Capital City Council Heim Pál

Children’s Hospital

Department of Dermatology

Mh.code t012210821
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F I N A L R E P O R T

Name : Bernadett Horváth

Place of birth: **Budapest**
Date of birth: **01.10, 1996.**
Social security number: 110260246

Address :
No 39 Tegzes str. XXII. district,1222 Budapest
Blood type:

Document No: 1200513786

Time of admission: 22:03 hrs, 08.27, 2005.
Released: 09:00 hrs, 09.19, 2005.

DIAGNOSIS:
Kindler syndrome in obs
Scleroderma, PPS in ibs
Lichen sclerosus bullosa in obs

EPICRISIS:
Anamnesis of the 9 yr old female patient includes virtiligo on the father’s side. At 3 months of age after receiving active Hib vaccine patient developed extensive and unresponsive soor oris in addition to failing to thrive. Patient developed a variety of dermatological symptoms afterwards.
Wide range of allergologic examinations was performed. Patient’s parents mentioned allergy to several extrinsic allergenes (agents)(for example: steroids, local intrinsic immune-stimulants (immunolulars), internal use of neurofen, etc.)
Patient was hospitalized on numerous occasions; however apart from test results no final reports were prepared .
Main dermatological symptoms include photosensitivity, avoidance of light, poikiloderma-like lesions, hypopigmentation with extensive interconnected atrophic lesions and with occasional bullas and erosions. The aforementioned symptoms are accompanied by extensive pain, restricted movement and contractures.
Based on the clinical symptoms diagnosis of Kindler syndrome was considered by the treating physicians at the university clinic in the USA where the family lived at the time. DNA analysis was performed and was negative for the mutation, according to the verbal information received by the family. Our hospital did not succeed in getting in touch with the American clinic.
Based on the progrediating symptoms histological analysis was performed and an additional diagnosis of bullosus lichen and morphea was made. In the histological sample: ……..
Haematological examinations were required.
Desinficient and hydrating externas were used as local therapy.

Autoimmune positivity eosinophilia, histology

CONSULT:
08.29, 2005 Ophthalmology:
Peaceful bulbi. Corneas are resistant to staining, but multiple old spherical blurs and cloudiness are apparent on both sides, back side is free.
Therapy: Actovegin in the evening
Dr Véghseő

08.30, 2005 Cardiology:
Sharp, rhythmical heart sounds. No audible murmur.
ECG: Sinus rhythm. Normal heart axis.
Signs from thoracic leads are age-appropriate.
No pathology.

No cardiologic irregularity can be found.
Dr Kiss

09.06, 2005. Ophthalmology:
Patient is difficult to examine. Eyelids are inflamed, barely examinable with slit lamp, epithelial disruption suspected on the left side of the cornea, papillas are normal.
Therapy: 5x artificial tears on both sides, 3x Vitamin A oil
E: Tobrex eye-oinment.
Check-up in 1-2 days.
Dr Gottlieb

09.08, 2005 Surgery:
Biopsy
Check-up + bandage tomorrow.
Dr Hegyi

09.09, 2005 Surgery:
Good general state. Feverless.
Bandage: Epithelial proliferation in the wound.
Check-up on Monday.
Dr Hegyi

09.09, 2005. Ophthalmology:
St. on. Upper eyelids are livid, conjunctivas are hyperaemic.
Cornea is not brownish with irregular surface (right side > left side), rich angiogenesis.
Staining is only adsorbed on the right side.
Therapy: Tobrex in the evening and during the day, Vitamin A.
Recommendation: Tears Naturale II. or free.
Dr Véghseő

09.12, 2005.. Surgery:
Bandage: peaceful wound.
Check-up: 19th
Dr Hegyi

Imaging examinations:
08.29, 2005. X-ray results:
Hiluses are mildly richer, no other radiological irregularity.
dr.Bitvai/sz ... .

09.02, 2005. X-ray results:
Oesophagus is normally spacious and peristaltic movement is regular.
No apparent rigid parts.
DR Bitvai/G,

09.05, 2005. X-ray results:
Thick arch bones are normal, sutures, vessels, impression pattern are fine.
Sella is smaller, but with intact outlines, no apparent pathological lime shadow in the projection.
Thicker dorsum sellae, no apparent intracranial lime.
Opinion: smaller sella DerKádas

