

Case Report
T-B-SCID AR
R.V. 11.12.02
Diagnosis:

Severe Combined Immunodeficiency
T-B-SCID. Mutation not found.

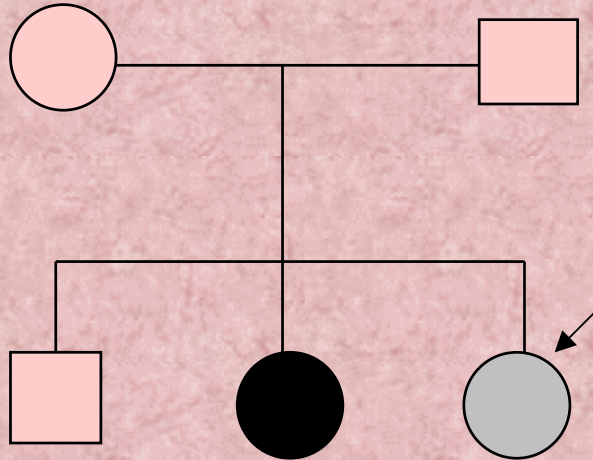
Generalized BCG-it is. Allogenic BMT
28.11.02, 14.01.03. EBV-lymphoma.

Hemophagocytosis. Sepsis Ps. Aeruginosa, in
blood culture 27.10.03.

Immune reconstitution.

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Anamnesis



- BCG-vaccination at first week
- At 2 weeks - candidiasis, conjunctivitis
- At 3 month- pneumonia, first hospitalization
- At 4 month- diarrhea, in stool identified klebsiella, failure to thrive,
 - hospitalization in ICU (1 month) treated with antibiotics, not irradiated blood transfusion from father
- In blood lymphopenia from first month
- At 8 month - SCID was diagnosed

First hospitalization in clinical immunology department

- At 9 month - first hospitalization in Russian State Clinical Hospital
- At the admission to Russian State Children Hospital - weight-4600g, height - 62 cm, failure to thrive, generalized BCG-itis
- Immunophenotype: lymphocytes- $0.4 \times 10^9/l$
CD3+ 22%, CD4+ 13%, CD8+19%, CD19+ 1%, Ig profoundly decreased
- Mutation was not found (evaluated in Erasmus University Rotterdam Department of Immunology, Prof. J.J.M. van Dongen



19/09/02 - 16/08/03 - BMT Department

In BMT unit: low weight, fever, generalized BCG-it is, mix infection

Conditioning - flu 5 mg/kg+ ATG 90 mg/kg.

BMT from HLA-matched brother.

NC $5,5 \times 10^8$ /kg, MNC $1,4 \times 10^8$ /kg, CD34+ nd

After BMT- sepsis, fever, generalized BCG-itis, poor condition

antibiotics: imipenem, cefipim, vanko, amikacin,

antifungal: ambisome 3 mg/kg/day,

antivirus: gancyclovir

antimucobacterial: INH, PZA, ETB, RIF

methilprednisolone 2 mg/kg

+30 day-activation of mucobacterial infection,
hemophagocytosis in bone marrow.

EBV-limphoproliferative syndrome (PCR EBV++, IgM
6550 mg/dl - monoclonal)

Treatment: VP16 5 mg/kg/w N2, Mabthera 375 mg/m²

N3- without effect

In BMT unit

+48 day- second BMT, without conditioning

+53/+15 day fever, liver, spleen, cytopenias,
hemophagocytosis in bone marrow

Treatment VP 16 5mg/kg 2 times at week without
effect

+58/+20 day severe condition, fever, polyorganic
insufficiency

Treatment Dexamethasone 10 mg/m²

for GVHD prophylactic CsA 1 mg/kg IV

AB IL-2aR, TNF

after treatment good results: normal level of
leucocytes, platelets, lymphocytes 1800/mkl,
without fever

increase weight, without organic disfunction

In BMT unit

+143/+83day- activation of mycobacterial infection and hemophagocytosis

poor condition: fever, new elements of BCG-it is, increase liver, spleen

Treatment- 5 antimycobacterial drugs: INH, PZA, ETB, RIF, CIP; VP16 100 mg/m², N3; dexametasone 20 mg/m²-short course, then decrease dose-with good effect

+155/+95 day hemophagocytosis

Treatment:dexa 20 mg/m² - with good effect.

