WHOLE-BODY MRI IN FSHD

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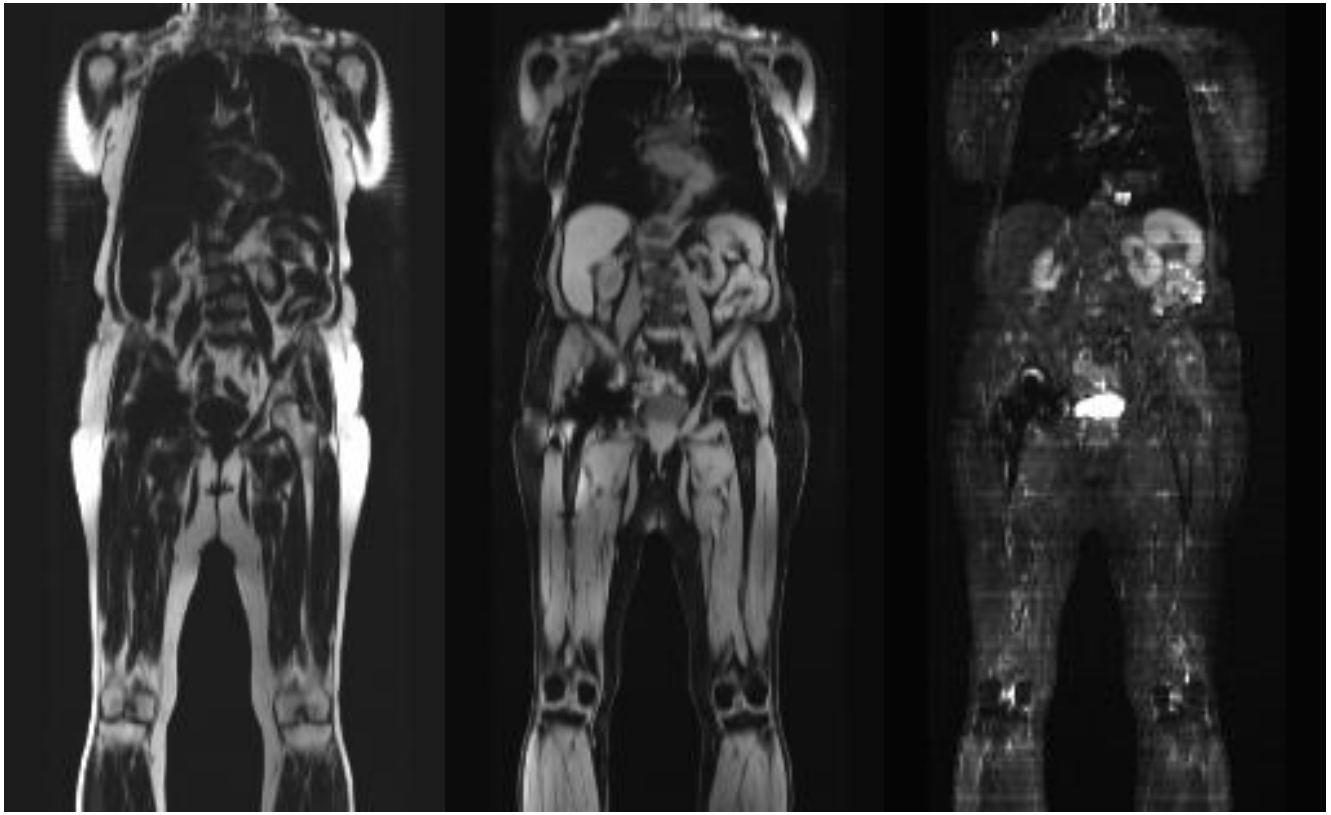
What is an MRI?

