

II ASBAI-BRAGID Meeting

2016

Case Presentation

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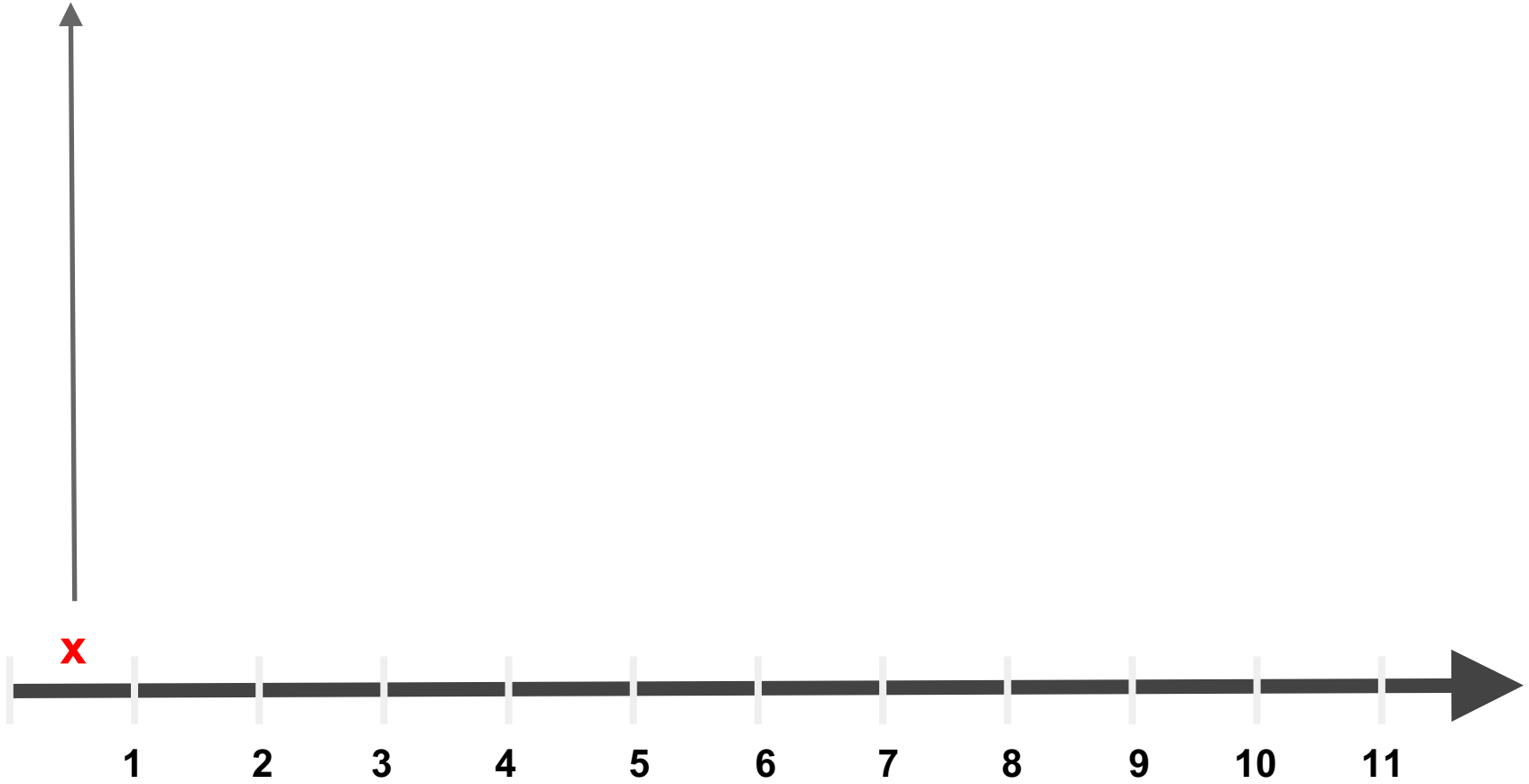
Curitiba - Brazil