Mucobacterial activation and hemophagocytosis when dexa was decreased

Good effect of high dose Dexa (20 mg/kg)

+261/+201 day-lymphocytes $1 \times 10^9/l$, without activation of mycobacterial infection and hemophagocytosis

treatment: 5 antimucobacterial drugs; dexa 8 mg/day; cefepime, itraconazole, Bactrim, valtrex

16/08/2003-30/06/2004 - Clinical Immunology Dept.

General: cushingoid baby, sitting in the bed,
elements of BCG-it is under the skin

Treatment: 5 antimycobacterial agents; dexamethasone-8mg/day
with decrease the dose; 3 antibiotics IV; antifungal;
antiviral therapy; IVIG 0,5g/kg/weekly

+320/+272 day- low count of lymphocytes, donor
chimerism of lymphocytes 7% - no lymphocytes
engraftment

+334/+284day- *pseudomonas aeruginosa* sepsis with
skin abscesses, fever, disseminated intravascular
coagulation

Treatment: piperacillin/tazobactam; amikacin; 5
antimycobacterial agents; antifungal; antiviral
therapy; Pentaglobin 0,4g/kg weekly N4;
dexamethasone 2 mg/day; heparin 100 ME/kg;
plasma 150 ml/day

transfusions of donors lymphocytes 10^5 /kg- 10^6 /kg
/weekly N10



Clinical Immunology department

+419/+371day - lymphocytes 1250 cells, CD3+ 84%, CD4+29%,
CD8+55%, CD19+3%, CD16+/56+ 10%, HLADR+29%

+449/+401day - +547/+499 day - semi sterile regimen, no signs of
mycobacterial and other infections and hemophagocytosis

Treatment: IVIG 0,5g/kg/month , 5 antimycobacterial drugs,
antibacterial, antifungal, antiviral prophylaxis, dexta 0,5 mg/day

+542/+495 day -lymphocytes 1450 cells, CD3+ 81%, CD4+33%,
CD8+34%, CD19+1%, CD16+/56+ 14%, HLADR+14%, low level of Ig

proliferation of lymphocytes	625	N	500-1500
answer on T-mitogen FHA	26935	N	20000-80000
Index of stimulation	43,1	N	20-75
answer on T,B-mitogen ML	9391	N	5000-15000
Index of stimulation	15,0	N	5-25

Left hospital 2 years 6 month:

Weight 11 kg, cushingoid baby, can go, can not speak

Duration of hospitalization from 09.09.02 to 30.06.04



**During last two years
(from 30/06/04-15/02/06)**

Clinical Status:

