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Prevalence of Osteochondromas in the Spine in Patients with Multiple Hereditary Exostoses

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Background: Multiple hereditary exostoses (MHE) is an autosomal-dominant disorder characterized by the development of multiple cartilage-capped exostoses originating from the physis that are known as osteochondromas. The potential for these osteochondromas to impinge on the spinal cord is a clinical concern. The aim of our study was to determine the prevalence of osteochondromas in the spine in individuals with MHE. Additionally, we aimed to identify any risk factors for neural-impinging osteochondromas.

Methods: We prospectively enrolled a cohort of patients and their family members with MHE at a single institution from 2010 to 2022. Demographics, osteochondroma location, and clinical outcomes were documented. Magnetic resonance imaging (MRI) scans were made and interpreted by a pediatric musculoskeletal radiologist. Patients were categorized based on osteochondroma location: no spinal involvement, on the spinal column, in the spinal canal but not impinging, or neural-impinging. We also noted when osteochondromas were present on the ribs and pelvis to assess if these were predictive of spinal involvement.

Results: Ninety-four patients with MHE (50% female; 78% White; mean age, 23 years) were enrolled. Fifty (53%) had no spinal involvement. Twenty-two (23%) had osteochondromas located on the spinal column, 18 (19%) had osteochondromas in the spinal canal, and 4 (4%) had an osteochondroma causing neural impingement. Of the 4 with neural impingement, 2 displayed paraparesis requiring immediate surgical intervention. The remaining 2 patients were observed clinically and monitored with use of serial MRI scans. One patient developed symptoms and underwent surgical excision of the osteochondroma. The remaining patient remained stable throughout the follow-up period. Age, gender, and the presence of osteochondromas on the ribs and pelvis were not associated with spinal involvement, osteochondromas in the canal, or neural impingement.

Conclusions: Although nearly half of the patients had spinal osteochondromas, neural impingement was rare (4%). Neither age, gender, nor the presence of rib and pelvic osteochondromas were associated with spinal involvement, osteochondromas in the canal, or neural impingement. This information can be used to guide clinical decision-making regarding the use of MRI scans for patient screening.

Level of Evidence: Prognostic Level II. See Instructions for Authors for a complete description of levels of evidence.

Multiple hereditary exostoses (MHE), an autosomal-dominant musculoskeletal disorder, is characterized by the development of multiple cartilage-capped exostoses originating from the physis that are known as osteochondromas[1,2]. Osteochondromas grow during skeletal maturation and then gradually ossify and cease their growth upon skeletal maturity[1-7]. Associated anomalies may include a shortened ulna with a bowed radius (39% to 60% of cases) and limb-length discrepancy (10% to 50% of cases)[2-7].

Spinal osteochondromas were historically considered an uncommon presentation. In 2009, Roach et al.[2] reviewed data for 44 patients and noted that 34 (68%) had osteochondromas arising from the spinal column, 12 (27%) exhibited osteochondromas within the spinal canal, and 3 (7%) experienced neurological deficits. Following the article by Roach et al., subsequent retrospective studies with smaller sample sizes have demonstrated varying rates of intracanal lesions, leading to questions about the necessity and frequency of advanced spinal imaging to identify patients at risk for osteochondromas that cause neurological manifestations[3-6].

Several studies have sought to identify predictive factors for spinal osteochondromas. Male gender and the co-occurrence

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of rib and pelvic osteochondromas have been identified as predictors of intracanal osteochondromas in some studies but not in others[2,4-6]. We sought to determine the prevalence of osteochondromas in the spine in individuals with MHE, distinguishing between intracanal osteochondromas and osteochondromas causing neural impingement. Additionally, we aimed to determine whether demographic factors and the presence of rib and pelvic osteochondromas served as predictors of spinal involvement and/or neural impingement.

Materials and Methods

With institutional review board approval, participants were prospectively enrolled in this study from 2010 to 2022. Patients were recruited from our clinical practice to undergo magnetic resonance imaging (MRI) of the spine. Additionally, if they had a family history of MHE, affected family members were also offered enrollment. The MRI examinations included a localizing sequence, coupled with sagittal spin-echo T1-weighted and axial fast spin-echo T2-weighted imaging across the entire spinal region. The interpretation of the MRI scans was performed by a pediatric musculoskeletal radiologist. Demographic information was collected from medical records and analyzed alongside the MRI data.

