

A newborn with a rare association of various congenital skin disorders – Question

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Case report

We report the case of a female neonate admitted to our department a few hours after birth. The pregnancy was physiological, and she was born by spontaneous full-term vaginal delivery without complications. Apgar scores were 9 at 1 minute and 9 at 5 minutes. Birth weight was 3,075 g (39th centile), the length was 50 cm (66th centile) and head circumference was 33.5 cm (37th centile). On physical examination, she had congenital skin anomalies (**Fig. 1**). We observed extensive homogeneous erythematous patches involving her face, the upper left side of the chest, left shoulder, left arm, and most of the surface of her right leg and knee. Additionally, we found reddish marble-like reticulated patches on the back and blue spots partly mixed with them. Consequently, an ocular examination was performed, and it showed buphthalmos of her right eye. Echocardiogram

revealed a structurally normal heart with good contractility. Blood tests, abdominal and cranial ultrasound were normal, and no other abnormalities were found.

Questions

1. What kind of skin lesions do you recognize?
2. What is your diagnosis?
3. Which further investigations would you suggest?

Statement of Ethics

The patient's parents provided written informed consent. The study adhered to the tenets of the Declaration of Helsinki.

Declaration of interest

The Authors declare that there is no conflict of interest.



Figure 1. Clinical presentation at 2 days of age. Skin anomalies involving the front (**A**) and the back side (**B**) of the body.