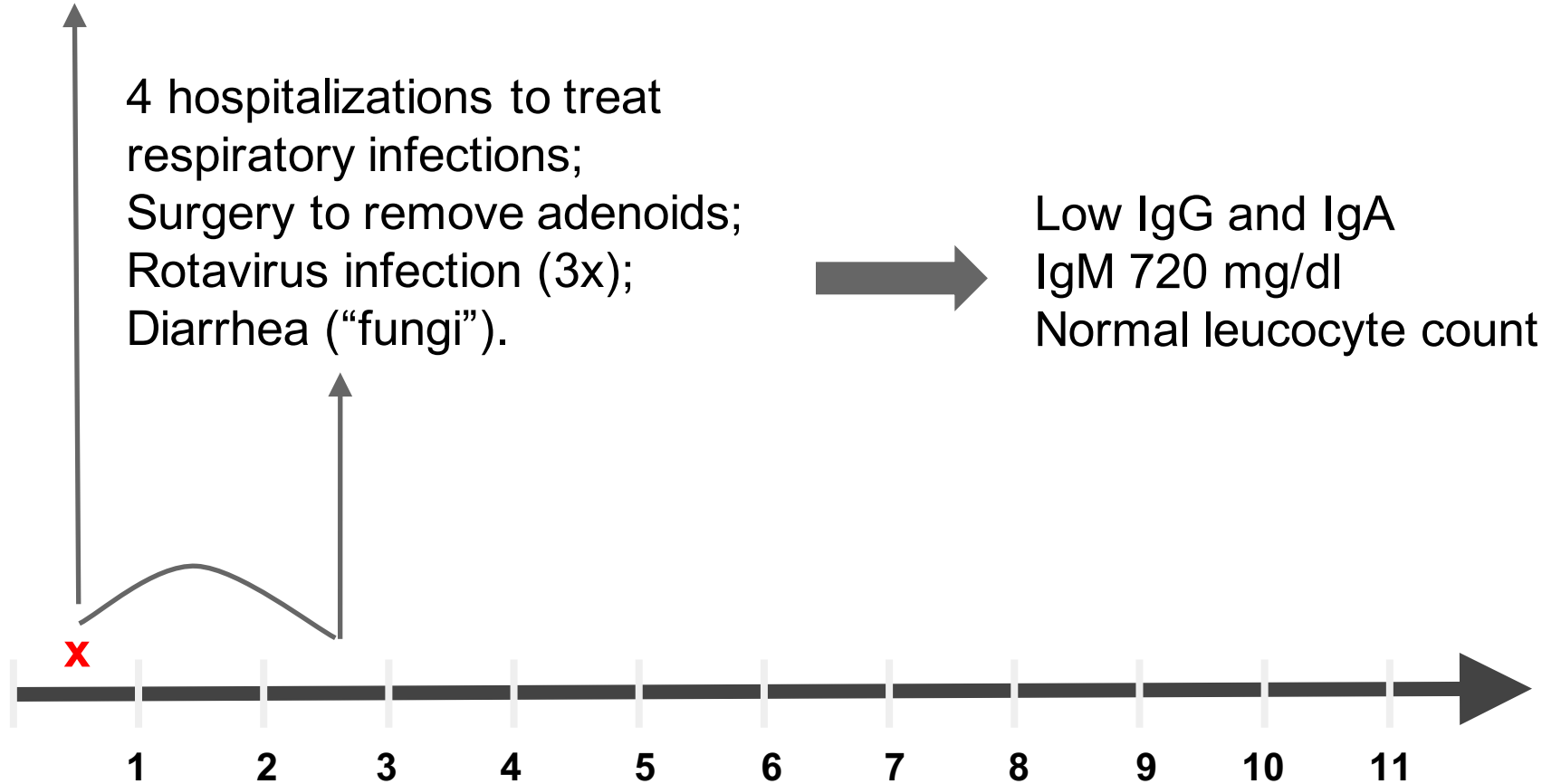


10 years old boy
non consanguineous parents
