

# An Update on Diagnostic Data from the Desmoid Tumor Research Foundation

## Natural History Study

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THE DESMOID TUMOR  
RESEARCH FOUNDATION



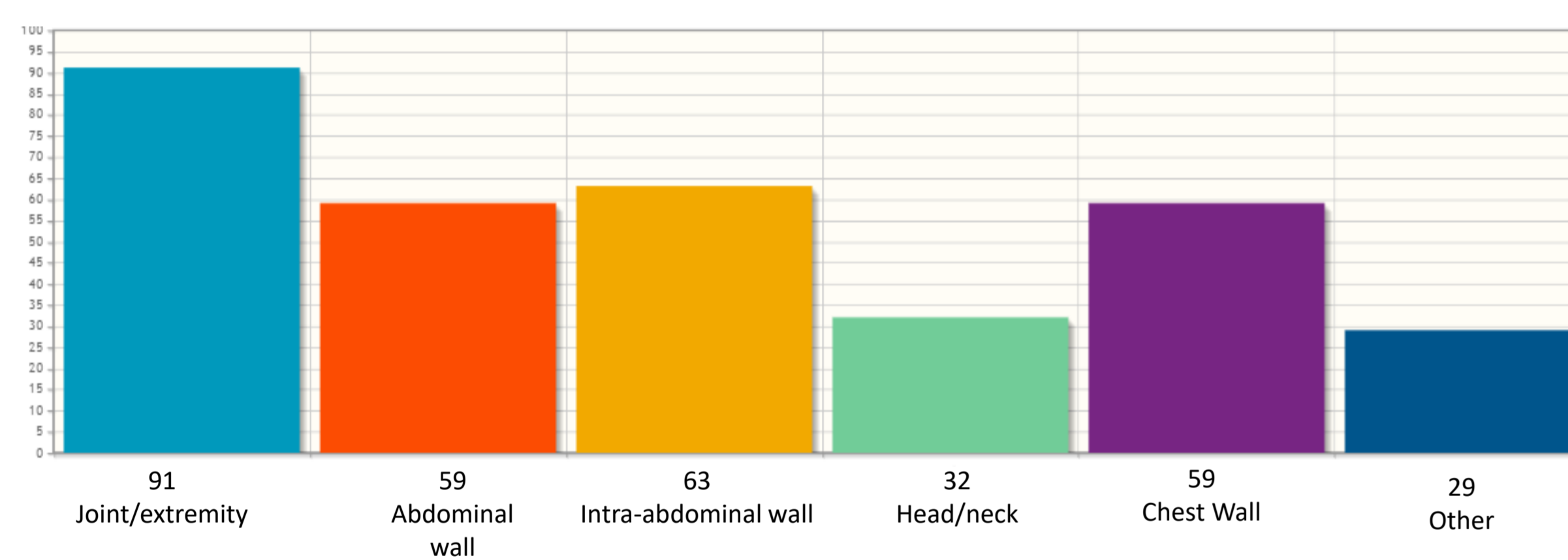
### 1. BACKGROUND AND AIMS

The Desmoid Tumor Research Foundation (DTRF) launched the patient registry and natural history study in 2017. Understanding the diagnostic process is of the upmost important for rare diseases. Here, we describe the longitudinal data, to include patient symptoms and diagnostic data for desmoid tumor participants.

