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**ABSTRACT #:10552**

**Background :** Multiple hereditary exostoses is an autosomal-dominant skeletal disorder that has a wide ranging reported risk of malignant degeneration to chondrosarcoma.

**Questions/purposes:** The aims of our study were to use a large, web-based survey approach to characterize (1) the demographic distribution of patients with multiple hereditary exostoses, (2) the number of surgeries performed related to one's diagnosis of multiple hereditary exostoses, and (3) the proportion of survey respondents who described experiencing malignant degeneration in a large international, heterogeneous cohort of patients with multiple hereditary exostoses.

**Methods:** An anonymous web-based survey was distributed to several online support groups and social media networks designed to support and educate patients with multiple hereditary exostoses and their families. The survey collected demographic and epidemiologic data on 779 respondents. Data were recorded to assess respondents' disease burden and the rate of malignant degeneration.

**Results:** Females represented a slightly greater proportion of those with multiple hereditary exostoses who responded (56% female; 419 of 742 patients). Median age for all respondents was 28 years (range, 1–85 years). Median age for males was 25 years (range, 1–85 years), while median age for females was 29 years (range, 1–82 years). The mean age at diagnosis of male and female respondents was in the mid-first decade (5.4 years  $\pm$  7.2 years). The mean number of surgeries a patient had undergone was 7.3 ( $\pm$  7.1 surgeries). The proportion of respondents who experienced malignant transformation was 2.7% (21 of 757 respondents), at a mean age of 28.6 years ( $\pm$  9.3 years). The most common sites of malignant change from benign exostoses included the pelvis (eight of 21 respondents) and scapula (four of 21 respondents).

**Conclusions:** In the largest and most geographically diverse study of patients with multiple hereditary exostoses of which we are aware, we found the proportion of patients with multiple hereditary exostoses who have undergone malignant degeneration to be consistent with those reported in prior studies. Our study perhaps more accurately assessed the proportion of patients who undergo malignant transformation of multiple hereditary exostoses. As with prior studies on this topic, the proportion of malignant change may be expected to represent a high-end estimate as recruitment and selection bias likely predisposes for patients with more severe disease, whereas patients with lesser disease may be unaware of their diagnosis. In discussing the sequelae of multiple hereditary exostoses, clinicians perhaps might use this study to offer an unspecific statement of risk of malignant degeneration of multiple hereditary exostoses among the population at large.

## Geographic Distribution of Respondents

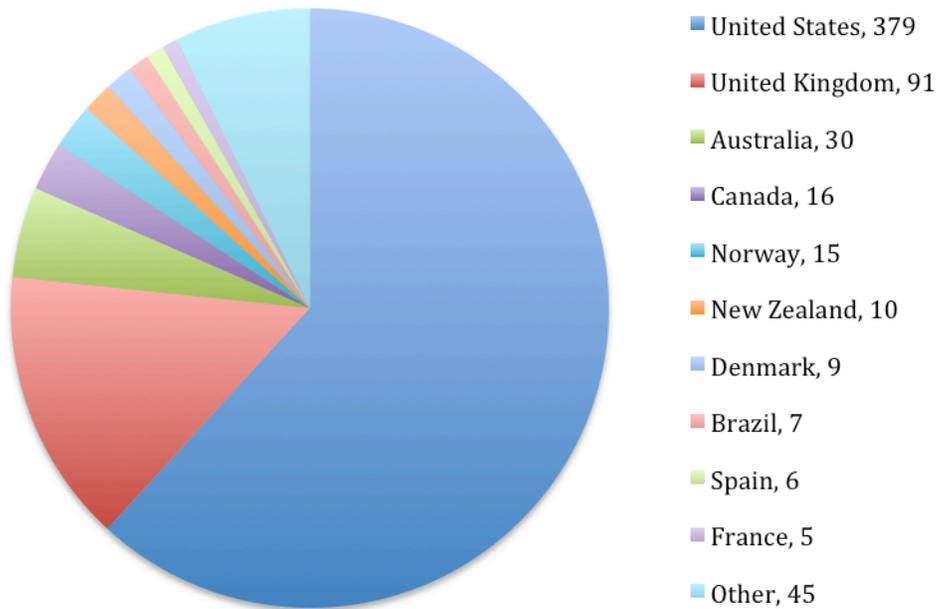


Figure 1: This figure demonstrates the geographic distribution of applicable respondents. Other locations include the following: Costa Rica (4), Israel (4), Portugal (4), Italy (3), Mexico (3), Sweden (3), Belgium (2), Germany (2), Guatemala (2), Indonesia (2), Jordan (2), Malta (2), Peru (2), Poland (2) Serbia (2), Algeria (1), Argentina (1), Bulgaria (1), Colombia (1), Finland (1), Haiti (1), India (1), Ireland (1), Japan (1,) Netherlands (1), Panama (1), Puerto Rico (1), Romania (1), Russia (1), Singapore (1), South Africa (1), South Korea (1).

- 1) Do you have a diagnosis of multiple hereditary exostoses (MHE)? (yes or no)
- 2) If you do NOT have a diagnosis of MHE, are you completing this survey on behalf of a relative (child, sibling, parent, grandparent, etc.) who is diagnosed, or on behalf of a relative who was diagnosed but is now deceased? (yes or no)
- 3) At what age were you or the patient diagnosed with MHE? (free text)
- 4) To the best of your knowledge, have you or the patient had any surgeries related to MHE? (yes or no)
- 5) If you answered yes to question 4, how many surgeries have you or the patient had? (free text)
- 6) To the best of your knowledge, have you or the patient been diagnosed with chondrosarcoma as a result of your MHE? (yes or no)
- 7) If yes, at what age were you or the patient diagnosed with chondrosarcoma? (free text)
- 8) To the best of your knowledge, at which bone(s) were you or the patient diagnosed with chondrosarcoma? (free text)
- 9) If you are completing this survey on behalf of someone else, what is your relationship to that patient (i.e. child, parent, sibling, grandparent)? (free text)
- 10) What is your current age (or that of the patient you are responding on behalf of)? (free text)
- 11) Sex of the patient: (male or female)
- 12) Country of birth: (free text)
- 13) Please use the space below to add any comments, suggestions, or questions. Thank you for your participation. (free text)

Table 1: This table represents the survey distributed to each respondent via [www.surveymonkey.com](http://www.surveymonkey.com).