The presence of axial osteochondromas, along with their anatomical location (spine, ribs, or pelvis), and the clinical outcomes were documented. Patients were stratified into 1 of 4 categories based on the location of their most infiltrative spinal osteochondroma: no spinal involvement, on the spinal column, in the spinal canal, or producing neural impingement. Spinal column osteochondromas referred to those that were exclusively on the vertebrae and projected away from neural structures, without intracanal extension. Intracanal osteochondromas were defined as the presence of a cartilage cap or osseous stalk intruding into the spinal canal (but not impinging on the neural elements). Neural impingement indicated an osteochondroma-produced deformation of a neural element (i.e., the spinal cord [associated with changes in cerebrospinal fluid] or nerve root). We determined the sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV) of the presence of rib and pelvic osteochondromas associated with osteochondromas arising from the spinal column, arising within the canal, or producing neural impingement.

Statistical analyses were performed with use of IBM SPSS (version 27.0) and the ROC package. The Mann-Whitney U test (for continuous variables) and the chi-square or Fisher exact test (for categorical variables) were utilized to identify differences between groups. Group comparisons included gender (male vs. female), age (<18 years versus ≥18 years), and osteochondroma anatomical site (no spinal involvement, spinal column, intracanal but not impinging, or neural impingement). Logistic regression was performed to identify combinations of demographic, clinical, and radiographic variables that were predictive of spinal involvement and neural impingement. The level of significance was set at $p < 0.05$ for all tests.

Results

A total of 94 patients (47 female) with MHE were enrolled and underwent MRI of the spine (Fig. 1). The mean age at MRI was 23 years (range, 4 to 66 years). Fifty-five patients (59%) were <18 years old, with a mean age of 13 years (range, 4 to 17 years), whereas 39 (41%) were >18 years old, with a mean age of 37 years (range, 18 to 66 years). The racial distribution of the patients was as follows: 78% White, 14% Hispanic, 5% Black, and 3% Asian. MRI findings revealed that 50 patients (53%) had no osteochondromas on the spine, 22 (23%) had osteochondromas on the spinal column, 18 (19%) had

