



The Natural History of Desmoid Tumors

*What we are learning from the DTRF
Patient Registry and Natural History Study*

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THE DESMOID TUMOR
RESEARCH FOUNDATION

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What is the DTRF Registry?

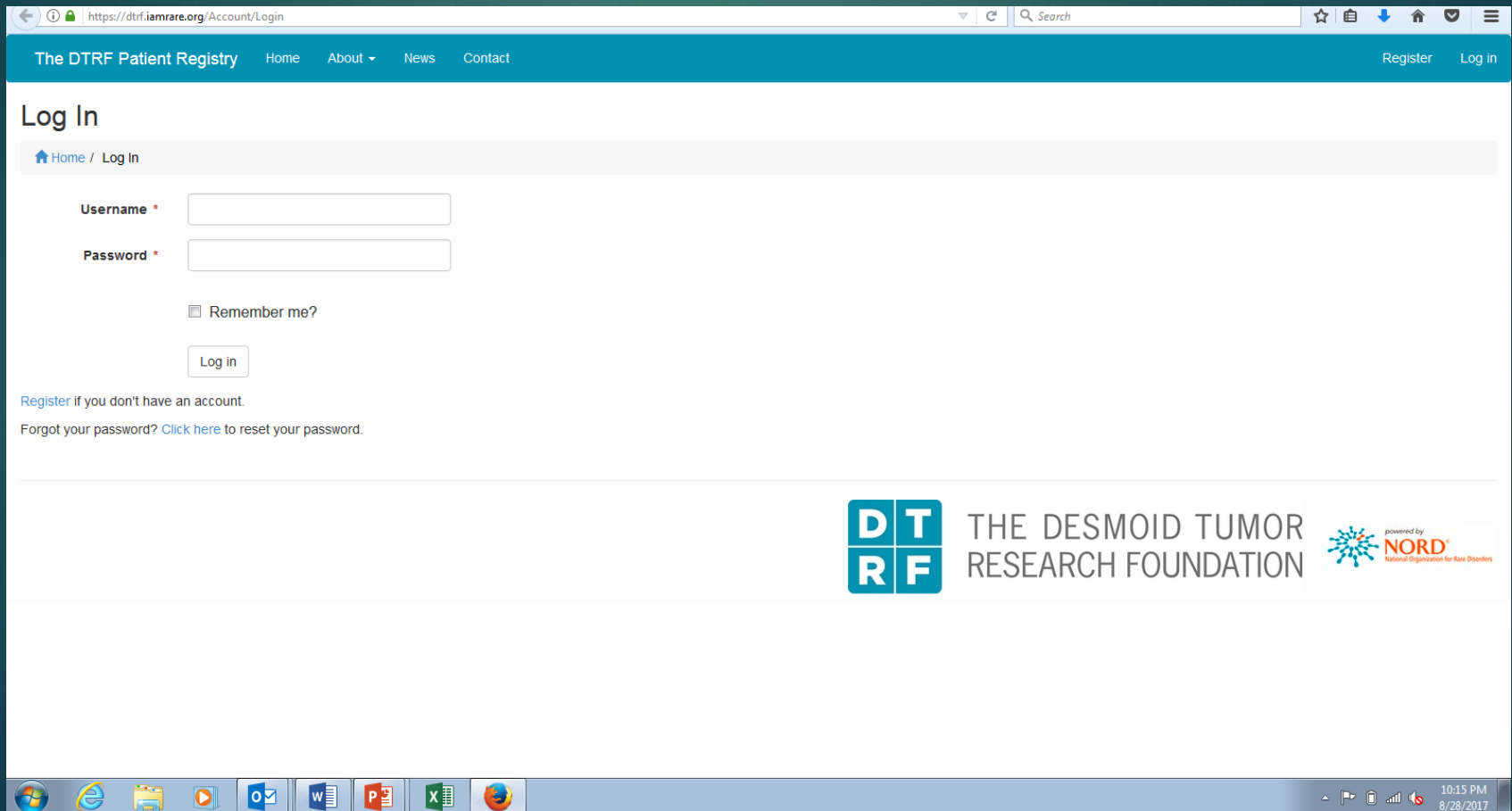
- *An organized system*
- *Observational study to collect data*
- *Evaluate specified outcomes for a population defined by a particular disease, condition, or exposure*
- *Serves a predetermined scientific, clinical, or policy purpose (s).*

What is the DTRF Natural History Study?

