CASE PRESENTATION
II Encontro ASBAI-BRAGID

MÁRCIA BUZOLIN, M.D.

COLABORADORA AMB. IMUNOLOGIA – UNICAMP

PROF. DR. ELI MANUSR, M.D., PHD.

Case Presentation

ID:

- Caucasian woman, 22yo, referred to the Allergy & Clinical Immunology service of the University of Campinas-Brazil

Chief Complaint :

- recurrent infections since childhood

Family history:

- She was the third child of young and non-consanguineous parents of caucasian Brazilian background who had a previous spontaneous miscarriage

Pregnancy and Birth History:

- poor fetal movements and maternal edema in the third trimester. Delivery was at term by cesarean section, weight was 2,750 g and length 46 cm, being detected with clubfeet and hypotonia in the neonatal period.

Case Presentation

History of Present Illness:

- She had been followed since the age of 11 yo by the Medical Genetic service of the same University, due to neurological disabilities, dyslexia, clubfeet, scoliosis, and chronic dermatitis.
- Early childhood: rubella and chicken pox, eight episodes of throat infection, two pneumonias and intermittent diarrhea; generalized eczema; recurrent furunculosis.
- several episodes of low respiratory tract infections.
- moderate/severe rhinitis and intermittent asthma
- In adulthood she was diagnosed with hypothyroidism

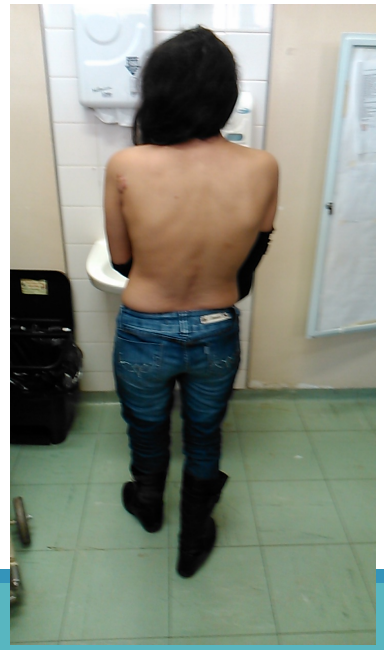
Immunization: up to date, unremarkable adverse reactions

Developmental history: global delay → referred to a special school since the age of 18 months.

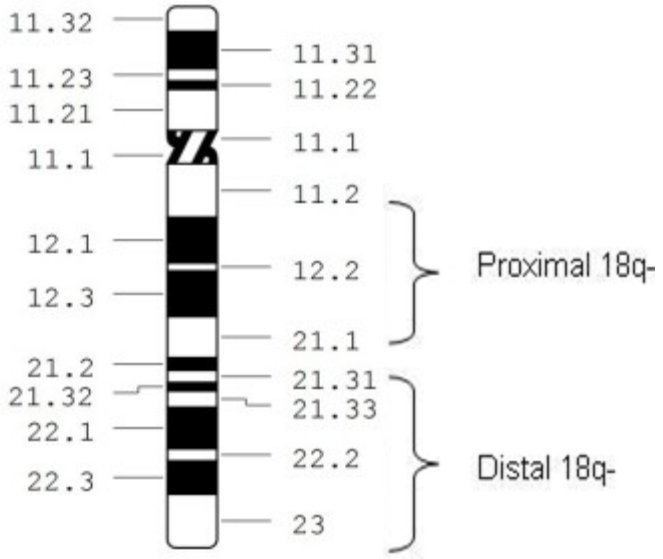
Case Presentation

Physical examination:

- 11 yo: short stature with small hear circumference
- high forehead, malar hypoplasia, triangular face, dysmorphic ears, synophrys, upslanting palpebral fissures, inner epicanthal folds, prominent nose, prognathism, bowed superior lip, short phyltrum, cleft uvula, asymmetric thorax with prominent sternum, scoliosis, short fourth and fifth metacarpals and metatarsals, clinodactyly of the third toes, hypotonia, and several small scars in the skin.
- Final height is 146 cm.



Case Presentation



Chromosomal analysis on lymphocytes with G-banding technique revealed a 46,XX,inv(9)(p12q13)pat,del(18)(q22)dn constitution–i.e, demonstrating a polymorphic pericentric inversion of a chromosome 9, paternally inherited, and a distal deletion of the long arm of a chromosome 18, *de novo*.