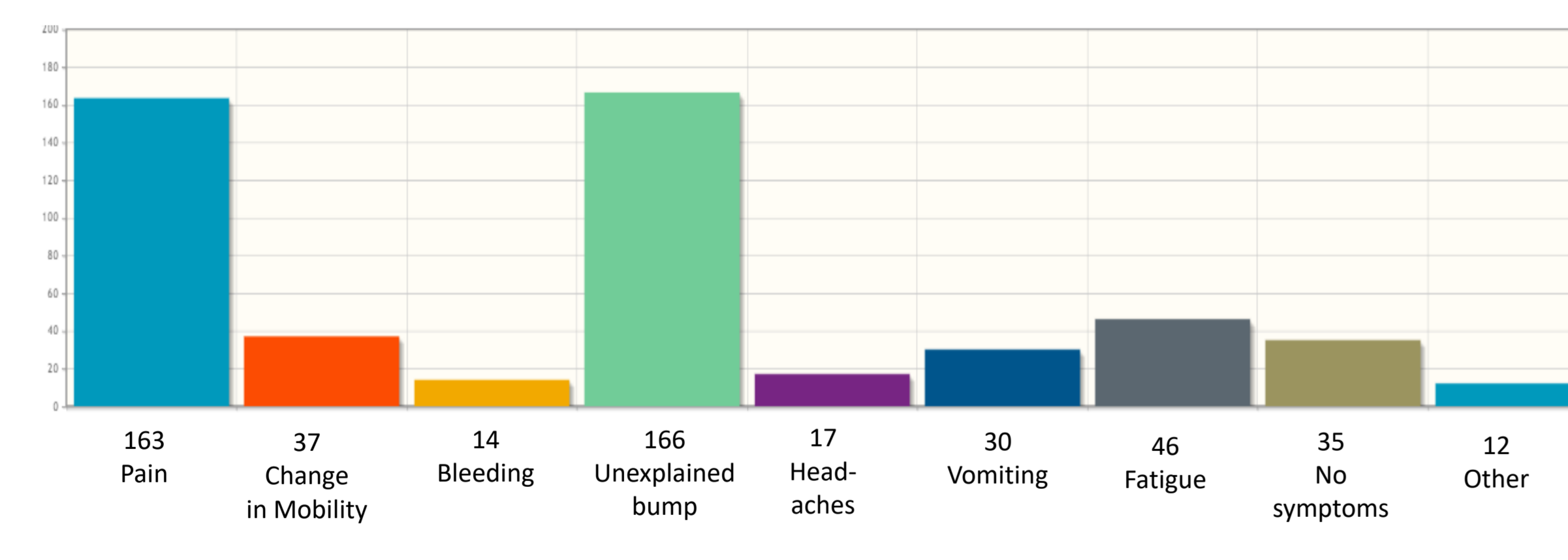
### 2. METHODS

The registry launched September 2017 and contains 15 surveys covering diagnostics, disease, treatment, care management, and quality of life. As of August 20, 1154 patients and 71 legally authorized representatives have enrolled in the study.

