Letter to the Editor

Monozygotic twins discordant for port wine stains support the post-zygotic mutation hypothesis

To the Editor:

Port wine stains (PWS) are flat-thick, red-purple, cutaneous lesions most frequently located in the head and neck (1). The molecular basis for PWS is unknown, but is believed to represent an error in vascular development occurring during embryogenesis. Here, we report on two pairs of discordant monozygotic (MZ) twins with PWS. We suggest that post-zygotic mutation origin may play a role in the etiology factors of PWS.

A pair of 18-year-old male twins (A) referred to our department because of lateral facial with red nevus since birth (Fig. 1). On the clinical examination, the affected twin presented with an extensive PWS lesion on his right face without skin temperature increased and cardiac movement. The lesion mainly distributes in the V2 dermatomes. He was healthy without glaucoma and MRI indicated that the leptomeningeal vascular abnormalities and cerebral atrophy were not present. He had never undergone any therapy previously. There was no consanguinity in his parents; the family history was negative for PWS, hemangioma and other vascular malformations.

Three-year-old female twin pair (B) were born after natural pregnancy and uneventful conception. Delivery at 30 weeks of gestation, both babies stayed in the cabinet for 20 days. Weight at birth was equal 1800 g, respectively. The twin with birthmarks went to our Vascular Anomaly Center at the age of 1 year. The lesion presented as flat pink stains on her left periconchal and neck (Fig. 2). No skin temperature increased and fast-flow by Doppler examination was found. Once laser therapy was conducted, part of the stain fades when she was 3 years old and both twins went to our center (Fig. 3). Family history with PWS was not found from parents recall.

The phenotypic features of the twin pairs were nearly same (their looks, blood types, hand preferences, fingerprints etc.). To prove MZ, we performed a haplotype analysis using polymerase chain reaction amplification of 15 polymorphic markers in peripheral DNA, the samples were analyzed on an ABI PRISM 310 genetic analyzer, fragment lengths were determined in Genemapper 4.0 (ABI, Foster City, CA).

As far as we know, this is the first special reports of MZ twin discordance for PWS published to date.

Fig. 1. Monozygotic twins (A) discordant for port wine stains. (Right) The stain mainly distribute in the V2 dermatome on his right face.

Fig. 2. Affected twin (B) showing port wine stains on her left periconchal and neck.
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Most of the PWS lesions were sporadic without gender differences and typical familial case reports were rare. Facial PWS appear in a segment corresponding to a trigeminal root distribution support the maturational defect in the local sympathetic nervous system can lead to the lesions (2). Error in vascular development occurring during embryogenesis was speculated. There was no condition to check the placentation and therefore no presumed timing of embryo splitting. But from the patients' parent recall, both of the twins were monochorionic diamniotic twinning. This implies that post-zygotic events at the end of the first week of gestation may be a possible mechanism. Discordant MZ twin pairs of van der Woude syndrome were used to search for modifiers and proved post-zygotic mutations (3). Reports of twins discordant in various disorders indicate the post-zygotic regulation of development separately, such as proteus syndrome (4), phacomatosis pigmentovascularis (5, 6) and Sturge-Weber syndrome (7, 8). We report two pairs of MZ twin discordance for PWS. The differential expression of the PWS phenotype reinforced the hypothesis of post-zygotic mutation as the best mechanism of pathogenesis.

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