Case Presentation

Lab findings:

- markedly reduced levels of plasma immunoglobulins
- severe reduction of CD19+ cell counts

Treatment:

- intravenous immunoglobulin s (IVIG) replacement therapy 25g (~580 mg/kg) every 21 days

	level (mg/dL)
IgG	21,8 (830-2040)
IgM	24,5 (57-212)
IgA	<27,5 (80-476)
IgE	<4,6

RBC	5,33 x 10^6 /mm^3	**CD3**	88,7 % – 709 cells/mm^3
Hemoglobin	14,50 g/dl	**CD3 + CD4**	67,0 % – 536 cells/mm^3
Hematocrit	42,80 %	**CD3 + CD8**	16,1 % – 128 cells/mm^3
WBC	7,43 x 10^3 /mm^3	**CD3/CD4**	4,16
Neutrophils	5,83 x 10^3 /mm^3	**CD19**	0,9 % – 7 cells/mm^3
Lymphocytes	1,06 x 10^3 /mm^3	**CD16 + CD56**	3,7 % – 29 cells/mm^3
Monocytes	0,37 x 10^3 /mm^3	**CD3/CD16 + CD56**	0,4 % – 3 cells/mm^3
Eosinophils	0,14 x 10^3 /mm^3		
Platlets	119 x 10^3 /mm^3		

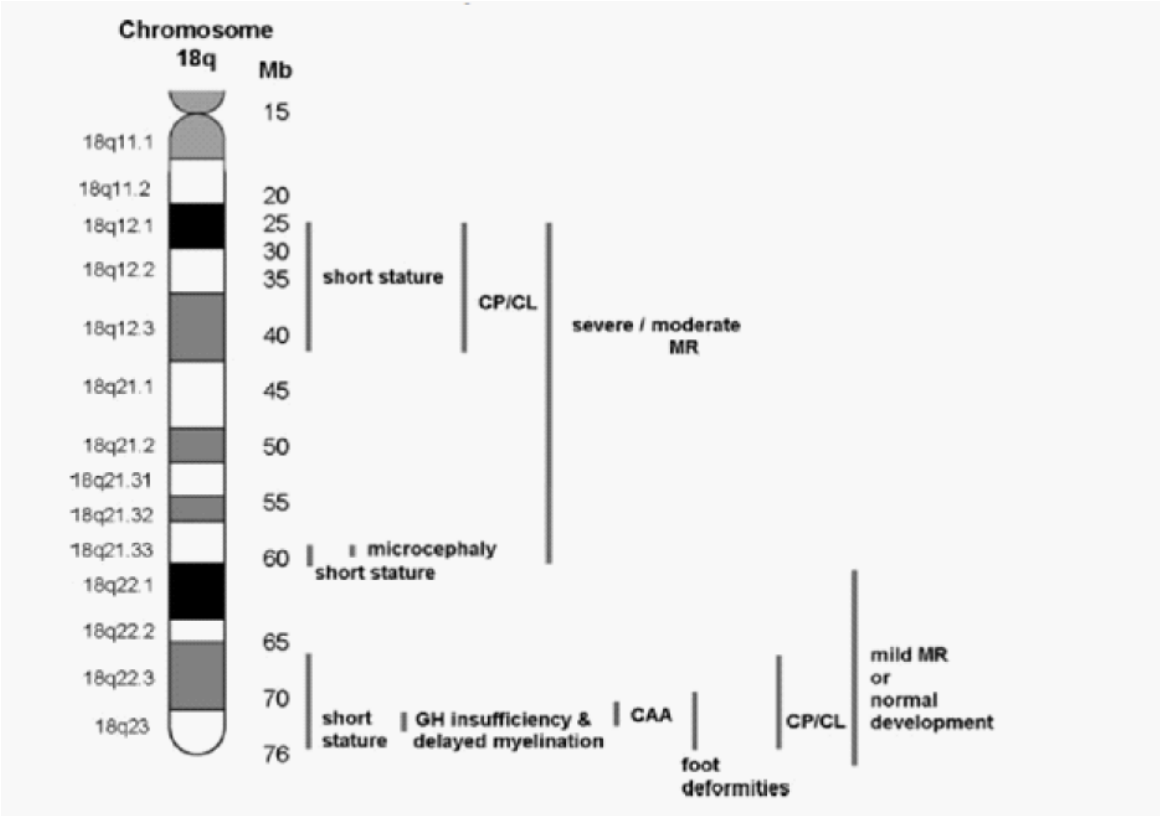
Discussion (MIM #601808)

Chromosome 18 abnormalities → 1:40,000 live births

- most frequent autosomal deletion syndromes in humans
- female predominance with a M:F ratio estimated 0.71

Immunological findings

- absence or markedly reduced level of serum IgA in almost half of the cases
- association with other autoimmune or immunodeficiency diseases such as thyroid abnormalities, common variable immunodeficiency (CVID), rheumatic arthritis, celiac disease, and insulin-dependent diabetes mellitus



Conclusion

Thus, this presents clinical evidence of the association of 18q deletion syndrome with hypogammaglobulinemia and CD19+ deficiency.

Our case report should encourage the evaluation of cell counts in patients with 18q deletion syndrome and immunodeficiency.