Pediatric Dermatology

OnlineEarly Articles

To cite this article: Theresa R. Pacheco M.D., Lisa S. Scatena M.D., Edward J. Hoffenberg M.D., Jane Gralla Ph.D, Lela A. Lee M.D.
Café au Lait Macules and Juvénile Polyps
Pediatric Dermatology (OnlineEarly Articles).
doi:10.1111/j.1525-1470.2007.00386.x

Next Article

Café au Lait Macules and Juvénile Polyps

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Abstract

Abstract: Several hereditary and nonhereditary gastrointestinal tract polyposis syndromes exhibit extra-intestinal manifestations, including cutaneous findings. However, a lack of information exists regarding cutaneous features of juvenile polyposis. Our objective was to document the prevalence of cutaneous hyperpigmented lesions in children with juvenile polyposis coli or juvenile polyposis coli and their first degree relatives. Children seen in the gastroenterology practice at The Children's Hospital in Denver, Colorado with polyps (juvenile polyposis coli, sporadic juvenile polyps, and familial adenomatous polyposis coli) and their first degree relatives were invited to participate in the study. A comprehensive skin examination was performed on those who consented to participate. We found that 8 of 14 patients (eight with juvenile polyposis coli, four with juvenile polyposis, and two with familial adenomatous polyposis coli) had at least one café-au-lait macule, compared with three of 27 relatives ($p = 0.003$). The prevalence of at least one café-au-lait macule in our patients (8/14 or 57.1%, CI: 28.9–82.3%) was significantly higher than the general population prevalence of 28.5% ($p = 0.023$). However, if the two patients with familial adenomatous polyposis coli were excluded, the comparison with the general population prevalence did not reach statistical significance ($p = 0.095$). The prevalence of multiple café-au-lait macules in our patients (4/14 or 28.6%; CI: 8.4–58.1%) was significantly higher than the general population prevalence of 5.2% ($p = 0.005$). A notable finding was the presence of multiple café-au-lait macules in 4 of 12 juvenile polyposis coli/juvenile polyposis patients. Two patients with juvenile polyposis coli also had lentigines. In this selected case series, we observed single or multiple café-au-lait macules in a high proportion of children with the three types of polyps. Further studies are needed to assess a possible common pathway for hamartomatous polyps and café-au-lait macules.