoft 1 nål, SC90 28 G 1 nål, Venofix 25G 1 nål, Venofix 23G 1 nål, Q-control 2 9 mm, Q-control 2 12 mm, Q-control 2 14 mm |
| Uppdragskanyler | Mini-Spike plus, Mini-Spike plus Micro, Uppdragningskanyl rosa, Uppdragningskanyl grön |

| Behandling, hjälpmedel - fortsätting | Registringsalternativ |
|---|---|
| Sprutor | Crono 10 ml, Crono 20 ml, Crono 50 ml, BD 05 ml, BD 10 ml, BD 20 ml, BD 30 ml, BD 50/60 ml, Terumo 05 ml, Terumo 10 ml, Terumo 20 ml, Terumo 50/60 ml, PF 05 ml, PF 10 ml, PF 20 ml, PF 30 ml, PF 50/60 ml |

| PROM | Registreringsalternativ |
|---|---|
| EQ5D5L | |
| RAND35 | |
| Sjukskrivning | Datum från/till. Antal %. Infektionsrelaterat: Ja/nej. Yrkesverksam Ja/Nej/Delvis |
| Vårddagar på sjukhus | Datum från/till. Infektionsrelaterat: Ja/nej/Delvis |
| Symtomdagbok: Snuva | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Torrhosta | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Öronvärk | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Sjukdomskänsla | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Feber över 38 grader | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Slemhosta | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Värk över bihålor | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Magbesvär | Veckonummer. Ja, (eller blank) |
| Symtomdagbok: Halsont | Veckonummer. Ja, (eller blank) |
| Sjukdoms- och behandlingsbörda: Så här mycket påverkar behandlingen mitt dagliga liv | Datum. 1 = Ingen påverkan alls, 2, 3, 4, 5, 6, 7, 8, 9, 10 = Mitt liv styrs helt och hållet av behandlingen |
| Sjukdoms- och behandlingsbörda: Så här mycket påverkar sjukdomen mitt dagliga liv | Datum. 0 = Ingen påverkan alls, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10 = Mitt liv styrs helt och hållet av sjukdomen |
| Immunglobulinregistrering | Datum, dos, batchnummer |
| Immunglobulinregistrering, biverkningar | Svullnad vid injektionsstället, Ömhet/smärta vid injektionsstället, Rodnad vid injektionsstället, Övergående förhårdnad vid injektionsstället, Värmekänsla vid injektionsstället, Klåda vid injektionsstället, Blåmärke vid injektionsstället, Utslag vid injektionsstället, Smärta som strålar ner i benet (efter intramuskulär injektion), Trötthet, Huvudvärk, Frossa, Sjukdomskänsla, Yrsel, Hjärtklappning, Andnöd, Illamående/kräkningar, Smärta i länd och/eller korsryggen, Generell klåda, Nässelutslag på andra delar av kroppen än injektionsstället, Övrigt |

**Versionshistorik**

| Dokumentnamn | | | |
|---|---|---|---|
| Variabellista | | | |
| **Version** | **Datum** | **Förändring och kommentar** | **Ansvarig** |
| 1.0 | 2021-05-03 | Uppdatering av tidigare dokument | Susanne Hansen |
| 2.0 | 2024-01-29 | Uppdatering: Diagnoser inkl bilaga 1, immunglobulinpreparat, infusionsset | Susanne Hansen |
| | | | |

# Bilaga 1: Diagnoser

Kategorisering enligt IUIS: Major category 1-10
Tilläggsdiagnoser för PIDcare: Major category 11-12

| Major category, subcategory, diagnos | ICD10 |
|---|---|
| **1 Immunodeficiencies affecting cellular and humoral immunity** | |
| Subtable 1 T-B+ SCID | |
| CD3d deficiency | D81.2 |
| CD3e deficiency | D81.2 |
| CD3z deficiency | D81.2 |
| CD45 deficiency | D81.2 |
| Coronin-1A deficiency | D81.2 |
| gc deficiency (common gamma chain SCID, CD132 deficiency) | D81.2 |
| IL7Ra deficiency | D81.2 |
| ITPKB deficiency | D81.2 |
| JAK3 deficiency | D81.2 |
| LAT deficiency | D81.2 |
| PAX1 deficiency | D81.2 |
| SLP76 deficiency | D81.2 |
| Subtable 2 T-B- SCID | |
| Activated RAC2 defect | D81.0 |
| Adenosine deaminase (ADA) deficiency | D81.3 |
| Cernunnos/XLF deficiency | D81.1 |
| DCLRE1C (Artemis) deficiency | D81.1 |
| DNA ligase IV deficiency | D81.1 |
| DNA PKcs deficiency | D81.1 |
| RAG1 deficiency | D81.1 |
| RAG2 deficiency | D81.1 |
| Reticular dysgenesis | D81.0 |
| Subtable 3 Combined Immunodeficiencies Generally Less Profound than Severe Combined Immunodeficiency | |
| BCL10 deficiency | D81.9 |
| CARD11 deficiency (LOF) | D81.9 |
| CD3g deficiency | D81.9 |
| CD40 deficiency | D80.5 |
| CD40 ligand deficiency (CD154) | D80.5 |
| CD8 deficiency | D81.9 |
| CHUK deficiency | |
| COPG1 deficiency | |
| c-Rel deficiency | D81.9 |
| DOCK2 deficiency | D81.9 |

