Title: BPAG1 in muscles: structure and function in skeletal, cardiac and smooth muscle

Authors: Masao Horie, Nozomu Yoshioka, Hirohide Takebayashi

PII: S1084-9521(16)30320-2
DOI: http://dx.doi.org/doi:10.1016/j.semcdb.2017.07.016
Reference: YSCDB 2279

To appear in: *Seminars in Cell & Developmental Biology*

Received date: 21-2-2017
Revised date: 12-7-2017
Accepted date: 14-7-2017

Please cite this article as: Horie Masao, Yoshioka Nozomu, Takebayashi Hirohide.BPAG1 in muscles: structure and function in skeletal, cardiac and smooth muscle.*Seminars in Cell and Developmental Biology* http://dx.doi.org/10.1016/j.semcdb.2017.07.016

Seminars in Cell & Developmental Biology

BPAG1 in muscles: structure and function in skeletal, cardiac and smooth muscle

Masao Horie, Nozomu Yoshioka, Hirohide Takebayashi*

Division of Neurobiology and Anatomy, Niigata University, Niigata 951-8510, Japan

*Corresponding author
E-mail address: takebaya@med.niigata-u.ac.jp (H. Takebayashi)

Present address of MH
Department of Morphological Sciences, Graduate School of Medical and Dental Sciences, Kagoshima University, Kagoshima 890-8544, Japan

Abstract

BPAG1, also known as Dystonin or BP230, belongs to the plakin family of proteins, which has multiple cytoskeleton-binding domains. Several BPAG1 isoforms are produced by a single *BPAG1* genomic locus using different promoters and exons. For example, BPAG1a, BPAG1b, and BPAG1e are predominantly expressed in the nervous system, muscle, and skin, respectively. Among BPAG1 isoforms, BPAG1e is well studied because it was first identified as an autoantigen in patients with bullous pemphigoid, an autoimmune skin disease. BPAG1e is a component of hemidesmosomes, the adhesion complexes that promote dermal-epidermal cohesion. In the nervous system, the role of BPAG1a is also well studied because disruption of *BPAG1a* results in a phenotype similar to that of *Dystonia musculorum* (*dt*) mutants, which show progressive motor disorder. However, the expression and function of BPAG1 in muscles is not well studied. The aim of this review is to provide an overview of and highlight some recent findings on the expression and function of BPAG1 in muscles, which can assist future studies designed to delineate the role and regulation of BPAG1 in the *dt* mouse phenotype and in human hereditary sensory and autonomic neuropathy type 6 (HSAN6).

Keywords: Bullous pemphigoid antigen 1 (BPAG1), BP230, Dystonin (Dst), *Dystonia musculorum (dt)* mice, skeletal muscle, myocardium.