- Magnetic resonance imaging
- Uses high-power magnets to measure the activity of protons in body tissues

https://usa.healthcare.siemens.com/

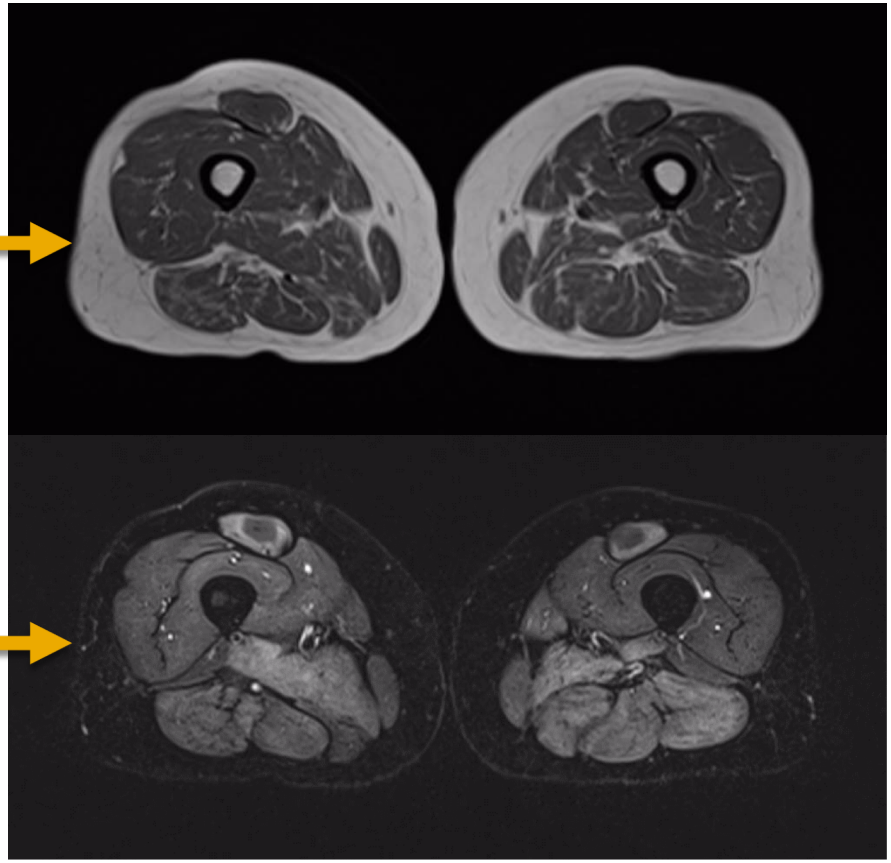
Multiple types of images can be acquired using MRI



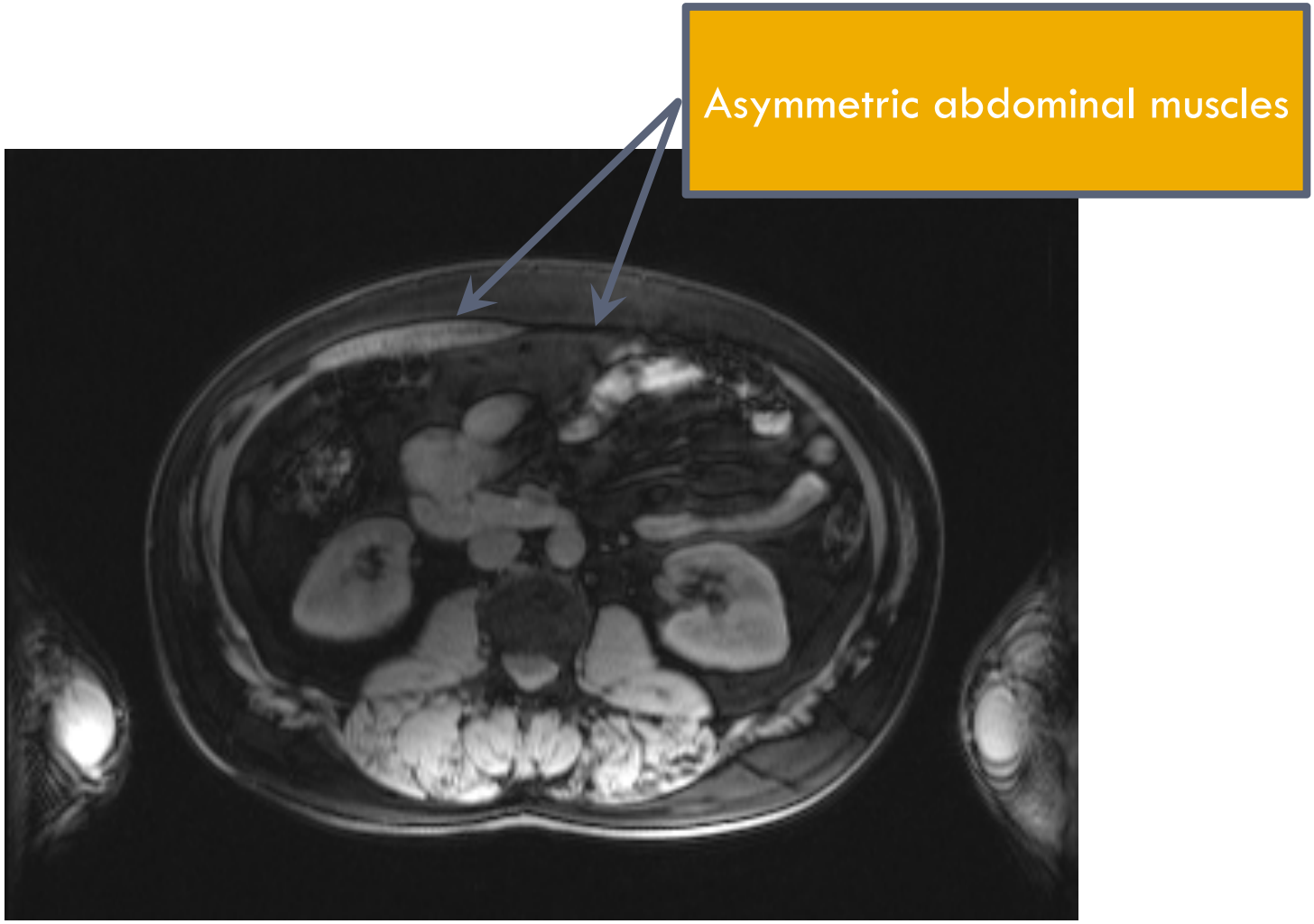