Curitiba- Brazil

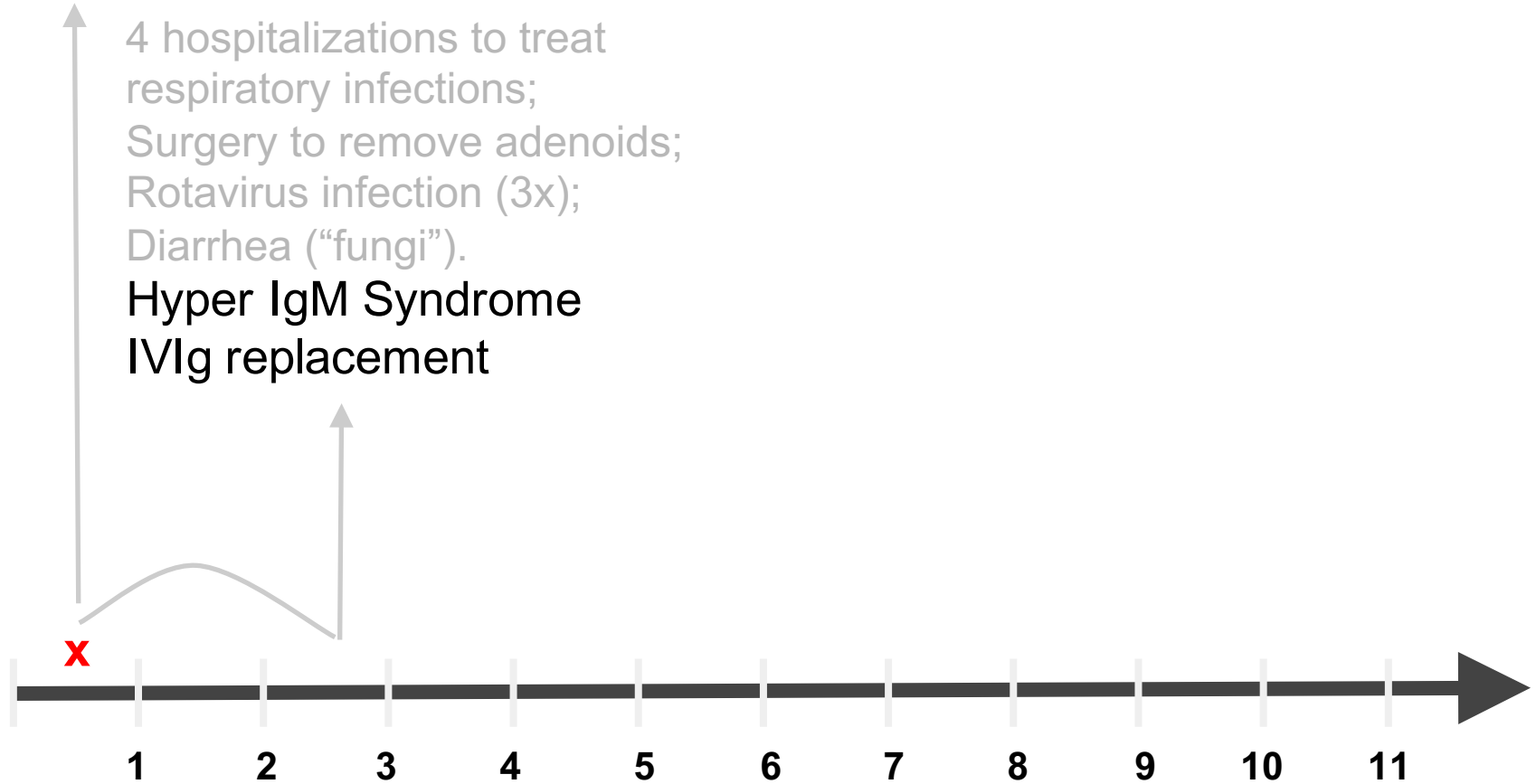
Bronchiolitis



