Letter to the Editor

Is there a genomic link and common pathogenesis (postzygotic mutations in beta-actin) for Poland syndrome and Becker nevus syndrome?

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I read with interest the excellent review of Poland syndrome by Hashim *et al.* (1). The investigators provide a current and comprehensive summary of all aspects of this extraordinary condition. Prominent features of Poland syndrome include pectoral muscle hypoplasia or aplasia, mammary hypoplasia and ipsilateral limb anomalies (1).

The researchers emphasize that several hypotheses have been suggested regarding the pathogenesis of Poland syndrome (1). The vascular disruption theory (resulting from a subclavian artery supply disruption sequence) is the favored mechanism of pathogenesis (2,3). However, a Poland syndrome patient with no arterial alteration has been described (4); therefore, at least in some patients with Poland syndrome, the vascular disruption theory may not be a uniform etiology for the development of this condition.

Becker nevus syndrome is a genodermatosis characterized by the presence of a Becker nevus (a circumscribed hyperpigmentation with hypertrichosis which often also has a concurrent smooth muscle hamartoma) associated with certain skin and extracutaneous abnormalities (5). Hypoplastic or absent breast and ipsilateral upper extremity skeletal abnormalities are present in several of the patients with Becker nevus syndrome (5). Indeed, it is rather remarkable that the morphologic presentation of Becker nevus syndrome has several phenotypic features similar to those observed in Poland syndrome.

The pathogenesis of Becker nevus syndrome has been demonstrated by Cai *et al.*: postzygotic mutations of the *ACTB* gene that codes for beta-actin (6). However, a definitive pathogenesis for Poland syndrome remains to be established (1,3). In addition to the other potential etiopathogeneses of Poland syndrome that Hashim *et al.* have summarized (1), based on the phenomenal clinical similarities demonstrated by Cai *et al.*'s Becker nevus syndrome patient who had unilateral left breast and pectoral muscle hypoplasia (6), I respectfully propose another etiology—that Poland syndrome may also result from postzygotic mutations in beta-actin (7).

In summary, Poland syndrome—similar to Becker nevus syndrome—has several systemic and dermatologic manifestations (1,8). In addition, both Poland syndrome and Becker nevus syndrome have identical phenotypic features (hypoplastic or absent breast and skeletal abnormalities of the ipsilateral upper extremity); these common features introduce the possibility that both syndromes share a common genomic aberration. Investigation as to whether or not patients with Poland syndrome have postzygotic mutations in beta-actin may be warranted.

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Footnote

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Ethical Statement: The author is accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

References

1. Hashim EAA, Quek BH, Chandran S. A narrative review of Poland's syndrome: theories of its genesis, evolution and its diagnosis and treatment. Transl Pediatr 2021;10:1008-19.
2. Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. Am J Med Genet 1986;23:903-18.
3. Shahrul Baharin N, Awadh Hashim E, Bin Huey Q, et al. Reinforcing the vascular disruption theory of the genesis of Poland's syndrome: a rare association of diaphragmatic eventration in a preterm infant with severe musculoskeletal defects. BMJ Case Rep 2021;14:238392.
4. Ferraro GA, Perrotta A, Rossano F, et al. Poland syndrome: description of an atypical variant. Aesthetic Plast Surg 2005;29:32-3.
5. Torchia D. Becker nevus syndrome: A 2020 update. J Am Acad Dermatol 2021;85:e101-3.
6. Cai ED, Sun BK, Chiang A, et al. Postzygotic Mutations in Beta-Actin Are Associated with Becker's Nevus and Becker's Nevus Syndrome. J Invest Dermatol 2017;137:1795-8.
7. Cohen PR. Poland's Syndrome: Are Postzygotic Mutations in β-Actin Associated with its Pathogenesis? Am J Clin Dermatol 2018;19:133-4.
8. Vazirnia A, Cohen PR. Poland's syndrome: a concise review of the clinical features highlighting associated dermatologic manifestations. Am J Clin Dermatol 2015;16:295-301.

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