| | |
|---|---|
| DOCK8 deficiency | D81.9 |
| FCHO1 deficiency | D81.9 |
| ICOLG deficiency | |
| ICOS deficiency | D81.9 |
| Ikaros deficiency | |
| IKBKB deficiency | D81.9 |
| IKK alfa deficiency | |
| IL-21 deficiency | D81.9 |
| IL-21R deficiency | D81.9 |
| ITK deficiency | D82.3 |
| LCK deficiency | D81.9 |
| MALT1 deficiency | D81.9 |
| MAN2B2 deficiency | D81.9 |
| MHC class I deficiency | D81.6 |
| MHC class II deficiency group A | D81.7 |
| MHC class II deficiency group B | D81.7 |
| MHC class II deficiency group C | D81.7 |
| MHC class II deficiency group D | D81.7 |
| Moesin deficiency | D81.9 |
| MST1 deficiency | D81.9 |
| NIK deficiency | D81.9 |
| OX40 deficiency | D81.9 |
| Polymerase d 1 deficiency | |
| Polymerase d 2 deficiency | |
| RelA haplosufficiency | D81.9 |
| RelB deficiency | D81.9 |
| Rhoh Deficiency | D81.9 |
| SASH3 deficiency | |
| TCRα deficiency | D81.9 |
| TFRC deficiency | D81.9 |
| ZAP70 combined hypomorphic GOF | D81.9 |
| ZAP-70 deficiency (ZAP70 LOF) | D81.9 |

## 2 Combined immunodeficiencies with associated or syndromic features

### Subtable 1 Immunodeficiency with Congenital Thrombocytopenia

| | |
|---|---|
| ARPC1B deficiency | D81.9 |
| WIP deficiency | D81.9 |
| Wiskott-Aldrich syndrome (WAS LOF) | D82.0 |

### Subtable 2 DNA Repair Defects other than those listed in 1

| | |
|---|---|
| Ataxia-telangiectasia | D81.9 |
| Bloom Syndrome | D81.9 |
| GINS1 deficiency | D81.9 |
| Immunodeficiency with centromeric instability and facial anomalies, ICF1 | D81.9 |

| | |
|---|---|
| Immunodeficiency with centromeric instability and facial anomalies, ICF2 | D81.9 |
| Immunodeficiency with centromeric instability and facial anomalies, ICF3 | D81.9 |
| Immunodeficiency with centromeric instability and facial anomalies, ICF4 | D81.9 |
| Ligase I deficiency | D81.9 |
| MCM4 deficiency | D81.9 |
| Nijmegen breakage syndrome | D81.9 |
| NSMCE3 deficiency | D81.9 |
| PMS2 Deficiency | D81.9 |
| POLE1 (Polymerase ε subunit 1) deficiency (FILS syndrome) | D81.9 |
| POLE2 (Polymerase ε subunit 2) deficiency | D81.9 |
| RNF168 deficiency (Radiosensitivity, Immune Deficiency, Dysmorphic features, Learning difficulties [RIDDLE] Syndrome) | D81.9 |
| X-linked reticulate pigmentary disorder-POLA1 | D81.9 |
| **Subtable 2 DNA Repair Defects other than those listed in Table 1** | |
| MCM10 deficiency | D81.9 |
| **Subtable 3 Thymic Defects with Additional Congenital Anomalies** | |
| 22q11 deletion syndrome (DiGeorge/velocardiofacial syndrome) | D82.1 |
| CHARGE syndrome | D81.9 |
| CHARGE syndrome due to CHD7 deficiency | D81.9 |
| CHARGE syndrome due to SEMA3E deficiency | D81.9 |
| Chromosome 10p13-p14 deletion Syndrome (10p13-p14DS) | D81.9 |
| Chromosome 11q deletion syndrome (Jacobsen syndrome) | D81.9 |
| Chromosome 22q11.2 deletion Syndrome (22q11.2DS) (AKA DiGeorge/velocardiofacial syndrome) | D82.1 |
| FOXN1 Haplosufficiency | D81.9 |
| TBX1 deficiency | D82.1 |
| Winged helix FOXN1 deficiency (Nude SCID) | D81.9 |
| **Subtable 4 Immuno-osseous Dysplasias** | |
| Cartilage hair hypoplasia (CHH) | D82.2 |
| Immunoskeletal dysplasia with neurodevelopmental abnormalities (EXTL3 Deficiency) | D81.9 |
| MOPD1 deficiency (Roifman syndrome) | D81.9 |
| MYSM1 deficiency | D81.9 |
| Schimke Immuno-osseous Dysplasia | D81.9 |
| **Subtable 5 Hyper IgE Syndromes (HIES)** | |
| AD-HIES Job syndrome | D82.4 |
| CARD11 DN LOF | D82.4 |
| Comel-Netherton syndrome | D82.4 |
| ERBIN deficiency | D82.4 |
| IL6 receptor deficiency | D82.4 |
| IL6 signal transducer (IL6DT) deficiency (partial) | |
| IL6 signal transducer (IL6ST) deficiency | D82.4 |
| IL6ST deficiency (partial) | |
| Loeys Dietz syndrome due to TGFBR1 deficiency | D82.4 |