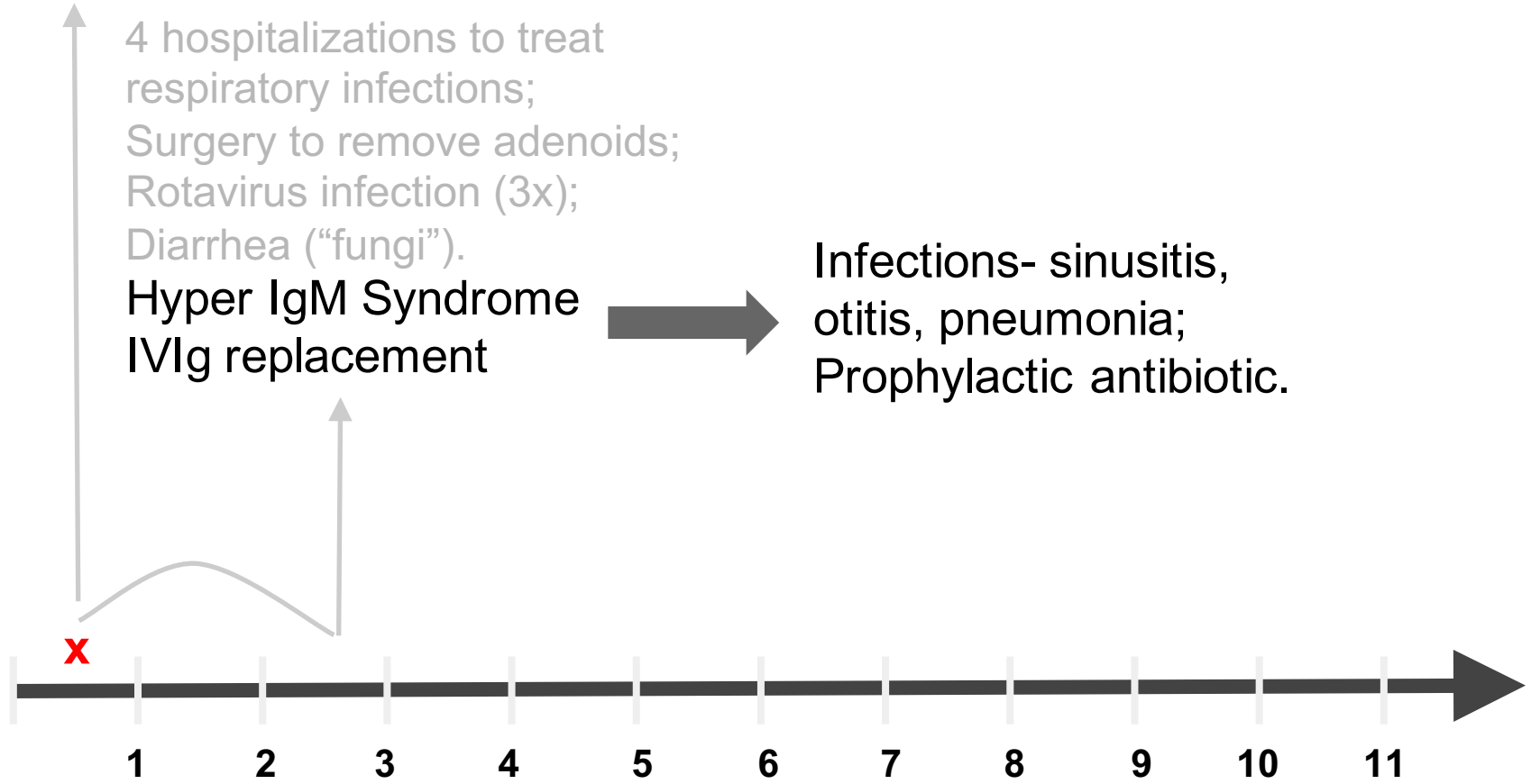
Bronchiolitis



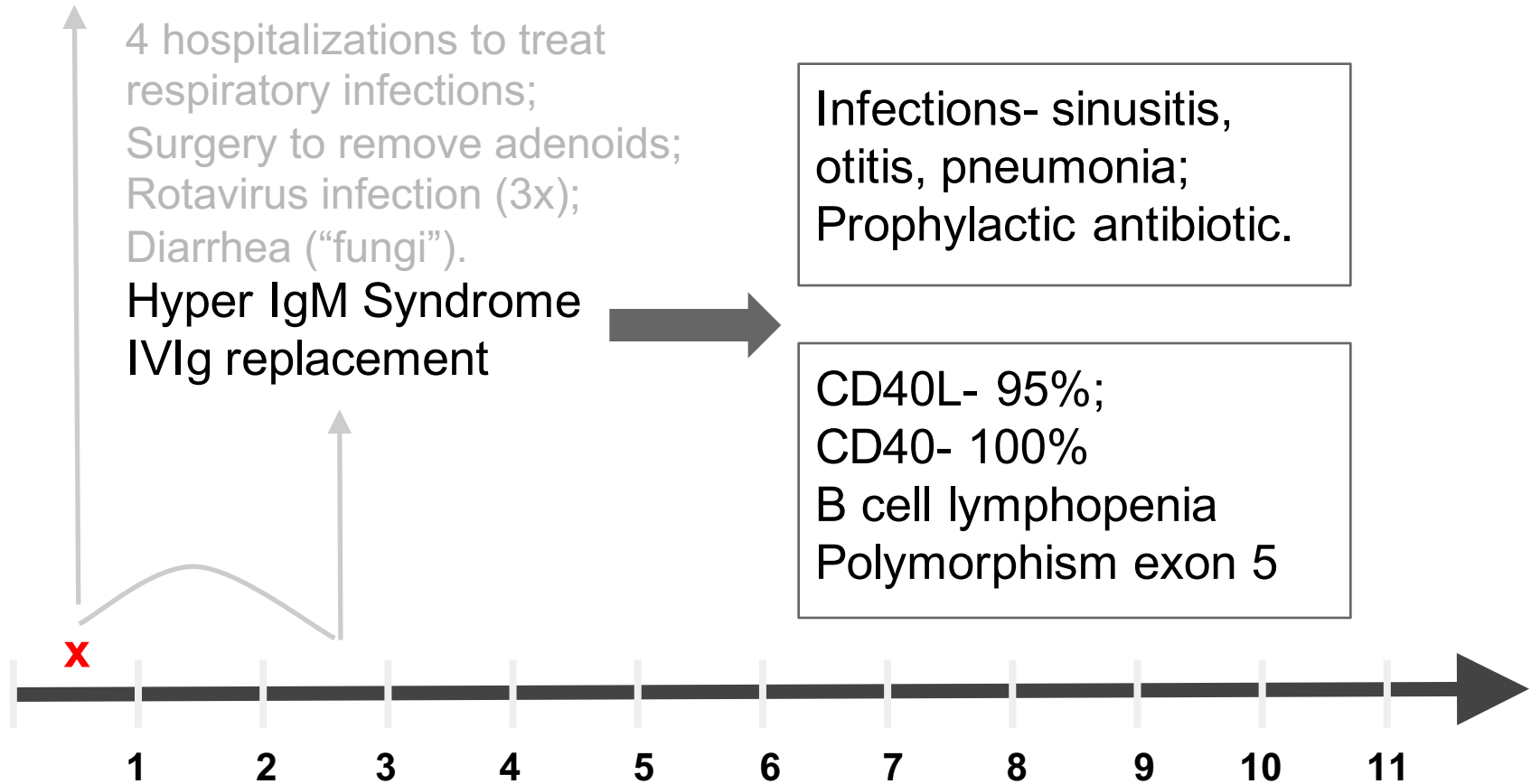