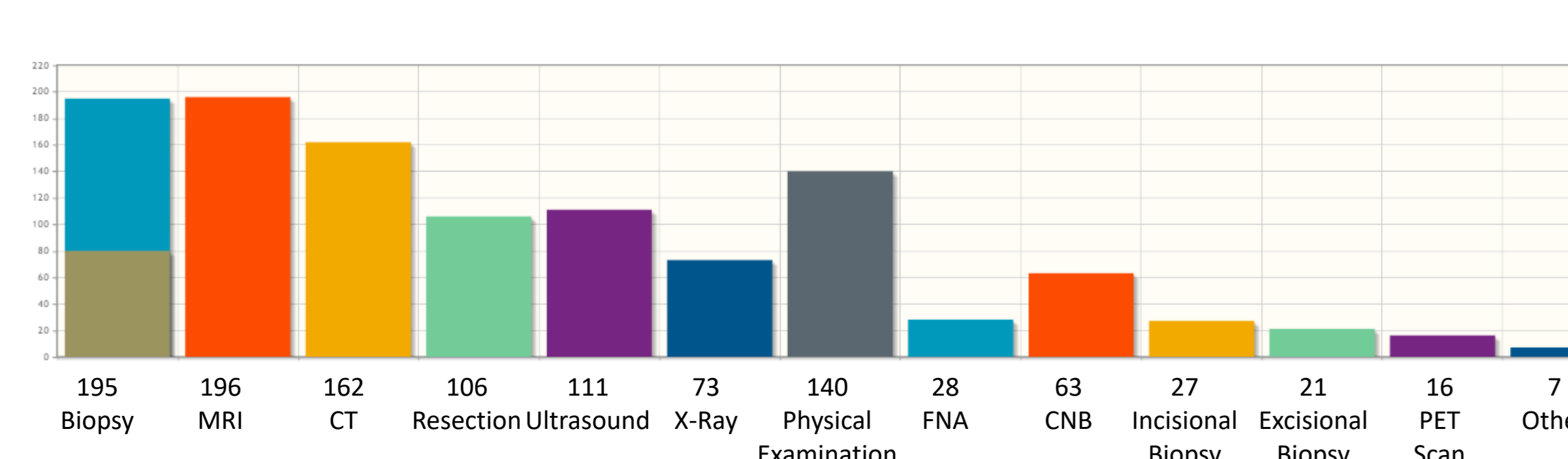
### 3. LOCATION OF DESMOID TUMORS N=333



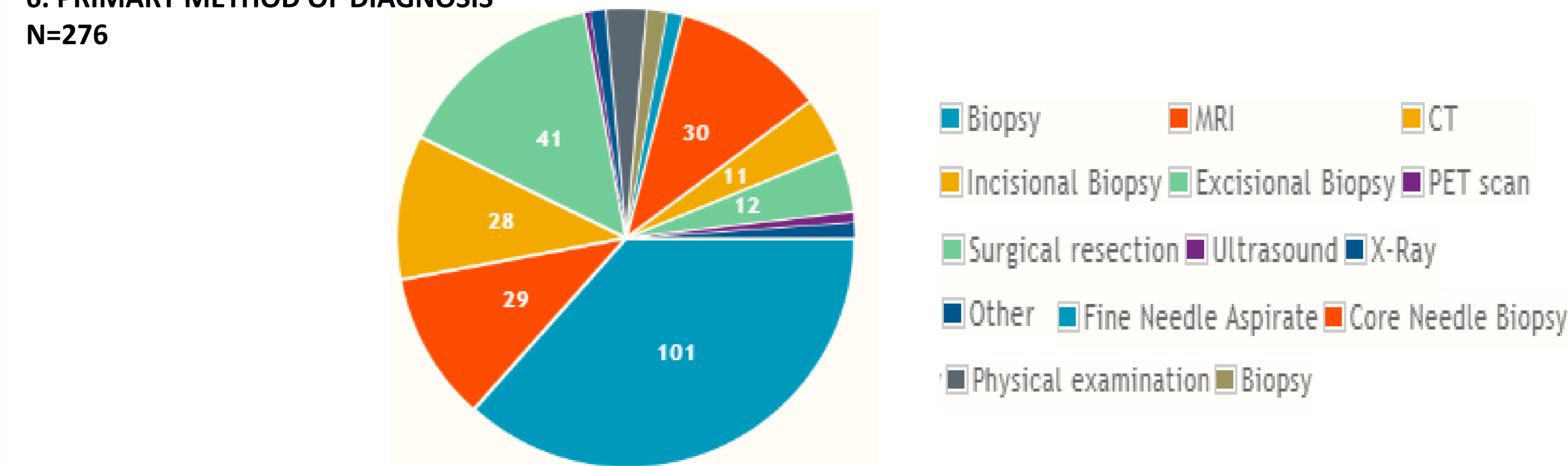
### 4. SYMPTOMS LEADING