Supplementary Table 1.

Patients with Identified PI Defects by Region

Region	Number of Surveys Received	Compliance Rate	Patients with Identifed PI Defects	% of Total
Western Europe	52	82.26%	22,701	37.61%
United States	56	67.06%	15,602	25.85%
Eastern Europe	26	85.19%	5,367	8.89%
Latin America	17	80.95%	4,275	7.08%
Middle East	6	54.55%	3,432	5.69%
Canada	8	88.89%	3,423	5.67%
Asia	17	68.97%	3,127	5.18%
Africa	7	87.50%	1,228	2.03%
Australia	3	100.00%	1,209	2.00%
Total	**192**	**75.59%**	**60,364**	**100.00%**

Supplementary Table 2.

Patients with Identified PI Defects – Global

TABLE I. Combined T and B–cell immunodeficiencies	
ADA Deficiency	298
Artemis Deficiency (DCLREIC)	101
CD3 δ / CD3 ε / CD3 ζ Deficiency	25
Cernunnos Deficiency	10
DNA Ligase IV Deficiency	14
γc Deficiency	454
IL-2Ralpha Deficiency (CD25 Deficiency)	34
IL-7R α Deficiency	98
JAK3 Deficiency	116
MHC Class I or II Deficiency	226
Omenn Syndrome	237
PNP Deficiency	38
RAG 1/2 Deficiency	312
Reticular Dysgenesis (AK2 Deficiency)	15
ZAP-70 Deficiency	51
Other SCID:	1,134
Total	3,163

TABLE II. Other well defined immunodeficiency syndromes	
Ataxia-Telangiectasia (A-T)	2,190
Ataxia-Telangiectasia like Disease (ATLD)	22
Bloom Syndrome	26
Cartilage Hair Hypoplasia	135
Chronic Mucocutaneous Candidiasis	456
Comel-Netherton Syndrome	59
DiGeorge Syndrome (DGS)	4,310
Dyskeratosis Congenita	37
Hyper IgE, AD (STAT3, Job Syndrome)	760
Hyper-IgE, AR (DOCK8, TYK2 Deficiency)	156
ICF-ID Centromeric Instability and Facial Anomalies	15
Nijmegen Breakage Syndrome	227
PMS2 Deficiency	4
Schimke Syndrome	30
Wiskott Aldrich Syndrome (WAS)	1,000
Total	9,427

TABLE III. Diseases of immune dysregulaton	
ALPS Type 1a, CD95 (fas) Defects	242
ALPS, other types	210
APECED	165
Chediak-Higashi Syndrome	174
Griscelli Syndrome Type 2	96
Hermansky Pudlak Syndrome (Type 2; AP3 deficiency)	11
HLH (other types, Munc13-4, Munc 18-2, STX11)	183
HLH Perforin Deficiency	101
IPEX (X-Linked)	80
IPEX-Like Syndrome	77
XLP1, SH2D1A Deficiency	189
XLP2, XIAP Deficiency	25
Total	1,553

TABLE IV. Congenital defects of phagocyte #s and function	
CGD, AR	964
CGD, XL	1,029
Glycogen Storage Disease Type 1b	31
Leukocyte Adhesion Deficiency I	168
Leukocyte Adhesion Deficiency II, III, and others	26
MSMD (IL-12p40, IL12RB, IFN-γR1/2, STAT1 Deficiency)	150
Neutropenia w/ Cardiac + Urogenital Malformations	12
Neutropenia, Cyclical	269
Neutropenia, Severe Congenital (ELA2, HAX1)	505
Neutropenia, XL (WASP mutation)	21
Papillon-Lefèvre Syndrome	14
Total	3,189

TABLE V. Predominantly antibody deficiencies	
Agammaglobulinemia, (XLA)	2,132
Agammaglobulinemia, AR	413
Common Variable Immunodeficiency (CVID)	7,613
Hyper IgM (X Linked, TNFSF5)	465
Hyper IgM, AR (AICDA, UNG, CD40)	304
Hypogammaglobulinemia of Infancy (Transient)	2,713
Hypogammaglobulinemia unspecified	3,148
Ig Heavy Chain Mutations and Deletions	10
IgA Deficiency, Selective	7,061
IgA with IgG Subclass Deficiency	1,055
IgG Subclass Deficiency, Isolated	3,878
Specific Antibody Deficiency (normal Ig and B cells)	2,138
TACI mutation (TNFRSF13B)	159
Thymoma with Immunodeficiency (Good Syndrome)	73
Total	31,162