09.05, 2005. US results: Liver is mildly enlarged with homogenous structure.
Bile ducts are not dilatated. Gall bladder is well-filled with no wall thickening.
No apparent focal irregularity of the pancreas.
Surrounding of the big vessels is partially shown without any specific irregularity.
Spleen is normal in size with homogenous structure.
Both adrenal gland regions are free.
Kidneys are normal in location and structure.
Right kidney is 89x34 mm. Left kidney is larger, 108x38 mm, with a mildly dilated

pyelon, 14 mm. No sign of stones.
Bladder is almost empty.
No gastro-oesophageal reflux found during the examination of the cardia. The pylorus is regular, stomach empties well.
Intestines are mostly gas-filled, no pathological feature is apparent in the visible areas. Opinion: Mild hepatomegaly, pyelectasia on the left side.
dr Héjj/I

EXAMINATION RESULTS:
09.01, 2005.. Body weight: 12kg.

Blood count:
08.29, 2005.
White blood cell: 13.38 G/l, Red blood cell: 4.87 T/l
Haemoglobin: 140.00 g/l, Hematocrit: 0.40 l/l, MCV: 81.10 fl
MCH: 28.70 pg, MCHC: 354.00 g/l, RDW: 12.50 %
Trombocyte: 428.00 G/l, MPV: 8.90 fl, Lymphocyte: 14.00 %
Intermedial cells: 22.70 %, Granulocyte: 63.30 %, Blood smear: *
Evaluation: mechanical , Stab: . %, Segment: 63.3 %, Lymphocyte: 14.0 %
Monocyte: 8.1 %, Eozinophil: 14.3 %, Basophil: 0.3 %, Other: . ,

09.01, 2005.
White blood cell: 6.97 G/l, Red blood cell: 4.08 T/l
Haemoglobin: 116.00 g/1, Hematocrit: 0.34 1/1, MCV: 83.10 fl
MCH: 28.40 pg, MCHC: 342.00 g/l, RDW: 12.40 %
Trombocyte: 327.00 G/l, MPV: 9.20 fl, Lymphocyte: 26,70 %
Intermedial cells: 39.40 %, Granulocyte: 33.90 %, Blood smear: *
Evaluation: mechanical, Stab: . %, Segment: 33.9 %, Lymphocyte: 26.7 %
Monocyte: 9.0 %, Eozinophil: 29*7'%, Basophil: 0.7 %, Other: . ,

Serum:
08.29, 2005.
Se Total protein: 77 g/l, Se Albumin: 45 g/l
Se Karbamide: 2.0 mmol/1, Se Creatinin: 36 umol/1, Glucose: 6.1 mmol/1
Se Bilirubin: 8.0 umol/1, GOT: 31 IU/1, GPT: 11 IU/1, GGT: 9 IU/1
CRP: 21 mg/l, Antistreptolysin titer: 51 IU/ml, IgM: 1.26 g/l

09.01, 2005.
CRP: 25 mg/1

09.07, 2005.
Se Total IgE: 37 kU/1

09.07, 2005.
Tree mixture I sp Ige: 0 RAST, Tree mixture II sp Ige: 0 RAST
Grass mixture sp IgE: 0 RAST, Bermuda grass (Cynodon dactylon) sp IgE: 0 RAST
Natural rye sp Ige: 0 RAST, Ragweed mixture sp Ige: 0 RAST
Ribwort plantain sp IgE: 0 RAST, Mugwort sp IgE: 0 RAST
Parietaria sp Ige: 0 RAST, Cat epithelium sp Ige: 0 RAST
Dog epithelium sp IgE: 0 RAST, Dust mite sp IgE: 0 RAST
Dust mite sp IgE: 0 RAST, Imperfect fungi sp IgE: 0 RAST
Aspergillus niger sp Ige: 0 RAST, Grime mold sp.IgE: 0 RAST
Vitellus sp Ige: 0 RAST, Egg-white sp IgE: 0 RAST
Milk sp IgE: 0 RAST, Casein sp Ige: 0 RAST, Sesame seed: 0 RAST
Wheat sp Ige: 0 RAST, Rye sp Ige: 0 RAST, Soy beans sp Ige: 0 RAST
Almond sp IgE: 0 RAST, Hazelnut sp IgeE: 0 RAST
Peanut sp Ige: 0 RAST, Walnut sp IgE: 0 RAST
Peach sp IgE: 0 RAST, Celery sp IgE: 0 RAST
Tomato IgE: 0 RAST, Potato sp IgE: 0 RAST, Apple sp IgE: 0 RAST
Carrot sp IgE: 0 RAST, Cod sp IgE: 0 RAST, Crab sp IgE: 0 RAST

09.07, 2005.
Se Total IgE: 37 kU/1

Urine:
08.30, 2005.
Urine specific gravity: 1005 , Urine pH: 8 , Urine WBC: Negative
Urine Nitrite: Negative , Urine Protein: Negative
Urine Glucose: Negative , Urine Ketones: Negative
Urine Ubg: Normal , Urine Bilirubin: Negative
Urine RBC, Hgb: Negative, Urine sediment: Negative

Stool:
09.01, 2005 Microbiological results:
Negative for worm eggs and protozoa.
With floating enrichment.

