MISDIAGNOSIS OF DESMOID TUMORS; INSIGHT FROM THE DESMOID TUMOR RESEARCH FOUNDATION (DTRF) NATURAL HISTORY STUDY

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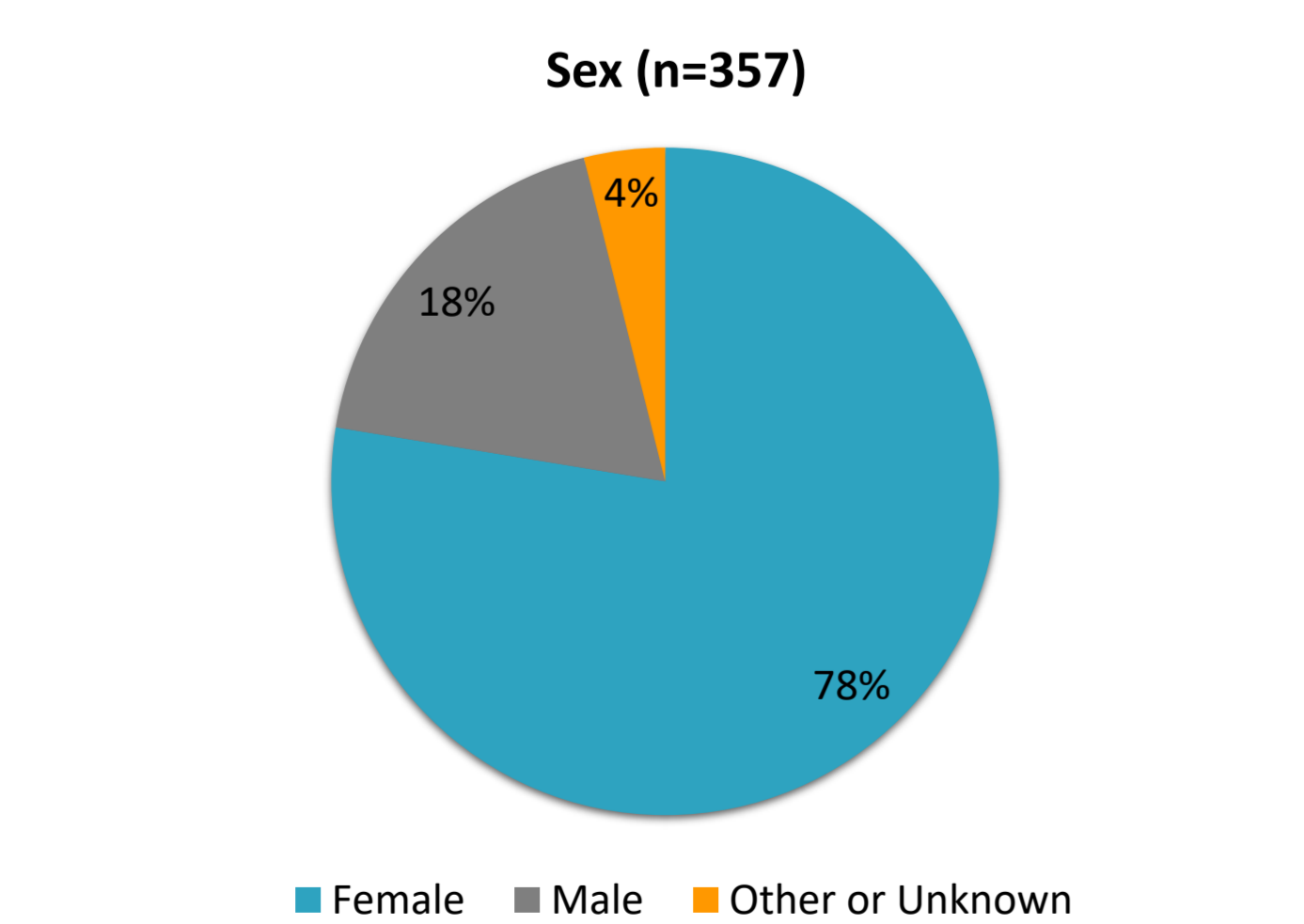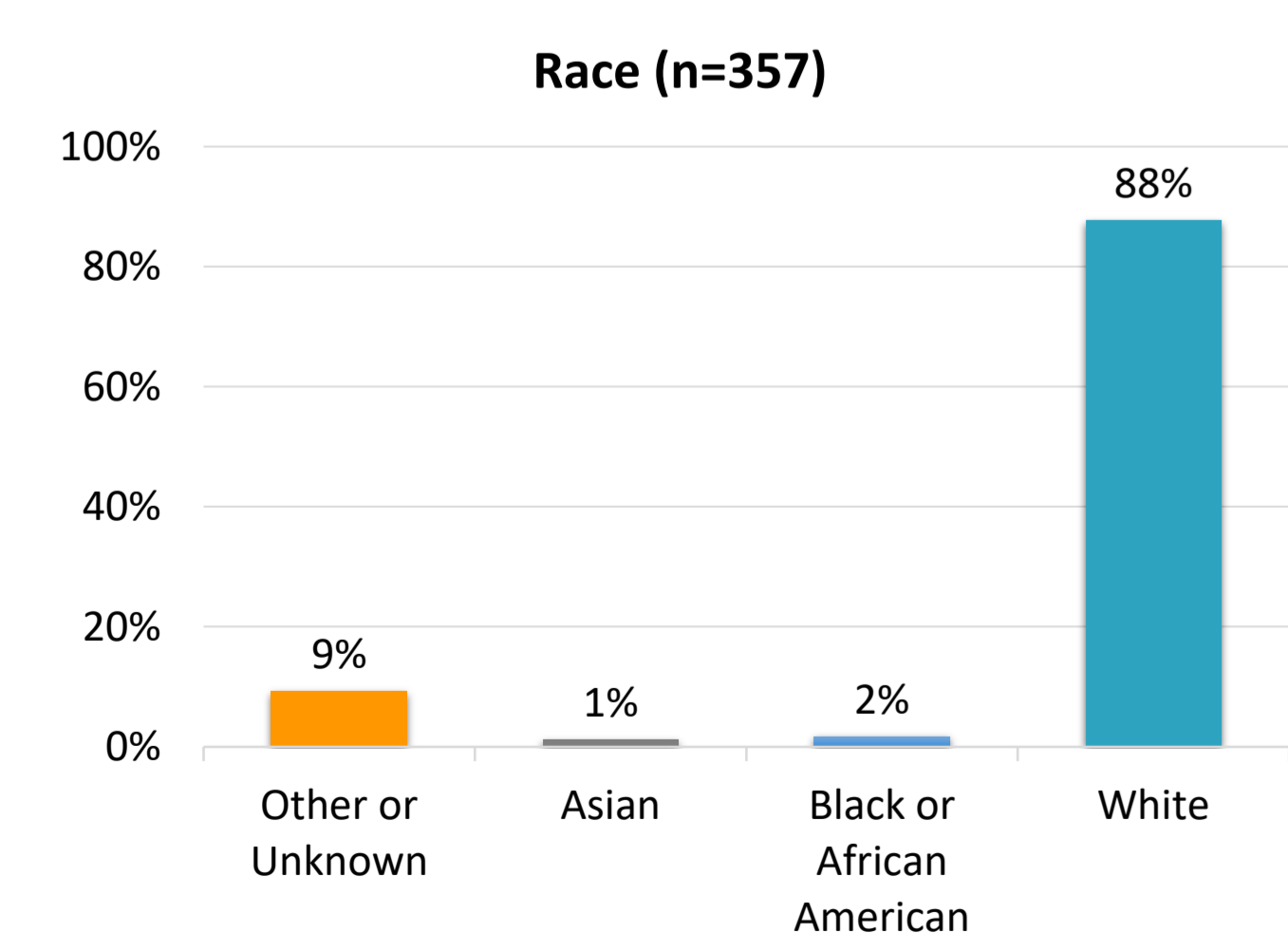