

**LAUGHTER IS NOT ALWAYS FUNNY - BREATH HOLDING IN FAMILIAL
DYSAUTONOMIA**

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Autonomic mechanisms are believed to be involved in breath-holding spells (BHS); therefore, we investigated this phenomenon in Familial Dysautonomia (FD) patients who exhibit various symptoms and signs due to the dysregulation of their autonomic nervous system.

Ninety FD patients were retrospectively evaluated for BHS in their childhood.

Our results showed that FD patients exhibited BHS while laughing (10%) as well as while crying (88.8%). (BHS with laughter has not yet been described in medical literature) Five patients experienced BHS while doing Pulmonary Function Testing. The incidence of BHS in FD patients was 53.3% in comparison to 4%-21% in normal populations. The mean age when BHS started was 1.03y (SD \square 0.72y), the mean age at which symptoms stopped was 5.66y (SD \square 2.66y), the mean duration of BHS was 4.65y (SD \square 2.5 y), and the family history of BHS was mostly negative. Excessive vagal tone may play a role in pathogenesis of BHS. Assessment of heart rate variability as a function of vagal stimulation proved to be increased in FD patients.

We conclude that BHS in FD patients is more prevalent, prolonged and severe and can occur in crying as well as in laughter or while doing lung function tests. This emphasizes the importance of the autonomic nervous system in the mechanisms underlying this phenomenon.