Bronchiolitis



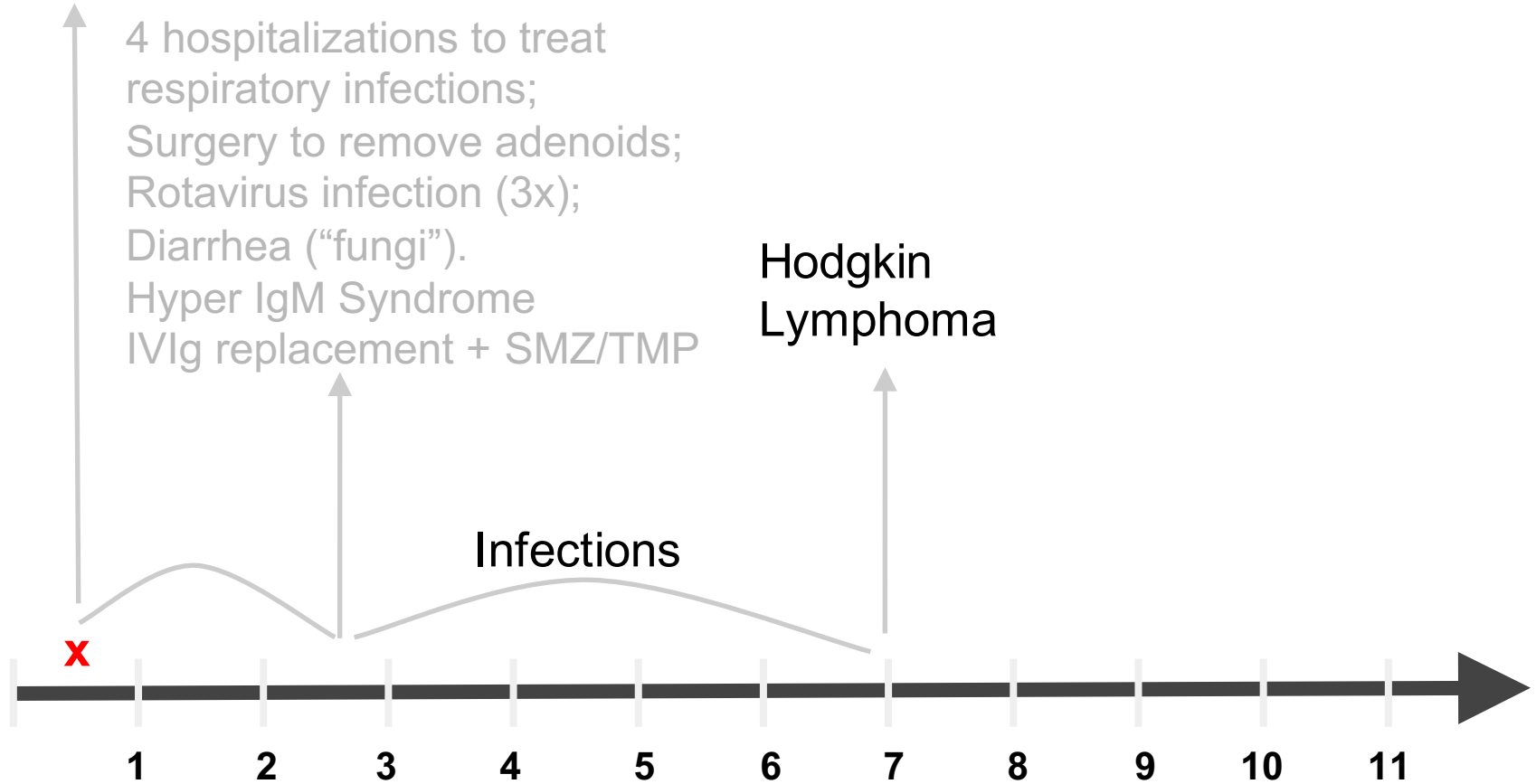