- Track the course of a disease over time
- Identify demographic, genetic, environmental and other variables that are associated with the disease
- Define the disease population, including a description of the full range of disease manifestations and subtypes

How will data being collected?

<https://dtrf.iamrare.org>



The screenshot shows a web browser window displaying the login page for the DTRF Patient Registry. The browser's address bar shows the URL <https://dtrf.iamrare.org/Account/Login>. The page has a blue header with the text "The DTRF Patient Registry" and navigation links for "Home", "About", "News", and "Contact". On the right side of the header, there are links for "Register" and "Log in".

The main content area is titled "Log In" and includes a breadcrumb trail "Home / Log In". Below this, there are two input fields: "Username" and "Password", both marked with an asterisk. A checkbox labeled "Remember me?" is positioned below the password field. A "Log in" button is located below the checkbox. Below the login form, there are two links: "Register if you don't have an account." and "Forgot your password? Click here to reset your password."

At the bottom of the page, there is a logo for "THE DESMOID TUMOR RESEARCH FOUNDATION" (DTRF) and a logo for "powered by NORD National Organization for Rare Disorders".












The Windows taskbar is visible at the bottom of the screenshot, showing icons for various applications and the system clock displaying "10:15 PM 8/28/2017".

Data Access



- Critical analysis by DTRF NHS committee, removing all personal health identifiers
- Critical analysis of de-identified patient history by external researchers
 - must have approval by DTRF NHS
- Patient recruitment for other research
 - must have IRB and DTRF NHS approval

How are we collecting this information?

Initial Surveys (13)	Updatable (1)
Surveys Not Taken	
Survey Title	
Medical and diagnostic data	 Take
Treatment and Review of Systems	 Take
Quality of Life Adult Part 1	
Quality of Life A	
	 Take
	 Take
	 Take
	 Take
	 Take
	 Take
	 Take
	 Take
	 Take

New survey added 26 April 2018
Other Medical Conditions

Rare Diseases

Desmoid Tumors

Incentive to participate



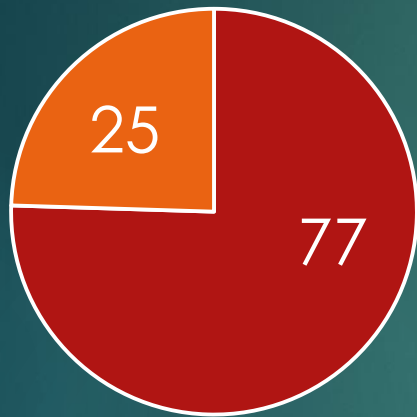
- ▶ This is a time consuming task.
- ▶ The DTRF values your time and information.
- ▶ The first 200 people to complete all of the study questionnaires will receive a \$25 Amazon card as a small token of appreciation.
- ▶ The BIG thank you will come from the clinicians and researchers who will leverage your information to better understanding of desmoid tumor natural history.

Current Numbers

- ▶ 392 registered users
- ▶ 249 completed participant profiles (first survey)
- ▶ 102 completed all of the surveys