TABLE VI. Defects in innate immunity	
EDA-ID, AD (NFKBIA mutation)	25
Epidermodysplasia verruciformis (EVER mutation)"	11
Herpes simplex encephalitis HSE (Unc93B, TLR3, others	36
IRAK4 (IL-1 Receptor Associated Kinase 4)	36
MyD88 Deficiency	12
NEMO Deficiency	96
NK cell Deficiency	69
WHIM Syndrome	43
Total	328

TABLE VII. Autoinflammatory disorders	
Blau Syndrome (NOD2 or CARD15)	28
DIRA (IL1RN)	4
Familial Cold Autoinflammatory Syndrome	27
Familial Mediterranean Fever	1,715
Hyper IgD Syndrome	96
Majeed Syndrome (Mutation of LPIN2)	21
Muckle-Wells syndrome	32
NOMID or CINCA	45
PFAPA Syndrome	1,518
TRAPS-TNF Receptor-Associated Periodic Fever	114
Total	3,600

TABLE VIII. Complement deficiencies	
C1 Inhibitor Deficiency	2,045
C1q Deficiency	29
C1r Deficiency	6
C2-C9 Deficiency	580
CD46 Deficiency	10
CR3-Complement Receptor 3 Deficiency	3
Factor D Deficiency	1
Factor H Deficiency	22
Factor I Deficiency	33
MBP/MBL or MASP Deficiency	881
Paroxysmal Nocturnal Hemoglobinuria(PIGA)	22
Properdin Deficiency	20
Total	3,652

"Unspecified" Tables I through VIII:	4,290

Total: 60,364

Supplementary Table 3.

Patients with Identified PI Defects – United States

TABLE I. Combined T and B–cell immunodeficiencies	
ADA Deficiency	99
Artemis Deficiency (DCLREIC)	15
CD3 δ / CD3 ε / CD3 ζ Deficiency	2
Cernunnos Deficiency	1
DNA Ligase IV Deficiency	5
γc Deficiency	135
IL-2Ralpha Deficiency (CD25 Deficiency)	5
IL-7R α Deficiency	34
JAK3 Deficiency	30
MHC Class I or II Deficiency	15
Omenn Syndrome	62
PNP Deficiency	4
RAG 1/2 Deficiency	42
Reticular Dysgenesis (AK2 Deficiency)	6
ZAP-70 Deficiency	18
Other SCID:	135
Total	608

TABLE II. Other well defined immunodeficiency syndromes	
Ataxia-Telangiectasia (A-T)	1,005
Ataxia-Telangiectasia like Disease (ATLD)	11
Bloom Syndrome	1
Cartilage Hair Hypoplasia	57
Chronic Mucocutaneous Candidiasis	90
Comel-Netherton Syndrome	13
DiGeorge Syndrome (DGS)	1,858
Dyskeratosis Congenita	11
Hyper IgE, AD (STAT3, Job Syndrome)	144
Hyper-IgE, AR (DOCK8, TYK2 Deficiency)	19
ICF-ID Centromeric Instability and Facial Anomalies	2
Nijmegen Breakage Syndrome	17
PMS2 Deficiency	0
Schimke Syndrome	4
Wiskott Aldrich Syndrome (WAS)	181
Total	3,413

TABLE III. Diseases of immune dysregulaton	
ALPS Type 1a, CD95 (fas) Defects	43
ALPS, other types	33
APECED	28
Chediak-Higashi Syndrome	19
Griscelli Syndrome Type 2	4
Hermansky Pudlak Syndrome (Type 2; AP3 deficiency)	1
HLH (other types, Munc13-4, Munc 18-2, STX11)	38
HLH Perforin Deficiency	19
IPEX (X-Linked)	28
IPEX-Like Syndrome	26
XLP1, SH2D1A Deficiency	34
XLP2, XIAP Deficiency	9
Total	282