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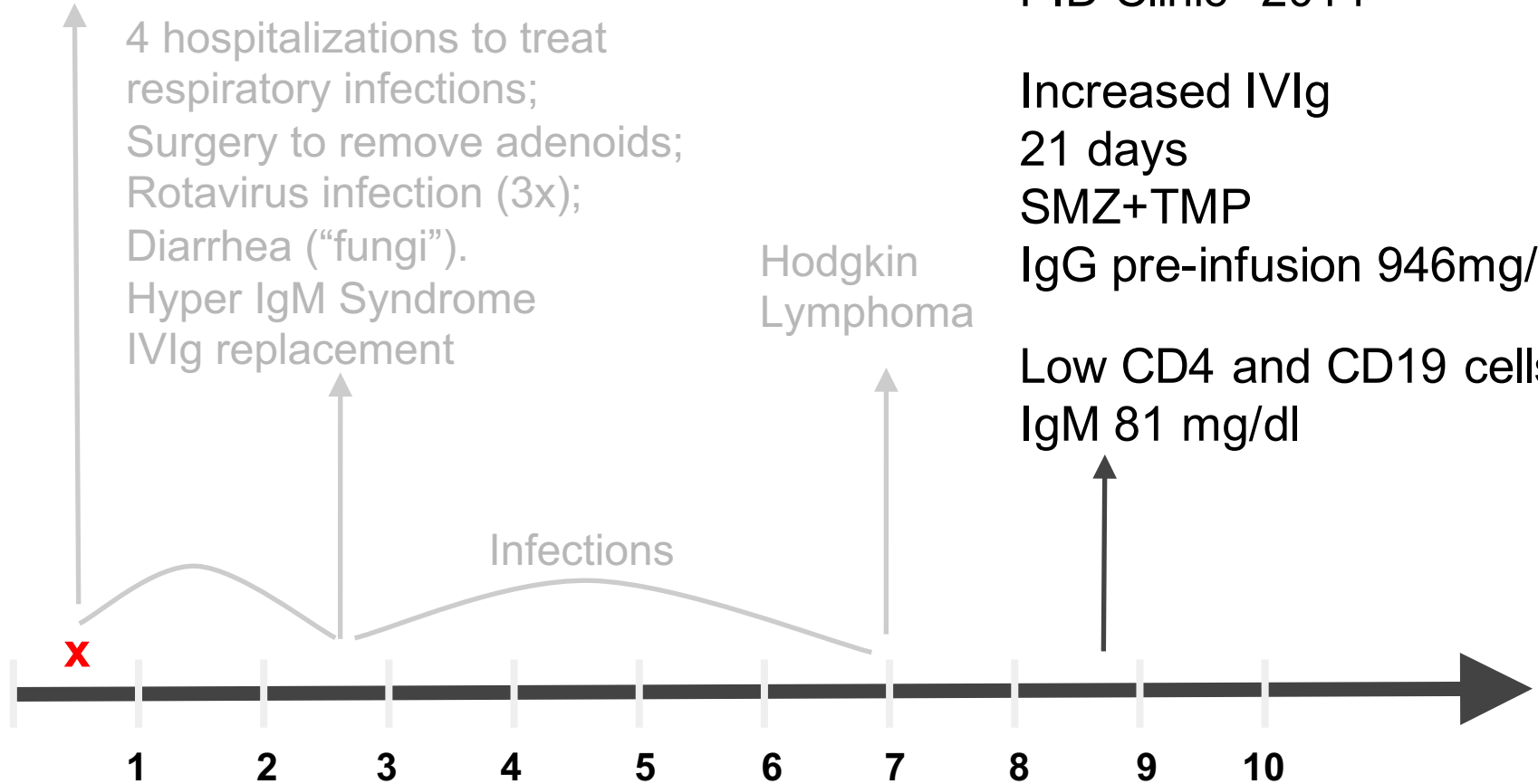
Bronchiolitis