Registry Demographics

Gender



Female Male

Age Ranges

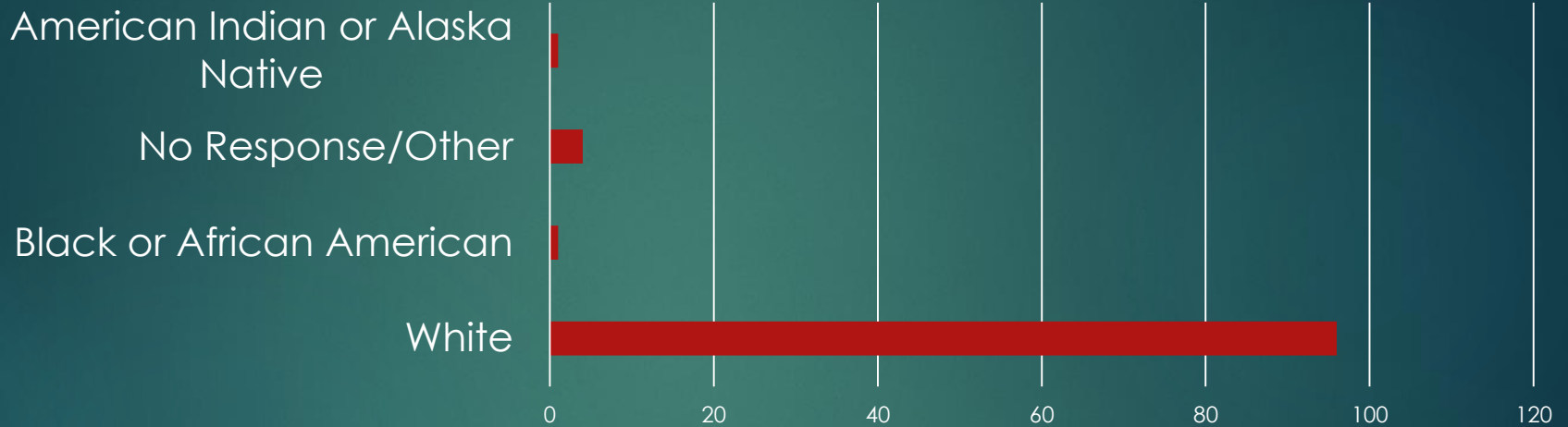


As of 4/8/18

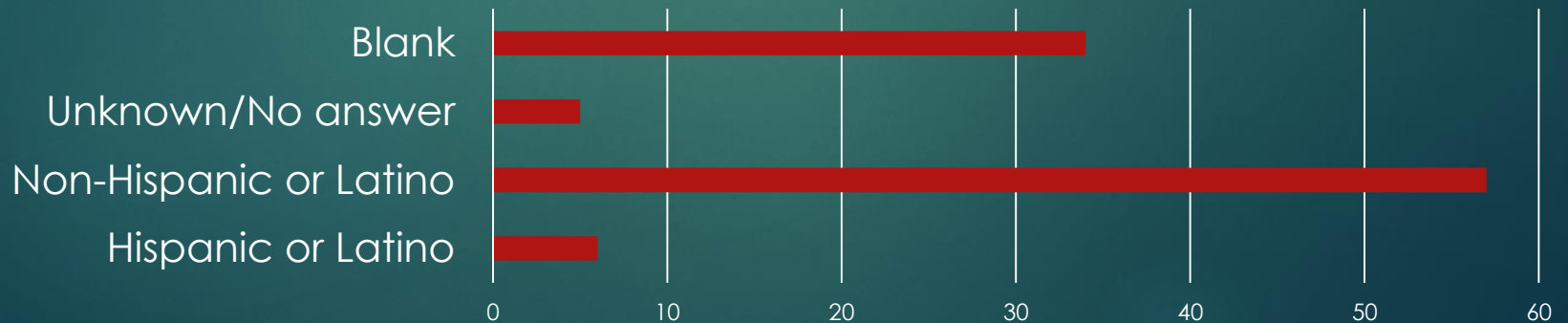
Registry Race/Ethnicity



Race



Ethnicity



As of 4/8/18

Age is important

Years since First Symptoms

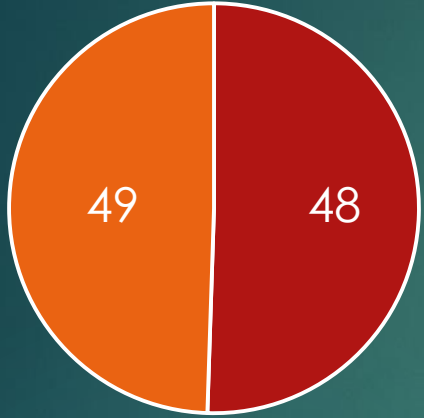


Years since Diagnosis



As of 4/8/18

Misdiagnosis?