PARTICIPANT TO SEEK MEDICAL CARE AND A DIAGNOSIS N=520



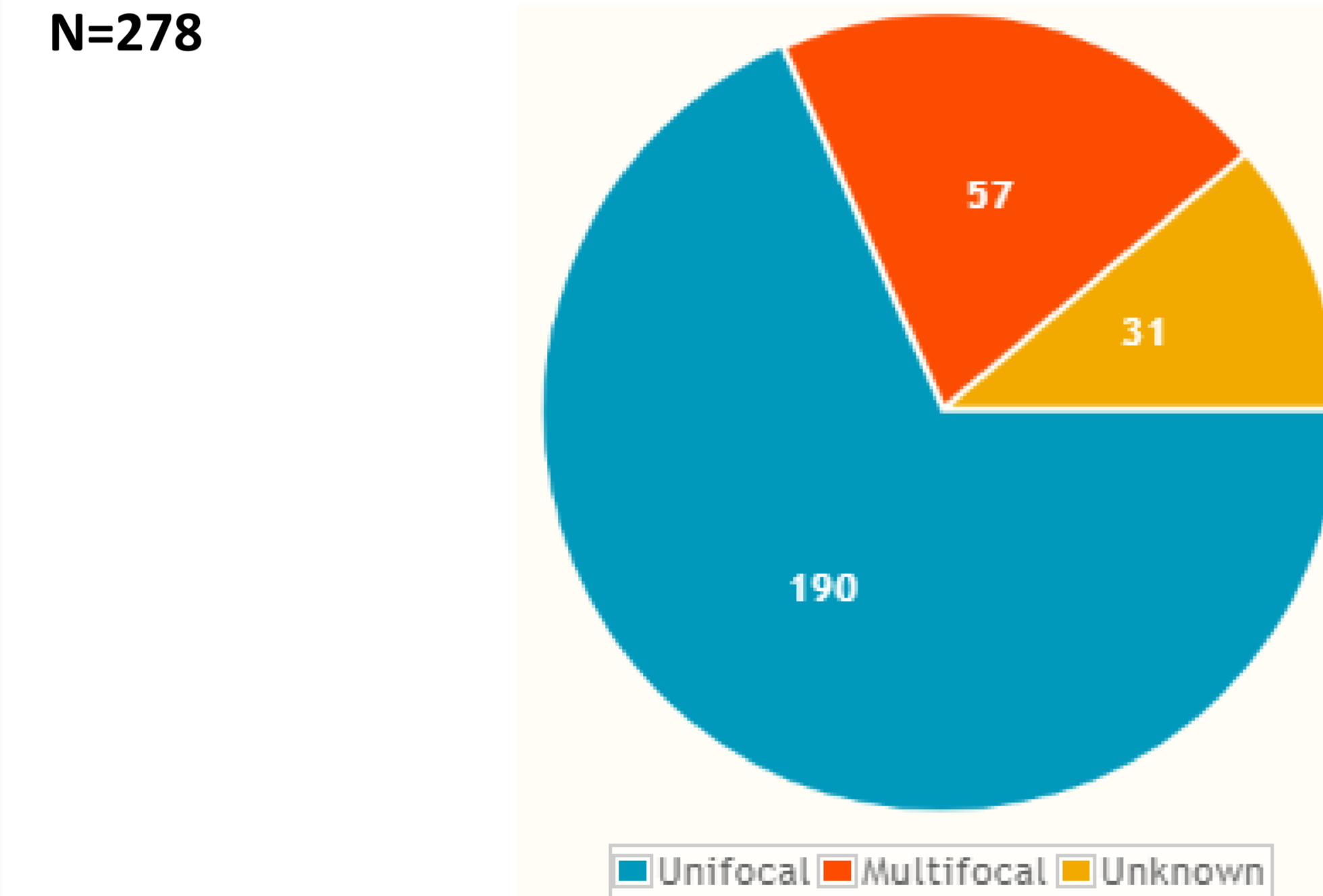
### 5. TYPES AND FREQUENCY OF DIAGNOSIS TECHNIQUES N=1225