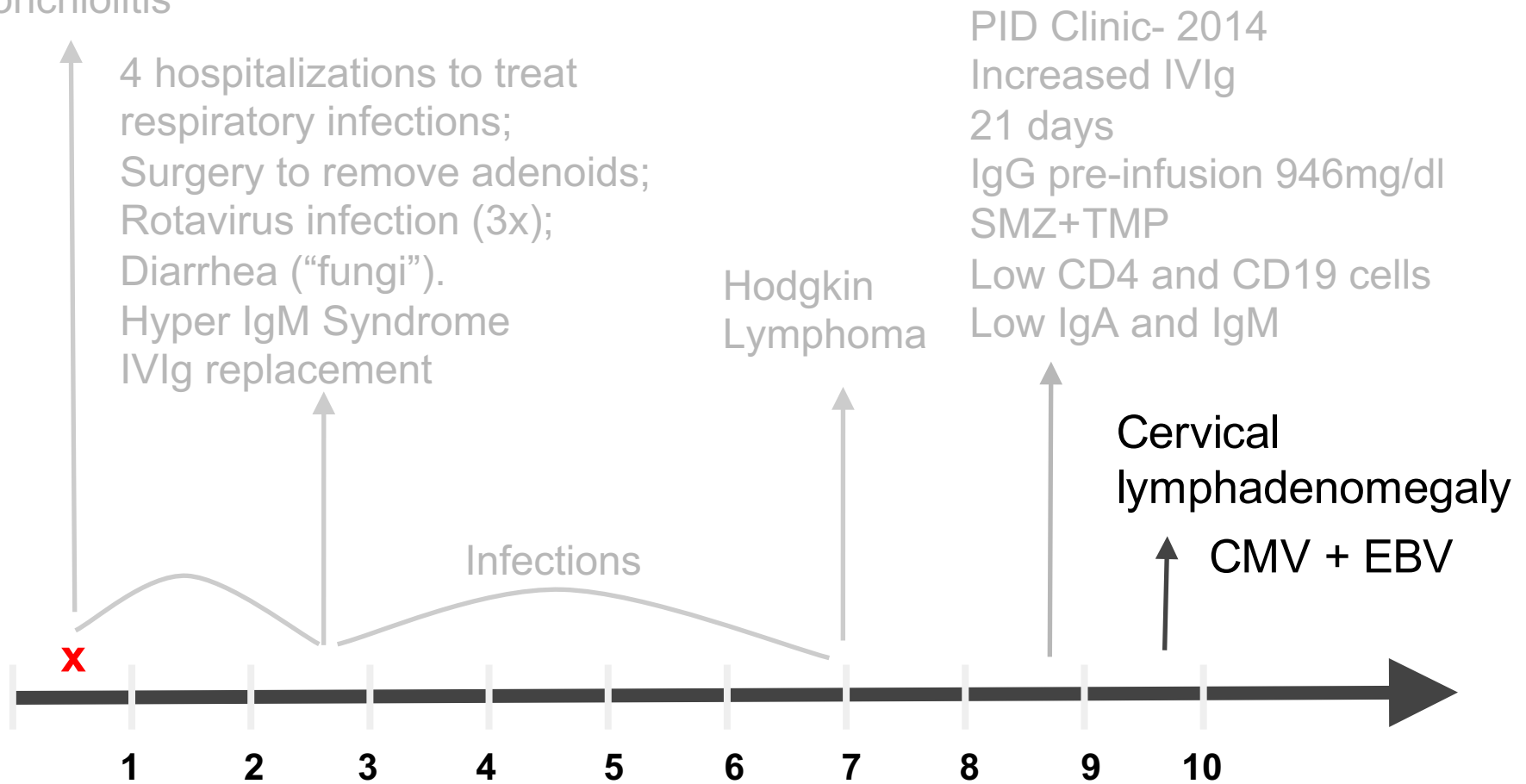
Bronchiolitis



Bronchiolitis



Bronchiolitis



Never free of infections

Hodgkin Lymphoma + CMV/EBV

T and B cell lymphopenia

Normal expression CD40/CD40L

Physical malformation

Acrosyndactyly

Absence of 2 toes on the foot

Chromosomal Abnormality??

Total Leucocytes- 10630

Lymphocytes- 2561 (24,1%)

	% LINFÓCITOS	N° Absol. (µl)
LINFÓCITO T		
CD3+	84,43	2.162,9
CD3CD4+	13,1	335,6
CD3CD8++	64,8	1.660,0
CD3+CD8+	2,7	69,2
CD3CD4+CD8+	0,03	0,8
CD3CD4(-)CD8(-)	3,8	97,3
RELAÇÃO CD4/CD8	0,20	
CD27+CD45RO(-)CD4+	0,4	10,2
CD27+CD45RO+CD4+	10,5	269,0
CD27(-)CD45RO+CD4+	2,2	56,4
CD27(-)CD45RO(-)CD4+	<0,1	----
CD27+CD45RO(-)CD8+	3,8	97,3
CD27+CD45RO+CD8+	16,8	430,4
CD27(-)CD45RO+CD8+	20,2	517,5
CD27(-)CD45RO(-)CD8+	24,0	614,8
CD45RA+CD31+CD4+	0,4	10,2
CD45RA+CD31+CD8+	23,9	612,3
CD57+CD28(-)CD4+	NR	----
CD57+CD28(-)CD8+	NR	----
CD25GITRCD4+	0,3	7,7
CD3+CD16+CD56+	10,4	266,4

