Preterm baby suspected of Omenn syndrome

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- 3rd uncomplicated pregnancy, preterm delivery (31. week, 1580 g) at 14.5.03
- 2 older brothers healthy
- Intubated at the delivery room for RDS, diffuse erythrodermia, fragile thin skin, hair detaching when washing
- Relative lymphopenia, eosinophilia (up to 43%) in initial blood tests
- Severe diarrhoea and malabsorption at three weeks of age

Differential diagnosis

- Netherton syndrome: skin biopsy - hair normal, lymphocytic infiltration
- GVHD: HLA typing – 2 alleles for each locus
- Omenn syndrome – supportive signs:
  - IgE 1749 … 2453 IU/ml
  - lymphopenia, CD4/CD8 T-ly: 3,9
  - nearly all CD8+ activated (CD 38+), CD 25 increased, HLA II expression increased x CD 69 very low, decreased response to mitogen stimulation
  - B-ly decreased (below normal in absolute counts)
  - increased spontaneous PMN activation (90%)
  - no thymus

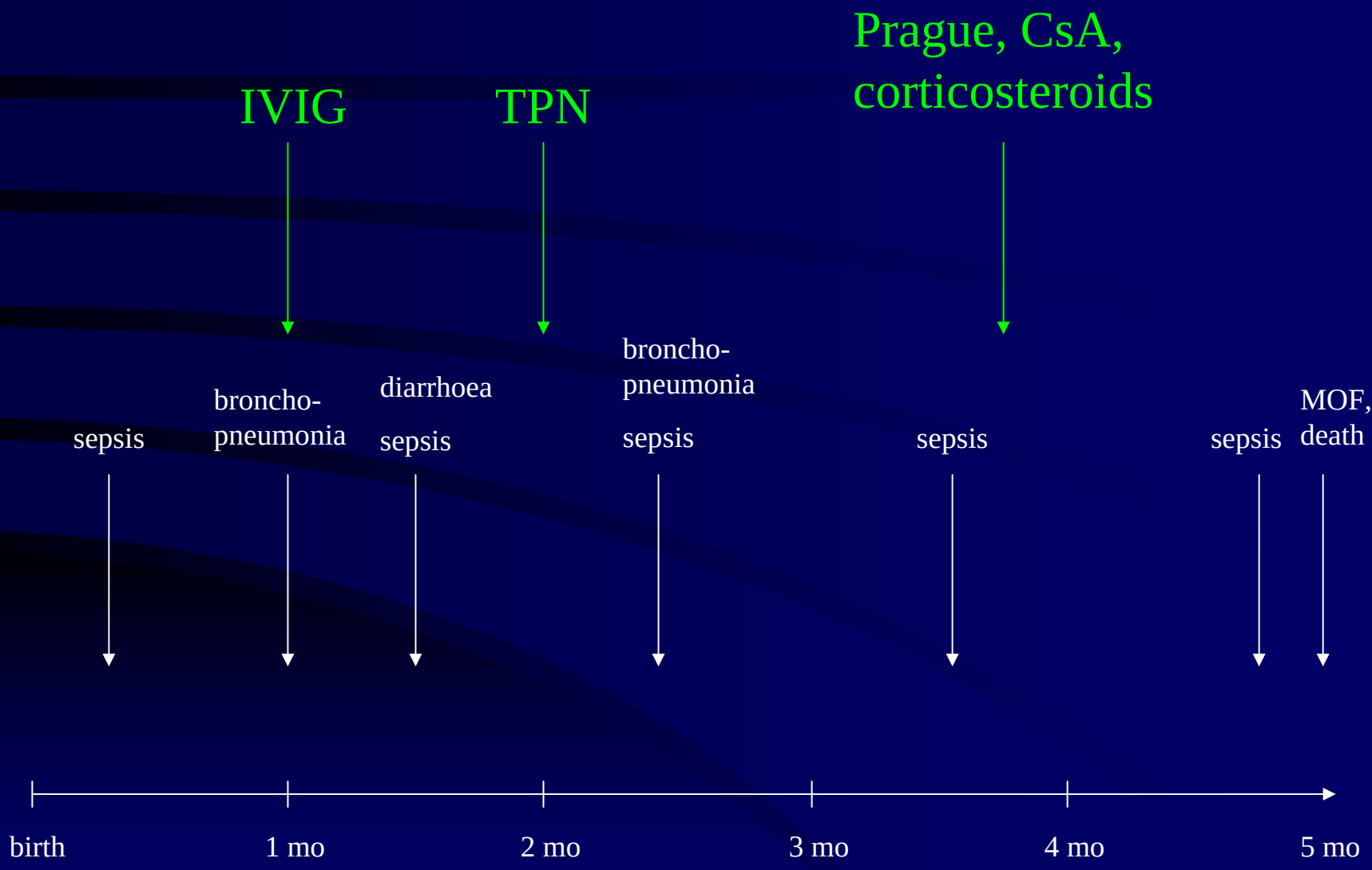
Differential diagnosis

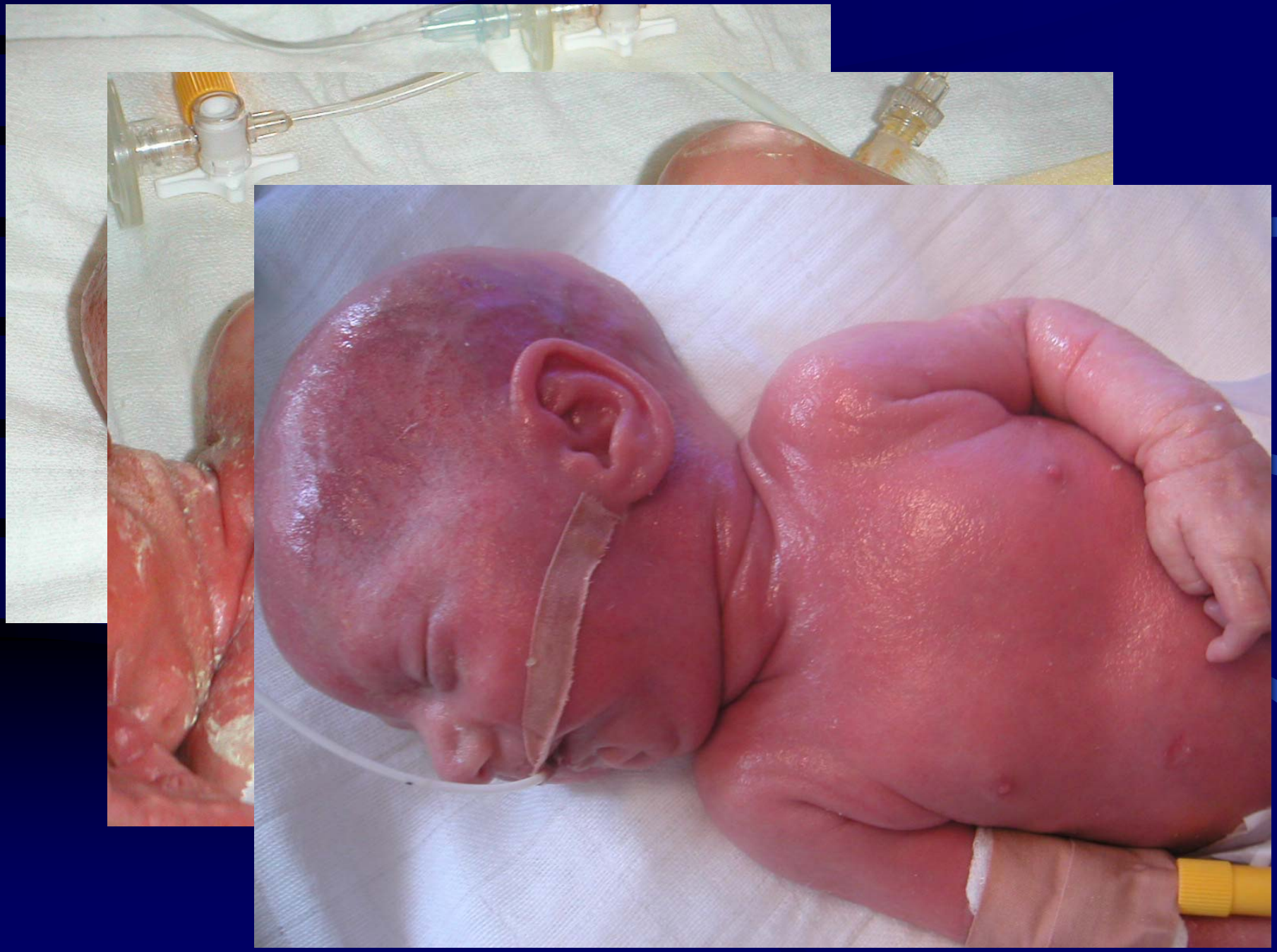
- Omenn syndrome – contrary signs:
  - no hepato/splenomegaly
  - traces of IgA and IgM on electrophoresis

    (IgG not tested before substitution)
  - negative RAG gene analysis
  - Artemis gene analysis underway

    (Prof. van Dongen, Rotterdam)

Clinical course





Conclusion

- Preterm baby with SCID condition closely resembling Omenn syndrome
- Negative RAG gene analysis does not preclude diagnosis – Artemis gene analysis pending
- Further follow-up of the family desirable

Thanks for your attention.