A Case of Agammaglobulinemia and Pneumocystis Jiroveci Pneumonia

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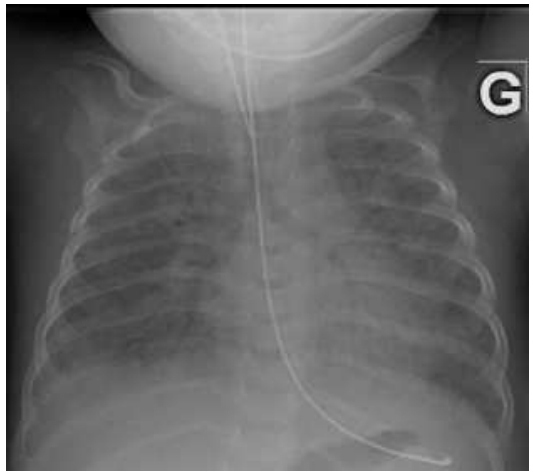
♀ 3 months old

ARDS 2nd PJP pneumonia

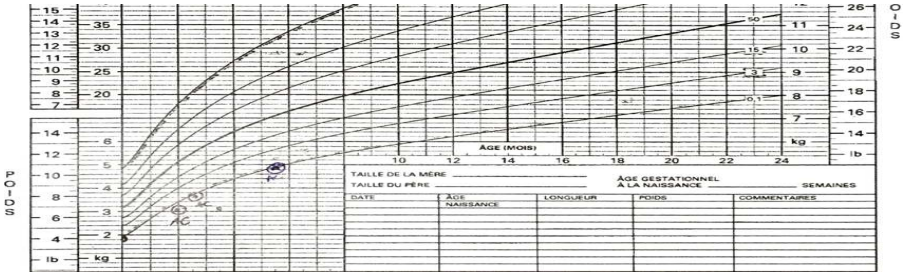


PMH

- Born prematurely at 33 2/7
  - Symmetrical IUGR (BW = 1080g)
- Chronic vomiting & intermittent diarrhea with FTT
- Hypotonia, global developmental delay

Family history

- ½ French Canadian, ½ Republic of Benin
- No consanguinity
- All healthy



Immunogical investigations

ANC (x10^9/L)	0.9 – 1.5
ALC (x10^9/L)	0.7 – 3.9
Hb (g/L)	73
Plt (x10^9/L)	N

IgG	0,41 g/l
IgA	< 0,01 g/l
IgM	< 0,01 g/l

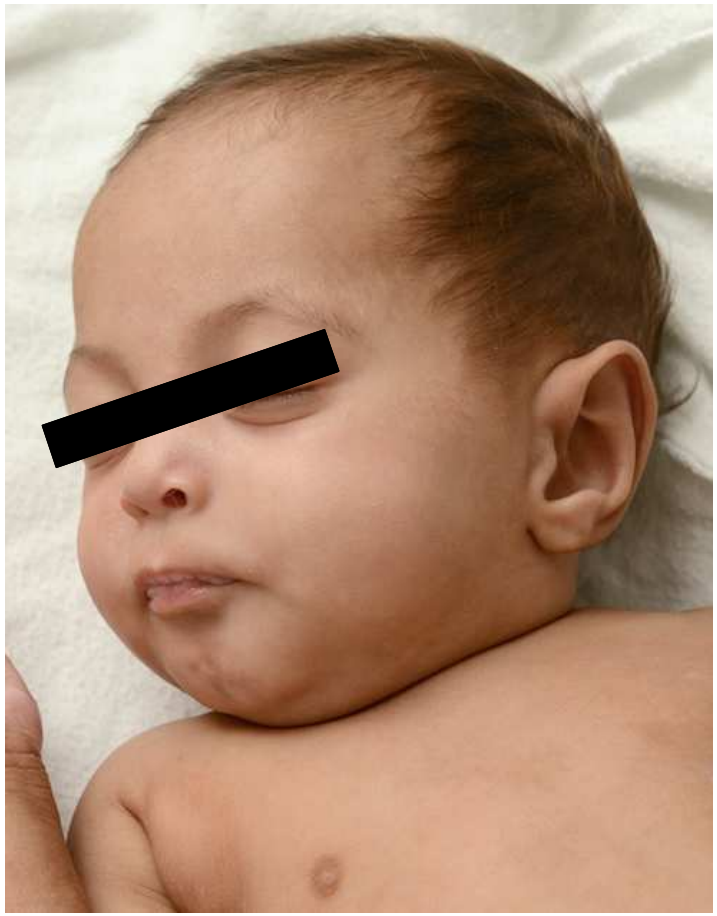
- Telomeres: Normal
- HIV: negative
- AFP: 1140 mcg/L ↑