No evidence of infections

Mucobacterial infection didn't activated

Hemophagocytosis was not activated

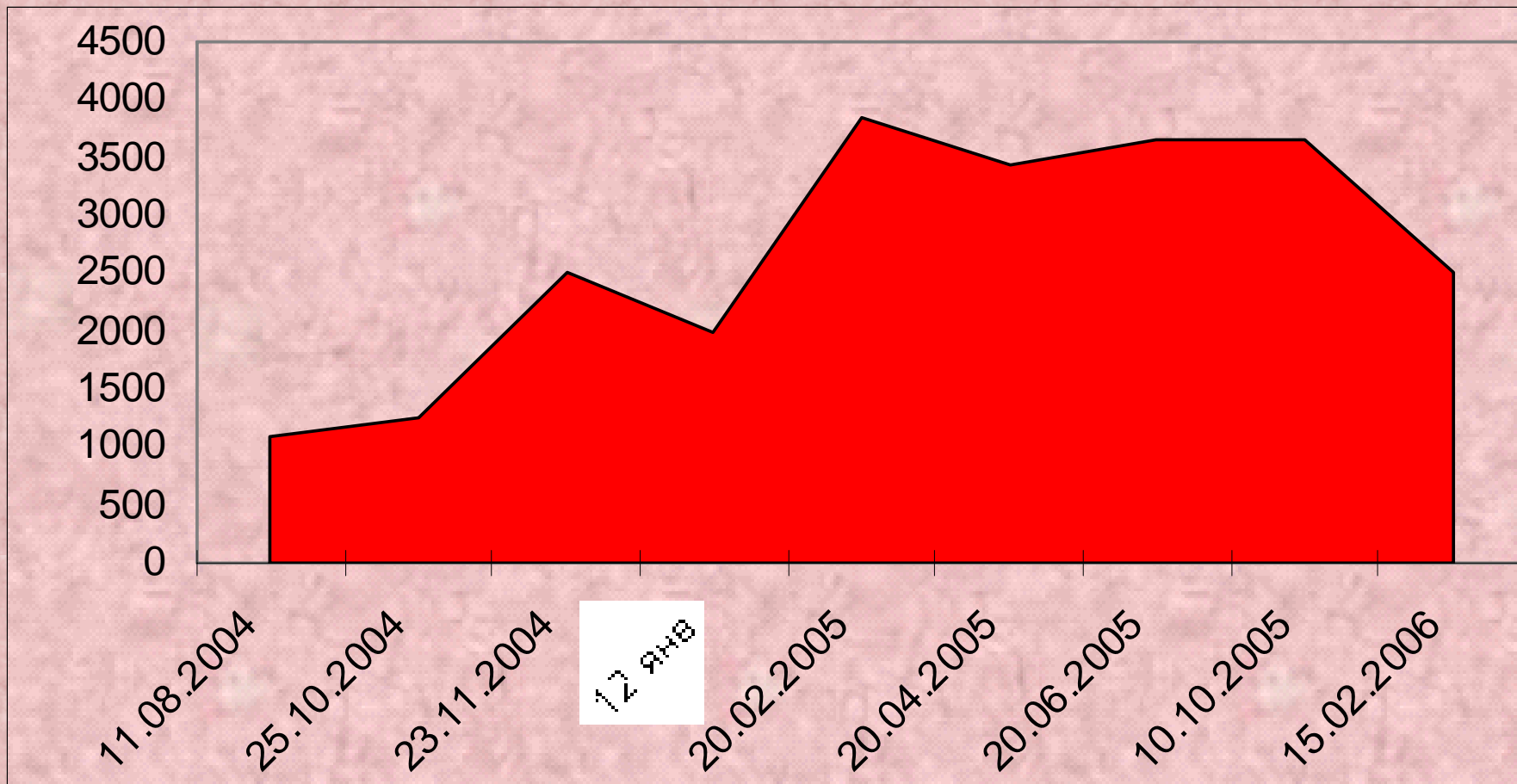
She is not cushongoig baby without osteoporosis

**Treatment: IVIG, bactrim, antivirius, antifungal stop
in 11/2005**

Decreased dexta and stop in 02/06

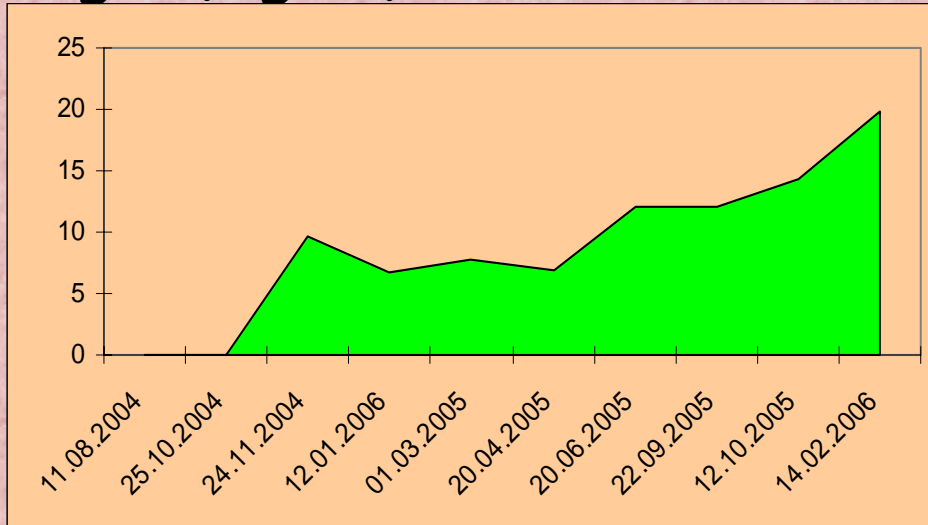
Cortisol 172 nmol/l - N

Lymphocytes count (30/06/04-15/02/06)