Fat sensitive | Fat suppressed | Fluid sensitive

MRI of muscle

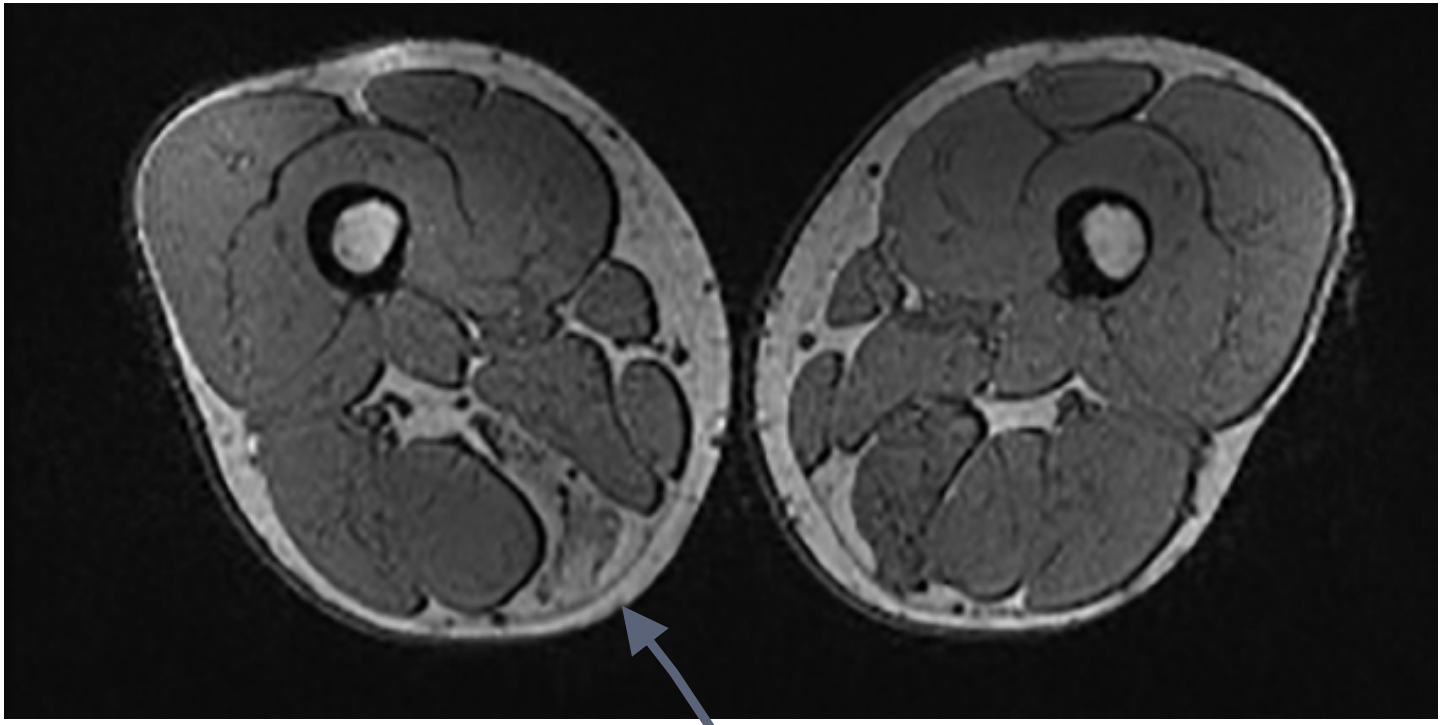
- T1-weighted
  - Contrasts fat and water
