The Role of Skeletal Muscle Hyperplasia

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Abstract

Beckwith-Wiedemann Syndrome is a rare congenital overgrowth disorder characterized by various clinical features, including macroglossia, gigantism, abdominal wall defects, and increased risk of childhood tumors. While the molecular basis of BWS has been extensively studied, the specifc mechanisms underlying the development of macroglossia remain unclear. This study aims to investigate the potential role of skeletal muscle hyperplasia in causing macroglossia in BWS. The keyhole-shaped partial resection revealed a harder posterior side than the anterior. Microscopically, the posterior side consisted of dense subepithelial eosinophilic areas composed of an abundance of tightly packed skeletal muscle fbers that were arranged in a fascicular or storiform pattern. BWS-associated macroglossia results from skeletal muscle hyperplasia, consistent with true macroglossia.

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B -W, S.

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IGF2, . U, BWS. F

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