■ Yes ■ No

Misdiagnosis by Age Range



■ Yes ■ No

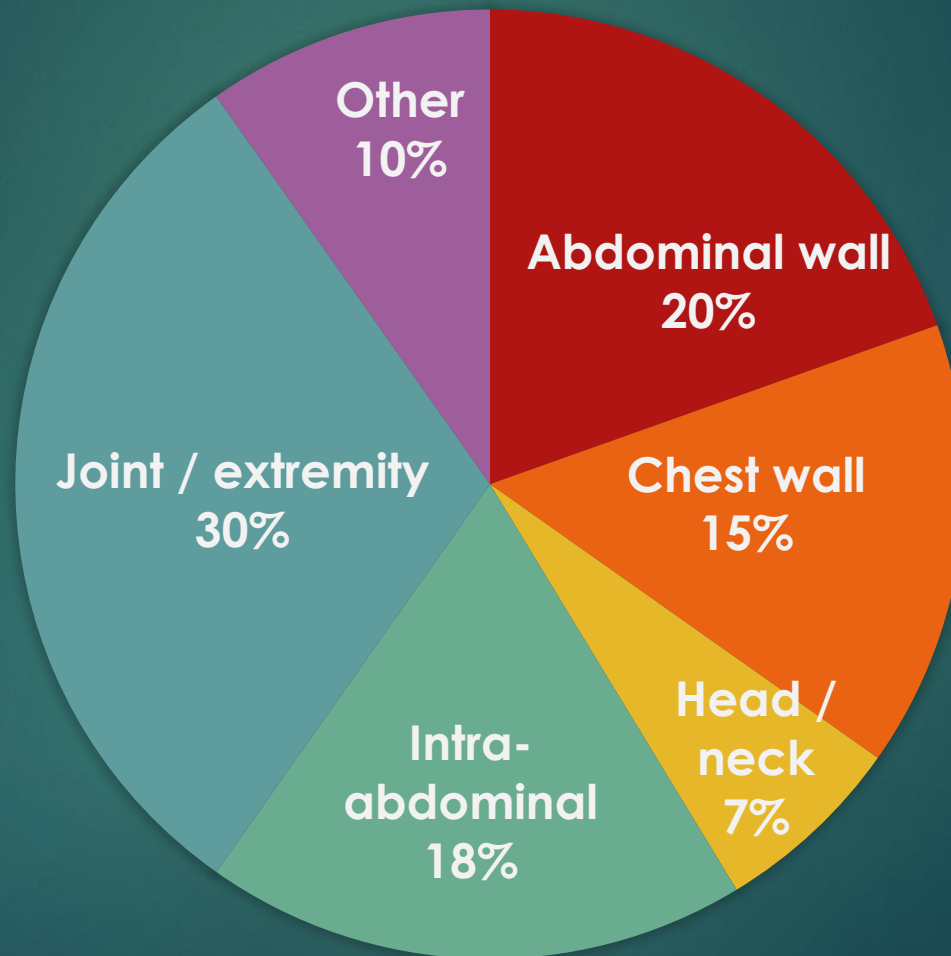
Females Misdiagnosis



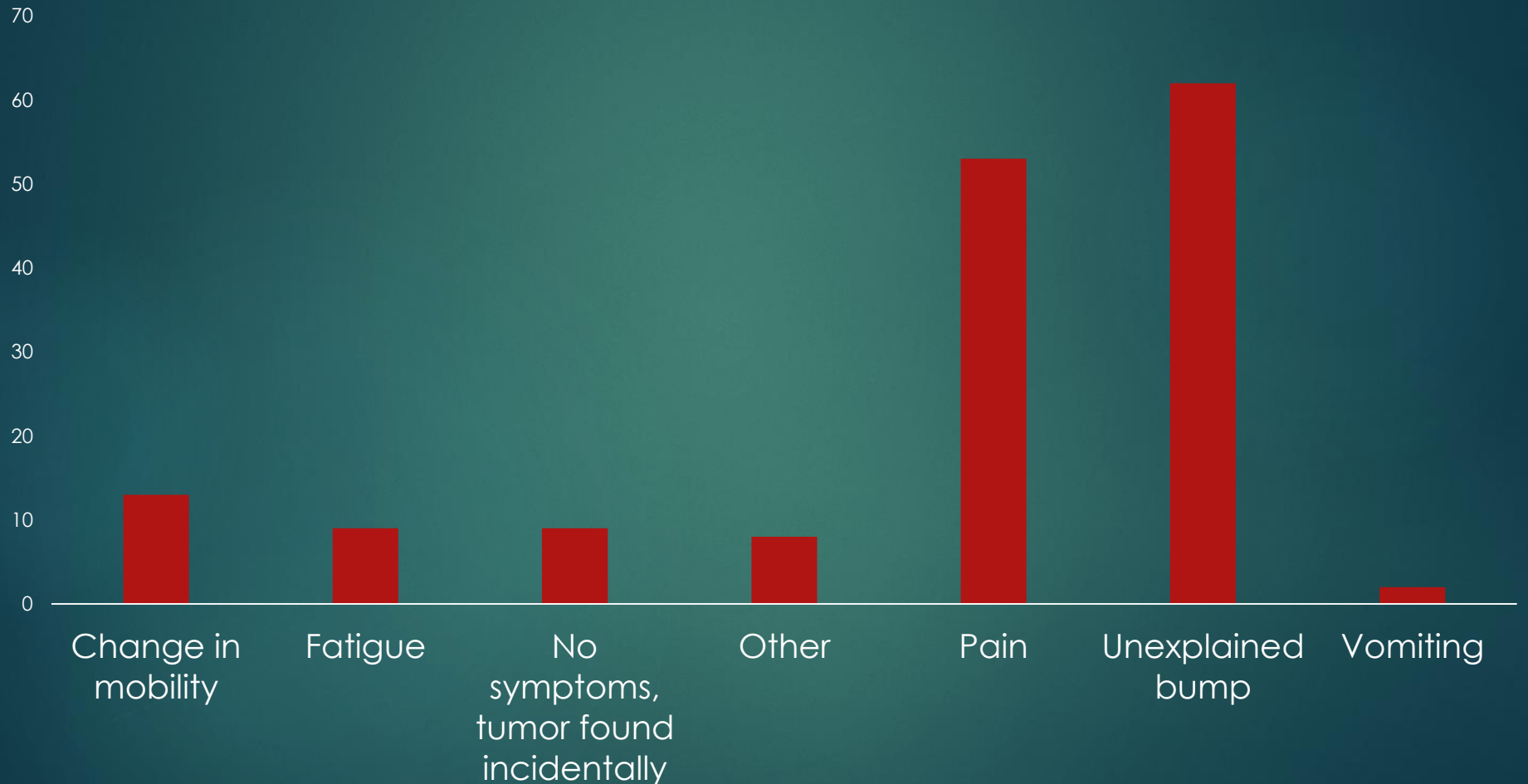
Males Misdiagnosis



Anatomical location

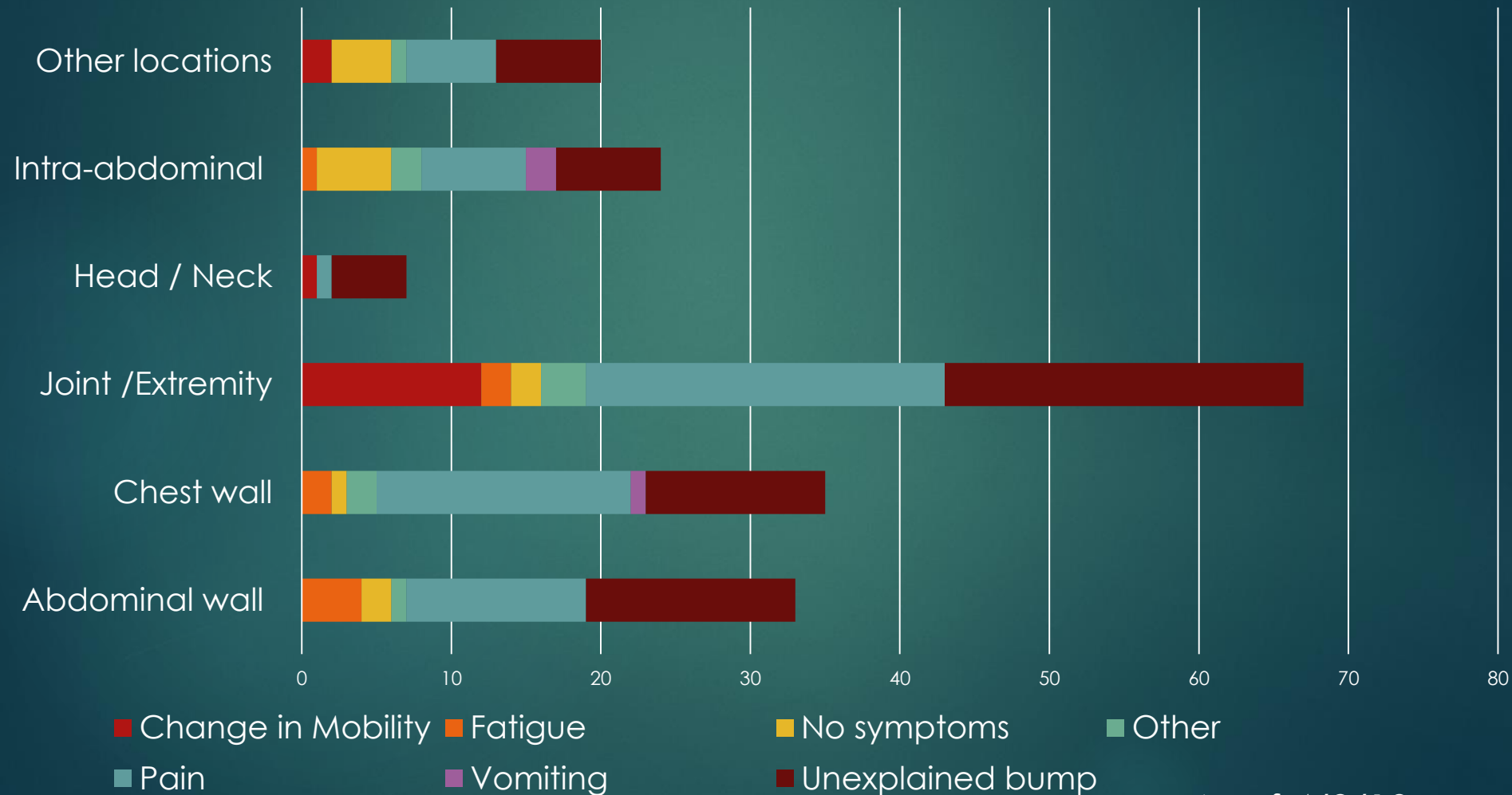


Symptoms leading to diagnosis



As of 4/8/18

Symptoms at diagnosis by anatomical location

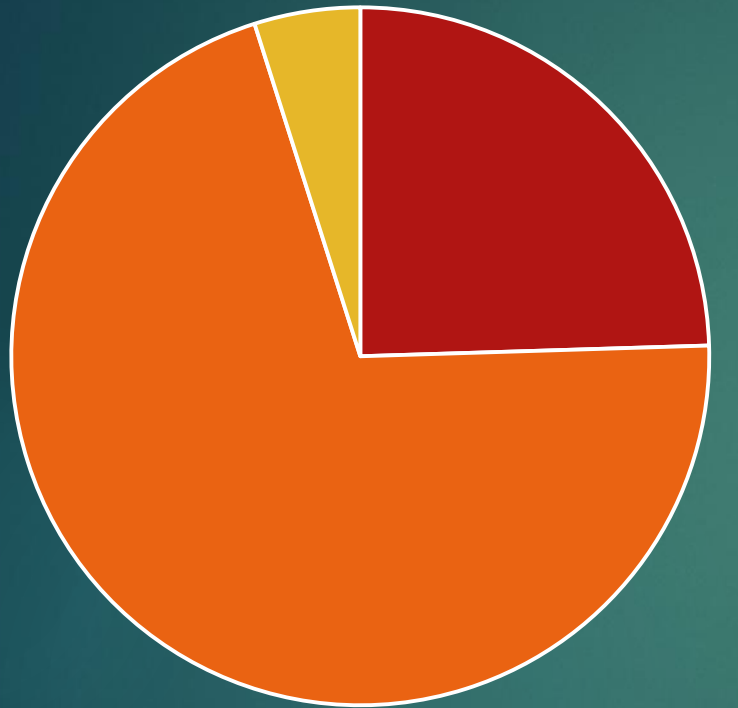