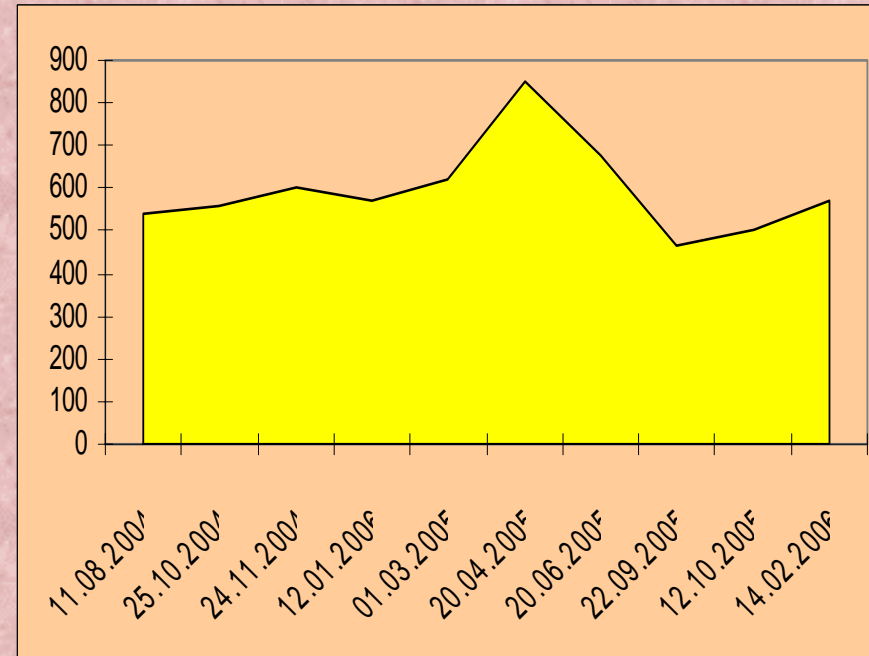


Level of immunoglobulins (11/08/04-15/02/06)

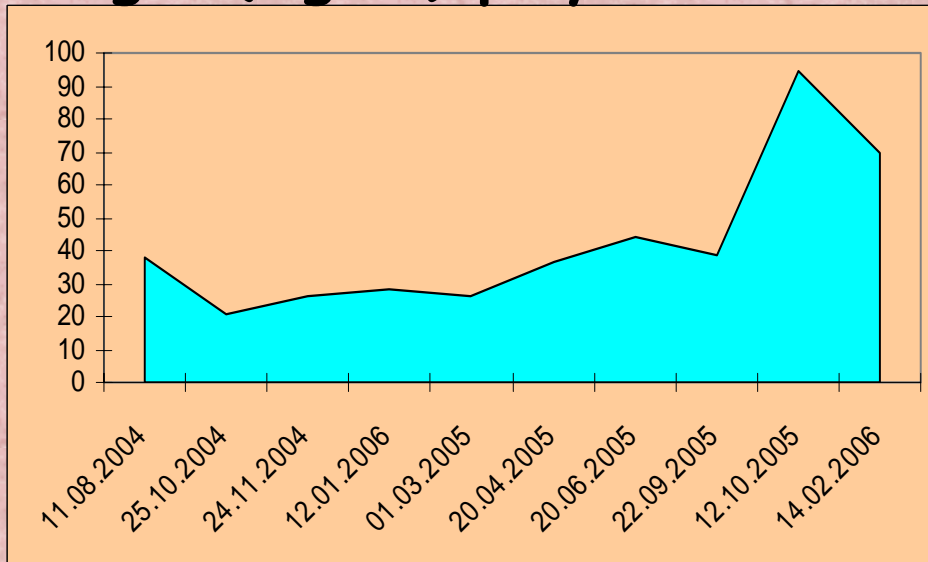
IgA (mg/dl)



IgG (mg/dl)



IgM (mg/dl) polyclonal



11/2004





02/2006

Four year old