| | |
|---|---|
| Loeys Dietz syndrome due to TGFBR2 deficiency | D82.4 |
| PGM3 deficiency | D82.4 |
| ZNF341 deficiency AR-HIES | D82.4 |
| **Subtable 6 Defects of Vitamin B12 and Folate Metabolism** | |
| Methylene-tetrahydrofolate dehydrogenase 1 (MTHFD1) deficiency | D81.9 |
| SLC46A1/PCFT deficiency causing hereditary folate malabsorbtion | D81.9 |
| Transcobalamin 2 deficiency | D81.9 |
| **Subtable 7 Anhidrotic Ectodermodysplasia with Immunodeficiency (EDA-ID)** | |
| EDA-ID due to IKBA GOF | D81.9 |
| EDA-ID due to IKBKB GOF mutation | |
| EDA-ID due to NEMO /IKBKG deficiency (ectodermal dysplasia, immune deficiency) | D81.9 |
| **Subtable 8 Calcium Channel Defects** | |
| CRACR2A deficiency | |
| ORAI-1 deficiency | D81.9 |
| STIM1 deficiency | D81.9 |
| **Subtable 9 Other Combined immunodeficiencies wwith syndromic features** | |
| AIOLOS deficiency | |
| BCL11B deficiency | D81.9 |
| CD28 deficiency | |
| DIAPH1 deficiency | |
| Hennekam-lymphangiectasia-lymphedema syndrome due to CCBE1 deficiency | D81.9 |
| Hennekam-lymphangiectasia-lymphedema syndrome due to FAT4 deficiency | D81.9 |
| Hepatic veno-occlusive disease with immunodeficiency (VODI) | D81.9 |
| HOIL1 deficiency | D81.9 |
| HOIP deficiency | D81.9 |
| Immunodeficiency with multiple intestinal atresias | D81.9 |
| Kabuki Syndrome 1 due to KMT2D deficiency | D81.9 |
| Kabuki Syndrome 2 due to KDM6A deficiency | D81.9 |
| NFE2L2 GOF | D81.9 |
| Purine nucleoside phosphorylase (PNP) deficiency | D81.9 |
| STAT5b deficiency | D81.9 |
| Tricho-hepato-enteric syndrome due to SKIV2L mutations | D81.9 |
| Tricho-hepato-enteric syndrome due to TTC37 mutations | D81.9 |
| Vici syndrome due to EPG5 deficiency | D81.9 |
| Widemann-Steiner syndrome | |
| **3 Predominantly Antibody Deficiencies** | |
| **Subtable 1 Agammaglobulinemia** | |
| BLNK deficiency | D80.0 |
| BTK deficiency, X-linked agammaglobulinemia (XLA) | D80.0 |
| E47 transcription factor deficiency | D80.0 |
| FNIP1 deficiency | D80.0 |