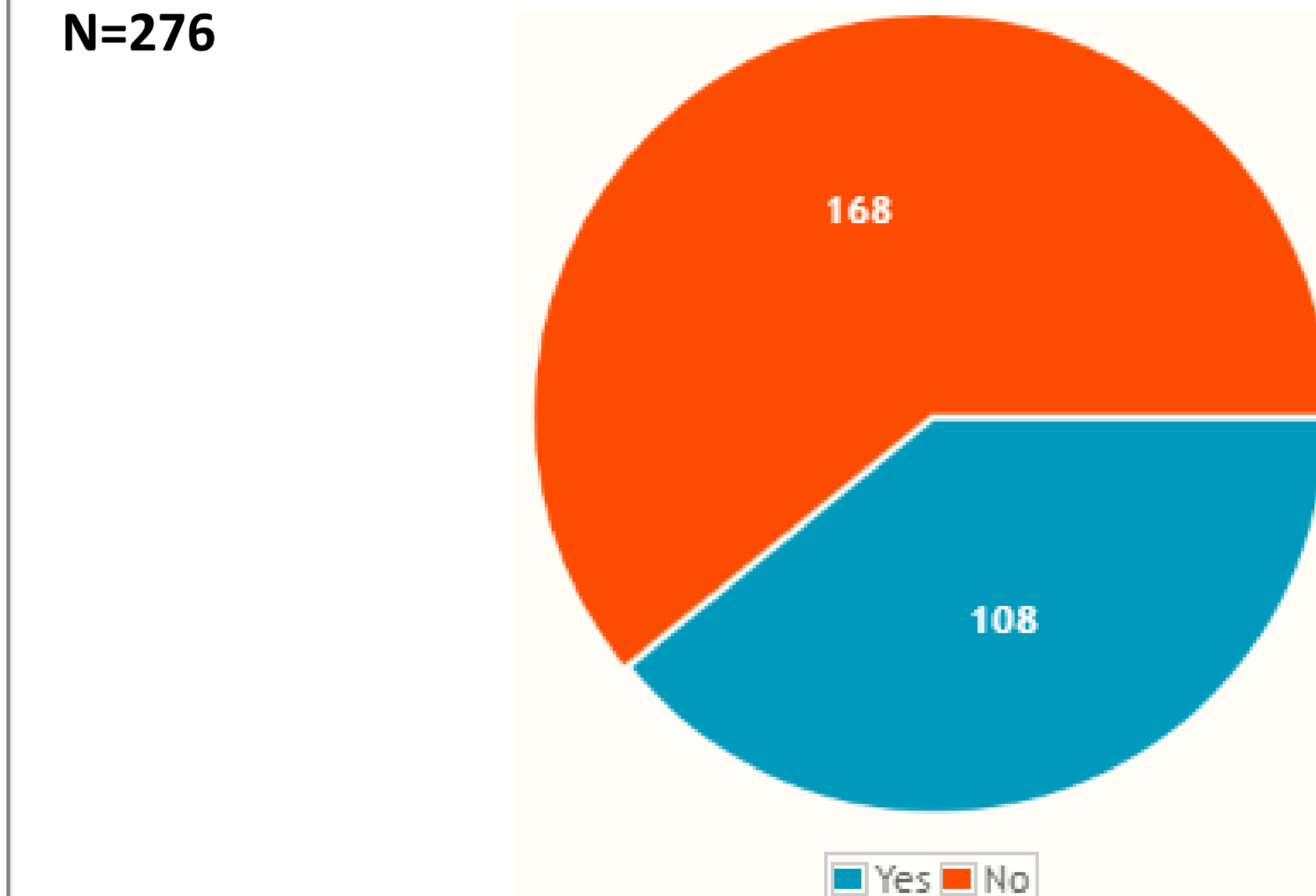
### 6. PRIMARY METHOD OF DIAGNOSIS N=276