- T2-weighted
  - Sensitive to free water



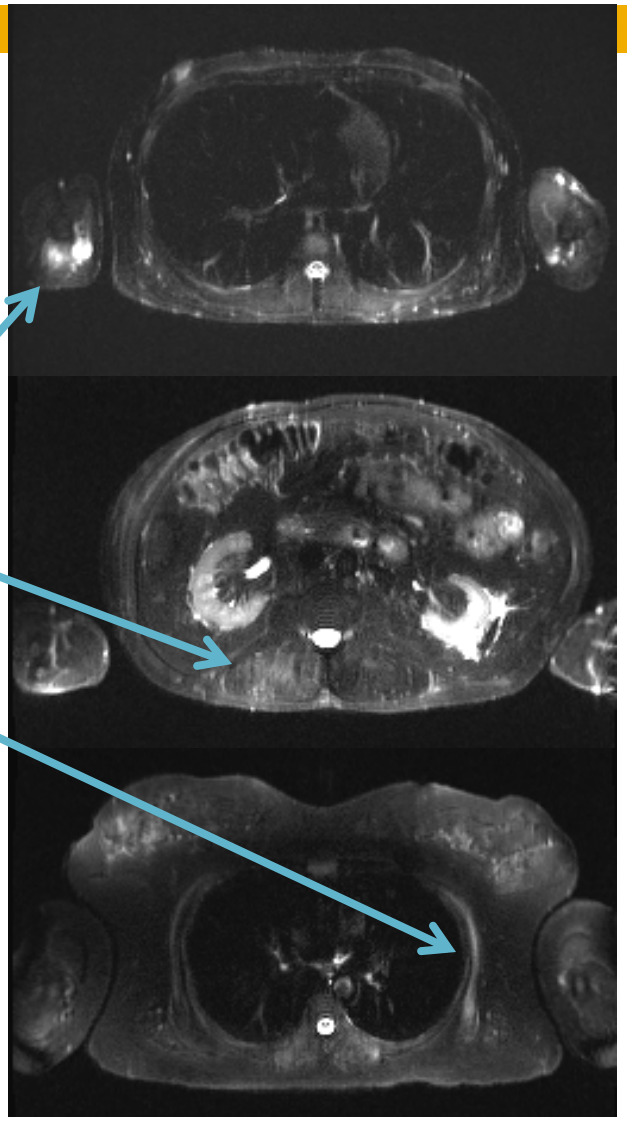
Evaluating muscles that can't be tested easily