TABLE IV. Congenital defects of phagocyte #s and function	
CGD, AR	105
CGD, XL	198
Glycogen Storage Disease Type 1b	4
Leukocyte Adhesion Deficiency I	20
Leukocyte Adhesion Deficiency II, III, and others	4
MSMD (IL-12p40, IL12RB, IFN-γR1/2, STAT1 Deficiency)	5
Neutropenia w/ Cardiac + Urogenital Malformations	0
Neutropenia, Cyclical	39
Neutropenia, Severe Congenital (ELA2, HAX1)	78
Neutropenia, XL (WASP mutation)	8
Papillon-Lefèvre Syndrome	0
Total	461

TABLE V. Predominantly antibody deficiencies	
Agammaglobulinemia, (XLA)	425
Agammaglobulinemia, AR	66
Common Variable Immunodeficiency (CVID)	2,501
Hyper IgM (X Linked, TNFSF5)	93
Hyper IgM, AR (AICDA, UNG, CD40)	62
Hypogammaglobulinemia of Infancy (Transient)	551
Hypogammaglobulinemia unspecified	1,445
Ig Heavy Chain Mutations and Deletions	6
IgA Deficiency, Selective	1,209
IgA with IgG Subclass Deficiency	172
IgG Subclass Deficiency, Isolated	533
Specific Antibody Deficiency (normal Ig and B cells)	1,278
TACI mutation (TNFRSF13B)	33
Thymoma with Immunodeficiency (Good Syndrome)	14
Total	8,388

TABLE VI. Defects in innate immunity	
EDA-ID, AD (NFKBIA mutation)	5
Epidermodysplasia verruciformis (EVER mutation)"	1
Herpes simplex encephalitis HSE (Unc93B, TLR3, others	3
IRAK4 (IL-1 Receptor Associated Kinase 4)	4
MyD88 Deficiency	6
NEMO Deficiency	49
NK cell Deficiency	44
WHIM Syndrome	6
Total	118

TABLE VII. Autoinflammatory disorders	
Blau Syndrome (NOD2 or CARD15)	7
DIRA (IL1RN)	0
Familial Cold Autoinflammatory Syndrome	16
Familial Mediterranean Fever	100
Hyper IgD Syndrome	22
Majeed Syndrome (Mutation of LPIN2)	1
Muckle-Wells syndrome	5
NOMID or CINCA	3
PFAPA Syndrome	171
TRAPS-TNF Receptor-Associated Periodic Fever	27
Total	352

TABLE VIII. Complement deficiencies	
C1 Inhibitor Deficiency	226
C1q Deficiency	2
C1r Deficiency	3
C2-C9 Deficiency	129
CD46 Deficiency	9
CR3-Complement Receptor 3 Deficiency	0
Factor D Deficiency	1
Factor H Deficiency	3
Factor I Deficiency	3
MBP/MBL or MASP Deficiency	162
Paroxysmal Nocturnal Hemoglobinuria(PIGA)	7
Properdin Deficiency	19
Total	564

"Unspecified" Tables I through VIII:	1,416

Total: 15,602

Supplementary Table 4.

Patients with Identified PI Defects – International

TABLE I. Combined T and B–cell immunodeficiencies	
ADA Deficiency	199
Artemis Deficiency (DCLREIC)	86
CD3 δ / CD3 ε / CD3 ζ Deficiency	23
Cernunnos Deficiency	9
DNA Ligase IV Deficiency	9
γc Deficiency	319
IL-2Ralpha Deficiency (CD25 Deficiency)	29
IL-7R α Deficiency	64
JAK3 Deficiency	86
MHC Class I or II Deficiency	211
Omenn Syndrome	175
PNP Deficiency	34
RAG 1/2 Deficiency	270
Reticular Dysgenesis (AK2 Deficiency)	9
ZAP-70 Deficiency	33
Other SCID:	999
Total	2,555