| | |
|---|---|
| Hoffman syndrome/TOP2B deficiency | |
| Iga deficiency | D80.0 |
| Igb deficiency | D80.0 |
| l5 deficiency | D80.0 |
| m heavy chain deficiency | D80.0 |
| PIK3CD deficiency | |
| PIK3R1 deficiency | D80.0 |
| Pu.1 deficiency | |
| SLC39A7 (ZIP7) deficiency | |
| Subtable 2 CVID Phenotype | |
| ARHGEF1 deficiency | |
| ATP6AP1 deficiency | D83.9 |
| BAFF receptor deficiency | D83.9 |
| CD19 deficiency | D83.9 |
| CD20 deficiency | D83.9 |
| CD21 deficiency | D83.9 |
| CD81 deficiency | D83.9 |
| Common variable immune deficiency with no gene defect specified (CVID) | D83.9 |
| IKAROS haplosufficiency | D83.9 |
| IRF2BP2 deficiency | D83.9 |
| Mannosyl-oligosaccharide glucosidase deficiency (MOGS) | |
| NFKB1 deficiency | D83.9 |
| NFKB2 deficiency | D83.9 |
| PIK3CD mutation (GOF) APDS1 | D83.9 |
| PIK3CG deficiency | D83.9 |
| PIK3R1 deficiency (LOF) APDS2 | D83.9 |
| POU2AF1 deficiency | |
| PTEN Deficiency (LOF) | D83.9 |
| RAC2 deficiency | |
| SEC61A1 deficiency | |
| SH3KBP1 (CIN85) deficiency | |
| TACI deficiency | D83.9 |
| TRNT1 deficiency | D83.9 |
| TWEAK deficiency | D83.9 |
| Subtable 3 Severe Reduction in Serum IgG and IgA with Normal/Elevated IgM and Normal Numbers of B cells, Hyper IgM | |
| AID deficiency | D80.5 |
| CTNNBL1 deficiency | D80.5 |
| INO80 | D80.5 |
| MSH6 | D80.5 |
| TNFSF13 (APRIL) deficiency | D80.5 |
| UNG deficiency | D80.5 |

| | |
|---|---|
| **Subtable 4 Isotype, Light Chain, or Functional Deficiencies with Generally Normal Numbers of B Cells** | |
| CARD11 GOF | D80.9 |
| Decreased IgA (excludes IgA deficiency) | |
| Decreased IgG (excludes CVID and IgG subclass deficiency) | |
| Decreased IgG with decreased IgA (excludes CVID, IgG subclass deficiency and IgA-deficiency) | |
| Ig heavy chain mutations and deletions | D80.3 |
| IgG subclass deficiency with decreased IgA (excludes IgA deficiency) | |
| IgG subclass deficiency with IgA deficiency | D80.3 |
| Isolated IgG subclass deficiency | D80.3 |
| Kappa chain deficiency | D80.3 |
| Selective IgA deficiency | D80.2 |
| Selective IgM deficiency | D80.4 |
| Specific antibody deficiency with normal Ig levels and normal B cells | D80.6 |
| Transient hypogammaglobuliemia of infancy | D80.7 |
| **4 Diseases of Immune Dysregulation** | |
| **Subtable 1 Familial Hemophagocytic Lymphohistiocytosis (FHL syndromes)** | |
| FAAP24 deficiency | D76.1 |
| Lysinuric protein intolerance SLC7A7 deficiency | |
| Perforin deficiency (FHL2) | D76.1 |
| RHOG deficiency | |
| STXBP2 / Munc18-2 deficiency (FHL5) | D76.1 |
| Syntaxin 11 deficiency (FHL4) | D76.1 |
| UNC13D / Munc13-4 deficiency (FHL3) | D76.1 |
| **Subtable 2 FHL Syndromes with Hypopigmentation** | |
| CEBPE neofunction | |
| Chediak-Higashi syndrome | D76.1 |
| Griscelli syndrome, type 2 | D76.1 |
| Hermansky-Pudlak syndrome, type 10 | D76.1 |
| Hermansky-Pudlak syndrome, type 2 | D76.1 |
| **Subtable 3 Regulatory T Cell Defects** | |
| BACH2 deficiency | D81.8 |
| CD122 deficiency | |
| CD25 deficiency | D81.8 |
| CTLA4 deficiency (ALPSV) | D81.8 |
| DEF6 deficiency | |
| FERMT1 deficiency (Kindler syndrome) | |
| IKZF1 GOF | D81.8 |
| IPEX, immune dysregulation, polyendocrinopathy, enteropathy X-linked | D82.9 |
| LRBA deficiency | D81.8 |
| STAT3 GOF mutation | D81.8 |