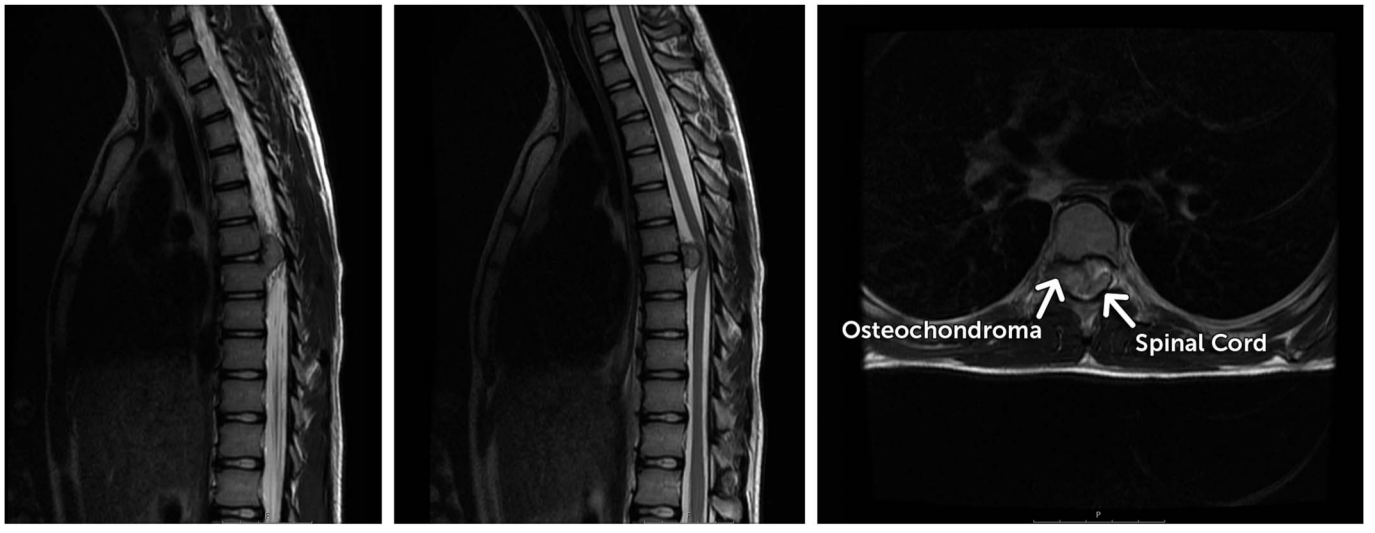

Fig. 1

T1-weighted MRI scans of a 12-year-old female patient with a history of MHE demonstrate an osteochondroma arising from the superior aspect of the right pedicle of T7 and extending into the spinal canal, compressing the spinal cord.

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TABLE I Patient Demographics (N = 94)*

Gender *(no. of patients)*	
Female	47
Male	47
Mean age at MRI *(yr)*	23 (4-66)
Age distribution *(no. of patients)*	
<18 years	55
≥18 years	39
Group distribution *(no. of patients)*	
No spinal involvement	50
Spinal column	22
Intracanal	18
Neural impingement	4

*Values are given as the count or as the mean, with the range in parentheses.

intracanal osteochondromas, and 4 (4%) had neural impingement (Table I).

Of the 55 patients who were <18 years of age, 28 (51%) had no spinal involvement, 18 (33%) had osteochondromas on the spinal column, 6 (11%) had osteochondromas in the canal, and 3 (5%) had an osteochondroma impinging on the spinal cord. Of the 39 adult patients, 22 (56%) had no spinal involvement, 4 (10%) had osteochondromas on the spinal column, 12 (31%) had osteochondromas in the canal, and 1 (3%) had had an osteochondroma impinging on the spinal cord that was excised at 18 years of age. Using logistic regression, there was no significant difference in the prevalence of neural impingement when controlling for gender, age, and extraspinal osteochondromas (p = 0.5) (Table II).

There were no significant differences in osteochondroma location between male and female patients (p = 0.5). Of the 47 female patients, 28 (60%) had no spinal involvement, 9 (19%) had osteochondromas on the spinal column, 9 (19%) had osteochondromas in the canal, and 1 (2%) had an osteochondroma impinging on the spinal cord. Of the 47 male patients, 22 (47%) had no spinal involvement, 13 (28%) had osteochondromas on the spinal column, 9 (19%) had osteochondromas in the canal, and 3 (6%) had an osteochondroma impinging on the spinal cord (Table II).

Of the 4 patients with osteochondromas causing neural impingement, 2 presented with paraparesis and underwent immediate surgical excision for neural decompression. Of the 2 remaining patients, 1 was found to have an osteochondroma impinging at C4-C5. After 2 years of clinical observation and serial MRI scans, the patient developed paretic symptoms and was treated with surgical excision of the osteochondroma. The final patient, who was found to have an osteochondroma that was mildly impinging on the spinal cord at T5, underwent serial MRI scans and remained neurologically stable for 5 years, requiring no surgical intervention. Three of these 4 patients were male, and 3 of the 4 were <18 years old (ages, 10.3, 12.3, 15.3, and 28.7 years).

In our cohort, using the presence of both rib and pelvic osteochondromas to determine spinal involvement yielded a sensitivity of 65%, a specificity of 59%, a PPV of 58%, and an NPV of 66% (Table III). Likewise, using the presence of both rib and pelvic osteochondromas to determine intracanal involvement demonstrated a sensitivity of 30%, a specificity of 79%, a PPV of 31%, and an NPV of 79% (Table III). When evaluating the accuracy of predicting neural impingement on the basis of rib and pelvic osteochondromas, the sensitivity was 50%, the specificity was 77%, the PPV was 8%, and the NPV was 97%. On logistic regression analysis, there was no significant difference in the prevalence of a spinal osteochondroma ($\chi^2[3] = 5.23$; p = 0.156) or the prevalence of neural impingement