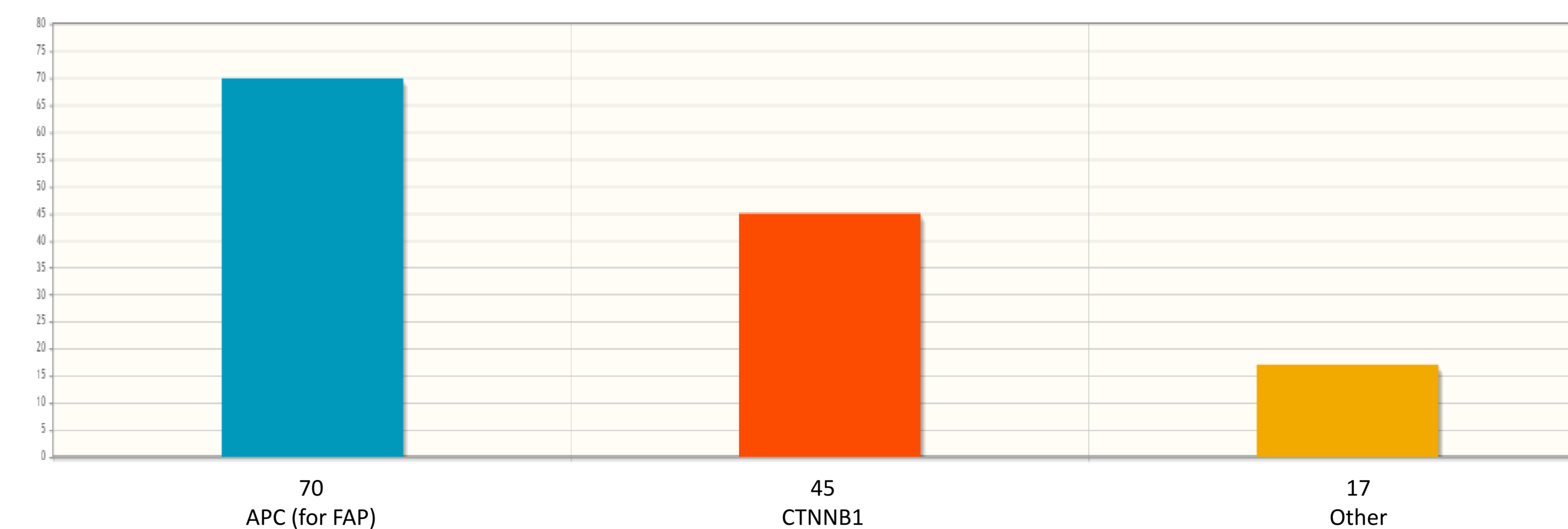
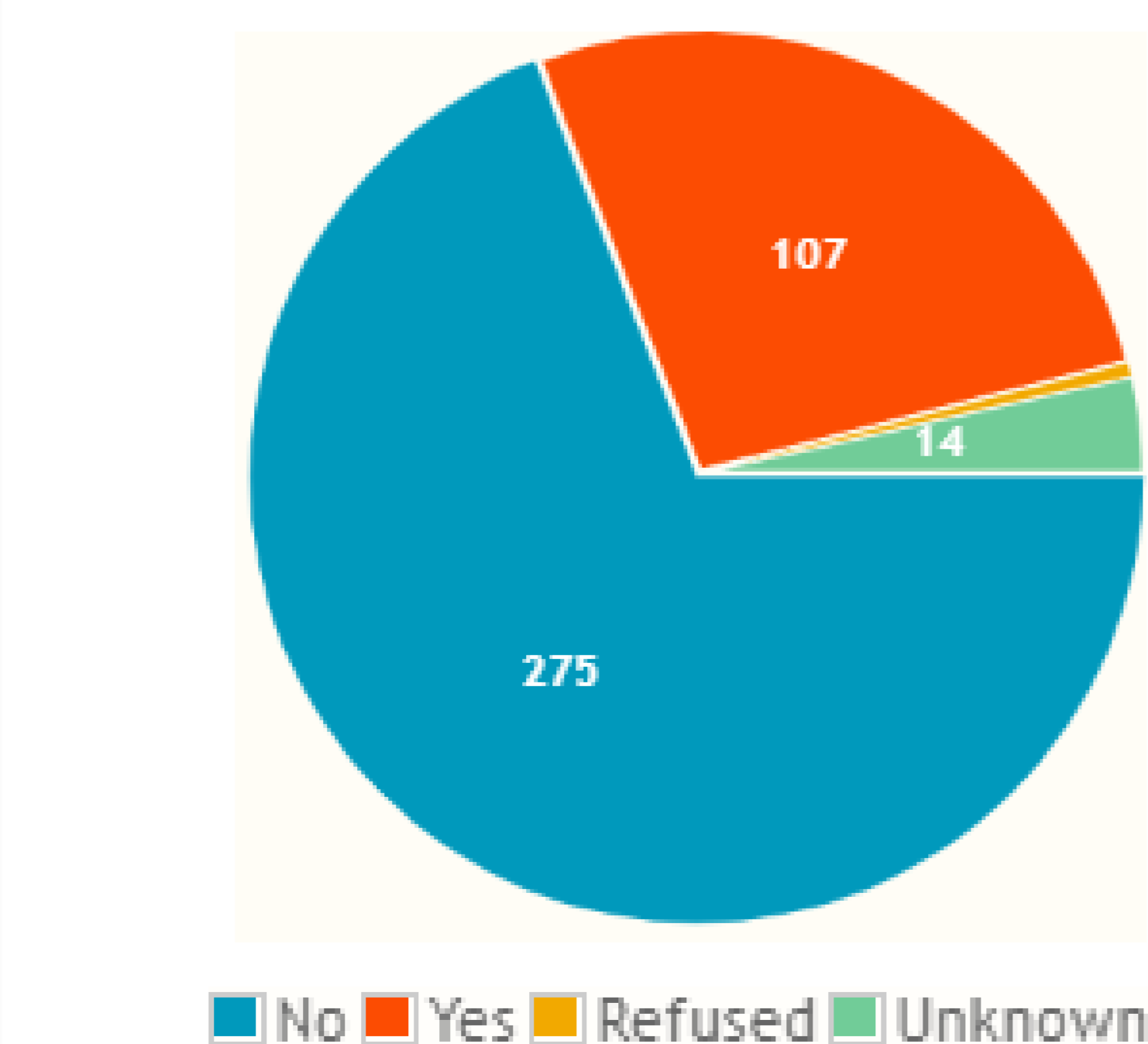
### 7. UNIFOCAL VS MULTIFOCAL TUMOR(S) N=278