| | |
|---|---|
| Subtable 4 Autoimmunity with or without Lymphoproliferation | |
| APECED (APS-1), autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy | D81.8 |
| ITCH deficiency | D81.8 |
| JAK1 GOF | D81.8 |
| PD1 deficiency | |
| Prolidase deficiency | D81.8 |
| SOCS1 deficiency | D81.8 |
| Tripeptidyl-Peptidase II Deficiency | D81.8 |
| Subtable 5 Immune Dysregulation with Colitis | |
| ELF4 deficiency | |
| IL-10 deficiency | D82.8 |
| IL-10Ra deficiency | D82.8 |
| IL-10Rb deficiency | D82.8 |
| NFAT5 haploinsufficiency | D82.8 |
| RIPK1 deficiency | D82.8 |
| TGFB1 deficiency | D82.8 |
| Subtable 6 Autoimmune Lymphoproliferative Syndrome (ALPS, Canale Smith syndrome) | |
| ALPS-Caspase 8 | D89.82 |
| ALPS-Caspase10 | D89.82 |
| ALPS-FAS | D89.82 |
| ALPS-FASLG | D89.82 |
| FADD deficiency | D89.82 |
| Subtable 7 Susceptibility to EBV and Lymphoproliferative Conditions | |
| CD137 deficiency (41BB) | |
| CD27 deficiency | D82.3 |
| CD70 deficiency | D82.3 |
| CTPS1 deficiency | D82.3 |
| MAGT1 deficiency (XMEN) | D82.3 |
| PRKCD deficiency | D82.3 |
| RASGRP1 deficiency | D82.3 |
| RLTPR (CARMIL2) deficiency | D82.3 |
| SH2D1A (SAP) deficiency (XLP1) | D82.3 |
| TET2 deficiency | D82.3 |
| XIAP deficiency (XLP2) | D82.3 |
| **5 Congenital defects of phagocyte number or function** | |
| Subtable 1 Congenital Neutropenias | |
| 3-Methylglutaconic aciduria | D70.0 |
| Barth Syndrome, (3-Methylglutaconic aciduria type II) | D70.0 |
| Clericuzio syndrome (Poikiloderma with neutropenia) | D70.0 |
| Cohen syndrome | D70.0 |
| CXCR2 deficiency | |
| Elastase deficiency (SCN1) | D70.0 |

| | |
|---|---|
| G6PC3 deficiency (SCN4) | D70.0 |
| G-CSF receptor deficiency | D70.0 |
| GFI 1 deficiency (SCN2) | D70.0 |
| Glycogen storage disease type 1b | D70.0 |
| HAX1 deficiency (Kostmann Disease) (SCN3) | D70.0 |
| HYOU1 deficiency | D70.0 |
| JAGN1 deficiency | D70.0 |
| P14/LAMTOR2 deficiency | D70.0 |
| Schwachman Diamond syndrome due to EFL1 deficiency | D71 |
| Shwachman Diamond syndrome due to DNAJC21 deficiency | D71 |
| Shwachman-Diamond Syndrome due to SBSD deficiency | D71 |
| SMARCD2 deficiency | D70.0 |
| Specific granule deficiency | D71 |
| SRP54 deficiency | |
| VPS45 deficiency (SCN5) | D70.0 |
| X-linked neutropenia/ myelodysplasia WAS GOF | D70.0 |
| **Subtable 2 Defects of Motility** | |
| b actin deficiency | D71 |
| Cystic fibrosis | D71 |
| Leukocyte adhesion deficiency type 1 (LAD1) | D84.0 |
| Leukocyte adhesion deficiency type 2 (LAD2) | D71 |
| Leukocyte adhesion deficiency type 3 (LAD3) | D71 |
| Localized juvenile periodontitis | D71 |
| Neutropenia with combined immune deficiency due to MKL1 deficiency | D71 |
| Papillon-Lefèvre Syndrome | D71 |
| Rac 2 deficiency | D71 |
| WDR1 deficiency (Lazy leukocyte) | D71 |
| **Subtable 3 Defects of Respiratory Burst** | |
| Autosomal recessive CGD EROS | |
| Autosomal recessive CGD p22phox | D71 |
| Autosomal recessive CGD p40phox | D71 |
| Autosomal recessive CGD p47phox | D71 |
| Autosomal recessive CGD p67phox | D71 |
| G6PD deficiency Class I | D71 |
| X-linked chronic granulomatous disease (CGD), gp91phox | D71 |
| **Subtable 4 Other Non-Lymphoid Defects** | |
| Congenital pulmonary alveolar proteinosis due to CSF2RA mutations | |
| Congenital pulmonary alveolar proteinosis due to CSF2RB mutations | |
| GATA2 deficiency (MonoMac syndrome) | D72.818 |

