



ASBAI

Associação Brasileira de
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Case report

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History

- V.S.L., female, DOB October/2004
- Second child of non-consanguineous parents of Brazilian family
- No family history of immunodeficiency
- Born at term, after a normal pregnancy
- She received all vaccines according to the national immunization program with no adverse reaction

History

- 2mo: pruritic skin lesions (recurrent) → dx = atopic eczema
- 8mo: UTIs (*Pseudomonas aeruginosa*, *E. coli*, *Shigella sp.*) – recurrent: absence of structural or functional abnormalities
- 1yr8mo: vomiting, diarrhea, distended stomach, weight loss → paralytic ileus + severe candidiasis esophageal + Knee arthritis (MRN) (hospitalization – 3mo: ICU 1mo): antifungal, antibiotics, TPN – enteral)
- 8yrs3mo: fever, diarrhea, dehydration with sudden loss of consciousness (recurrent) (ICU) *CPR 2x
- 8yrs6mo: dengue hemorrhagic fever + encephalitis (ICU) *CPR 1x
- 10yrs: pneumonia (ICU)

History

- Others:
 - candidiasis: oral (2 – 9mo, 7yrs) and genital (10yrs)
 - cervical cellulitis (1x), herpes zoster (1x)
 - recurrent skin rashes, recurrent arthritis (knees, ankle)
 - acute diffuse hair loss, sudden loss of nails, absence of baby teeth loss



History

- Age at evaluation: 11yrs
 - cushing face, partial white hair, dry skin xerosis, short stature, turner stage 2

- *Medications:
 - Prednisolone (0.5mg/kg/d) since 2yrs
 - Hydroxychloroquine 160mg
 - MTX 15mg
 - Dietary restriction of cow milk, soy and wheat

Laboratory Data

Blood Counts	11yrs
Hb	14,3g/dL
Leucocytes	7770/ μ l
Neutrophils	6061/ μ l (80%)
Lymphocytes	1088/μl (14%) ↓
Platelets	333.000

T/B/NK	7yrs
CD3+	3010/ μ l
CD4+	2080/ μ l
CD8+	773/ μ l
CD19+	1126/ μ l
CD16/56+	↑14%
CD4/CD8 ratio	2,7

Igs	10yrs
IgG	799mg/dl
IgM	139mg/dl
IgA	↓ 69mg/dl

*ANA and RF : negative
 * Ac anti-thyroid : negative

-Thyroid, Renal and Liver functions:
 normal

-GH : normal
 - **IGF1 : 596** ↑

Sequenciamento do gene AIRE

Gene	Notation	Frequency in population	Zygoty
AIRE	c.937G>A:p.G3135	0%	Heterozygoty

**Father and sister were found to have the same mutation

??

APECED in heterozygotous