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Journal
Dermatology Online Journal, 22(9)

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Publication Date
2016

DOI
10.5070/D3229032521

Volume 22 Number 9
September 2016

Abstract

Basal Cell Carcinoma Nevus Syndrome: Population of patients negative for Chromosome 9 mutations

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Dermatology Online Journal 22 (9)

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Patients with Basal Cell Carcinoma Nevus syndrome are assumed to have a chromosome 9 mutation, such as a PTCH1 [9q22] mutation, despite the majority of patients with BCCNS being diagnosed through the presence of major and minor diagnostic criterion. The main treatment for BCCNS, Hedgehog inhibitors (HhI), targets these mutations. The results of an internet based survey of self-identified adults with BCCNS which was launched through SurveyMonkey with access provided by the Basal Cell Carcinoma Nevus Syndrome Life Support Network (www.BCCNS.org). Of a total of 395 participants, 282 and 289 individuals responded to questions regarding PTCH1 and chromosome 9 genetic testing, respectively. Those who reported having been genetically tested for either PTCH1 and/or chromosome 9 mutations, were limited to 14-28 % of all BCCNS patient respondents. Of those genetically tested, 12% reported they were negative for PTCH1 and 11% were negative for chromosome 9 mutations. Biogenetic pathway testing needs to be performed on a higher percentage of phenotypical BCCNS patients and the results correlated with HhI treatment responses. These results suggest the need to recognize that phenotypical BCCNS consists of multiple subpopulations of patients with differing biogenetic signaling mutations making current treatment less than adequate for phenotypical BCCNS individuals.