

Patient	Genetic disorder	Age (years)	Sex	Immunophenotypic abnormalities	Infections	Non-infectious clinical features	Immunoglobulin replacement	Antibiotic prophylaxis	Immunosuppressive therapy	Other treatment	Mortality	Cause of death
1	Akaza telangiectasia	16.3	Female	Low T and B cells, low IgG, A and M.	Recurrent respiratory infections, ear infections, toe nail fungal infection.	Akasia, speech problem, mydriasis, eye telangiectasia, skin pruritus, thoracolumbar scoliosis, fatty liver, gastritis, gallbladder polyp, transaminitis, obesity, vitamin D deficiency, impaired glucose tolerance.	Yes	Yes			No	
2	Akaza telangiectasia	12.6	Female	Low T and B cells, low IgG and A.	Recurrent respiratory infections, impetigo.	Transaminitis, mydriasis, fatty liver, slow learner, delayed motor skills, bulbar compressed telangiectasia, scoliosis.	Yes	Yes			No	
3	Akaza telangiectasia	20.3	Male	Low T cells, low IgG, A and M.	Pneumonia, neutropenic sepsis.	Right cerebral infection, cerebral venous thrombosis, calf muscle tightness, vitamin D deficiency, transaminitis, lymphoma.	Yes	No	Chemotherapy		No	
4	Akaza telangiectasia	22.9	Male	Low T and B cells, low IgG, A and M.	Pneumonia, sepsis.	Acute necrosis of both legs, vitamin D deficiency, long QT syndrome, dystonia, thoracolumbar scoliosis, skin ataxia, myopia, lymphoma.	Yes	No	Chemotherapy		No	
5	DiGeorge syndrome	12.8	Male	Low T, B and NK cells, raised double negative T cells, low IgG and G4, inadequate vaccine responses, autoimmune neutropenia, lymphopenia and thrombocytopenia, MBL deficiency, allergic reaction to penicillin and penicillin.	Bronchiectasis, recurrent respiratory infections, recurrent skin infections, subacute bacterial endocarditis, gastroenteritis, C difficile infection.	Truncus arteriosus, ventricular septal defect, developmental delay, learning difficulty.	Yes	Yes	Rituximab, prednisolone, mycophenolate mofetil		Yes	NA
6	DiGeorge syndrome	13.3	Male	Severe combined immune deficiency, absent T cells, absent PHA responses, low IgG, A and M, autoimmune haemolytic anaemia, alopecia areata, autoimmune thrombocytopenia.	Oral candidiasis, recurrent respiratory infection, C difficile infection, sapovirus, adenovirus and norovirus gastrointestinal infection, enterovirus infection, parvovirus.	Hypoparathyroidism, femoral fracture, scoliosis, gene vilgum, raised alanine transaminase, hepatomegaly, iron overload due to frequent transfusion, myopic astigmatism, osteopenia, areolar developmental delay, tetralogy of fallot, pulmonary artery stenosis, urinary calculus, developmental delay.	Yes	Yes	Bortezomib, Rituximab, prednisolone, sirolimus, mycophenolate mofetil	Thymic transplant	No	
7	DiGeorge syndrome	21.9	Female	Low B cells, IgG, A and M, poor vaccine responses, autoimmune cytopaenias (neutrophil, platelet, anaemia), EBV driven T and B lymphoproliferative disease.	Bronchiectasis.	Cleft palate, pulmonary stenosis, ventricular septal defect, learning difficulty, ADHD, primary nocturnal enuresis, vertigo, thoracic scoliosis, autoimmune hypothyroidism, developmental delay.	Yes	Yes			No	
8	DiGeorge syndrome	21.4	Male	Low B cells, IgG, A and M, poor vaccine responses, autoimmune cytopaenias (neutrophil, platelet, anaemia), EBV driven T and B lymphoproliferative disease.	Bronchiectasis, otitis externa.	Cleft palate, iron deficiency anaemia, speech delay, learning difficulties, developmental delay.	Yes	Yes	Rituximab		No	
