PAEDIATRIC DERMATOLOGY

A GREYISH-BLUE, RED AND YELLOW CHINESE INFANT WITH ASYMMETRIC CRYING FACES: IS IT A DISTINCT UNKNOWN ASSOCIATION?

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Background: Phacomatosis pigmentovascularis (PPV) is a congenital syndrome characterized by the simultaneous occurrence of a pigmented nevus and a vascular nevus in an individual. Cayler cardiofacial syndrome is also a rare syndrome characterized by congenital unilateral hypoplasia of the depressor anguli oris muscle (DAOM) associated with congenital cardiac defects. We report a case of PPV type IIb with Cayler cardiofacial syndrome.

Observation: We report a case of a native Chinese infant. She was born at 37 weeks' gestation with extensive greyish-blue symmetric aberrant hyperpigmentation involving her trunk and extremities, and flat homogeneous erythematous patches with clear margins involving her face, cheek and planta pedis . She also has asymmetric crying faces in the form of drooping of the right angle of mouth when crying and normal faces while sleeping or being silent. Antenatal scan was normal and Apgar score was 10 at birth. Bilateral eye closure,wrinkling of the forehead and nasolabial furrows are intact and symmetrical. Thus, congenital facial nerve palsy is excluded and left-sided hypoplasia of DAOM is indicated. Echocardiogram confirmed the congenital heart disease of patent ductus arteriosus (2.0mm), patent foramen ovale (3-3.5mm). These findings are suggestive of Cayler cardiofacial syndrome. Complete blood count, basic metabolic panel, hearing screen, cranial magnetic resonance angiography and magnetic resonance imaging are normal. Neither family history nor consanguineous marriages were detected. So far, she has not been investigated with fluorescence in situ hybridization (FISH) analysis.

Key message: Our literature review indicates that this is the first report of PPV type IIb together with Cayler cardiofacial syndrome.