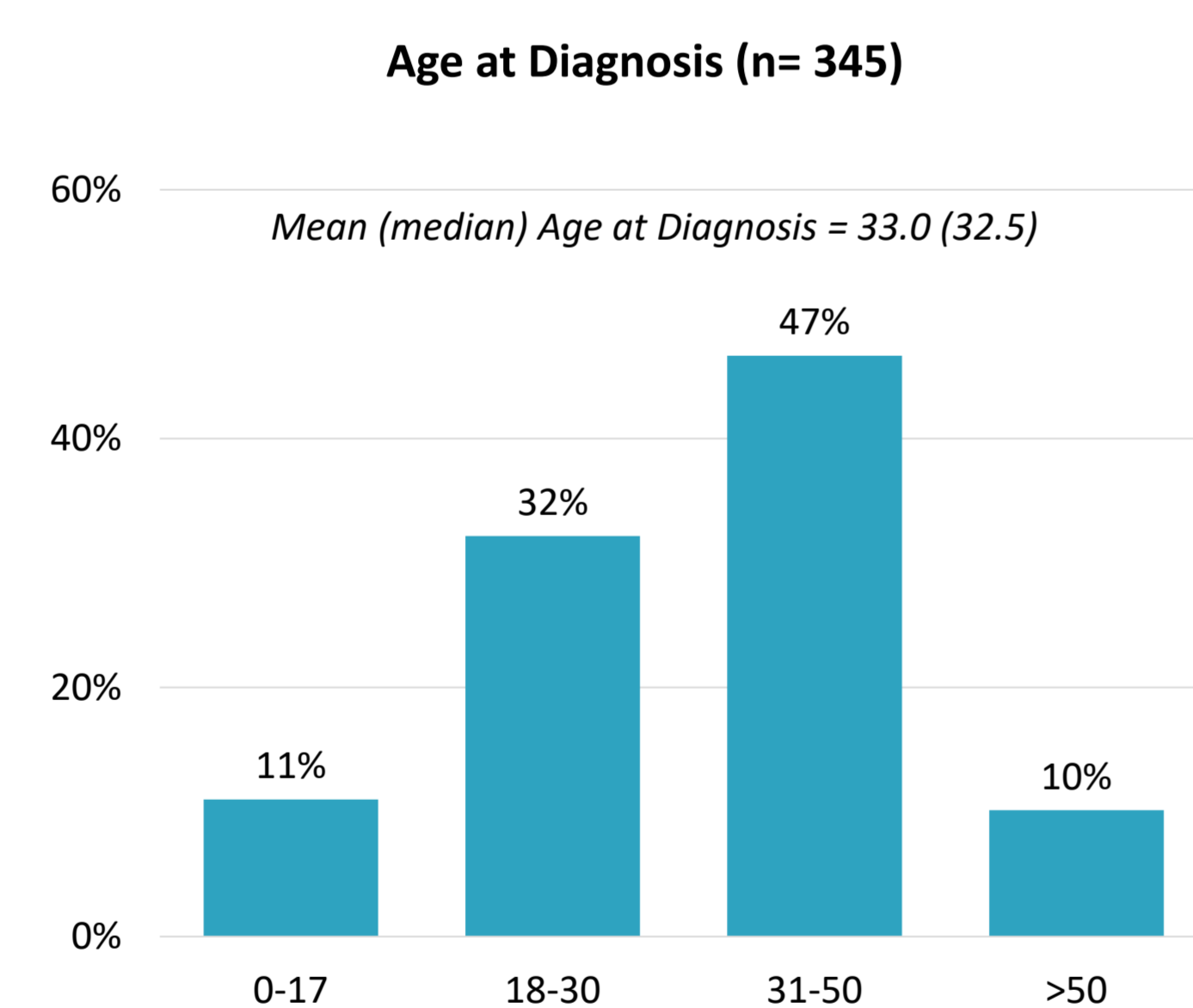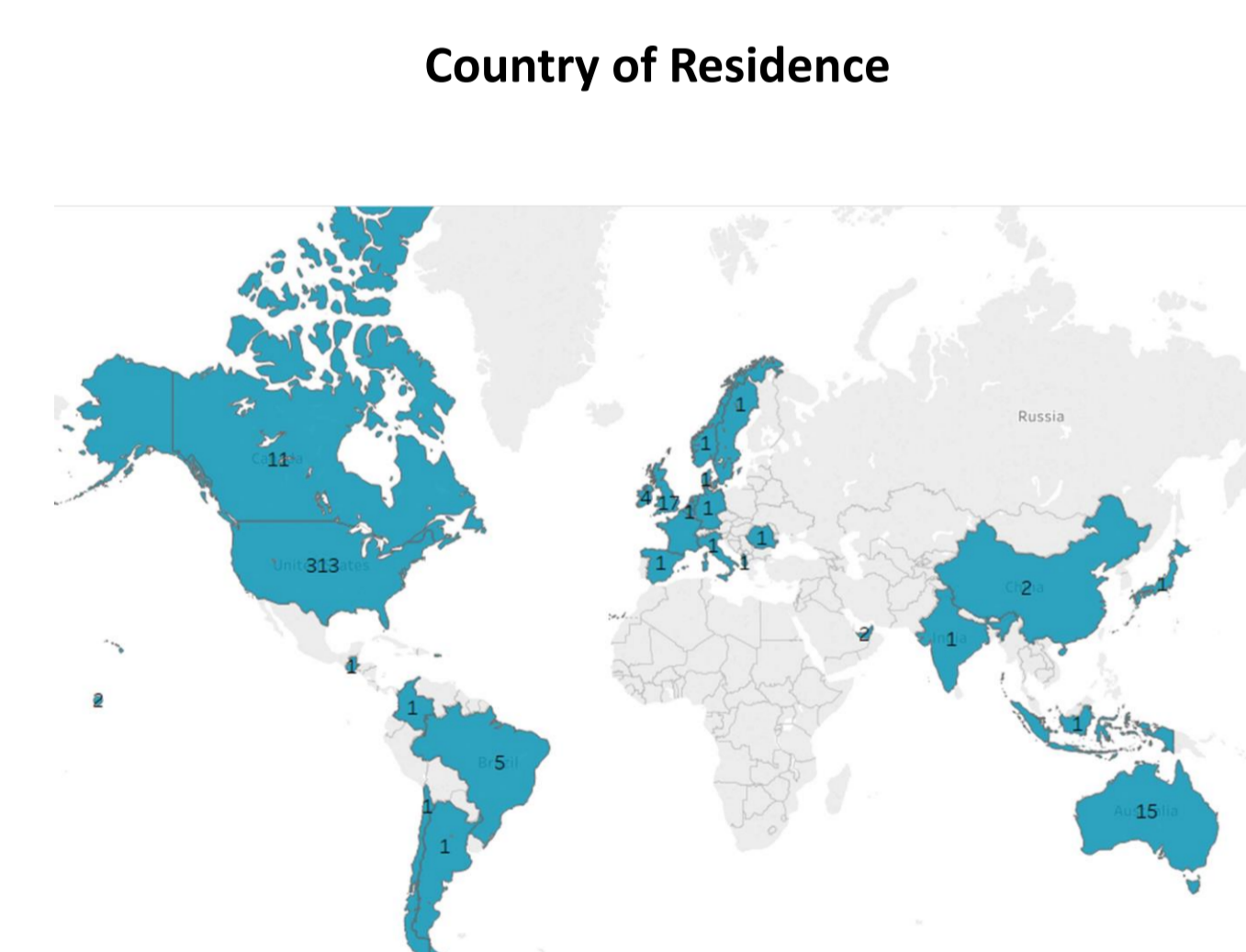
1. BACKGROUND AND AIMS