9	DiGeorge syndrome	23.0	Female	Autoimmune haemolytic anaemia and thrombocytopenia, autoimmune hepatitis, polymorphic duct lymphoproliferative disease, low B cells, IgG and M (following lymphoproliferative disease treatment).	Grain infection, otitis media, cellulitis.	Interrupted aortic arch, ventricular septal defect, tricuspid regurgitation, bicuspid aortic valve, cleft palate, lymphangitis, liver fibrosis, learning difficulties, allergic rhinitis.	Yes	Yes	Prednisolone, mycophenolate mofetil, cyclosporin, tacrolimus, Rituximab, cyclophosphamide.		No	
10	Trichophaga telangiectasia	5.1	Male	Poor vaccine responses, interferonopathy.	Recurrent respiratory infections.	Cerebral perforation, inflammatory bowel disease, chronic diarrhoea, recurrent fracture, visual impairment, patent ductus arteriosus, developmental delay, horseshoe kidney, scalp eczema, right exotropia, hernia, renal tubular acidosis, vitamin D deficiency, hepatomegaly, congenital jaundice.	Yes	No	Prednisolone, baricitinib, mycophenolate mofetil		No	
11	Trichophaga telangiectasia	17.5	Male	Low B cells, low IgG, A and M, poor vaccine responses.	Recurrent ear infections, recurrent respiratory infections, skin infection, conjunctivitis.	Mouth ulcers, chronic diarrhoea, nocturnal enuresis, pancreatitis, joint pain and hypermobility, eczema, splenomegaly.	Yes	Yes	Mycophenolate mofetil		No	
12	Noonan syndrome	13.4	Male	Low IgG, absent MBL, common variable immune deficiency.	Recurrent respiratory infections, urinary tract infection.	Primary amenorrhea, long QT syndrome, neurobehavioural problem, asthma, allergic rhinitis, type 1 diabetes mellitus, sleep problem, arthralgia, easy bruising, dyspraxia, sensory processing disorder, autism, learning difficulties, cleft au fat spots, multiple drug allergy, cyclical neutropenia.	Yes	Yes			No	
13	Trisomy 22	15.8	Male	Low T cells, low IgG, poor vaccine response.	Recurrent respiratory infections, recurrent urinary tract infections.	Cleft lung hypoplasia, imperforate anus, asplenia, total anomalous pulmonary venous drainage, pulmonary stenosis, left atrial isomerism, gut malrotation, megacolonitis, ureteric dilatation, hypertension, ADHD, asthma.	Yes	Yes			No	
14	Trichophaga telangiectasia/ Cockayne syndrome complex	8.6	Female	Low IgG, intermittent neutropenia.	Recurrent respiratory infections, adenovirus gastroenteritis, Enterococcus urinary tract infection.	Trichophaga telangiectasia, ichthyosis, myopia, exotropia, microphthalmia, horizontal nystagmus, cataract, delayed brain maturation, developmental delay, myoclonic movements, photosensitivity, osteosclerosis, raised alanine transaminase, lymphitis.	Yes	Yes			No	
15	Myhre syndrome	11.8	Male	Low IgG.	Recurrent respiratory infections, recurrent severe norovirus infection, purulent rhinitis, scalp fungal infection, conjunctivitis, dental abscess.	ADHD, global developmental delay, stiff joints, thumb deformity, sleep problem, coarctation of aorta, precocious puberty, vitamin D deficiency.	Yes	Yes			No	
16	Arfoides Tham syndrome	13.0	Female	T cell lymphopenia, low IgG, MBL deficiency, intermittent neutropenia, poor vaccine response.	Recurrent/persistent wet cough & respiratory infections, otitis media, mastoiditis, recurrent urinary tract infections.	ADHD, learning difficulty, diarrhoea, constipation, sleep problem, recurrent seizures, long QT syndrome, hypermobility, conductive hearing loss, myopia, vitamin B deficiency, humeral fracture, incoordination, myelochondrial cytopathy, swallowing problem, developmental delay, motor difficulties.	Yes	No			No	
17	Sideroblastic anaemia with B cell immunodeficiency, periodic fevers and developmental delay (SFD syndrome)	4.9	Male	Low B cells, low IgG, A and M.	HHV8 viraemia, sepsis, skin and soft tissue infection.	Anaemia, splenomegaly, eczema, arthritis, chronic diarrhoea, hepatitis, developmental delay, refractive error, dacrylitis, raised amyloid, thrombocytosis.	Yes	No	Anakinra, etanercept		No	
