**Table E3:** phenotypes and outcomes of PID patients, identified to harbor predicted pathogenic variants

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **pat.ID** | **age** | **gender** | **clinical diagnosis** | **genetic defect** | **infections** | **immundyregulation** | **other manifestations** | **follow-up** |
| P.204 | 21 | m | agammaglobulinemia | *IGLL1*, AR | recurrent bronchitis (*H. influenza*), bronchiectasis | no | sensorineural deafness, neuropathy and muscle degenerative diseases, exocrine pancreatic insufficiency | alive |
| P.71 | 45 | f | CVID | *LRBA*, AR | recurrent bronchitis, HSV pneumonia, bronchiectasis | ITP, ILD, psoriasis | hypoxemic respiratory failure | deceased |
| P.219\* | 32 | m | CID | *CARMIL2*, AR | cutaneous infections with *S. aureus*, pneumonia, verrucae vulgares, molluscum contagiosum | eczema | no | alive |
| P.285\* | 4 | m | CID | *CARMIL2*, AR | recurrent bronchitis, cutaneous infections with *S. aureus* and *S. dysgalactiae equisimillis*, verrucae vulgares, molluscum contagiosum | eczema, psoriasis | no | alive |
| P.289\* | 9 | f | CID | *CARMIL2*, AR | recurrent pneumonia, mucocutaneous infections through *S. aureus*, *S. dysgalactiae equismilis*, beta-hemolytic streptococci, *Candida parapsilosis* and *Candida albicans*, verrucae vulgares, molluscum contagiosum | eczema, psoriasis | no | alive |
| P.208 | 24 | m | CVID | *PIK3CD*, AR | osteomyelitis, recurrent sinusitis and bronchitis, recurrent pneumonias, *C. difficile* colitis, *C. jejuni* enteritis | Crohn disease, psoriasis | no | alive |
| P.273 | 5 | m | atypical SCID (CID) | *DCLRE1C* (ARTEMIS), AR | recurrent infections, chronic diarrhea, CMV pneumonitis | no | no | allogeneic stem cell transplantation 2017 |
| P.280 | 9 | m | atypical SCID (CID) | *DCLRE1C* (ARTEMIS), AR | recurrent bronchitis and pneumonias, granuloma annulare due to rubella virus, bronchiectasis | vitiligo | no | alive |
| P.114 | 51 | m | CVID | *NFKB1*, AD | recurrent gastrointestinal infections (*S. enteritis*), verrucae vulgares, recurrent bronchitis and sinusitis | no | no | alive |
| P.183 | 56 | m | CVID | *NFKB1*, AD | recurrent bronchitis and sinusitis | no | no | alive |
| P.122 | 61 | m | CVID | *NFKB1*, AD | recurrent pneumonias, bronchitis, sinutitis, CMV-colitis (during R-CHOP) | splenomegaly, nodular lymphoid hyperplasia of the gastrointestinal tract, nodular regenerative hyperplasia of the liver, Addison’s disease | NHL, diabetes mellitus type 2 | alive |
| P.150 | 62 | f | CVID | *NFKB1*, AD | recurrent pneumonias, bronchitis, sinusitis, generalized shingles | ITP, lymphadenopathy, splenomegaly | no | alive |
| P.200 | 36 | m | CID | *NFKB1*, AD | recurrent bronchitis and sinusitis | adult-onset Still’s disease, AIHA; lymphadenopathy | no | alive |
| P.210 | 43 | m | CVID | *NFKB1*, AD | recurrent bronchitis and sinusitis, pneumonia and sepsis through *S.pnemoniae* | no | no | alive |
| P.232 | 22 | m | CVID | *NFKB1*, AD | meningococcal meningitis with sepsis, parechovirus meningitis | no | no | alive |
| P.233 | 20 | m | CVID | *NFKB1*, AD | meningococcal meningitis with sepsis | no | no | alive |
| P.254 | 55 | m | unclassified antibody deficiency | *NFKB1*, AD | recurrent bronchitis, recurrent oral and genital herpes | no | no | alive |
| P.26 | 59 | m | CVID | *NFKB2*, AD | pneumonia, recurrent sinutitis | lymphadenopathy, hepatosplenomegaly, psoriasis | no | alive |
| P.181 | 33 | f | CVID | *NFKB2*, AD | recurrent bronchitis, severe pneumonia | hypothyroidism, neurodermatitis | bronchial asthma | alive |
| P.236 | 37 | m | CID | *NFKBIA*, AD | recurrent gastrointestinal infections (*S. flexneri*, *C. jejuni*), recurrent bronchitis, sinusitis, otitis media, recurrent pneumonias (*S. pneumoniae, H. influenza*), meningitis (*N. meningitidis*), CNS tuberculosis with brain abscess, verruca vulgaris (HPV 9 and 57), bronchiectasis | juvenile seropositive rheumatoid arthritis | no | alive |
| P.154 | 26 | m | CVID | *STAT3*-GOF, AD | bacterial tracheitis | multiple inflammatory flocks in brain, lungs and on the thorax wall | sun allergy | alive |
| P.278 | 17 | m | CID | *STAT3*-GOF, AD | recurrent pneumonia, bronchiolitis | severe eczema, inflammatory bowel disease, polyarthritis, advanced lung fibrosis; systemic lymphadenopathy, alopecia, , insulin-dependent diabetes | failure to thrive | decaesed |