As few as 5-6 per 1 million people are diagnosed with desmoid tumors annually, which may be an underestimate of the actual affected population due to difficulty in correctly diagnosing the disease. These locally invasive, noncancerous tumors can yield a wide range of symptoms including no symptoms at all. Conclusive diagnosis requires biopsy and a pathologist experienced in or knowledgeable of rare cancers. To improve awareness of desmoid tumors, DTRF in partnership with the National Organization for Rare Disorders launched the DTRF natural history study. Here, we describe patient-reported data on their experience with misdiagnosis.

2. METHODS

The registry initiated February 2017 and contains 15 surveys covering diagnostics, disease, treatment, care management, and quality of life. As of January 2019, 357 patients have completed 2,371 surveys.

Country of Residence

Age at Diagnosis (n= 345)
Mean (median) Age at Diagnosis = 33.0 (32.5)

Race (n=357)

Sex (n=357)

Data regarding the diagnosis of their desmoid tumor was provided by 171 registrants. Comparisons of properly diagnosed vs initially misdiagnosed groups were made by chi-square or Fisher's Exact test (categorical) or Student T test (continuous) in SPSS 25.

3. DEMOGRAPHICS

49% (84/171) of respondents indicated initial misdiagnosis of their desmoid tumor. Differences between the groups were not broadly identified by gender, age, location of residence, race, payer coverage, or known relative with same diagnosis.

| DISTRIBUTION WITHIN GROUPS UNLESS NOTED	CORRECTLY DIAGNOSED n=87	MISDIAGNOSED n=84	TOTAL N=171	p
GENDER				
FEMALE	79%	80%	80%	0.942
MALE	21%	20%	20%	
AGE - MEAN (SD)				
AT SYMPTOM ONSET	31.4 (16.0)	29.5 (14.3)	30.5 (15.1)	0.411
AT DIAGNOSIS	33.6 (14.9)	31.7 (13.6)	32.6 (14.2)	0.391
RESIDES				
OUTSIDE US	18%	10%	14%	0.095
US	82%	90%	86%	
RACE				
BLACK	2%	2%	2%	0.25
WHITE	90%	95%	92%	
OTHER	5%	1%	3%	
UNKNOWN	3%	1%	2%	
PAYER				
MEDICAID	3.4%	7.1%	5.3%	0.542
MEDICARE	11.5%	14.3%	12.9%	
PRIVATE	65.5%	65.5%	65.5%	
OTHER	8.0%	8.3%	8.2%	
UNKNOWN	11.5%	4.8%	8.2%	
RELATIVE WITH DIAGNOSIS	10.3%	10.7%	10.5%	0.790

