IRIS AperTO

UNIVERSITÀ DEGLI STUDI DI TORINO

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De novo 13q12.3-q14.11 deletion involving BRCA2 gene in a patient with developmental delay, elevated IgM levels, transient ataxia, and cerebellar hypoplasia, mimicking an A-T like phenotype

This is the author's manuscript

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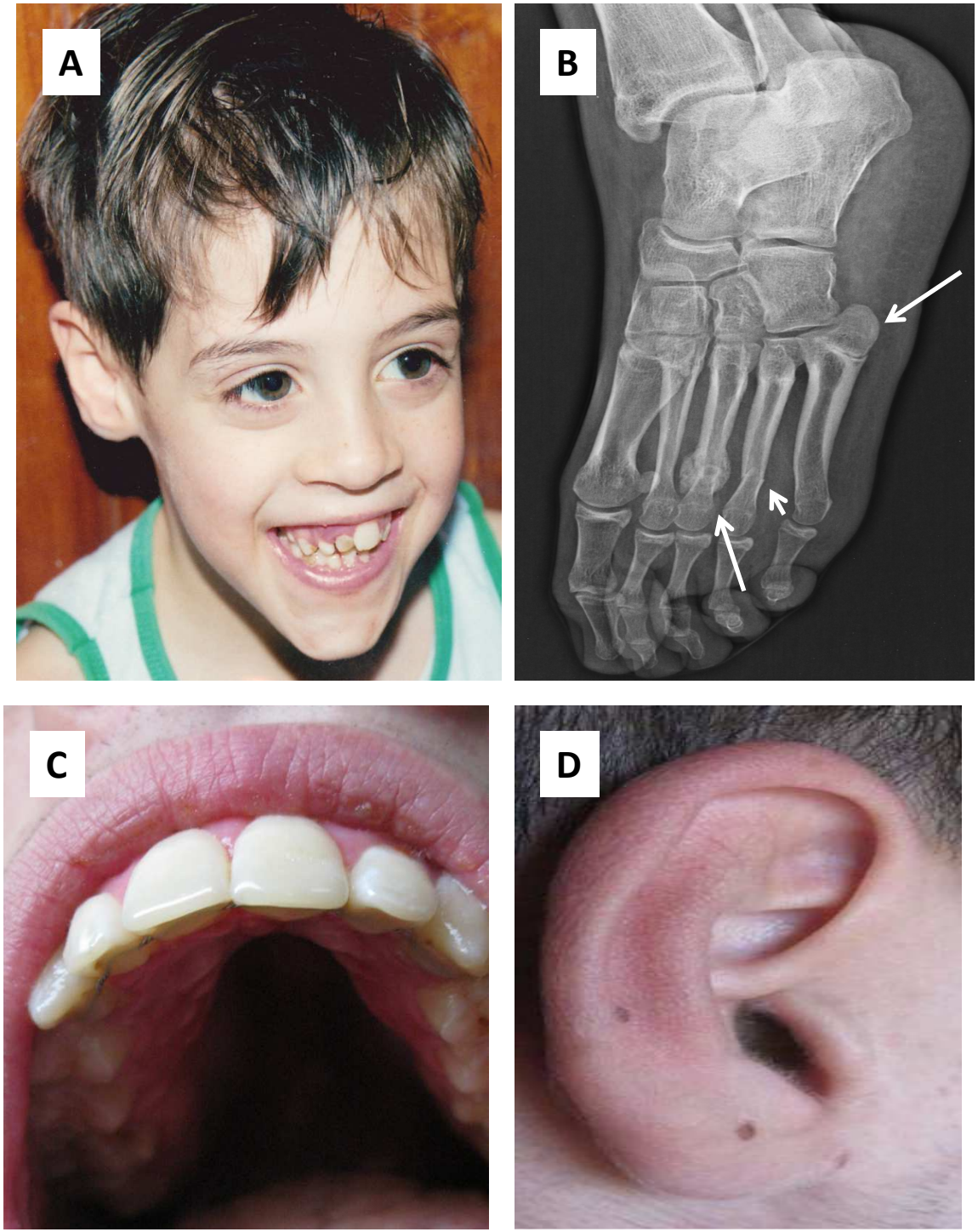
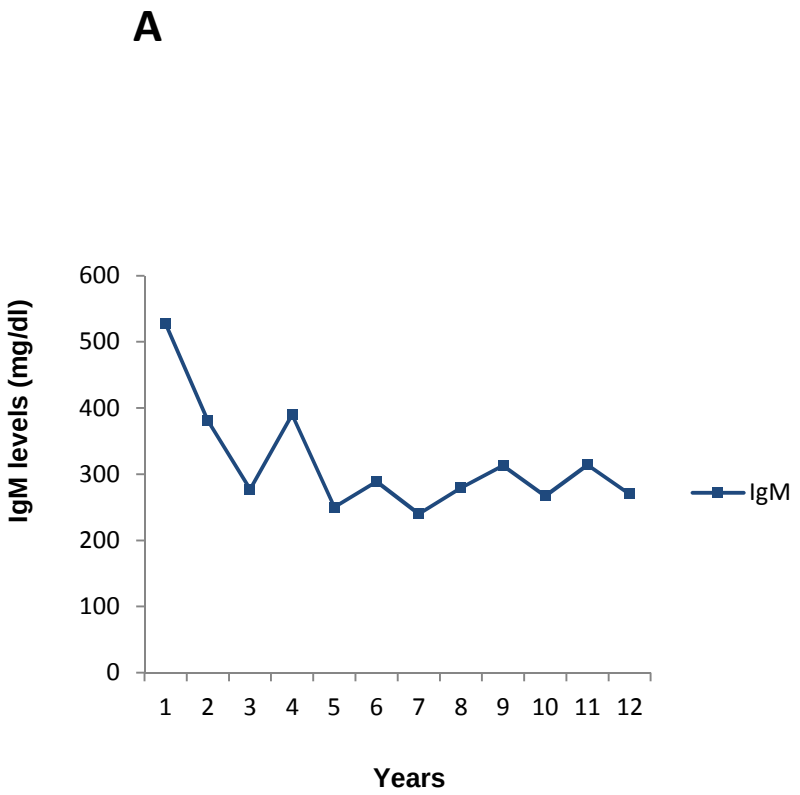