| 6 Defects in intrinsic and innate immunity | |
|---|---|
| Subtable 1 Mendelian Susceptibility to mycobacterial disease (MSMD) | |
| IFN gamma deficiency | D72 |
| IFN-g receptor 1 deficiency | D72 |
| IFN-g receptor 2 deficiency | D72 |
| IL-12 and IL-23 receptor b1 chain deficiency | D72 |
| IL-12p40 (IL-12 and IL-23) deficiency | D72 |
| IL-12Rb2 deficiency | |
| IL-23R deficiency | |
| IRF8 deficiency (AD) | D72 |
| IRF8 deficiency (AR) | D72 |
| ISG15 deficiency | D72 |
| JAK1 (LOF) | D72 |
| Macrophage gp91 phox deficiency | D72 |
| P1104A TYK2 homozygosity | |
| RORgt deficiency | D72 |
| SPPL2a deficiency | |
| STAT1 deficiency (AD LOF) | D72 |
| TBX21 deficiency | D72 |
| Tyk2 deficiency | D72 |
| Subtable 2 Epidermodysplasia verruciformis (HPV) | |
| CIB1 deficiency | |
| EVER1 deficiency | D89.9 |
| EVER2 deficiency | D89.9 |
| WHIM (Warts, Hypogammaglobulinemia, infections, Myelokathexis) syndrome | D89.9 |
| Subtable 3 Predisposition to Severe Viral Infection | |
| CD16 deficiency | D89.9 |
| IFNAR1 deficiency | |
| IFNAR2 deficiency | D89.9 |
| IRF7 deficiency | D89.9 |
| IRF9 deficiency | |
| MDA5 deficiency (LOF) | D89.9 |
| NOS2 deficiency | D89.9 |
| RNA polymerase III deficiency due to POLR3A defects | D89.9 |
| RNA polymerase III deficiency due to POLR3C defects | D89.9 |
| RNA polymerase III deficiency due to POLR3F defects | D89.9 |
| STAT1 deficiency (AR LOF) | D89.9 |
| STAT2 deficiency | D89.9 |
| ZNFX1 deficiency | |

| | |
|---|---|
| **Subtable 4 Herpes Simplex Encephalitis (HSE)** | |
| ATG4A | D89.9 |
| DBR1 deficiency | |
| IRF3 deficiency | D89.9 |
| MAP1LC3B2 | D89.9 |
| SNORA31 deficiency | D89.9 |
| TBK1 deficiency | D89.9 |
| TLR3 deficiency | D89.9 |
| TRAF3 deficiency | D89.9 |
| TRIF deficiency | D89.9 |
| UNC93B1 deficiency | D89.9 |
| **Subtable 5 Predisposition to invasive fungal infections** | |
| CARD9 deficiency | D89.9 |
| **Subtable 6 Predisposition to Mucocutaneous Candidiasis** | |
| ACT1 deficiency | D89.9 |
| IL-17F deficiency | D89.9 |
| IL-17RA deficiency | D89.9 |
| IL-17RC deficiency | D89.9 |
| MAPK8 deficiency | D89.9 |
| STAT1 GOF | D89.9 |
| **Subtable 7 TLR Signaling Pathway Deficiency with Bacterial Susceptibility** | |
| IRAK1 deficiency | D89.9 |
| IRAK4 deficiency | D89.9 |
| MyD88 deficiency | D89.9 |
| TIRAP deficiency | D89.9 |
| TLR7 deficiency | D89.9 |
| TLR8 GOF | |
| **Subtable 8 Other Inborn Errors of Immunity Related to Non-Hematopoietic Tissues** | |
| Acute liver failure due to NBAS deficiency | D89.9 |
| Acute necrotizing encephalopathy | D89.9 |
| CLCN7 deficiency associated osteopetrosis | D89.9 |
| Isolated congenital asplenia (ICA) due to HMOX deficiency | D89.9 |
| Isolated congenital asplenia (ICA) due to RPSA deficiency | D89.9 |
| NCSTN deficiency hidradenitis suppurativa | D89.9 |
| OSTM1 deficiency associated osteopetrosis | D89.9 |
| PLEKHM1 deficiency associated osteopetrosis | D89.9 |
| PSEN deficiency hidradenitis suppurativa | D89.9 |
| PSENEN deficiency hidradenitis suppurativa | D89.9 |
| SNX10 deficiency associated osteopetrosis | D89.9 |
| TCIRG1 deficiency associated osteopetrosis | D89.9 |
| TNFRSF11A deficiency associated osteopetrosis | D89.9 |
| TNFSF11 deficiency associated osteopetrosis | D89.9 |
| Trypanosomiasis susceptibility | D89.9 |