4. SYMPTOMS LEADING PATIENT TO SEEK CARE

Symptoms were not significantly different between groups. However, a significantly larger fraction of misdiagnosed patients had multiple symptoms compared to the correctly diagnosed group (57% v 43%, p≤0.05).
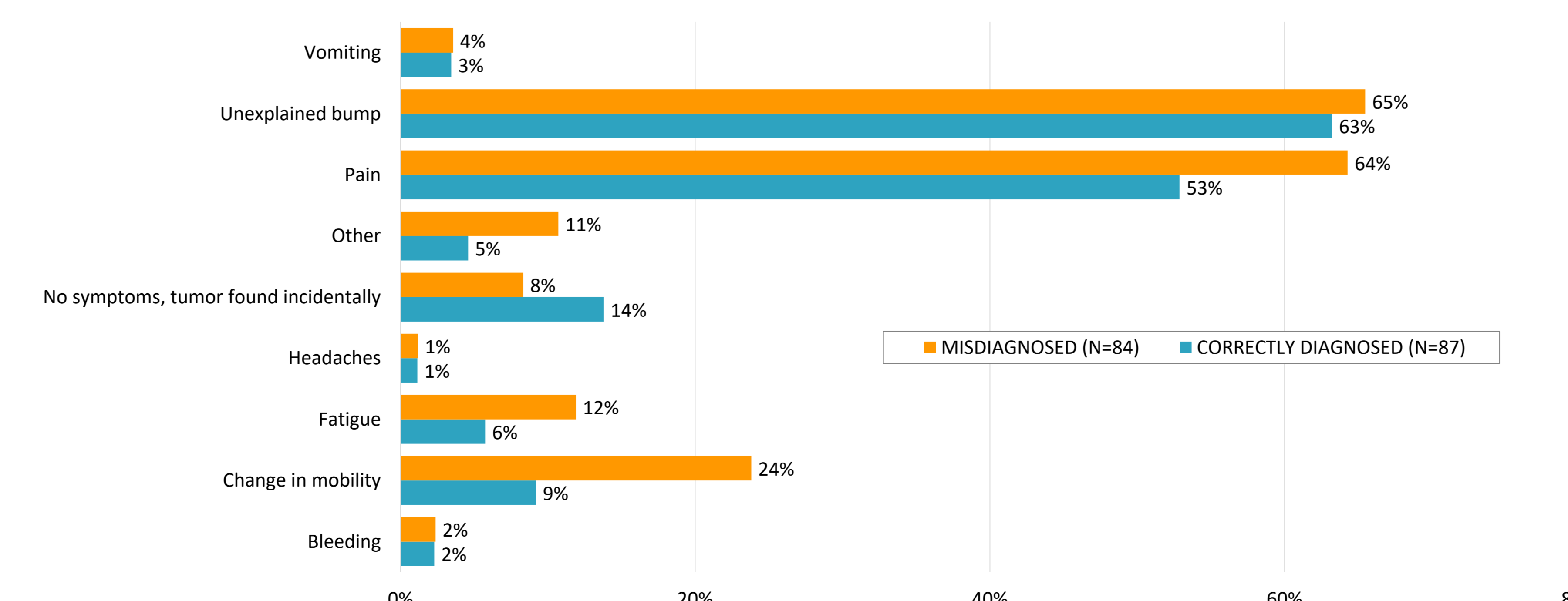
5. MISDIAGNOSIS BY INITIAL TUMOR LOCATION