TABLE II. Other well defined immunodeficiency syndromes	
Ataxia-Telangiectasia (A-T)	1,185
Ataxia-Telangiectasia like Disease (ATLD)	11
Bloom Syndrome	25
Cartilage Hair Hypoplasia	78
Chronic Mucocutaneous Candidiasis	366
Comel-Netherton Syndrome	46
DiGeorge Syndrome (DGS)	2,452
Dyskeratosis Congenita	26
Hyper IgE, AD (STAT3, Job Syndrome)	616
Hyper-IgE, AR (DOCK8, TYK2 Deficiency)	137
ICF-ID Centromeric Instability and Facial Anomalies	13
Nijmegen Breakage Syndrome	210
PMS2 Deficiency	4
Schimke Syndrome	26
Wiskott Aldrich Syndrome (WAS)	819
Total	6,014

TABLE III. Diseases of immune dysregulaton	
ALPS Type 1a, CD95 (fas) Defects	199
ALPS, other types	177
APECED	137
Chediak-Higashi Syndrome	155
Griscelli Syndrome Type 2	92
Hermansky Pudlak Syndrome (Type 2; AP3 deficiency)	10
HLH (other types, Munc13-4, Munc 18-2, STX11)	145
HLH Perforin Deficiency	82
IPEX (X-Linked)	52
IPEX-Like Syndrome	51
XLP1, SH2D1A Deficiency	155
XLP2, XIAP Deficiency	16
Total	1,271

TABLE IV. Congenital defects of phagocyte #s and function	
CGD, AR	859
CGD, XL	831
Glycogen Storage Disease Type 1b	27
Leukocyte Adhesion Deficiency I	148
Leukocyte Adhesion Deficiency II, III, and others	22
MSMD (IL-12p40, IL12RB, IFN-γR1/2, STAT1 Deficiency)	145
Neutropenia w/ Cardiac + Urogenital Malformations	12
Neutropenia, Cyclical	230
Neutropenia, Severe Congenital (ELA2, HAX1)	427
Neutropenia, XL (WASP mutation)	13
Papillon-Lefèvre Syndrome	14
Total	2,728

TABLE V. Predominantly antibody deficiencies	
Agammaglobulinemia, (XLA)	1,707
Agammaglobulinemia, AR	347
Common Variable Immunodeficiency (CVID)	5,112
Hyper IgM (X Linked, TNFSF5)	372
Hyper IgM, AR (AICDA, UNG, CD40)	242
Hypogammaglobulinemia of Infancy (Transient)	2,162
Hypogammaglobulinemia unspecified	1,703
Ig Heavy Chain Mutations and Deletions	4
IgA Deficiency, Selective	5,852
IgA with IgG Subclass Deficiency	883
IgG Subclass Deficiency, Isolated	3,345
Specific Antibody Deficiency (normal Ig and B cells)	860
TACI mutation (TNFRSF13B)	126
Thymoma with Immunodeficiency (Good Syndrome)	59
Total	22,774

TABLE VI. Defects in innate immunity	
EDA-ID, AD (NFKBIA mutation)	20
Epidermodysplasia verruciformis (EVER mutation)"	10
Herpes simplex encephalitis HSE (Unc93B, TLR3, others	33
IRAK4 (IL-1 Receptor Associated Kinase 4)	32
MyD88 Deficiency	6
NEMO Deficiency	47
NK cell Deficiency	25
WHIM Syndrome	37
Total	210

TABLE VII. Autoinflammatory disorders	
Blau Syndrome (NOD2 or CARD15)	21
DIRA (IL1RN)	4
Familial Cold Autoinflammatory Syndrome	11
Familial Mediterranean Fever	1,615
Hyper IgD Syndrome	74
Majeed Syndrome (Mutation of LPIN2)	20
Muckle-Wells syndrome	27
NOMID or CINCA	42
PFAPA Syndrome	1,347
TRAPS-TNF Receptor-Associated Periodic Fever	87
Total	3,248

TABLE VIII. Complement deficiencies	
C1 Inhibitor Deficiency	1,819
C1q Deficiency	27
C1r Deficiency	3
C2-C9 Deficiency	451
CD46 Deficiency	1
CR3-Complement Receptor 3 Deficiency	3
Factor D Deficiency	0
Factor H Deficiency	19
Factor I Deficiency	30
MBP/MBL or MASP Deficiency	719
Paroxysmal Nocturnal Hemoglobinuria(PIGA)	15
Properdin Deficiency	1
Total	3,088

"Unspecified" Tables I through VIII:	2,874

Total: 44,762

Supplementary Table 5.

