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Inborn errors of immunity in Latvia: Analysis of data from 1994 to 2020

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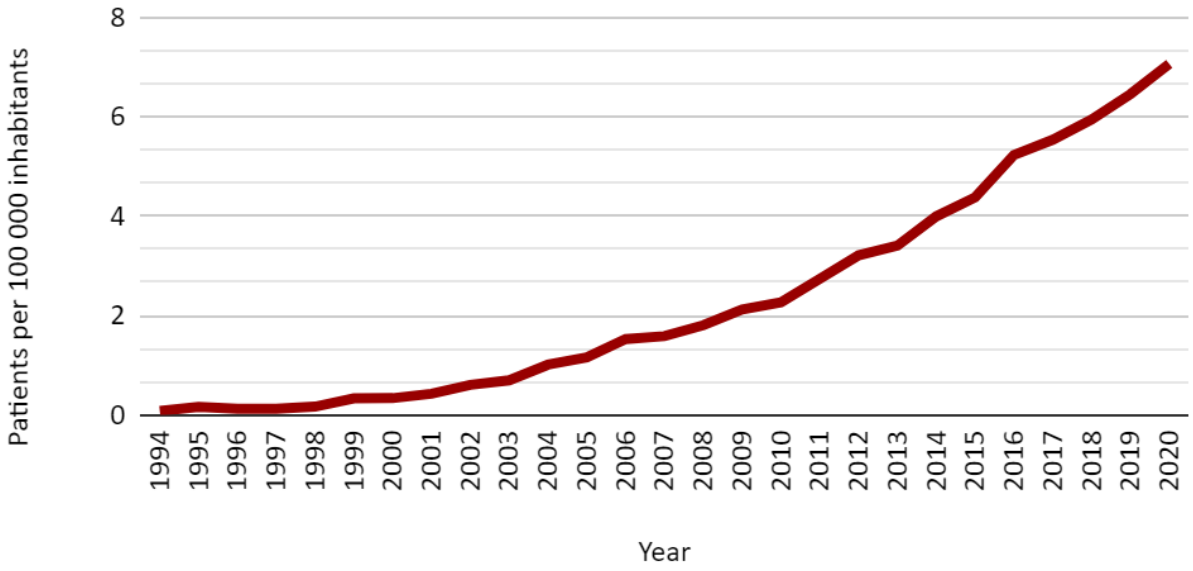
Supplementary Figure 1.



Supplementary Figure 1. Alive patients per 100 000 inhabitants in Latvia

Supplementary Table 1. Results of genetic testing in IEI patients in Latvia

Immunodeficiencies affecting cellular and humoral immunity
Severe combined immunodeficiency, undifferentiated phenotype NM_000536.4(RAG2):c.595G>T (p.Glu199Ter) (1)
T-B+ severe combined immune deficiency NM_000206.3(IL2RG):c.670C>T (p.Arg224Trp) (1) Direct fluorescent sequencing of the *CD3Z* and *CD3G* genes did not show a mutation (1)
T-B- severe combined immune deficiency Homozygous deletion DCLRE1C, EX1-3DEL,(?_-38)_(246-?)del (2) *DCLRE1C* gene. Information about mutation is not available (1)
CD40 ligand deficiency

CD40L c.511C>A, (p.S184X) (2)
Combined immunodeficiencies with associated or syndromic features
Wiskott-Aldrich syndrome NM_000377.3(WAS):c.631C>T (p.Arg211Ter) (1)
Ataxia-telangiectasia Heterozygous NM_000051.4(ATM):c.1564_1565del (p.Glu522fs) (1) NM_000051.4(ATM):c.5932G>T (p.Glu1978Ter); NM_000051.4(ATM):c.8731A>C (p.Thr2911Pro) (1)
Nijmegen breakage syndrome NM_002485.5(NBN):c.657_661del (p.Lys219fs) (5)
22q11 deletion syndrome 22q11.2 deletion (based on FISH analysis) (35)
Comel-Netherton syndrome NM_006846.4(SPINK5):c.1048C>T (p.Arg350*) (3) Compound heterozygous NM_006846.4(SPINK5):c.1048C>T (p.Arg350*); c.1430+4A>G (1)
Kabuki syndrome KMT2D c.3809C>G, p.(Ser1207*) (1) KTM2D c.11377c>T, p.(Gln379*) (1) KMT2D c.2728_2729dup, p.(Pro912Cysfs*) (1) Duplication in Xp11.3 region (based on array comparative genomic hybridization data) (KDM6A gene) (1) Information about mutations is not available (3)
Predominantly antibody deficiency
BTK deficiency, X-linked agammaglobulinaemia (XLA) NM_000061.3(BTK):c.1082A>G (p.Tyr361Cys) (1) BTK c.391+1G>A, IVS5+1G>A (1)
Common variable immune deficiency with no gene defect specified (CVID) PID panel inconclusive (1)
Activated p110δ syndrome (APDS) NM_005026.5(PIK3CD):c.1573G>A (p.Glu525Lys) (1) Heterozygous NM_005026.3(PIK3CD):c.2905C>T, p.(Arg969Cys) (1)
Diseases of immune dysregulation
Familial haemophagocytic lymphohistiocytosis, Perforin deficiency 2 – Compound heterozygous NM_001083116.3(PRF1):c.658G>A (p.Gly220Ser); NM_001083116.3(PRF1):c.666C>A (p.His222Gln) (2)
X-linked lymphoproliferative disease (XLP1) [GRCh37]Xq25(123479999_123672135)x0 (SH2D1A gene) (2) NM_002351.5(SH2D1A):c.5A>G (p.Asp2Gly) (1)
Congenital defects of phagocyte number or function

Shwachman-Diamond Syndrome NM_016038.4(SBDS):c.258+2T>C; c.183-184TA>CT (1)
X-linked chronic granulomatous disease NM_000397.4(CYBB):c.676C>T (p.Arg226Ter) (3) CYBB c.1166G>T, (p.Gly389Val) (1) CYBB c.577T>C, p.(Ser193Pro) (1)
Congenital neutropenia Information about mutations is not available
Defects in intrinsic and innate immunity
NM_007315.4(STAT1):c.812A>C (p.Gln271Pro) (1) NM_007315.4(STAT1):c.1154C>T (p.Thr385Met) (1)
Auto-inflammatory disorders
Familial Mediterranean fever Heterozygous NM_000243.3(MEFV):c.2080A>G (p.Met694Val) (1) Heterozygous NM_000243.3(MEFV):c.2177T>C (p.Val726Ala) (1) Heterozygous NM_000243.3(MEFV):c.442G>C (p.Glu148Gln) (1) Compound heterozygous NM_000243.3(MEFV):c.2080A>G (p.Met694Val); NM_000243.3(MEFV):c.2177T>C (p.Val726Ala) (2)
Unclassified inborn error of immunity PID panel inconclusive (1)

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