On-line Table: Summary of abnormal brain CT/MRI findings in 30 patients with ATR-X syndrome

			High Signal Intensity on T2WI/FLAIR in WM			GM Abnormality			
Case	Exon	Mutation	Peritrigonal Area	Widespread, Scattered	Delayed Myelination	Atrophy	Progressive Atrophy	Others	Evaluated Age
1	5′-UTR	Nucleotide substitution		+					1 yr
2	2–5	Exon 2–5 del				+			5 mo
3	5	R81fs				+			20 yr
4	6	E131fs	+		+	+		HCC	9 yr
5	8	c.536A>G				+			13 yr
6	8	c.536A>G	+					HCC	3 yr
7	8	P190L				+			5 yr
8	8	V194I				+			11 yr
9	8	V194A				+			21 yr
10	8	P190L	+		+			HCC	8 yr
11	10	C223F	+						9 yr
12	10	L229F				+			21 yr
13	10	L245P				+			7 yr
14	10	R246C				+			26 yr
15	10	R246C	+			+			11 yr
16	10	Y266C				+			15 yr
17	10	R246C				+			14 yr
18	10	T278P				+			54 yr
19	10	Ser576X	+						35 mo
20	18	1 a.a. del.	+		+	+			16 mo
21	19	A1622V	+						35 mo
22	19	V1624M	+					CVA	4 yr
23	19	L1645S				+			31 mo
24	29	R2085C	+						21 mo
25	31	M2171V	+			+		VE, HCC	5 yr
26	35	R7386X			+				4 mo
27	Int 35	43 a.a. del					+	VE	6–34 mo
			11	1	4	17	1		

Note:—HCC indicates hypoplasia of the corpus callosum; CVA, cerebellar vermis atrophy; UTR, untranslated region; VE, ventricular enlargement; +, present.