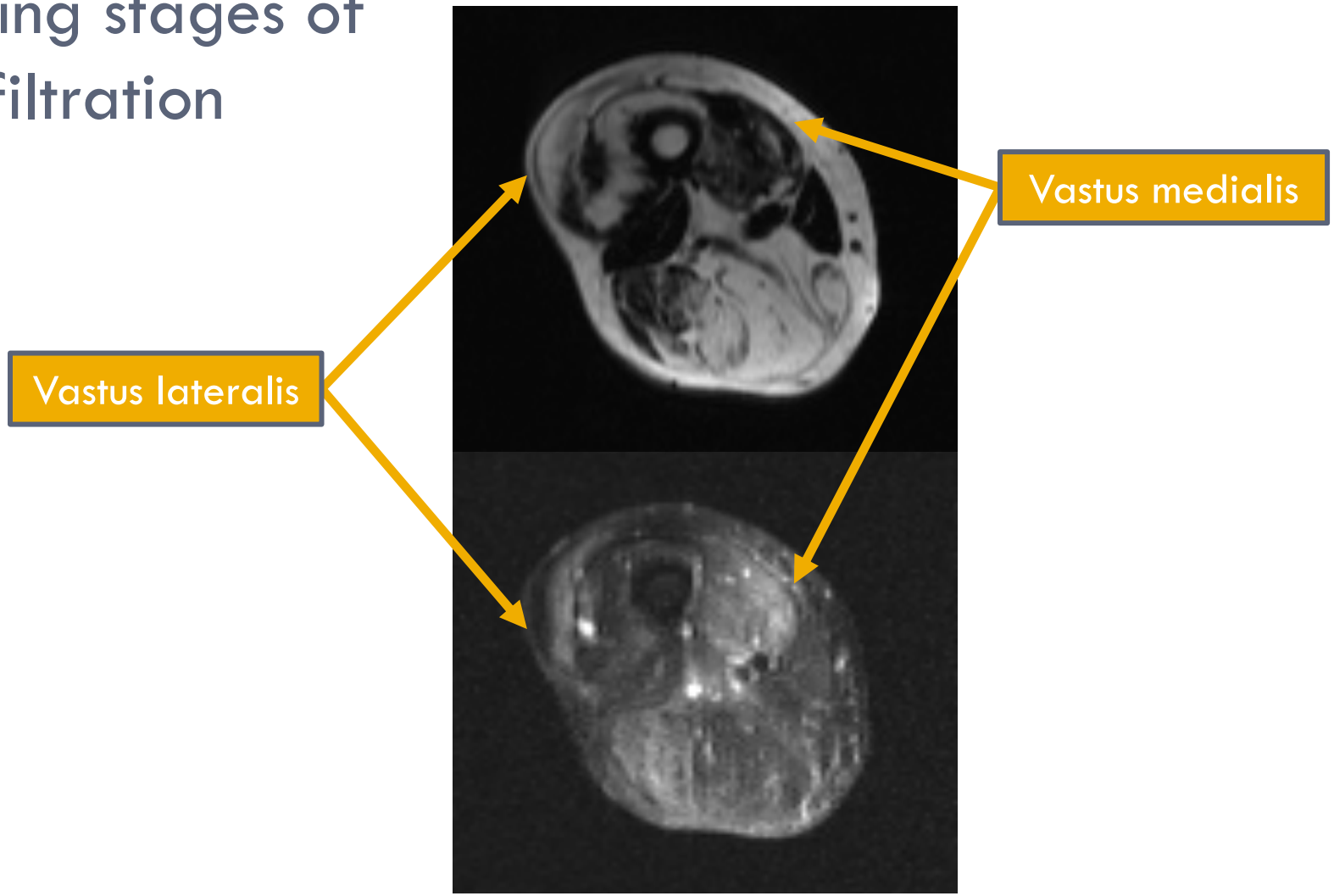
Identifying subclinical disease



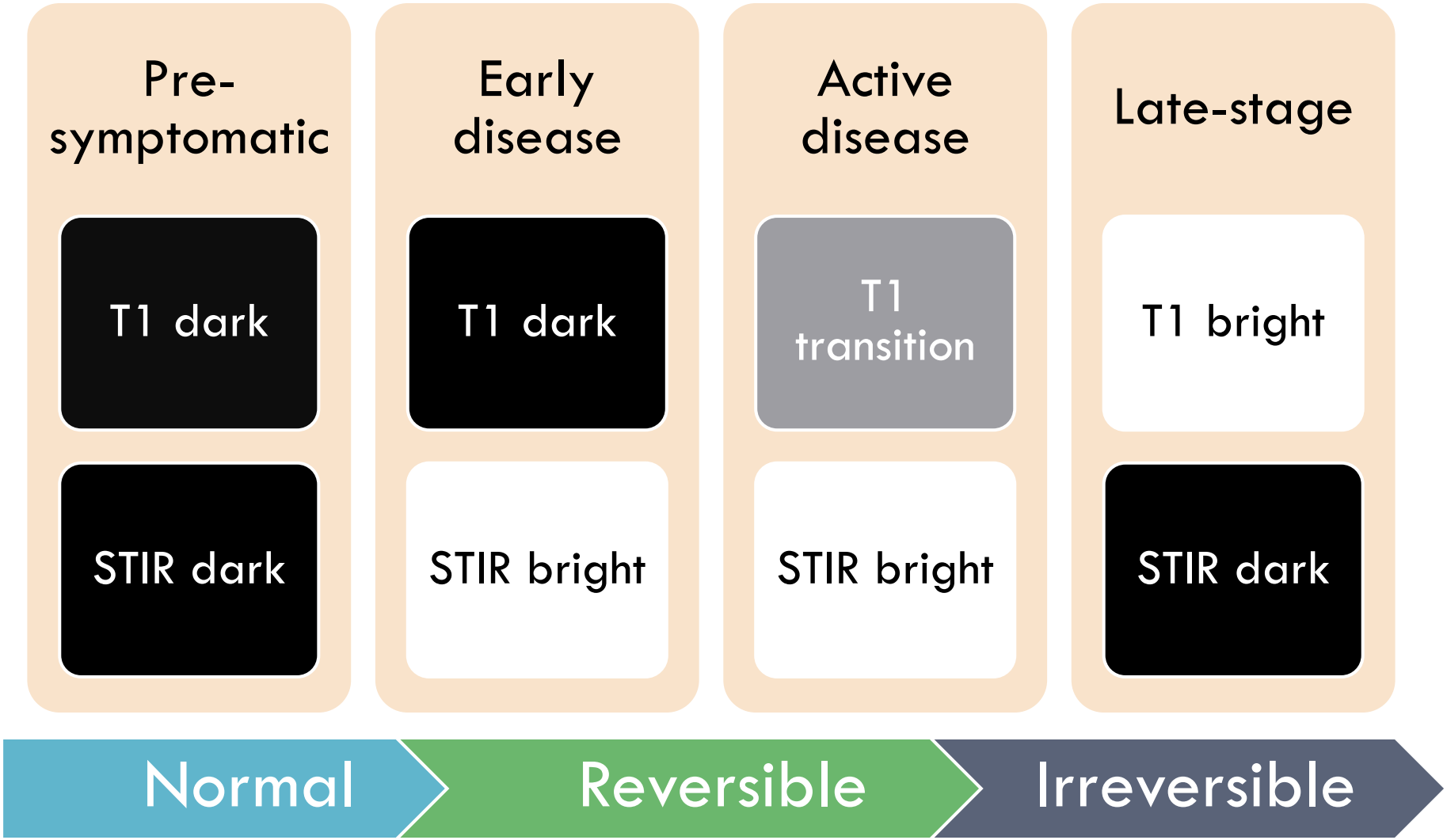
Identifying areas of active disease



