Table S1. **Genotype and phenotype information on the patients in the study**

Age/sex	Disease Mutation Protein Domain	CMC?	Other infectious history	Other clinical features
25/M	STAT3 LOF c.1027 G>T DNA binding	No	Recurrent skin abscess Pneumonia with pneumatocele formation Recurrent otitis media Cellulitis	Newborn rash Retained primary teeth Osteopenia with multiple fractures Scoliosis
37/M	STAT3 LOF c.1110delAG DNA binding	Yes	Recurrent skin abscesses Recurrent sinopulmonary infections with pneumatocele formation requiring right lower lobectomy	Scoliosis Retained primary teeth Asthma Osteoporosis Minimal trauma fractures
31/M	STAT3 LOF c.1144 C>T DNA binding	No	Recurrent skin abscesses Recurrent pneumonia and sinusitis Herpes zoster	Newborn rash Eosinophilic esophagitis Alopecia areata Raynaud Hypothyroidism Allergic asthma Scoliosis Multiple fractures
11/F	STAT3 LOF c.1144 C>T DNA binding	Yes	Recurrent pneumonia Skin abscesses	Newborn rash
31/M	STAT3 LOF c.1144 C>T DNA binding	No	Recurrent skin abscesses Recurrent sinusitis and PNA	Newborn rash Hypothyroidism Esophageal ulcers Multiple fractures Retained primary teeth Asthma
19/F	STAT3 LOF c.1387delGTG DNA binding	No	Pneumonia Warts Recurrent sinus and ear infections Skin abscesses	Newborn rash Retained primary teeth Asthma Warts Hyperextensibility
46/F	STAT3 LOF c.1393 T>G DNA binding	Yes	Recurrent sinopulmonary infections Recurrent skin infections	Esophageal strictures Retained primary teeth Fractures

Age/sex	Disease Mutation Protein Domain	CMC?	Other infectious history	Other clinical features
14/F	STAT3 LOF c.1394 C>T DNA binding	Yes	Recurrent pneumonia Mediastinal abscess	Allergic asthma
46/F	STAT3 LOF c.1861 T>G SH2	Yes	Recurrent PNA with pneumatocele Osteomyelitis	Recurrent minimal trauma fractures Scoliosis
20/F	STAT3 LOF c.1909G>A SH2	Yes	Staphylococcal skin infections Recurrent otitis media Recurrent sinusitis Invasive *Candida* sinusitis Infectious Mononucleosis	Newborn rash Minimal trauma fractures Retained primary teeth
47/F	STAT3 LOF c.1909 G>A SH2	Yes	Chronic sinusitis Fungal pneumonia Recurrent bacterial pneumonia with cavitation s/p right upper lobe resection	Osteopenia Coronary aneurysms
13/F	STAT3 LOF c.1909G>A SH2	Yes	Recurrent skin infections Recurrent otitis media	Newborn rash Delayed dental eruption Retained primary teeth Recurrent minimal trauma fractures Recurrent OM
25/M	STAT3 LOF c.1909 G>T SH2	Yes	Recurrent pneumonia Skin abscesses	None
16/F	STAT3 LOF c.1972 A>G SH2	Yes	Recurrent otitis media Recurrent sinusitis Recurrent pneumonia Septic arthritis Cervical lyphadenitis	Pulmonary stenosis Hip dysplasia Systemic lupus erythematosus
12/F	STAT3 LOF c.2117 T>C Transactivation	Yes	Recurrent pneumonia Recurrent upper respiratory infections Recurrent otitis media Orbital abscess Subdural empyema Skin infections Invasive *Candida* pneumonia	Newborn rash Retained primary teeth Osteopenia Scoliosis Eosinophilic GI disease Allergic rhinitis Food allergy
13/F	STAT1GOF c.467 T>C coiled-coil	Yes	Recurrent urinary tract infections Molluscum contagiosum	Crohn's disease

Age/sex	Disease Mutation Protein Domain	CMC?	Other infectious history	Other clinical features
36/F	STAT1 GOF c.467 T>C coiled-coil	Yes		Congenital midgut malrotation Hypothyroidism Recurrent UTIS, sinopulmonary infections
30/M	STAT1 GOF c.493 G>C coiled-coil	Yes	Recurrent otitis media Shingles	Type 1 diabetes mellitus
34/F	STAT1 GOF c.704 A>C coiled-coil	Yes	Recurrent pneumonia with bronchiectasis Recurrent sinusitis	None
60/F	STAT1 GOF c.704 A>C coiled-coil	Yes	Recurrent sinusitis Recurrent urinary tract infections Pneumonia vaginitis Recurrent otitis media Skin boils Herpes simplex virus oral ulcers Shingles Molluscum contagiosum	basal cell and squamous cell skin cancer hemangiomas aneurysms
9/M	STAT1 GOF c.983 A>G DNA binding	No	Recurrent lung infections	Hypothyroidism Insulin dependent diabetes mellitus Growth hormone deficiency Failure to thrive Enteropathy
27/F	STAT1 GOF c.1057 G>A DNA binding	No	Disseminated coccidiomycosis Herpes simplex virus oral ulcers	
4/M	STAT1 GOF c.1073 T>G DNA binding	Yes	Herpes simplex virus otitis media Recurrent sinusitis Skin infections	Type 1 diabetes mellitus Hypothyroidism Enteropathy Bronchiectasis Celiac disease Developmental delay Cerebral aneurysm
13/F	STAT1 GOF c.1154 C>T DNA binding	Yes	Respiratory syncytial virus bronchiolitis Mycobacterial skin infection Shingles Recurrent pneumonia Recurrent otitis media	Insulin dependent diabetes mellitus Vasculopathy/aneurysms Bronchiectasis

Age/sex	Disease Mutation Protein Domain	CMC?	Other infectious history	Other clinical features
26/F	STAT1 GOF c.1885C>T SH2	Yes	Recurrent sinusitis and necrotizing pneumonia complicated by empyema and bronchopleural fistulae Recurrent herpes zoster	CD4 and B cell lymphopenia Hypogammaglobulinemia on IVIG Retained primary dentition Hypothyroidism