As of 4/8/18

Unifocal vs Multifocal vs Multinodular

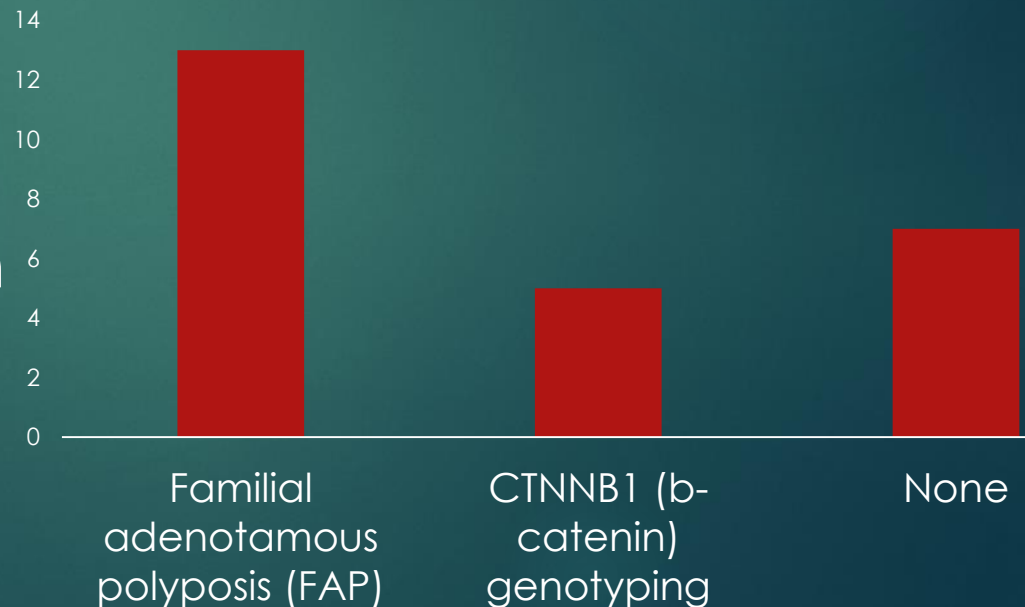
	Unifocal	Multifocal	Multinodular	Unknown
Abdominal wall (e.g. quite superficial on your belly muscle)	4	13	3	0
Chest wall	18	0	0	0
Intra-abdominal (e.g. deep in your stomach/abdomen/pelvis, wrapping up your bowels, kidney, pelvis)	20	0	0	0
Joint / extremity (e.g. hip, knees, shoulders, arms, hands, feet, legs)	30	0	0	3
Other (areas that do not fit with any of the above)	2	0	0	11

Was the tumor tested for molecular markers?



■ Yes ■ No ■ Unknown

Which mutations were present?



Conclusion

- ▶ We are starting to piece together:
 - ▶ Who is participating in the study
 - ▶ What the diagnostic process looks like
 - ▶ What symptoms are leading to a diagnosis
 - ▶ Correct vs incorrect diagnoses
- ▶ What is next?
 - ▶ Continued enrollment
 - ▶ Continued participant engagement for questionnaire completion
 - ▶ Continued evaluation of preliminary results
 - ▶ Engagement with clinicians and researchers

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