Supplementary Table 1: Type of NF1 pathogenic variant

	A Series (H. Ramon & Cajal)	B Series (CSUR)	Total (%)	Expected proportion*	p-val	Statistical Method^
Sporadic NF1						
Nonsense	107	21	128 (0,25)	0,207	0,043	1
Frameshift	124	21	145 (0,29)	0,256	**0,229**	1
Splicing	95	18	113 (0,22)	0,270	0,030	1
Missense / in frame variants	47	17	64 (0,13)	0,159	**0,069**	1
First Methionine	1	1	2 (0.004)	1,734	**0,621**	2
Single or multiple exon deletions	16	4	20 (0,04)	0,022	0,047	1
Whole NF1 gene deletion	34	5	39 (0,07)	0,050	0,032	1
Familial NF1 (index cases)						
Nonsense	38	12	50 (0,23)	0,207	**0,506**	1
Frameshift	50	18	68 (0,31)	0,256	**0,091**	1
Splicing	52	9	61 (0,13)	0,270	**0,822**	1
Missense / in frame variants	18	11	29 (0,13)	0,159	**0,363**	1
First Methionine	1	0	1 (0.005)	2,034	**0,441**	2
Single or multiple exon deletions	2	0	2 (0,01)	0,411	**0,310**	2
Whole NF1 gene deletion	5	2	7 (0,03)	0,050	**0,311**	1

* Messiaen & Wimmer, 2008 ^ 1) 2-sample test for equality of proportions with continuity correction; 2) Fisher's Exact Test for Count Data