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Mode of inheritance and molecular studies in the 184 patients with known genetic defect.

Gene	Mode of inheritance	Number of patients	Molecular defects		Genetic analysis performed to first detect mutation
			cDNA	Protein	
IMMUNODEFICIENCIES AFFECTING CELLULAR AND HUMORAL IMMUNITY (*N* = 75)					
RAG1	AR	7	c.1361T>A	L454Q	6 SS−1 WES
		1	–	R404Q	WGS
		1	–	R394W	SS
RAG2	AR	6	–	G35A	4 SS−2 WES
DCLRE1C	AR	3	–	G135R	SS
		2	Ex 1-9 del/Ex 1-3 del	–	SS
		1	–	K157KfsX13	SS
		1		G6E	WES
JAK3	AR	2	c.1019C>A	S340X	WES
		2	–	A573P	1 WES– 1 SS
		1	c.1744C>T	R582W	SS
AK2	AR	5	c.524G>A	R175Q	SS
CD3D	AR	3	c.56-3T>G	–	WES
ADA	AR	2	c.428dupA	–	SS
DOCK8	AR	6	Ex 1-5 del	–	SS
		1	c.4070C>A	S1357X	SS
		1	Ex 2-12 del	–	SS
		1	Ex 1-23 del	–	SS
		1	Ex 1-2 del	–	SS
DOCK2	AR	2	c.1868G>A	W623X	WES
		1	–	Y1242YfsX33	WES
RFXANK	AR	6	insTCAC.IVS4+1	–	SS
		4	c.362A>T	D121V	SS
		2	c.564G>A	W188X	SS
ZAP70	AR	1	c.1606G>A	G536S	SS
TFRC	AR	8	–	Y20H	WGS
IKBKB	AR	1	c.736A>G	S246G	WES
ICOS	AR	2	c.90delG	M30fsX26	WES
GM2A	AR	1	c.Chr5q33.1 del	–	CMA
COMBINED IMMUNODEFICIENCIES WITH ASSOCIATED OR SYNDROMIC FEATURES (*N* = 54)					

Gene	Mode of inheritance	Number of patients	Molecular defects		Genetic analysis performed to first detect mutation
			cDNA	Protein	
WAS	XL	1	c.400G>A	A134T	SS
		1	c.91G>A	E31K	SS
		1	–	–	SS
ATM	AR	3	c.381delA/ IVS 44+1 G>A c.2932T>C (Het)	– S978P	SS
		2	c.7082T>C	L2360P	SS
		1	c.748C>T	R250X	SS
22q11.2DS	AD	30	22q11.2 del	–	FISH–CMA
STAT3	AD LOF	2	c. 1144C>T	R382W	SS
		1	c.1910T>C	V637A	SS
		1	c. 1868G>T	W623L	SS
STAT5B	AR	2	c.1643-1delG	–	SS
DNMT3B	AR	1	c.Chr20:31390243G>A	–	WES
ZBTB24	AR	1	c.1492C>A	–	WES
RMRP	AR	1	c.27G>A	–	SS
TTC7A	AR	1	c.1919+1G>A	–	WES
HOIP (RNF31)	AR	1	c.215T>C	L72P	WES
SP110	AR	2	c.617C>T (homozygous) c.642delC (homozygous)	A206V S215fs	SS
MYSM1	AR	2	c.1168G>T	E390X	WES
PREDOMINANTLY ANTIBODY DEFICIENCIES (*N* = 15)					
BTK	XL	1	c.82C>T	R28C	SS
		1	c.982C>T	Q328X	SS
		1	Ex 4–5 del	–	WES
		1	–	–	SS
AICDA	AR	7	c.254G>A	S85N	SS
		2	c.169G>A	V57M	WES
NFKB2	AD	2	c.2596_2597delAG	S866fs	WES
DISEASES OF IMMUNE DYSREGULATION (n=31)					
LYST	AR	2	c.1902dupA	A635SfsX4	1 WES–1 SS
		1	IVS19 c.5784+5 G>A	–	SS
		1	c.2311C>T	Q771X	WES