09.02, 2005.. Microbiological results:
Salmonella, Shigella, Y.enterocolitica, Campylobacter didn’t vegetate.

09.02, 2005 Microbiological results:
Negative for worm eggs and protozoa.
With floating enrichment.

09.05, 2005.
Stool worm eggs: Negative, Stool Giardia: Negative

09.03, 2005.
Rotavirus antigen: Negative
Adenovirus AG result: Negative

Bacteriology:
08.31, 2005.. Urine:
Mixed bacterial flora, probably contamination, repetition is recommended.

08.31, 2005.. Throat swab:
No pathological bacteria vegetated.

08.31, 2005.. Nasal swab:
Staphylococcus aureus:
Resistant: penicillin
Sensitive: oxacillin, amoxicillin/clavulanic acid, gentamicin, erythromycin, tetracycline, sumetrolim, vancomycin, clindamycin, ciprofloxacin

Carrier!

09.01, 2005. Wound swab:
Escherichia coli:
Sensitive: propicillin, ampicillin/sulbactam, amoxicillin/clavulanic acid, cefalexin, cefuroxim, ceftazidime, cefixim, gentamicin, sumetrolim, ofloxacin

Enterococcus SP::
Moderate: penicillin, gentamicin
Resistant: ampicillin, erythromycin, vancomycin, levofloxacin

Staphylococcus aureus:
Resistant: penicillin
Sensitive: oxacillin, amoxicillin/clavulanic acid, gentamicin, erythromycin, tetracycline, sumetrolim, vancomycin, clindamycin, ciprofloxacin

Pseudomonas aeruginosa:

Sensitive: cefoperazone, ceftazidime, tazocin, imipenem, meropenem, gentamicin,

tobramycin, amikacin, ofloxacin, ciprofloxacin, cefepime,

Microscopy results:
Stained smear examination
Gram + coccus
Gram + rod

Other results:
08.31, 2005.. Microbiological results:
Borrelia burgdorferi IgG+IgM VIDAS ELFA: negative

09.09, 2005. Immunology results:
Anti-nuclear Ab 1:40 : strongly positive gran.
Anti-nuclear Ab 1:160: positive
Centromere Ab : negative
Histon Ab : 5 0.00-40.00
Clq at : negative

SS-A (Ro) ELISA	1.00 U/ml	0.00-15.00
SS-B (La) ELISA	1.00 ü/ml	0.00-15.00
Scl-70 ELISA	1.00 U/ml	0.00-15.00
Jo-1 ELISA Sm	1.00 U/ml	0.00-15.00
ELISA RNP/SM	1.00 U/ml	0.00-15.00
ELISA Anti-DNS	1.00 U/ml	0.00-15.00
ELISA	5 IU/ml	0.00-20.00
Nucleosoma at.	3 U/ml	0.00-20.00

10.04, 2005. Histological results:
Flattened skin-part with atrophic epithelium, with general hyperkeratosis and focal parakeratosis on the surface. Histological picture is uncommon and complex with the most conspicuous feature being a ribbon-like, strongly epidermotrophic lymphoid lesion in the upper part of the dermis, as a result of which it was sent to the 1st Institute of Pathology for standardization (see results in the attachment). In the basal epithelial layer mostly only moderate vacuolisation is apparent with a occasional necrotic keratinocytes (this resembles GVHD).
Based on the findings to this point, I would categorize the disease as piokiloderma congenitale, but in other views the histological view is completely different, it resembles LSA, maybe superficial morphea, with regards to the compactness and the lack of cells. In these latter parts the epithelium seems to be detaching from the connective tissue, in fact, even follicular keratosis is apparent, thus by all means the primary option is LSA. In the epidermotrophic regions small clusters of multi-nuclear giant cells were seen in the upper part of the dermis, which was thought to be the result of elastolysis and were stained with orcein, but neither on these slides, nor on further serial slides were these cells found again.
I request continuous further consulting!
Type of excision: diagnostic excision
Histological dg.: See above.
Other specified dermatitises.
Dr Hársing Judit

10.04, 2005. Histopathological results:
Microscopic description:
Histology:
The biopsy sample contains a part of the skin without subcutis. Epidermis is atrophic with signs of ortokeratotic hyperkeratosis and focal parakeratosis. The basal cell-line is vacuolised with a significant number of lymphocytes penetrating between the cells. Branch-like, moderate lymphoid infiltration is apparent in the papillary dermis with periodic interface activity. Moderate perivascular infiltration is apparent in the superficial dermis.