| P.282 | 16 | m | CVID | *STAT3*-GOF, AD | recurrent pneumonia, bronchiolitis, otitis externa | collagenous colitis, autoimmune cytopenia, alopecia, lymphadenopathy, splenomegaly | failure to thrive | alive |
| P.265 | 72 | m | CID | *STAT3*-GOF, AD | no | GLILD, arthritis, lymphadenopathy | no | alive |
| P.227 | 36 | f | CID | *STAT3*-GOF, AD | chronic sinusitis, bronchiectasis | GLILD, lymphadenopathy, AIH | no | alive |
| P.246 | 32 | f | HIES | *STAT3*-LOF, AD | recurrent cutaneous abscesses | severe atopic dermatitis | no | alive |
| P.56 | 32 | m | CVID | *CTLA4*, AD | chronic sinusitis, bronchiectasis | enteropathy, AIHA, ITP, splenomegaly, granulomatous colitis | osteoporosis; obesity | deceased |
| P.165 | 33 | f | CID | *CTLA4*, AD | chronic diarrhea (*C. jejuni*), warts, recurrent herpes labialis | enteropathy, ITP, JIA, neurodermitis | no | alive |
| P.215 | 23 | f | CVID | *CTLA4*, AD | recurrent bronchiolitis; chronic diarrhea (*G. lamblia*) | pangastritis | no | alive |
| P.263 | 29 | f | CVID | *IRF2BP2*, AD | recurrent bronchitis and sinusitis, bronchiectasis | no | no | alive |
| P.159 | 27 | f | CVID | *IRF2BP2*, AD | recurrent pneumonias | psoriasis, allergy | idiopathic fulminant hepatitis | deceaced |
| p.257 | 55 | f | CVID | *RELA*, AD | recurrent infections | diverticulitis | vulvar carcinoma | alive |
| P.101 | 61 | m | CID | *STAT1*, AD | CMC, recurrent herpes zoster | diabetes mellitus, AIHA | multiple aneurysms; plattenepithelkarzinom | alive |
| P.262 | 53 | m | CID | *STAT1*, AD | CMC, *mycobacterium genavense*, recurrent sinusitis | no | no | alive |
| P.164 | 59 | m | CVID | *TCF3*, AD | pneumonia, CMV pneumonitis, enterocolitis (*C. difficile*) | lymphadenopathy, splenomegaly | no | lost to follow-up |
| P.166 | 26 | m | CVID | *TCF3*, AD | recurrent sinusitis; herpes zoster; | no | chronic eczema | alive |
| P.270 | 18 | m | CVID | *MYH9*, AD | pseudomonas aeruginosa pneumonia | IDDM | failure to thrive, nephrotic syndrome | deceased |
| P.253 | 48 | f | CVID | *VAV1*, AD | recurrent bronchitis, bronchiectasis | GLILD, ITP, splenomegaly, hypothyroidism, alopecia | no | alive |
| P.294 | 26 | f | unclassified antibody deficiency | *GATA2*, AD | recurrent bronchitis | lymphadenopathy | no | alive |
| P.267 | 20 | f | CID | *SEC61A2*, AD | pneumonia, fungal infections, onchomycosis, dermatomycosis, verrucae vulgares | no | failure to thrive, retardation, exanthema | alive |
| P.63 | 55 | f | CVID | *PIK3R1*, AD | recurrent sinopulmonary infections | no | no | alive |
| P.127 | 30 | m | unclassified antibody deficiency | *PIK3R4* | recurrent sinopulmonary infections | lymphadenopathy | no | lost to follow-up |
| P.124 | 31 | f | CID | *SOCS1*, AD | recurrent bronchitis, sinusitis, bronchiectasis | ITP, arthritis, enteropathy, lymphadenopathy, splenomegaly | panniculitis | alive |
| P.135 | 52 | f | CVID | *PIK3R1*, AD | recurrent bronchitis, recurrent fungal skin infections, rheumatic fever | hypothyroidism | allergy | alive |
| P.182 | 23 | m | CVID | 2x *RELB* variants | recurrent pneumonias and bronchitis | no | no | alive |
| P.264 | 68 | m | unclassified antibody deficiency | *REL*, AD | chronic bronchitis; recurrent herpes infections | no | no | alive |
| P.266 | 15 | f | CVID | *PIK3R4* | recurrent pneumonias and bronchitis | no | AML |  |
| P.279 | 18 | m | agammaglobulinemia | *BTK*, X-linked | chronic cough, chronic diarrhea (*G. lamblia*), bronchiectasis | no | no | alive |
| P.64 | 48 | m | CID | *RAG1*, compound heterozygous | PML (JC-virus), bronchiectasis | AIHA, splenomegaly, vasculitis, vitiligo | no | deceased |
| P.108 | 31 | f | CVID | 2x *USP8* variants | recurrent infections | no | no | alive |
| AD, autosomal dominant; AIH, autoimmune hepatitis; AIHA, autoimmune hemolytic anemia; AML, acute myeloid leukemia; AR, autosomal recessive; f, female; CID, combined immunodeficiency; CMC, chronic mucocutaneous candidiasis; CVID, common variable immunodeficiency; GLILD, Granulomatous–lymphocytic interstitial lung disease; GOF, gain-of-function; IDDM, insulin-dependent diabetes mellitus; ILD, interstitial lung disease; ITP, idiopathic thrombocytopenic purpura; JIA, juvenile idiopathic arthritis; LOF, loss-of-function; m, male; Pat. ID, patient’s identification number; PML, progressive multifocal leukoencephalopathy; SCID, severe combined immunodeficiency  \*patients from same consanguineous family (Atschekzei F et al., J Clin Immunol. 2019) | | | | | | | | |