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Differentiated effectors

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LINFÓCITO B	%	
CD19++ CD20++	1,5	38,4
CD19+CD10+	1,0	25,6
CD19+IgM+	<0,1	----
LINFÓCITO NK:		
NK	14,07	360,5
CD3-CD16+CD56+	12,87	329,7
CD3-CD16+CD56-	0,8	20,5
CD3-CD16-CD56+	0,4	10,2
CD45	100	2.561,8

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CGH

Exome Sequence

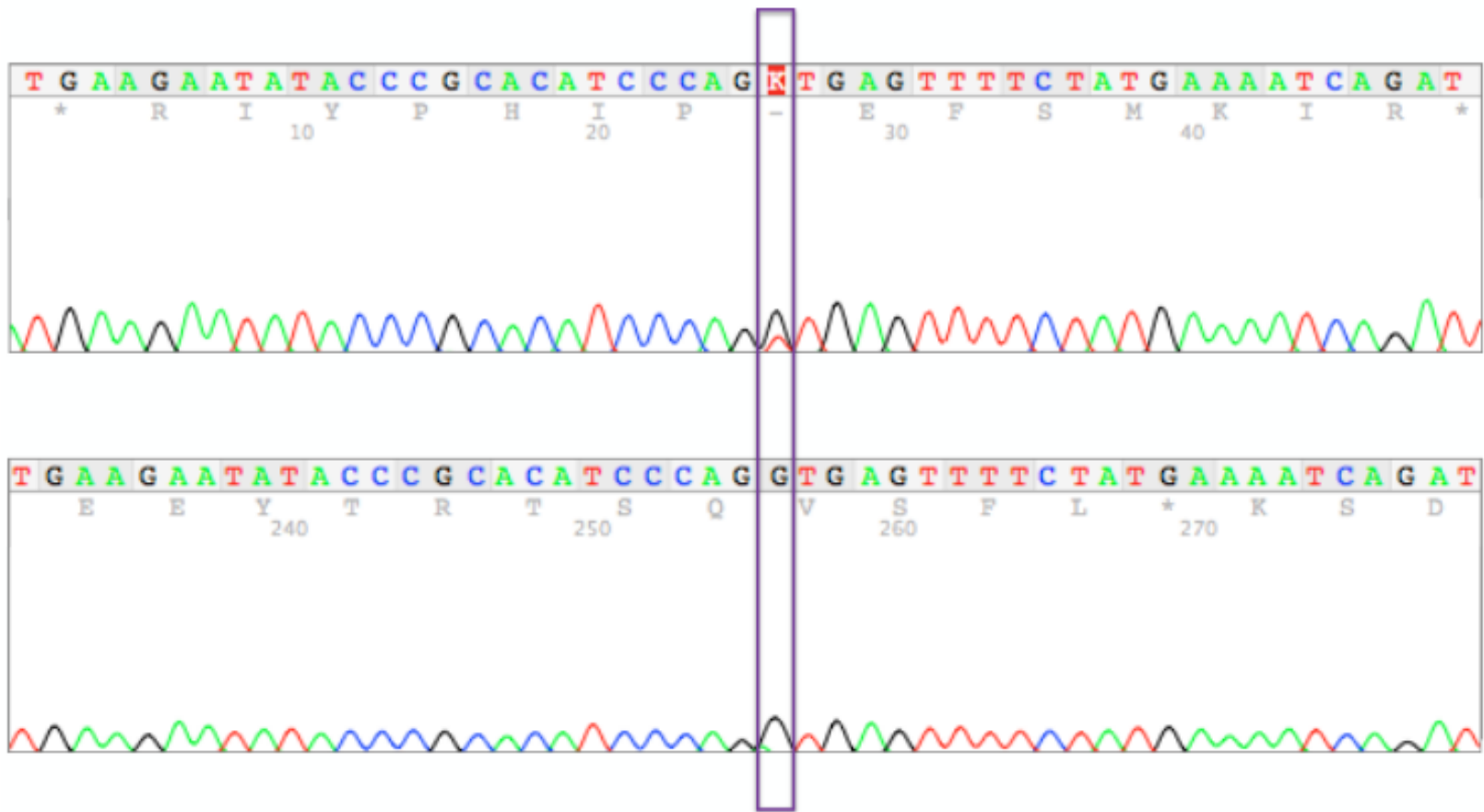
CGH

Exome Sequence

Single nucleotide substitution

PIK3R1 gene

c.1425+1 G>T



Both parents:
no mutations in PIK3R1

Suggestions for management are very welcome:

HSCT?

mTOR inhibitors?

others?

Thank you!

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