

Supplementary material

Table S1. Literature revision of patients with aminoacids substitution in R288.

Patient	IgG (486-1211mg/dl)	IgA (30-182mg/dl)	IgM (45-211)	B cells (%)	Age at diagnosis (months)	Symptoms onset (months)	Mutation
P6 ¹	134	22.1	16.8	0	36	NR	R288W
P19 ¹	33.3	6.67	10	1.1	144	NR	R288W
8a ²	NR	NR	NR	NR	NR	NR	R288W
1 ³	576	<7	29	0.1	115	48	R288W
once /37 ⁴	NR	NR	NR	1	NR	NR	R288W
276 ⁵	NR	NR	NR	<1	NR	NR	R288W
index case ⁶	331	<7	18	0.8	492	NR	R288Q
17a ⁷	<200	NR	NR	NR	NR	NR	R288W
17b ⁷	<200	NR	NR	NR	NR	NR	R288W
18a ⁷	<200	NR	NR	NR	NR	NR	R288W
18b ⁷	<200	NR	NR	NR	NR	NR	R288W
19 ⁷	<200	NR	NR	NR	NR	NR	R288W
20 ⁷	<200	NR	NR	NR	NR	NR	R288Q
1110 ⁸	NR	NR	NR	NR	NR	NR	R288Q
1111 ⁸	NR	NR	NR	NR	NR	NR	R288Q
9a ⁹	NR	NR	NR	NR	180	NR	R288W
9b ⁹	NR	NR	NR	NR	156	NR	R288W
9c ⁹	NR	NR	NR	NR	132	NR	R288W
10 ⁹	NR	NR	NR	NR	84	NR	R288W
276 ¹⁰	NR	NR	NR	NR	NR	NR	R288W
P9 ¹¹	7	3	2	NR	48	NR	R288W
P10 ¹¹	70	12	16	NR	72	NR	R288W
Patient 2 ¹²	33	<6	4	0	8	NR	R288W
8 ¹³	13	0	1	0	134	24	R288W
9 ¹⁴	96	28	17	1.3	96	NR	R288Q
27/23 ¹⁵	443	197	16	<2	188.64	42	R288Q
28/23 ¹⁵	955	15	21	<2	84	12	R288Q

Table S3. Literature revision of patients with aminoacid substitutions in R28.

Patient	IgG (486-1211mg/dl)	IgA (30-182mg/dl)	IgM (45-211 mg/dl)	B cells (%)	Age at diagnosis (months)	Age of Syntoms months	Mutation
P1 ¹⁶	2.67	<0.1	<0.1	0.1	209	NR	R28C
P5 ¹⁷	160	5	10	<1%	48	NR	R28C
P3 ¹⁸	2.7	0.38	0.5	1	36	9	R28C
3 ¹⁴	267	<10	<10	0.1	NR	NR	R28C
4 ¹⁴	7	49	58	0.2	NR	NR	R28S
40 ¹⁹	NR	NR	NR	NR	NR	NR	R28H
4 ²⁰	83	13	68	0	132	NR	R28H
2 ¹³	430	0.6	0.52	0.12	127	NR	R28C
3 ¹³	320	1.5	11.5	0.04	204	NR	R28C
1 ²¹	NR	NR	NR	NR	NR	NR	R28H
Patient 1 ²²	360	27	25	0	768	240	R28H
P05 ²³	NR	NR	NR	<1	168	NR	R28H
Index case ²⁴	480	185	26	0.11	300	276	R28H
Patient1 ¹²	50	53	16	0.1	72	NR	R28C
P1 ¹¹	174	<29	<18	NR	48	48	R28C
P23 ²⁵	NR	12	15	0.2	96	NR	R28P
P5 ²⁶	298	20	6	0.04	72	48	R28L
P12 ¹⁵	1970	29	37	0.3	210	180	R28H