TABLE II Risk Factors: Gender, Age, Rib and Pelvic Osteochondromas*

Variable	Total (N = 94)	No Spinal Involvement (N = 50)	Spinal Column (N = 22)	Intracanal (N = 18)	Neural Impingement† (N = 4)
Gender					
Female	47 (50%)	28 (56%)	9 (41%)	9 (50%)	1 (25%)
Male	47 (50%)	22 (44%)	13 (59%)	9 (50%)	3 (75%)
Age					
<18 years	55 (59%)	28 (56%)	18 (82%)	6 (33%)	3 (75%)
≥18 years	39 (41%)	22 (44%)	4 (18%)	12 (67%)	1 (25%)
Rib and pelvic lesions					
No	71 (76%)	42 (84%)	14 (64%)	13 (72%)	2 (50%)
Yes	23 (24%)	8 (16%)	8 (36%)	5 (28%)	2 (50%)

*Values are given as the number of patients, with the percentage in parentheses. †Logistic regression of the presence of neural impingement with respect to these 3 risk factors produced a p value of 0.505. The univariate p values for each covariate were as follows: gender, p = 0.3; age, p = 0.5; rib and pelvic lesions, p = 0.2.

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TABLE III Literature Reporting the Prevalence of Spinal Osteochondromas*

Study	Year	Design	No. of Patients with MHE and MRI	Osteochondroma Location *(no. of patients)*		
				Spinal Column, Non-Canal	Intracanal	Neural Impingement
Bess et al.[7]	2005	Retrospective	5	0 (0%)	2 (40%)	3 (60%)
Roach et al.[2]	2009	Prospective	44	18 (41%)	9 (20%)	3 (7%)
Ashraf et al.3	2013	Retrospective	9	0 (0%)	0 (0%)	2 (22%)
Jackson et al.[4]	2019	Retrospective	21	4 (19%)	3 (14%)	1 (5%)
Vu et al.[5]	2020	Retrospective	43	7 (16%)	5 (12%)	3 (7%)
Wininger et al.[6]	2021	Retrospective	39	6 (15%)	2 (5%)	3 (8%)
Current study	–	Prospective	94	22 (23%)	18 (19%)	4 (4%)

*PPV = positive predictive value, NPV = negative predictive value. †The univariate p values for each covariate were as follows: gender, p = 0.2; age, p = 0.6; rib and pelvic lesions, p = 0.042. ‡The univariate p values for each covariate were as follows: gender, p = 0.3; age, p = 0.5; rib and pelvic lesions, p = 0.2.

($\chi^2[3] = 2.34$; p = 0.505) when controlling for gender, age, and the presence of rib and pelvic osteochondromas (Tables II and III).

Discussion

In 2005, Bess et al.[7] retrospectively reported on 12 cases of spinal osteochondromas and compared them with 165 previously documented cases. Five (42%) of the 12 patients had a diagnosis of MHE (the other 7 had isolated osteochondromas). Two patients had intracanal lesions, and 3 had neural impingement requiring excision of the lesion. These findings revealed a higher prevalence of spinal osteochondromas than previously noted in smaller case series and case reports[8-26]. In 2009, Roach et al.[2] attempted to determine the prevalence of spinal osteochondromas in patients with MHE. They performed MRI evaluations of 44 consecutive patients and reported that 68% had osteochondromas with spinal involvement. More specifically, 18 (41%) of the 44 patients in their cohort had osteochondromas involving the spinal column, 9 (20%) had osteochondromas in the spinal canal, and 6 (14%) went on to have surgical excision. Of the latter 6 patients, 3 displayed neurological symptoms associated with spinal cord impingement, whereas the other 3 had normal neurological findings.

Subsequent studies aimed to retrospectively report on the prevalence of spinal osteochondromas and to determine the necessity of routine spinal imaging screening protocols. In 2013, Ashraf et al.[3] reported on 9 patients with MHE who underwent clinically indicated axial imaging. Two (22%) displayed neurological symptoms brought on by cervical stenosis caused by osteochondromas encroaching into the spinal canal. Both patients underwent emergency laminectomies, resulting in the complete resolution of their neurological symptoms. In 2019, Jackson et al.[4] reviewed 21 patients undergoing spinal imaging as part of their clinical assessment. Osteochondromas were detected on the spinal column in 4 (19%) of the screened patients. Three (14%) had lesions that were encroaching into the spinal canal, whereas only 1 (5%) presented with neurological symptoms that required surgical excision of the lesion.