PI Defects Listed by Prevalence

1	Common Variable Immunodeficiency (CVID)	7,613	2,501	5,112
2	IgA Deficiency, Selective	7,061	1,209	5,852
3	DiGeorge Syndrome (DGS)	4,310	1,858	2,452
4	IgG Subclass Deficiency, Isolated	3,878	533	3,345
5	Combined T and B–cell immunodeficiencies	3,163	66	2,555
6	Hypogammaglobulinemia unspecified	3,148	1,445	1,703
7	Hypogammaglobulinemia of Infancy (Transient)	2,713	551	2,162
8	Ataxia-Telangiectasia (A-T)	2,190	1,005	1,185
9	Specific Antibody Deficiency (normal Ig and B cells)	2,138	1,278	860
10	Agammaglobulinemia, (XLA)	2,132	425	1,707
11	C1 Inhibitor Deficiency	2,045	226	1,819
12	Familial Mediterranean Fever	1,715	100	1,615
13	PFAPA Syndrome	1,518	171	1,347
14	IgA with IgG Subclass Deficiency	1,055	172	883
15	CGD, XL	1,029	198	831
16	Wiskott Aldrich Syndrome (WAS)	1,000	181	819
17	CGD, AR	964	105	859
18	MBP/MBL or MASP Deficiency	881	162	719
19	Hyper IgE, AD (STAT3, Job Syndrome)	760	144	616
20	C2-C9 Deficiency	580	129	451
21	Neutropenia, Severe Congenital (ELA2, HAX1)	505	78	427
22	Hyper IgM (X Linked, TNFSF5)	465	93	372
23	Chronic Mucocutaneous Candidiasis	456	90	366
24	Agammaglobulinemia, AR	413	66	347
25	Hyper IgM, AR (AICDA, UNG, CD40)	304	62	242
26	Neutropenia, Cyclical	269	39	230
27	ALPS Type 1a, CD95 (fas) Defects	242	43	199
28	Nijmegen Breakage Syndrome	227	17	210
29	ALPS, other types	210	33	177
30	XLP1, SH2D1A Deficiency	189	34	155
31	HLH (other types, Munc13-4, Munc 18-2, STX11)	183	38	145
32	Chediak-Higashi Syndrome	174	19	155
33	Leukocyte Adhesion Deficiency I	168	20	148
34	APECED	165	28	137
35	TACI mutation (TNFRSF13B)	159	33	126
36	Hyper-IgE, AR (DOCK8, TYK2 Deficiency)	156	19	137
37	MSMD (IL-12p40, IL12RB, IFN-γR1/2, STAT1 Deficiency)	150	5	145
38	Cartilage Hair Hypoplasia	135	57	78
39	TRAPS-TNF Receptor-Associated Periodic Fever	114	27	87
40	HLH Perforin Deficiency	101	19	82
41	Griscelli Syndrome Type 2	96	4	92
42	NEMO Deficiency	96	49	47
43	Hyper IgD Syndrome	96	22	74
44	IPEX (X-Linked)	80	28	52
45	IPEX-Like Syndrome	77	26	51
46	Thymoma with Immunodeficiency (Good Syndrome)	73	14	59
47	NK cell Deficiency	69	44	25
48	Comel-Netherton Syndrome	59	13	46
49	NOMID or CINCA	45	3	42
50	WHIM Syndrome	43	6	37
51	Dyskeratosis Congenita	37	11	26
52	Herpes simplex encephalitis HSE (Unc93B, TLR3, others)	36	3	33
53	IRAK4 (IL-1 Receptor Associated Kinase 4)	36	4	32
54	Factor I Deficiency	33	3	30
55	Muckle-Wells syndrome	32	5	27
56	Glycogen Storage Disease Type 1b	31	4	27
57	Schimke Syndrome	30	4	26
58	C1q Deficiency	29	2	27
59	Blau Syndrome (NOD2 or CARD15)	28	7	21
60	Familial Cold Autoinflammatory Syndrome	27	16	11
61	Bloom Syndrome	26	1	25
62	Leukocyte Adhesion Deficiency II, III, and others	26	4	22
63	EDA-ID, AD (NFKBIA mutation)	25	5	20
64	XLP2, XIAP Deficiency	25	9	16
65	Ataxia-Telangiectasia like Disease (ATLD)	22	11	11
66	Factor H Deficiency	22	3	19
67	Paroxysmal Nocturnal Hemoglobinuria(PIGA)	22	7	15
68	Majeed Syndrome (Mutation of LPIN2)	21	1	20
69	Neutropenia, XL (WASP mutation)	21	8	13
70	Properdin Deficiency	20	19	1
71	ICF-ID Centromeric Instability and Facial Anomalies	15	2	13
72	Papillon-Lefèvre Syndrome	14	0	14
73	MyD88 Deficiency	12	6	6
74	Neutropenia w/ Cardiac + Urogenital Malformations	12	0	12
75	Epidermodysplasia verruciformis (EVER mutation)"	11	1	10
76	Hermansky Pudlak Syndrome (Type 2; AP3 deficiency)	11	1	10
77	CD46 Deficiency	10	9	1
78	Ig Heavy Chain Mutations and Deletions	10	6	4
79	C1r Deficiency	6	3	3
80	DIRA (IL1RN)	4	0	4
81	PMS2 Deficiency	4	0	4
82	CR3-Complement Receptor 3 Deficiency	3	0	3
83	Factor D Deficiency	1	1	0