| | |
|---|---|
| Subtable 9 Other inborn errors of immunity related to leukocytes | |
| IL-18BP deficiency | D89.9 |
| IRF4 haplo-insufficiency | D89.9 |
| **7 Autoinflammatory Disorders** | |
| Autoinflammatory disorder of unknown genetic origin | |
| PFAPA | |
| Subtable 1 Type 1 Interferonopathies | |
| ADA2 deficiency | D89.9 |
| ADAR1 deficiency, AGS6 | D89.9 |
| Aicardi-Goutieres syndrome 7 (AGS7) | D89.9 |
| ATAD3A deficiency | |
| C2orf69 deficiency | |
| CDC42 defects | D89.9 |
| DNASE1L3 deficiency | D81.8 |
| DNASE2 deficiency | D89.9 |
| LSM11 deficiency | D89.9 |
| OAS1 GOF | D89.9 |
| RNASEH2A deficiency, AGS4 | D89.9 |
| RNASEH2B deficiency, AGS2 | D89.9 |
| RNASEH2C deficiency, AGS3 | D89.9 |
| RNU7-1 deficiency | D89.9 |
| SAMHD1 deficiency, AGS5 | D89.9 |
| Spondyloenchondro-dysplasia with immune dysregulation (SPENCD) | D89.9 |
| STAT2 R148 LOF/regulation (prevents binding to USP18) | D89.9 |
| STING--associated vasculopathy, infantile-onset | D89.9 |
| STING-like disease | |
| TREX1 deficiency, Aicardi-Goutieres syndrome 1 (AGS1) | D89.9 |
| USP18 deficiency | D89.9 |
| Subtable 2 Defects Affecting the Inflammasome | |
| Familial cold autoinflammatory syndrome 1 | M04.2 |
| Familial cold autoinflammatory syndrome 2 | M04.2 |
| Familial Mediterranean fever | M04.1 |
| Mevalonate kinase deficiency (Hyper IgD syndrome) | M04.1 |
| Muckle-Wells syndrome | M04.1 |
| Neonatal onset multisystem inflammatory disease (NOMID) or chronic infantile neurologic cutaneous and articular syndrome (CINCA) | M04.2 |
| NLRC4-MAS (macrophage activating syndrome) or familial cold autoinflammatory syndrome 4 | M04.2 |
| NLRP1 deficiency | M04.8 |
| NLRP1 GOF | |
| PLAID (PLCg2 associated antibody deficiency and immune dysregulation) or familial cold autoinflammatory syndrome 3 or APLAID (c2120A>C) | M04.8 |
| RIPK1 deficiency | D82.8 |

| Subtable 3 Non-Inflammasome Related Conditions | |
|---|---|
| A20 deficiency | M04.8 |
| ADAM17 deficiency | M04.8 |
| ALPI deficiency | K52.3 |
| AP1S3 deficiency | M04.8 |
| Blau syndrome | M04.8 |
| CAMPS (CARD14 mediated psoriasis) | M04.8 |
| CANDLE (chronic atypical neutrophilic dermatitis with lipodystrophy) | L98.2 |
| Cherubism | M04.8 |
| Chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anemia (Majeed syndrome) | M04.8 |
| COPA defect | M04.8 |
| DIRA (Deficiency of the Interleukin 1 Receptor Antagonist) | M04.8 |
| DITRA (Deficiency of IL-36 receptor antagonist) | M04.8 |
| HCK disease | |
| HEM1 (NCKAPIL) deficiency | K52.3 |
| Hyperpigmentation hypertrichosis, histiocytosis-lymphadenopathy plus syndrome SLC29A3 mutation | M04.8 |
| NEMO-NDAS | M04.8 |
| Otulipenia/ORAS | M04.8 |
| PRAAS -like condition | 0 |
| Pyogenic sterile arthritis, pyoderma gangrenosum, acne (PAPA) syndrome, hyperzincemia and hypercalprotectinemia | M04.8 |
| SYK disease | |
| TBK1 disorder | M04.8 |
| Tim-3 deficiency | C86.3 |
| TNF receptor-associated periodic syndrome (TRAPS) | M04.8 |
| TRIM22 | K52.3 |
| **8 Complement Deficiencies** | |
| C 1 inhibitor deficiency (HAE II) | |
| C 1 inhibitor deficiency (HAE) | |
| C1 inhibitor deficiency | D84.1 |
| C1q deficiency due to defects in C1QA | D84.1 |
| C1q deficiency due to defects in C1QB | D84.1 |
| C1q deficiency due to defects in C1QC | D84.1 |
| C1r deficiency | D84.1 |
| C1r Periodontal Ehlers Danlos | D84.1 |
| C1s deficiency | D84.1 |
| C1s Periodontal Ehlers Danlos | D84.1 |
| C2 deficiency | D84.1 |
| C3 deficiency (LOF) | D84.1 |
| C3 GOF | D84.1 |
| C5 deficiency | D84.1 |