A
B
C
D


A
IgM levels (mg/dl)
600
500
400
300
200
100
0
1 2 3 4 5 6 7 8 9 10 11 12
Years
IgM

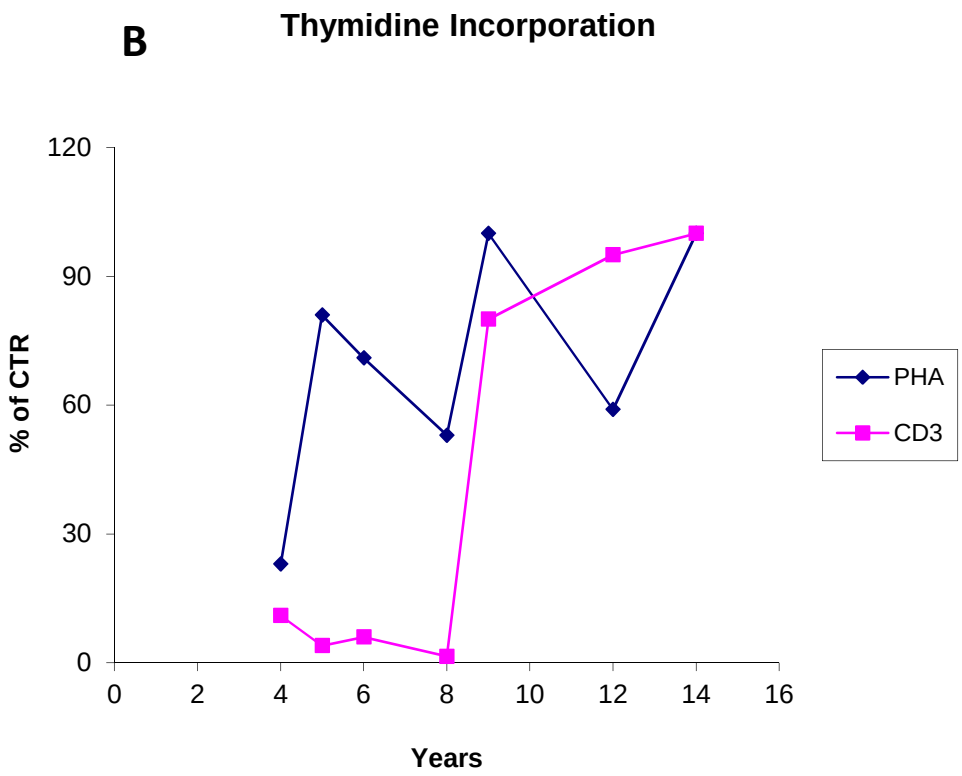
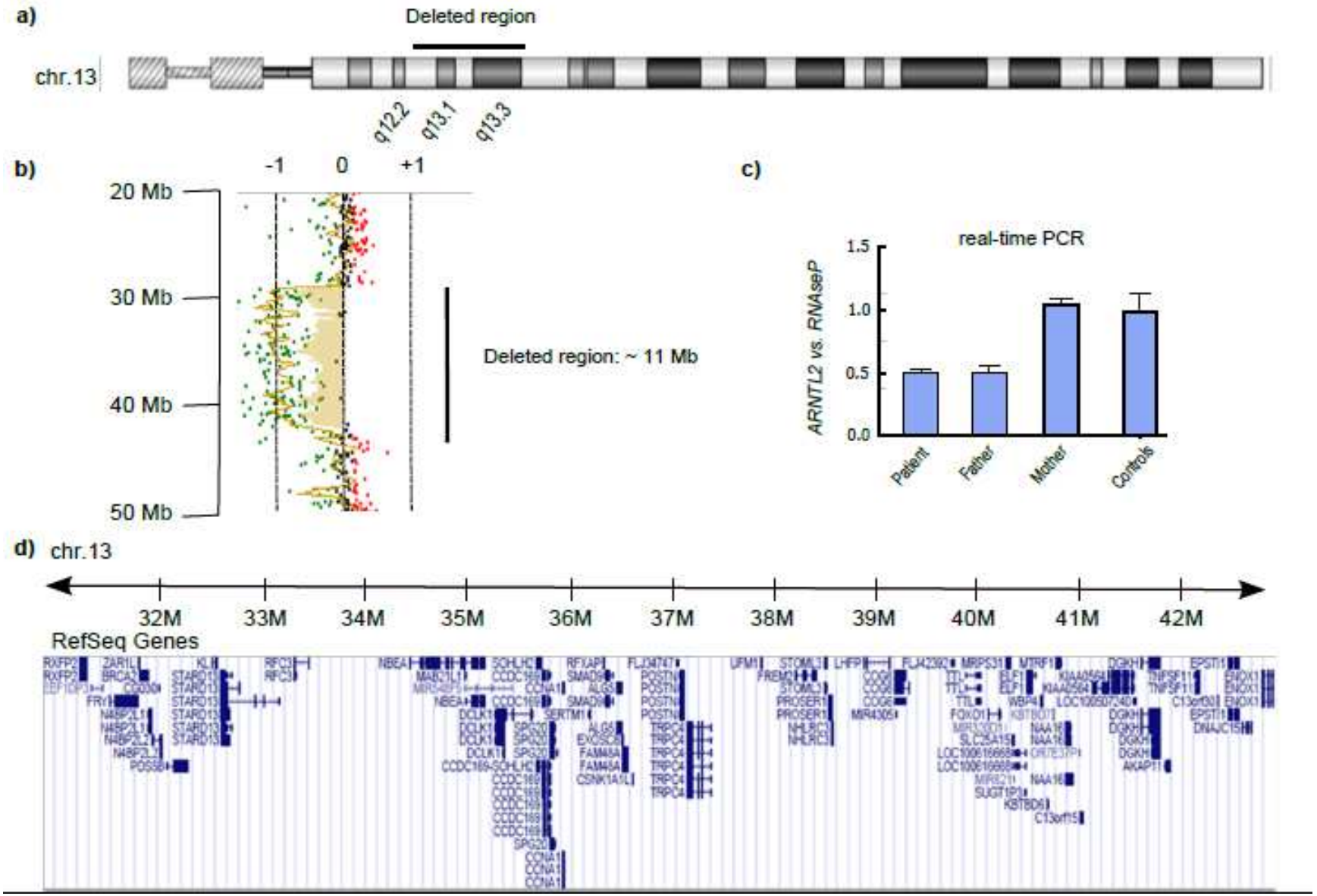

B
Thymidine Incorporation
% of CTR
120
90
60
30
0
0 2 4 6 8 10 12 14 16
Years
PHA
CD3

TABLE I. Immunophenotype and specific antibody production during the follow-up

Tests	3 y	8 y	17 y
Lymphocyte count (cells/mm^3)	7200	8150	7530
Lymphocyte subset (%)			
CD3	79	64	67
CD4	39	20	32
CD8	32	39	20
CD19	11	13	17
CD56+	6	19	5
CD3 HLA-DR	6	14	4
Specific IgG antibodies			
Toxoplasma	+	+	+
Rubeo	+	+	+
CMV	+	+	+
HSV	-		
EBV	+	+	+

y, year[s]; + positive; - negative.


a)
Deleted region
chr.13
q12.2
q13.1
q13.3
b)
-1
0
+1
20 Mb
30 Mb
40 Mb
50 Mb
Deleted region: ~ 11 Mb
c)
real-time PCR
ARNTL2 vs. RNAseP
1.5
1.0
0.5
0.0
Patient
Father
Mother
Controls
d)
chr.13
32M
33M
34M
35M
36M
37M
38M
39M
40M
41M
42M
RefSeq Genes