TABLE III (*continued*)

Surgical Excision *(no. of patients)*	Prediction of Spinal Lesion by Both Rib and Pelvic Lesions	Prediction of Intracanal Lesion by Both Rib and Pelvic Lesions	Logistic Regression (Gender, Age, and Rib and Pelvic Lesions)	
			Prediction of Spinal Involvement†	Prediction of Neural Impingement†
2 (40%)	–	–	–	–
6 (14%)	–	–	–	–
2 (22%)	–	–	–	–
1 (5%)	Sensitivity: 100% Specificity: 69.2% Prevalence: 38.1% PPV: 66.7% NPV: 100%	–	–	–
4 (9%)	–	Sensitivity: 63% Specificity: 69% Prevalence: 18.6% PPV: 31.3% NPV: 88.9%	–	–
2 (5%)	Sensitivity: 87.5% Specificity: 5.3% Prevalence: 29.6% PPV: 28% NPV: 50%	–	–	–
3 (3%)	Sensitivity: 65.2% Specificity: 59.2% Prevalence: 46.8% PPV: 58.4% NPV: 65.9%	Sensitivity: 30.4% Specificity: 78.9% Prevalence: 23.4% PPV: 30.6% NPV: 78.8%	0.156†	0.505†

In 2020, Vu et al.[5] conducted a study to determine the prevalence of MHE and to assess the utility of an MRI surveillance program. They reported on a cohort of 43 patients with MHE, all of whom underwent total MRI scans of the spine. Seven (16%) exhibited osteochondromas on the spinal column, 5 (12%) had intracanal osteochondromas, and 3 (7%) underwent surgical excision of the osteochondromas due to neural impingement.

In 2021, Wininger et al.[6] investigated the rate of spinal osteochondromas in patients with MHE. Of the 39 patients included in their study, 6 (15%) had spinal osteochondromas, 2 (5%) had encroaching intracanal osteochondromas, 3 (8%) had osteochondromas that were causing neural impingement, and 2 (5%) underwent surgical excision. These study findings, as well as ours, are summarized in Table III. It is not surprising that the work by Bess et al.[7]—which was primarily a case series of symptomatic patients—had the highest percentage of patients requiring surgical excision. It is also worth noting that, when excluding the 2 smallest studies, neural impingement was relatively uncommon, and the percentage of patients with such impingement was remarkably consistent across the studies (range, 4% to 8%).

Multiple studies have attempted to identify potential risk factors associated with the development of spinal osteochondromas causing neurological deficits[2,4-6]. Roach et al.[2] and Jackson et al.[4] identified male gender as a risk factor for spinal column involvement. Jackson et al. also identified a significant association between osteochondroma involvement in the spine and both rib and pelvic osteochondromas[4]. They concluded that the presence of both rib and pelvic lesions could be employed as a screening tool to predict spinal lesions with a sensitivity of 100%. However, Wininger et al.[6] reported in 2021 that the presence of both rib and pelvic osteochondromas had a sensitivity of 88% for predicting spinal involvement. Vu et al.[5] found that the presence of rib and pelvic osteochondromas was not strongly predictive of intracanal osteochondromas, reporting a PPV of 31% and a sensitivity of 63%. Similar to these later studies, we did not find the presence of rib and pelvic osteochondromas or any demographic data point to be predictive of spinal involvement and/or neural impingement (Table III).

The strengths of our study include its prospective design and enrollment of family members. When compared with multiple smaller retrospective studies, we believe that our study contributes to understanding the prevalence of spinal involvement and the

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potential for neurological manifestations. Our study is limited in that it only included patients from a single institution. Additionally, due to institutional review board restrictions on using sedation purely for research purposes, there were only 6 patients who were <10 years of age (none of whom had an osteochondroma in the canal).

In conclusion, we attempted to clarify the prevalences of spinal osteochondromas, particularly those projecting into the spinal canal and those causing neurological manifestations, in patients with MHE. Our findings align with those of earlier, smaller studies and confirm the rarity of neural impingement. However, we believe that it is imperative that physicians caring for patients with MHE recognize that neural-impinging osteochondromas can exist in any patient and avoid the pitfall of attributing subtle symptoms to more noticeable appendicular osteochondromas without considering and assessing the neural axis. We also concur with the recommendation by Roach et al. that patients with MHE should undergo "advanced imaging, preferably magnetic resonance imaging, as soon as they can cooperate and do not require general anesthesia."[2] ■

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