| | |
|---|---|
| C6 deficiency | D84.1 |
| C7 deficiency | D84.1 |
| C8a deficiency | D84.1 |
| C8b deficiency | D84.1 |
| C8g deficiency | D84.1 |
| C9 deficiency | D84.1 |
| CD55 deficiency (CHAPEL disease) | D84.1 |
| Complete C4 deficiency | D84.1 |
| Factor B GOF | D84.1 |
| Factor B LOF | D84.1 |
| Factor D deficiency | D84.1 |
| Factor H deficiency | D84.1 |
| Factor H –related protein deficiencies | D84.1 |
| Factor I deficiency | D84.1 |
| Ficolin 3 deficiency | D84.1 |
| MASP2 deficiency | D84.1 |
| MBL deficiency | |
| Membrane Attack Complex Inhibitor (CD59) deficiency | D84.1 |
| Membrane Cofactor Protein (CD46) deficiency | D84.1 |
| Properdin deficiency | D84.1 |
| Thrombomodulin deficiency | D84.1 |

## 9 Bone marrow failure

| | |
|---|---|
| BMFS3 HSP40 homolog | D61.89 |
| EVI1, MECOM deficiency | |

### Subtable 1 Fanconi anemia

| | |
|---|---|
| Ataxia Pancytopenia Syndrome | D61.89 |
| Fanconi Anemia Type A | D61.89 |
| Fanconi Anemia Type B | D61.89 |
| Fanconi Anemia Type C | D61.89 |
| Fanconi Anemia Type D1 | D61.89 |
| Fanconi Anemia Type D2 | D61.89 |
| Fanconi Anemia Type E | D61.89 |
| Fanconi Anemia Type F | D61.89 |
| Fanconi Anemia Type G | D61.89 |
| Fanconi Anemia Type I | D61.89 |
| Fanconi Anemia Type J | D61.89 |
| Fanconi Anemia Type L | D61.89 |
| Fanconi Anemia Type M | D61.89 |
| Fanconi Anemia Type N | D61.89 |
| Fanconi Anemia Type O | D61.89 |
| Fanconi Anemia Type P | D61.89 |

| | |
|---|---|
| Fanconi Anemia Type Q | D61.89 |
| Fanconi Anemia Type R | D61.89 |
| Fanconi Anemia Type S | D61.89 |
| Fanconi Anemia Type T | D61.89 |
| Fanconi Anemia Type U | D61.89 |
| Fanconi Anemia Type V | D61.89 |
| Fanconia Anemia Type W | D61.89 |
| SAMD9 | D46.9 |
| **Subtable 2 Dyskeratosis congenita** | |
| BMFS1 (SRP72-deficiency) | D61.89 |
| BMFS2 (Hebo deficiency) | D61.89 |
| BMFS5 | D61.89 |
| Coats plus syndrome due to CTC1 deficiency | D82.8 |
| Coats plus syndrome due to STN1 deficiency | D82.8 |
| DKCA1 | D61.89 |
| DKCA2 | D61.89 |
| DKCA3 | D61.89 |
| DKCA4 | D61.89 |
| DKCA5 | D61.89 |
| DKCA6 | D61.89 |
| DKCB1 | D61.89 |
| DKCB2 | D61.89 |
| DKCB3 | D61.89 |
| DKCB4 | D61.89 |
| DKCB5 | D61.89 |
| DKCB6 | D61.89 |
| DKCB7 | D61.89 |
| DKCX1 | D61.89 |
| **10 Phenocopies of PID** | |
| **Subtable 1 Associated with somatic mutations** | |
| Autoimmune lymphoproliferative syndrome (ALPS–SFAS) | |
| Cryopyrinopathy, (Muckle-Wells/CINCA/NOMID-like syndrome) | |
| Hypereosinophilic syndrome due to somatic mutations in STAT5b | |
| RAS-associated autoimmune leukoproliferative disease (RALD) | |
| TLR8 GOF | |
| VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome | D89.9 |
| **Subtable 2 Associated with autoantibodies** | |
| Acquired angioedema | |
| Adult-onset immunodeficiency with susceptibility to mycobacteria | |
| Atypical hemolytic uremic syndrome | |
| Chronic mucocutaneous candidiasis | |
| Pulmonary alveolar proteinosis | |

| | |
|---|---|
| Recurrent skin infection | |
| Severe Covid-19 | D89.9 |
| Thymoma with hypogammaglobulinemia (Good syndrome) | |

## 11 Secondary immunodeficiency

| |
|---|
| Drugs |
| Haematological malignancy |
| Increased loss of immunoglobulin or lymphocytes |
| Nutritional |
| Thymectomy |
| Transplantation |

## 12 No specific PID diagnosis

| |
|---|
| Increased susceptibility to infections, unremarkable work-up |
| On-going immunodeficiency work-up |