Misdiagnosis rates varied by initial tumor location and are highest for abdominal wall and joint / extremity. Assessments of proportions using z-test with Bonferroni correction identified only joint / extremity as more prevalent (e.g. reaching significant of p≤0.05) in misdiagnosed patients.
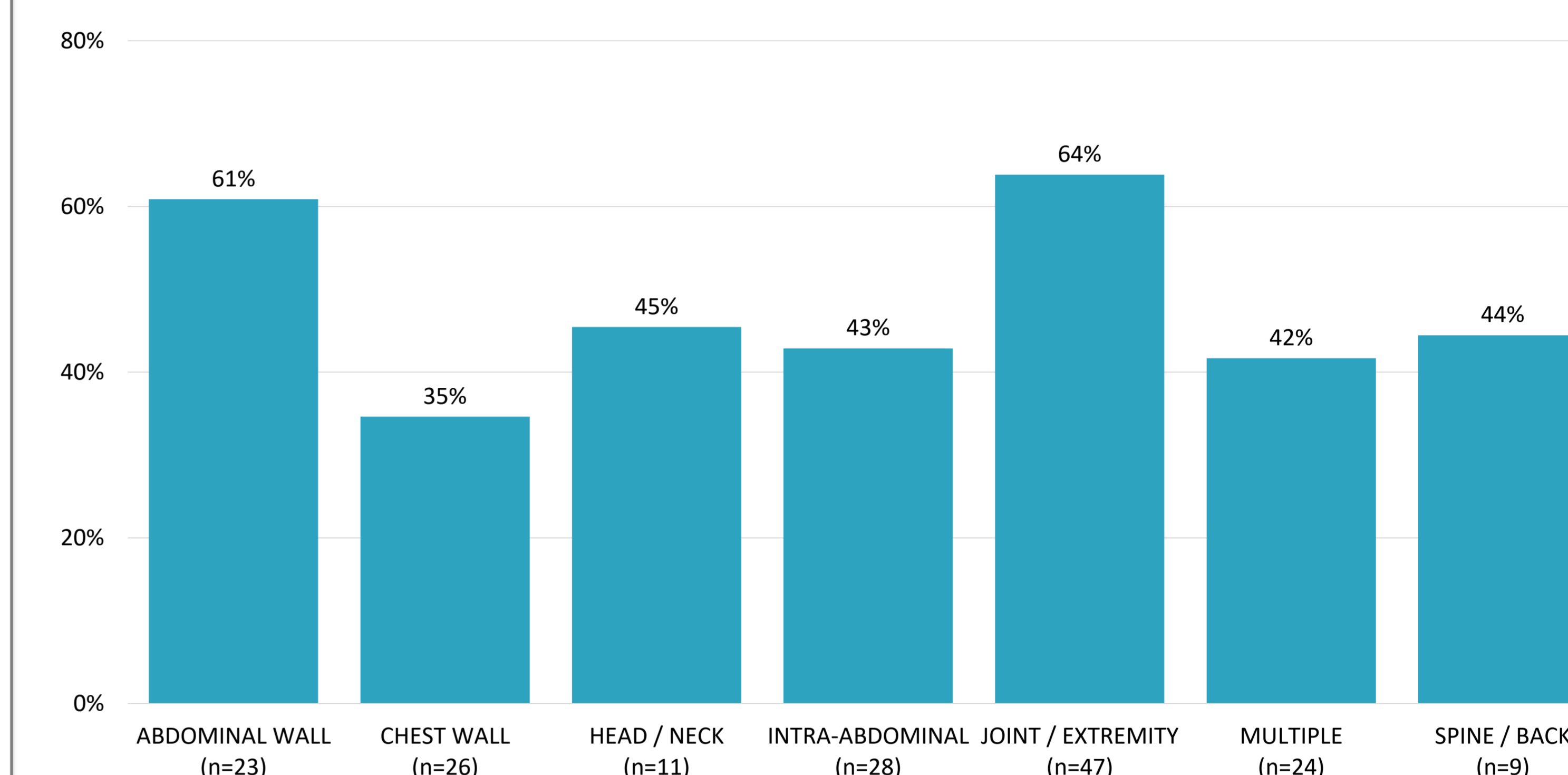
6. SUMMARY

In this study of patient registry data, nearly half (49% (84/171) of respondents indicated initial misdiagnosis. Patient demographics at a top level were not significantly different, though further studies will continue to evaluate the role of age, location, and payer on diagnosis.

Patients in this study originally sought care for wide range of symptoms, with unexplained bump and pain being the most common issues. While individual symptoms were not associated with misdiagnosis, having multiple symptoms was more associated with initial misdiagnosis.

Tumor location varied as expected based upon the nature of the disease. While both abdominal wall and joint / extremity locations were more often diagnosed than other locations, only joint / extremity reached statistical significance.

The challenge of correct initial diagnosis remains. Data collection through the DTRF patient registry continues with increasing awareness of desmoid tumors.