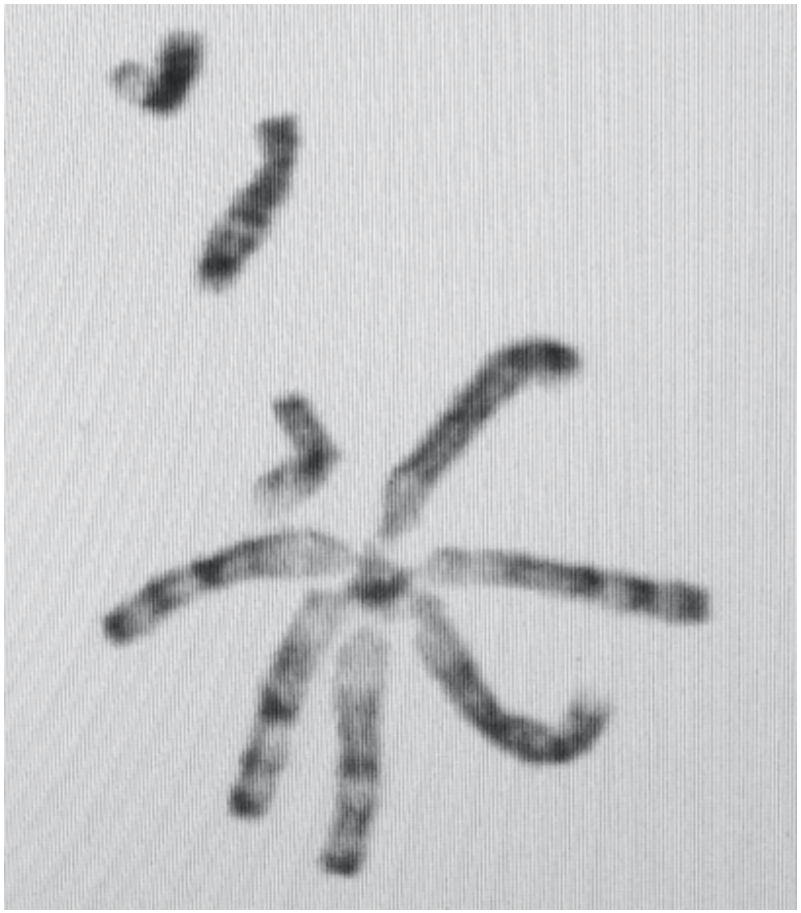
Total lymphocytes	4600
CD3+	2852
CD3+CD4+	2476
CD3+CD4+CD45RA+CD31+	32% ↓
CD3+CD8+	230
CD19+	966
CD19+CD27+	<0.01%
CD16+CD56+	262

- Vβ2 repertoire: Normal
- PHA proliferation : Normal
- OKT3 proliferation: LLN, complete restauration by IL-2

DDx?
Other investigations?

Karyotype





- Multiradial figures and centromere decondensation of chromosomes 1, 2, 10 & 16

ICF syndrome

Immunodeficiency, Centromeric instability and Facial dysmorphism

Immunodeficiency

Low Ig in the presence of B cells

Centromeric instability = hallmark of the disease

Characteristic rearrangement of the juxtacentromeric heterochromatin of chromosomes 1, 16 (±9)

Facial dysmorphism

Hypertelorism, flat nasal bridge, epicanthus

✧ DNMT3B & ZBTB24 sequencing: negative
✧ HELLS mutation: pending

Management

Options:

- IVIG and ATB ppx / HSCT/ Eventually: GT?

Our patient:

- Initially put on IVIG, septra, Synagis
- At 11 months: UCB transplant 8/10
  - Conditioning regimen: Bu + Cy + ATG
  - GvHD ppx: MMF & CsA
- Evolution:
  - Capillary leak syndrome at day +10 and day +100
  - GI GvHD
  - Chimerism 100% donor
  - Very slow immune reconstitution