### 8. FREQUENCY OF MISDIAGNOSIS N=276



### 9. GENETIC TESTING N=398, 133



### 10. SUMMARY

#### Location of tumors

- The joint/extremity was the most common tumor location reported by the participants (27%), followed by the intra-abdominal wall (19%).

#### Symptoms leading to seek a diagnosis

- 32% of participants reported that an unexplained bump was the symptom that led the participant to seek medical care and diagnosis.
- Pain was the second most common symptom to lead to a medical care and diagnosis (31%).

#### All diagnostic techniques

- Participants reported that different techniques have been used to diagnose the desmoid tumors.
- The most common and frequent techniques reported were MRI (16%), biopsy (16%), CT scan (13%) and physical examination (11%).

#### Primary diagnostic techniques

- 37% of participants reported that biopsy was the primary method of diagnosis of their desmoid tumors.

#### Unifocal vs multifocal tumors

- 68% of participants reported that they have had unifocal tumor.

#### Frequency of Misdiagnosis

- 39% of participants reported that their tumor was misdiagnosed.

#### Genetic testing

- 69% of participants reported that they have not had a genetic testing done on their desmoid tumors.
- Out of the participants who have had genetic testing done, 53% reported that the mutation was located at the APC gene.
- The second most common location reported was at the CTNNB1 gene (35%).

### 12. ACKNOWLEDGEMENTS

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