18	Supernumerary ring chromosome 10 syndrome	2.0	Male	T cell dysfunction, failure of T cell proliferation to mitogens, low CD8 and naive T cells, absent S cells, high NK cells, absent IgG, A and M, thrombocytopenia.	Recurrent respiratory infections, sapovirus, rotavirus infection, MRSA colonization, fungal urinary tract infection, E. coli sepsis.	Ecema, failure to thrive, congenital hypothyroidism, bilateral developmental dysplastic hips, bilateral small kidneys, pancreatic insufficiency, dermatitis, developmental delay, microphthalmia, bilateral cryptorchidism, raised alanine transaminase, gastroesophageal reflux disease, recurrent diarrhoea, thrombocytopenia.	Yes	Yes			Yes	NA
19	Kabuki syndrome	22.8	Female	Low IgG, undetectable IgM, thrombocytopenia.	Bronchiectasis, recurrent respiratory infections, chronic glue ear, urinary tract infection, severe chicken pox, shingles.	Scoliosis, learning difficulty, horseshoe kidney, hyperostosis, cystic liver with transaminitis, short stature, cleft palate, thrombocytopenia.	Yes	Yes			No	
20	Cartilage Hair Hypoplasia	15.3	Male	T cell cytopenia, y6 T cell expansion, very low TRECs, absent PHA response, anaemia.	Bronchiectasis, oral/bonophagal candidiasis, otitis media, shingles, norovirus gastroenteritis.	Fatty liver, hypocalcaemia, chronic diarrhoea, biliary destruction, malabsorption, iron deficiency anaemia, subcutaneous granulomatous, arthralgia, sleep disorder, short stature, uveitis, intussusception.	Yes	Yes			No	Pneumonia
21	Sideroblastic anaemia with B cell immunodeficiency, periodic fevers and developmental delay (SFD syndrome)	1.6	Female	Absent B cells, agammaglobulinemia, severe anaemia, recurrent red cell transfusion.	Norovirus gastroenteritis, E. faecium sepsis, norovirus pneumonitis.	Chronic diarrhoea, pulmonary hypertension, distal renal tubular acidosis, developmental delay, anaemia.	Yes	Yes	Anakinra	HSC	Yes	Norovirus infection, global encephalitis with widespread infection
22	Rothman syndrome	6.6	Female	Low B cells, low IgG, A and M.	Ecema herpeticum, shingles, norovirus gastroenteritis.	Cow's milk protein intolerance, eczema herpeticum, intrauterine growth retardation, dacrylitis, developmental delay, right hemiplegia, hypermobility, acute disseminated encephalomyelitis.	Yes	No			No	
23	Chromosome 2p36.3 deletion	8.2	Male	Low IgG.	Recurrent respiratory and skin infections.	Tourette syndrome, Asperger syndrome, mouth ulcers, motor and sensory difficulties.	No	Yes			No	
24	Wiskott-Aldrich syndrome	21.8	Male	Low IgG, thrombocytopenia.	Norovirus gastroenteritis, severe chicken pox, pinna cellulitis and pre-auricular abscess, chronic cough, pneumonia, S. aureus central ear abscess.	Recurrent scleritis, splenomegaly, allergic rhinitis and eczema, recurrent arthralgia, thrombocytopenia, glomerulonephritis.	Yes	Yes	Lentiviral gene therapy		No	
25	Wiskott-Aldrich syndrome	5.7	Male	Low IgG, thrombocytopenia.	Recurrent respiratory infections, norovirus and sapovirus gastroenteritis, otitis media and externa, adenovirus, chicken pox, corditis cap, oral thrush, conjunctivitis.	Charcot-Marie-Tooth syndrome (asymptomatic), hepatitis, eczema, chronic diarrhoea, and ulcer, speech delay, cow's milk protein allergy, thrombocytopenia.	Yes	Yes	HSC (complicated by grade 3 GVHD)		No	
26	Wiskott-Aldrich syndrome	3.1	Male	Low IgG, autoimmune thrombocytopenia.	EBV viraemia, E. coli and Enterococcus central line sepsis, C difficile infection.	Ecema, anaemia.	Yes	Yes	HSC		No	