Supplementary Table 6.

Patients with Identified PI Defects – Global

2004 Baseline vs. 2009 Published vs. 2011 Report

TABLE I. Combined T and B–cell immunodeficiencies

	2004	2009	2011
ADA Deficiency	60	166	298
Artemis Deficiency (DCLREIC)	N/A	90	101
CD3δ / CD3ε / CD3ζ Deficiency	N/A	8	25
Cernunnos Deficiency	N/A	N/A	10
DNA Ligase IV Deficiency	N/A	N/A	14
γc Deficiency	88	312	454
IL-2Ralpha Deficiency (CD25 Deficiency)	N/A	N/A	34
IL-7Rα Deficiency	N/A	74	98
JAK3 Deficiency	N/A	54	116
MHC Class I or II Deficiency	10	110	226
Omenn Syndrome	6	110	237
PNP Deficiency	N/A	14	38
RAG 1/2 Deficiency	N/A	118	312
Reticular Dysgenesis (AK2 Deficiency)	N/A	N/A	15
ZAP-70 Deficiency	N/A	27	51
Other SCID:	287	847	1,134
Total	446	1,930	3,163

TABLE II. Other well defined immunodeficiency syndromes

	2004	2009	2011
Ataxia-Telangiectasia (A-T)	324	1,487	2,190
Ataxia-Telangiectasia like Disease (ATLD)			22
Bloom Syndrome	N/A	N/A	26
Cartilage Hair Hypoplasia	N/A	62	135
Chronic Mucocutaneous Candidiasis	N/A	253	456
Comel-Netherton Syndrome	N/A	N/A	59
DiGeorge Syndrome (DGS)	864	2,665	4,310
Dyskeratosis Congenita	N/A	N/A	37
Hyper IgE, AD (STAT3, Job Syndrome)	N/A	528	760
Hyper-IgE, AR (DOCK8, TYK2 Deficiency)	N/A		156
ICF-ID Centromeric Instability and Facial Anomalies	N/A	N/A	15
Nijmegen Breakage Syndrome	6	145	227
PMS2 Deficiency	N/A	N/A	4
Schimke Syndrome	N/A	N/A	30
Wiskott Aldrich Syndrome (WAS)	151	706	1,000
Total	1,345	5,846	9,427

TABLE III. Diseases of immune dysregulaton

	2004	2009	2011
ALPS Type 1a, CD95 (fas) Defects	97	333	242
ALPS, other types			210
APECED	N/A	127	165
Chediak-Higashi Syndrome	12	113	174
Griscelli Syndrome Type 2	3	46	96
Hermansky Pudlak Syndrome (Type 2; AP3 deficiency)	N/A	N/A	11
HLH (other types, Munc13-4, Munc 18-2, STX11)	N/A	N/A	183
HLH Perforin Deficiency	N/A	N/A	101
IPEX (X-Linked)	15	62	80
IPEX-Like Syndrome			77
XLP1, SH2D1A Deficiency	40	140	189
XLP2, XIAP Deficiency			25
Total	167	821	1,553