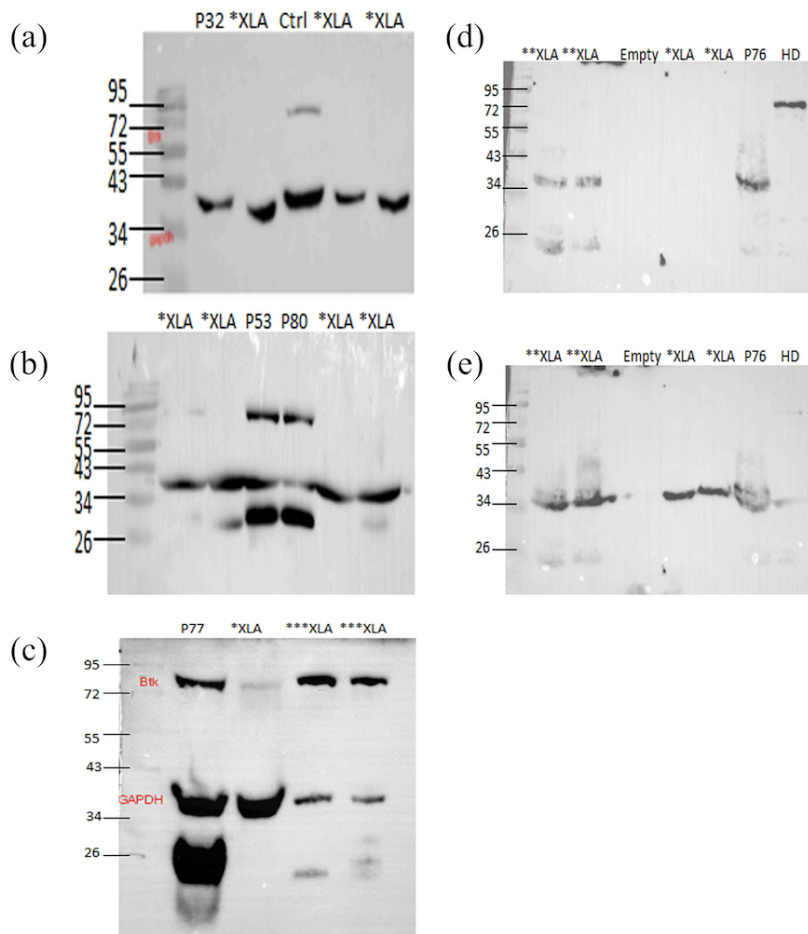


Fig. S1. BTK and GAPDH expression. Individual Western-Blot are showed. (a) P32, (b) P53 and P80, (c) P77 and (d) BTK for P76 (e) GAPDH for P76. Each blot was revealed using mouse-anti-human BTK and rabbit-anti-human GAPDH at the same time, with their respective secondary antibodies. For P76, an additional figure for BTK expression is showed since a band was observed very close to GAPDH.

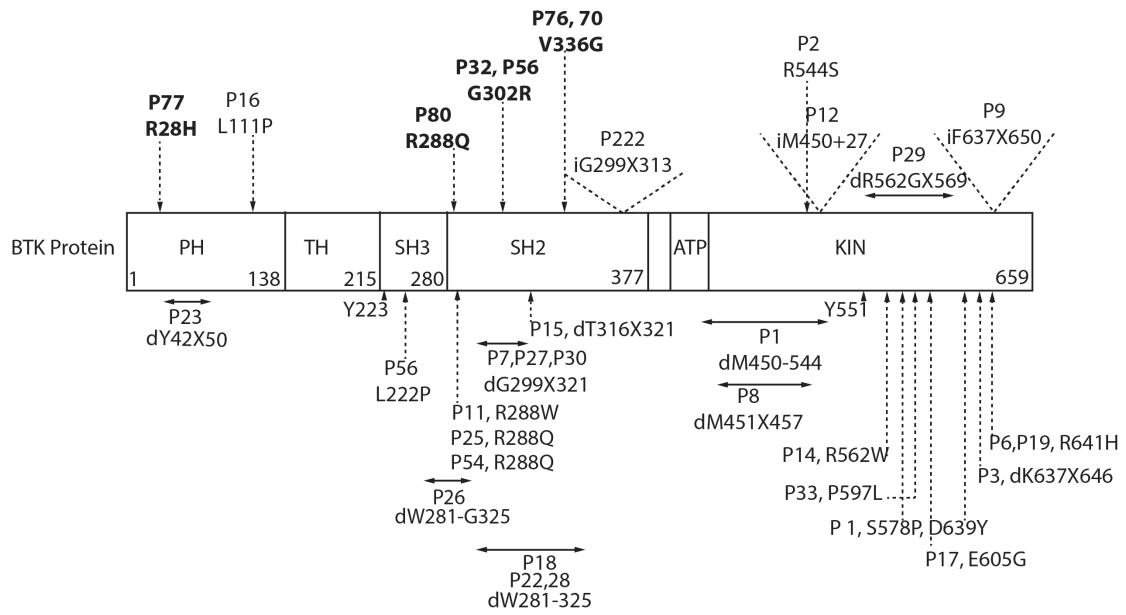


Fig. S2. Schematic representation of the distribution of mutations observed in the Mexican XLA cohort. Mutations in bold are those reported in the main manuscript. L111P and R544S are two additional patients with atypical data for XLA.

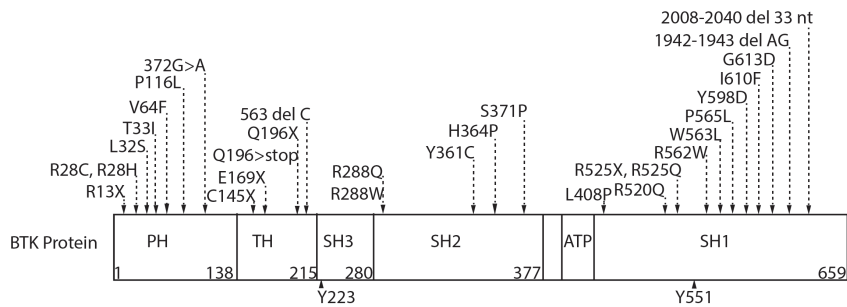


Fig. S3. Mutation distribution for atypical XLA patients revised from the literature.

Supplementary references

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