**Additional material descriptions:**

**Additional file 1. Overview of all clinical characteristics of the patients included in our diagnostic PID cohort, including all immunophenotype characteristics.**

**Bacterial acronyms:** AB = Acinetobacter Baumannii; BCG = Bacillus Calmette-Guérin; BAF = Bacteroide fragalis; BHSC = Beta-hemolytic Streptococcus group C; BOP = Bortedella Pertussis; BP = Burkholderia Pseudomallei; CDF = Clostridium Difficile; COB = Coxiella burnetii (Q-fever); EC = Escherchia Coli; ENC = Enterobacter Cloacae; ENG = Enterobacter Gergoviae; EFM = Enterococcus Faecium; EFS = Enterococcus Faecalis; GBS = Group B Streptococcus; HI = Heamophilus Influenzae; HPI = Heamophilus Parainfluenzae; KOX = Klebsiella Oxytoca; KP = Klebsiella Pneumoniae; MC = Moraxella catarrhalis; MRSA = Methicilin-Resistant Staphylococcus Aureus; MY = Mycobacteria; NM = Neisseria Meningitidis; PRB = Proprionibacterium ; PRT = Proteus; PSA = Pseudomonas Aeruginosa; SA = Staphylococcus Aureus; SE = Staphylococcus Epidermidis; SH = Staphylococcus Hominis; SHC = Staphylococcus Heamolyticus; SM = Serratia Marcescens; SAL = Salmonella; SO = Streptococcus Oralis; SP = Streptococcus Pneumoniae; SV = Streptococcus Viridans; TW = Tropheryma Whipplei; X = Not specified

**Viral acronyms:** CMV = CytoMegaloVirus; EBV = Epstein Barr Virus; HAdV = Human AdenoVirus; HBoV = Human BocaVirus; HCoV = Human CoronaVirus; HPV = Human Pappiloma Virus; HPIV = Human ParaInfluenzaVirus; HRV = Human RhinoVirus; HSV = Herpes Simplex Virus; IFV = Influenza Virus; RSV = Respiratory Syncytial Virus; RV = Rotavirus; VZV = Varicella Zoster Virus; A = Type A; B = Type B; 1 = Type 1; 2 = Type 2; 3 = Type 3; 4 = Type 4; X = Not specified

**Fungal or parasitic acronyms:** A = Candida Albicans; G = Glabrata; CAT = Tropicalis; DF = Dientamoeba Fragalis; MF = Malassezia Furfur; PJ = Pneumocystis Jirovecii; X = Not specified

**Autoimmune or autoinflammatory acronyms:** AA = Alopecia Areata ; AIM = Autoimmune; AIF = Autoinflammatory; ANCASV = Anti-Neutrophil Cytoplasmic Antibody systemic vasculitis ; ALPS = Autoimmune Lymphoproliferative Syndrome; APS = Antiphospholipid syndrome; ARTH = Arthritis; ARTHA = Aseptic Arthritis ; ARTHPJ = Polyarthritis Juvenalis; CD = Celiac Disease ; DM1 = Diabetes Mellitus type 1; EnP = Enteropathy; GLILD = Granulomatous-Lymphocytic Interstitial Lung Disease ; Hypo = Hypothyroidism; IBD = Inflammatory Bowel Disease; IleitisT = Ileitis Terminalis; LAD = Lymphadenopathy; NP = Nephropathy; SLE = Systemic Lupus Erythematosus; SSc = Systemic Sclerosis; X = Not specified

**Infection site acronyms:** CNS = Central Nervous System; ENT = Ear Nose Throat

**Additional features acronyms:** ADHD = Attention Deficit Hyperactivity Disorder; ASD = Autism Spectrum Disorder; BMF = Bone Marrow Failure; CHD = Coronary Heart Disease; DD = Developmental Delay; EPI = Exocrine Pancreatic Insufficiency; FD = Feeding Difficulties; FTT = Failure To Thrive; GERD = Gastroesophagal Reflux Disease; †= patient deceased

**Expected disease acronyms:** ALPS = Autoimmune LymphoProliferative Syndrome; APECED = Autoimmune PolyEndocrinopathy-Candidiasis-Ectodermal-Dystrophy; CVID = Common Variable Immunodeficiency; CMC = Chronic Mucocutaneous Candidiasis; GLILD= Granulomatous Lymphocytic Interstitial Lung Disease; HGG = Hypogammaglobulinemia; HLH = Hemophagocytic Lymphohistiocytosis; HSV = Herpes Simplex Virus; IPEX = Immunodysregulation Polyendocrinopathy Enteropathy X-linked syndrome; MSMD = Mendelian Susceptibility to Mycobacterial Diseases; PAPA = Pyogenic Arthritis Pyoderma gangrenosum and Acne; PFAPA = Periodic Fever Aphthous Stomatitis Pharyngitis Adenitis Syndrome; SCID = Severe Combined Immunodeficiency; SLE= Systemic Lupus Erythematosus; TORCH = toxoplasmosis, other infections, rubella, cytomegalovirus infection, and herpes simplex

**Indications of Cytopenias:** A = Aplastic anemia; AIH = AutoImmune Hemolytic Anemia; CN = Cyclic neutropenia; EO = Eosinopenia; H = Hemolytic anemia; IP = Immune thrombocytopenic purpura; N = Neutropenia; NC = Normocytic anemia; TR = Transient; X = Yes, type not specified

**Additional file 2. Shows all causative mutations identified in 72 patients from 68 families suffering from primary immunodeficiencies.** These 72 are part of a diagnostically tested cohort of 254 PID patients from 249 families. The “variant class” column also indicates variants that were identified by exome-wide analysis. ‡the homozygous mutation was identified in one of the large homozygous regions. Abbreviations used: AR = Autosomal Recessive; AD = Autosomal Dominant; XL = X-linked; hom = homozygous; CH = Compound heterozygous; APECED = Autoimmune PolyEndocrinopathy-Candidiasis-Ectodermal-Dystrophy; CVID = Common Variable Immunodeficiency; HLH = Hemophagocytic Lymphohistiocytosis; PAPA = Pyogenic Arthritis Pyoderma gangrenosum and Acne; SCID = Severe Combined Immunodeficiency; TORCH = toxoplasmosis, other infections, rubella, cytomegalovirus infection, and herpes simplex

**Additional file 3. Shows variants of unknown significance (class 3) and variants in TRAF3 identified in 17 patients suffering from primary immunodeficiencies.** These 17 are part of a diagnostically tested cohort of 254 PID patients from 249 families.The “variant class” column also indicates variants that were identified by exome-wide analysis.\*Only 1 (76.1) of 4 patients carrying a variant in TRAF3 suffered from infections with herpes simplex virus, which might also result form a mutation in GATA2 p.(R86fs/wt). ‡the homozygous mutation was identified in one of the large homozygous regions. Abbreviations used: AR = Autosomal Recessive; AD = Autosomal Dominant; XL = X-linked; hom = homozygous; CH = Compound heterozygous

**Additional file 4.** Provides **(A)** the number of patients with isolated or combined infections, and **(B)** the number of patients with isolated or combined immunophenotypes, and the percentage for which we have reported a genetic diagnosis

**Additional file 5.**

Provides quality information of the WES technology, with the mean target coverage, and the % of bases with >20X coverage.

**Additional file 6.**

Provides information on all large >5mb homozygous regions per patient, detected in the exome. Of each region, the genomic location, size, % homozygous variants, and the detected mutation is provided.

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