TABLE IV. Congenital defects of phagocyte #s and function

	2004	2009	2011
CGD, AR	186	1,399	964
CGD, XL			1,029
Glycogen Storage Disease Type 1b	N/A	N/A	31
Leukocyte Adhesion Deficiency I	33	127	168
Leukocyte Adhesion Deficiency II, III, and others			26
MSMD (IL-12p40, IL12RB, IFN-γR1/2, STAT1 Deficiency)	N/A	N/A	150
Neutropenia w/ Cardiac + Urogenital Malformations	82	553	12
Neutropenia, Cyclical			269
Neutropenia, Severe Congenital (ELA2, HAX1)			505
Neutropenia, XL (WASP mutation)			21
Papillon-Lefèvre Syndrome	N/A	N/A	14
Total	301	2,079	3,189

TABLE V. Predominantly antibody deficiencies

	2004	2009	2011
Agammaglobulinemia, (XLA)	369	1,347	2,132
Agammaglobulinemia, AR	N/A	247	413
Common Variable Immunodeficiency (CVID)	745	4,292	7,613
Hyper IgM (X Linked, TNFSF5)	83	327	465
Hyper IgM, AR (AICDA, UNG, CD40)	88	285	304
Hypogammaglobulinemia of Infancy (Transient)	127	1,771	2,713
Hypogammaglobulinemia unspecified	N/A	N/A	3,148
Ig Heavy Chain Mutations and Deletions	N/A	N/A	10
IgA Deficiency, Selective	867	3,072	7,061
IgA with IgG Subclass Deficiency	N/A	N/A	1,055
IgG Subclass Deficiency, Isolated	326	3,239	3,878
Specific Antibody Deficiency (normal Ig and B cells)	144	2,261	2,138
TACI mutation (TNFRSF13B)	N/A	N/A	159
Thymoma with Immunodeficiency (Good Syndrome)	N/A	N/A	73
Total	2,749	16,841	31,162

TABLE VI. Defects in innate immunity

	2004	2009	2011
EDA-ID, AD (NFKBIA mutation)	N/A	N/A	25
Epidermodysplasia verruciformis (EVER mutation)"	N/A	N/A	11
Herpes simplex encephalitis HSE (Unc93B, TLR3, others	N/A	N/A	36
IRAK4 (IL-1 Receptor Associated Kinase 4)	N/A	N/A	36
MyD88 Deficiency	N/A	N/A	12
NEMO Deficiency	3	72	96
NK cell Deficiency	N/A	N/A	69
WHIM Syndrome	N/A	26	43
Total	3	98	328

TABLE VII. Autoinflammatory disorders

	2004	2009	2011
Blau Syndrome (NOD2 or CARD15)	N/A	N/A	28
DIRA (IL1RN)	N/A	N/A	4
Familial Cold Autoinflammatory Syndrome	N/A	N/A	27
Familial Mediterranean Fever	N/A	N/A	1,715
Hyper IgD Syndrome	N/A	N/A	96
Majeed Syndrome (Mutation of LPIN2)	N/A	N/A	21
Muckle-Wells syndrome	N/A	N/A	32
NOMID or CINCA	N/A	N/A	45
PFAPA Syndrome	N/A	N/A	1,518
TRAPS-TNF Receptor-Associated Periodic Fever	N/A	N/A	114
Total	0	0	3,600

TABLE VIII. Complement deficiencies

	2004	2009	2011
C1 Inhibitor Deficiency	7	602	2,045
C1q Deficiency			29
C1r Deficiency			6
C2-C9 Deficiency			580
CD46 Deficiency			10
CR3-Complement Receptor 3 Deficiency			3
Factor D Deficiency			1
Factor H Deficiency			22
Factor I Deficiency			33
MBP/MBL or MASP Deficiency			881
Paroxysmal Nocturnal Hemoglobinuria(PIGA)			22
Properdin Deficiency			20
Total	7	602	3,652

	2004	2009	2011
"Unspecified" Tables I through VIII:	402	2,066	4,290

Total:	**5,420**	**30,283**	**60,364**