27	Hyper IgE syndrome (STAT3 deficiency)	27.0	Female	Low IgA, poor vaccine responses, reduced Th17, raised IgE, reduced CD4HL17 and CD4HL17.	Recurrent respiratory infections, skin and soft tissue infections and abscesses, otitis media and externa, orbital cellulitis, dental infection, mediastinal abscess, otitis myringoid abscess.	Paratracheal air cyst/diaphragmatic, pathological fracture, retention of deciduous teeth, pulmonary embolism and internal jugular thrombosis.	Yes	Yes			No	
28	Hyper IgE syndrome (STAT3 deficiency)	25.0	Female	Low IgA.	MRSA recurrent skin and soft tissue infection and abscesses, fungal nail infection, recurrent otitis.	Ecema, scoliosis, joint hypermobility, retained primary teeth, fractures, ostheitis.	Yes	Yes			No	
29	Hyper IgE syndrome (STAT3 deficiency)	24.0	Male	IgE >5000 kU/L.	Recurrent otitis media infections, corditis cap, S. aureus skin and soft tissue infection and abscess.	Fracture, hypermobility, dermatitis/eczema, chronic rhinitis, osteopenia, constipation, hypotonia, delayed motor milestones.	Yes	Yes			No	
30	Sideroblastic anaemia with B cell immunodeficiency, periodic fevers and developmental delay (SFD syndrome)	1.3	Male	Low B cells and low IgG.	S. epidermidis central line infection.	Proximal renal tubulopathy, dilated pulmonary artery and aortic root with small ventricular septal defect, carnitine deficiency, chronic diarrhoea, pancreatic insufficiency, meconium ileus, developmental delay.	Yes	No	Etanercept		Yes	Covid-19, cerebral edema
31	Wiskott-Aldrich syndrome	2.8	Male	Normal immunology. Persistent and profound thrombocytopenia - probable autoimmune thrombocytopenia (some increase in platelet count after steroid treatment, and HIV; rapid loss of platelets following platelet transfusion).		Ecema, thrombocytopenia.	No	No	Rituximab, prednisolone.		No	
32	Hyper IgE syndrome (STAT3 deficiency)	17.0	Female	IgE >2000 kU/L.	Recurrent unopulmonary infections, skin infections and abscesses, otitis media, lymphadenitis, lung abscess and empyema.	Pathological fractures, thoracic lumbar scoliosis, joint laxity, follicular hyperkeratosis/acrokeratosis pilaris, eczema.	No	No			No	
33	MRK22 syndrome	16.8	Male	Low B cells, low IgG, suboptimal vaccine responses.	Persistent wet cough.	Intrauterine growth restriction, glomerulonephritis, ambiguous genitalia, developmental delay, gastroesophageal reflux, got dysmimotony.	No	Yes			No	
34	Kabuki syndrome	9.2	Female	Low IgA.	Pyelonephritis, otitis media and recurrent otitis and respiratory infections.	Dysplastic kidneys, chronic kidney disease, hypertension, global developmental delay, bilateral dysplastic hips, joint hypermobility.	No	No			No	
35	Kabuki syndrome	6.6	Female	Slightly reduced CD8 and B cells, low IgG.	Pneumonia.	Complex congenital heart disease, developmental delay, developmental dysplasia of the hip, congenital hypothyroidism.	No	No			No	
36	Rothman syndrome	8.0	Male	Low B cells, reduced T cell proliferation to PHA/Concanavalin, low IgG, A and M, poor vaccine response (Diphtheria, Tetanus, Measles, Mumps).	Severe RSV pneumonia, enterovirus meningitis, recurrent respiratory infections, candidiasis.	Hypothyroidism, developmental delay, tracheobronchomalacia, paronychia, syndactyl/phalangial dysplasia, hypermetriosis, mydriasis, ventriculomegaly.	Yes	No			No	

#### ABBREVIATIONS

ADHD Attention deficit hyperactivity disorder  
EBV Epstein-Barr virus  
GVHD graft versus host disease  
HHV Human herpesvirus  
HCT Haematopoietic stem cell transplantation  
Ig immunoglobulin  
intereven immunoglobulin  
MBL mannose binding lectin  
NA not available  
PHA phytohemagglutinin  
PMA Phorbol myristate acetate  
RSV Respiratory syncytial virus  
TREC T cell receptor excision circles