Omenn syndrome in siblings

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Family history

BOY, 31 weeks gestation
- 31 weeks gestation, 2nd gestation, 2nd childbirth, spontaneous birth
- Oligohydramnios
- APGAR 2, 2, 2, 3, birth weight 2420g (5.3lbs)
- CPR at delivery room
- Ventilation support DUOPAP

GIRL, 39 weeks gestation
- 39 weeks gestation, 3rd gestation, 3rd childbirth, spontaneous birth
- HSV infection in 2nd and 3rd trimester of pregnancy
- Minor infection (cold) during the 3rd trimester of pregnancy
- Mother: vaginal culture- E.coli ESBL(-)
- APGAR 10, birth weight 3420g (7.53 lbs)

Family history
Healthy mother, healthy father
First baby born prematurely at 36 weeks of gestation - healthy
No congenital immunodeficiency syndromes and skin diseases in the family
Physical examination

BOY, 31 weeks gestation
- Respiratory failure (critical condition)
- Erythema with extensive skin defects
- Maceration of the skin
- Bleeding erosions
- Without ectropion and eclabium

GIRL, 39 weeks gestation
- Good adaptation to extrauterine life
- Erythema with many blisters
- Epidermal/superficial desquamation
BOY, 31 WEEK GESTATION
Physical examination

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- Respiratory failure (critical condition)
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**GIRL, 39 WEEKS GESTATION**
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GIRL, 39 WEEK GESTATION
BOY, 31 WEEKS GESTATION

GIRL, 39 WEEKS GESTATION
Laboratory tests

BOY, 31 weeks gestation
- Leukocytosis - WBC 45 K/uL
- Eosinophilia (31,2%; 14,14 K/uL; N=0-0,5 K/uL)
- Monocytosis (10,7%; 4,8 K/uL; N=0-1,2 K/uL)
- Thrombocytopenia (16 K/uL, 37 K/uL)
- Hypoglycaemia

GIRL, 39 weeks gestation
- WBC 6,34 K/uL
- Eosinophilia (34,9%, 2,21 K/uL; N=0-0,5 K/uL)
- Monocytosis (18%; 1,14 K/uL; N=0-1,1k/uL)
- Lymphopenia (10,4%, 0,66 K/uL; N=2-17 K/uL)
- Skin culture- E.Coli ESBL(-), [Mother: vaginal culture- E.coli ESBL(-)]
Dermatological consultation

BOY, 31 weeks gestation
CLINICAL DIAGNOSIS:
ICHTHYOSIS

GIRL, 39 weeks gestation
CLINICAL DIAGNOSIS:
CONGENITAL DERMATOSIS
FOLLOW-UP

BOY, 31 weeks gestation
baby died at 3rd day of life
due to rapidly developing
multi-organ failure caused
by EOS.

GIRL, 39 weeks gestation
baby was discharged from the
Neonatal Department in good
condition at the 9th day of
life. Superficial desquamation
was still visible on the skin

Vaccination against
tuberculosis was postponed!!!
FOLLOW-UP
GIRL, 39 weeks gestation

- 4th week of life- readmitted to Pediatric Unit because of severe pneumonia.
- Severe skin desquamation
- Lack of hair and eyebrows
- Eosynophilia, lymphopenia
- Extremely low level of lymphocytes B
- Malabsorption, diarrhea
- Hepatosplenomegaly.

DIAGNOSIS: OMENN SYNDROME
RESULTS

The girl died at the age of 3 months expecting bone marrow transplantation.

Taking into consideration blood abnormalities and similarities of skin signs in both children, it is highly probable that second child, born prematurely, also suffered from congenital immunodeficiency syndrome.
TAKE HOME MESSAGE

- It must be remembered that combined immunodeficiency syndromes are inherited conditions and might manifest in the neonatal period.
- It is particularly important to be vigilant in case of erythrodermic skin changes or skin desquamation combined with abnormalities in blood tests.
- In the presence of severe skin lesion, vaccination with attenuated vaccines should be postponed.
- Differential diagnosis should include atopic dermatitis and ichthyosis.
Thank You for Your attention.