No apparent vesicle or bulla formation or single-cell epithelial necrosis in the sample.

Immunohistochemistry:
Vast majority of the infiltrating cells are CD3 positive T-lymphocytes with CD4 predominance.

Molecular examination:
T-cell receptor gene rearrangement examination was performed on the reisolated DNA from the implanted skin and no clonal gene product was detected.

Opinion:
Based on the patient’s anamnesis, age, lack of T-cell receptor gene-rearrangement and histological image, where extensive epidermotrophism is apparent, lymphoproliferative processes can most probably be excluded. No definitive diagnosis can be given, but apart from the previously mentioned diseases, pellagra and Hartnup disease should also be considered.

Diagnosis: Chronic dermatitis
Prof.Dr Matolcsy András
Dr Csomor Judit

10.09, 2005. EmA results:
IgA class endomysium autoantibody (EmA): negative
IgÁ class reticulin autoantibody (ARA): negative
IgA class coeliakia-specic jejunal autoantibody (JeA): negative

IgG class endomysium autoantibody (EmA): negative
IgG class reticulin autoantibody (ARA): negative
IgG class coeliakia-specic jejunal autoantibody (JeA): negative

Opinion and recommendation:
Results contra-indicate gluten-sensitive enteropathy. If small intestinal biopsy is performed, please refer the histological results to me.
Dr Korponay-Szabó Ilma

10.06, 2005.. Histopathological results:
Macroscopic description:
I. Native smear
II, 1 tube with EDTA for peripheral blood-lymphocytes, DNA, 1 tube with citrate for peripheral blood.

Microscopic description:
Cytology: No pathological features are apparent in the peripheral smear apart from moderate eosinophilia. No apparent promyelocytes or other features indicating haematological diseases.

Diagnosis, opinion and recommended therapy:
Peripheral blood, moderate eosinophilia.

Flow cytometry:
During the repeated flow cytometry measurements the double population disappeared, probably as a result of cell degradation. Blood shows normal phenotype-distribution.

Flow cytometry results:
Controls (to detect aspecific immunoglobulin binding)
IgG FITC IgG RPE IgG RPE Cy5 among all the cells are below 1.0%

Cell-distribution based on the CD45-SSC diagram:
lymphoids: 25.2 %

monocytes: 8.3%
neutrophil granulocytes: 64%

T and B cells among all other cells:
CD 19 B cells: 3.8%
CD 3 T cells: 16.5%

Distribution of sub-populations among lymphocytes:

CD3 T cells:	65.6%
CD4 T cells:	45.9%
CD 5	74.8%
CD8 T cells :	17.9%
CD 10	5.2 %
CDI9 B cells:	14.9%
CD20	11.8%
CD22	16.2%
CD23	13.2%
CD38	63.7%
CD56	7.2 %
CDI 03	1.4 %
Kappa	15.6%
Lambda	8.9 %

B lymphocyte subpopulations:

CD19-CD5 double positive (pronounced CD5 expression)	31.3%
CD19-CD38 double positive (pronounced CD5 expression	81.4%
CDI9-CDI0 double positive	28.2%
CD23-CD5 double positive (pronounced CD5 expression)	25 %

T lymphocyte subpopulations:

CD4	: 71.9%
CD8	: 28.1%

Number of used triple stains: 7

Dr Matolcsy András, Dr Csomor Judit

THERAPY:
ung Hydrocortison-Buyow, gutt Fenistil, inj. Calcimusc, ung,
Chlorhexidin, sol Betadine, sol Corsodyl, ung Basis C, ung 5% Carbamide,
Cremor aquosum, ung Elocom-Chlorhexidin forte, ung Locoid-H, syr Erolin,
tbl Panadol, Tobrex eye-oinment, **Vitamin A oil**

PLEASE ALWAYS TAKE THIS FINAL REPORT WITH YOU TO CHECK-UPS, AND HAND THE SECOND COPY TO THE PEDIATRIC GP!

Budapest,09.19. 2005

Dr. Asbőth Dorottya
Department physician

Dr. Szalai Zsuzsanna
Head of Department

I acknowledge the receipt of a copy of the final report.

Signature of parent