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Abstract

Diagnostic parameters of children with basal cell carcinoma nevus syndrome

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Basal Cell Carcinoma Nevus Syndrome (BCCNS) is a multi-system genetic disease characterized by the development of multiple basal cell carcinomas (BCCs), macrocephaly, medulloblastomas, jaw keratocysts, and coarse facial features, amongst other symptoms. The major and minor criteria for adults with BCCNS are often extrapolated for children, however, little is known about the disease presentation of children with BCCNS. Our study focused on bringing the pediatric presentation of BCCNS to light. To the best of our knowledge, we are the first to investigate BCCNS and its medical impact on children. Using an internet accessible survey, we asked parents and guardians about the presenting symptoms of BCCNS in their children. It was found that at least 75% of children were diagnosed with BCCNS by the age of ten or earlier, which suggests that the presentation of disease starts much earlier than previously reported. Moreover, at least 19% of parents or guardians reported that their children had 50 or more BCCs by the age of diagnosis. It is our hope that these results will help clinicians be aware of the possible diagnosis of BCCNS at earlier ages in these children. An earlier diagnosis could provide the medical specialty-specific support services that may prevent the development of other medical consequences that arise from the burden of disease of BCCNS.