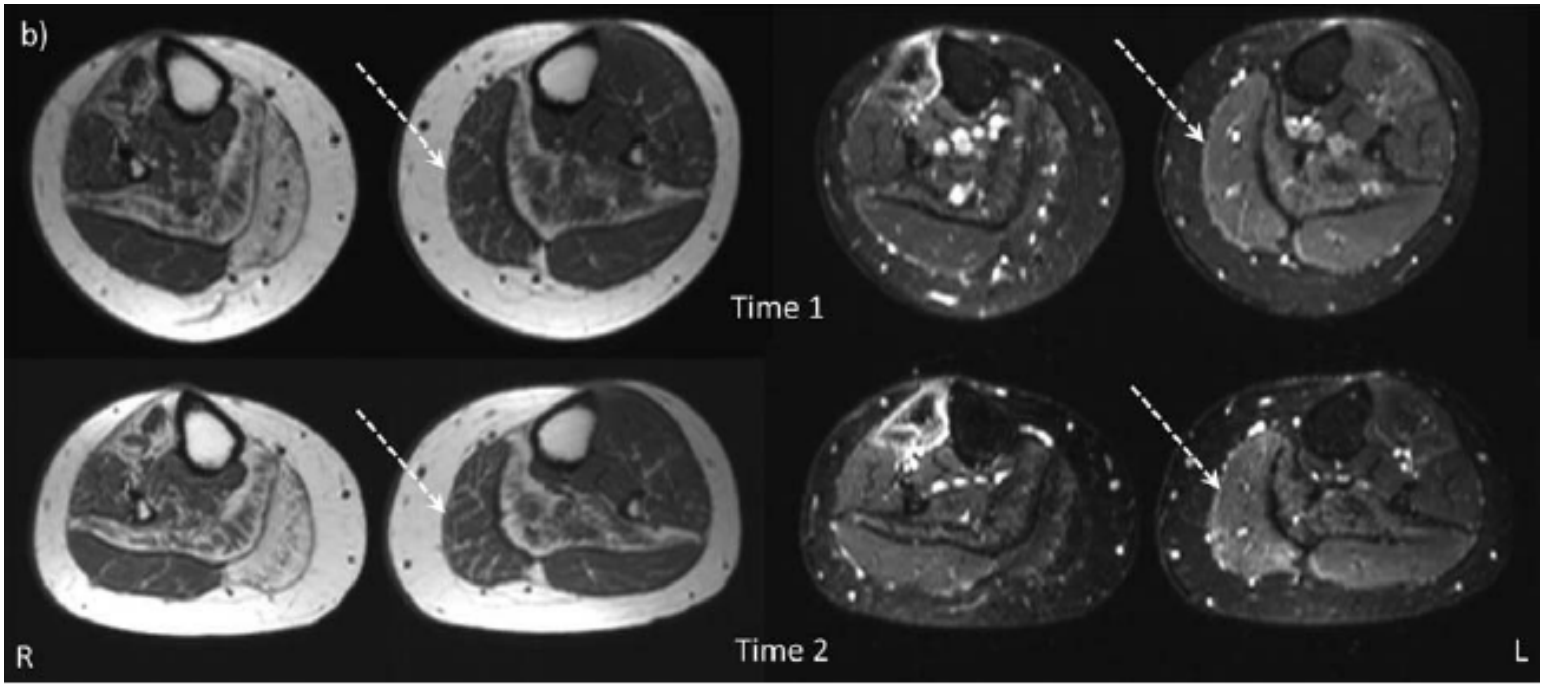
Muscle edema at differing stages of fat infiltration



Theory of disease progression

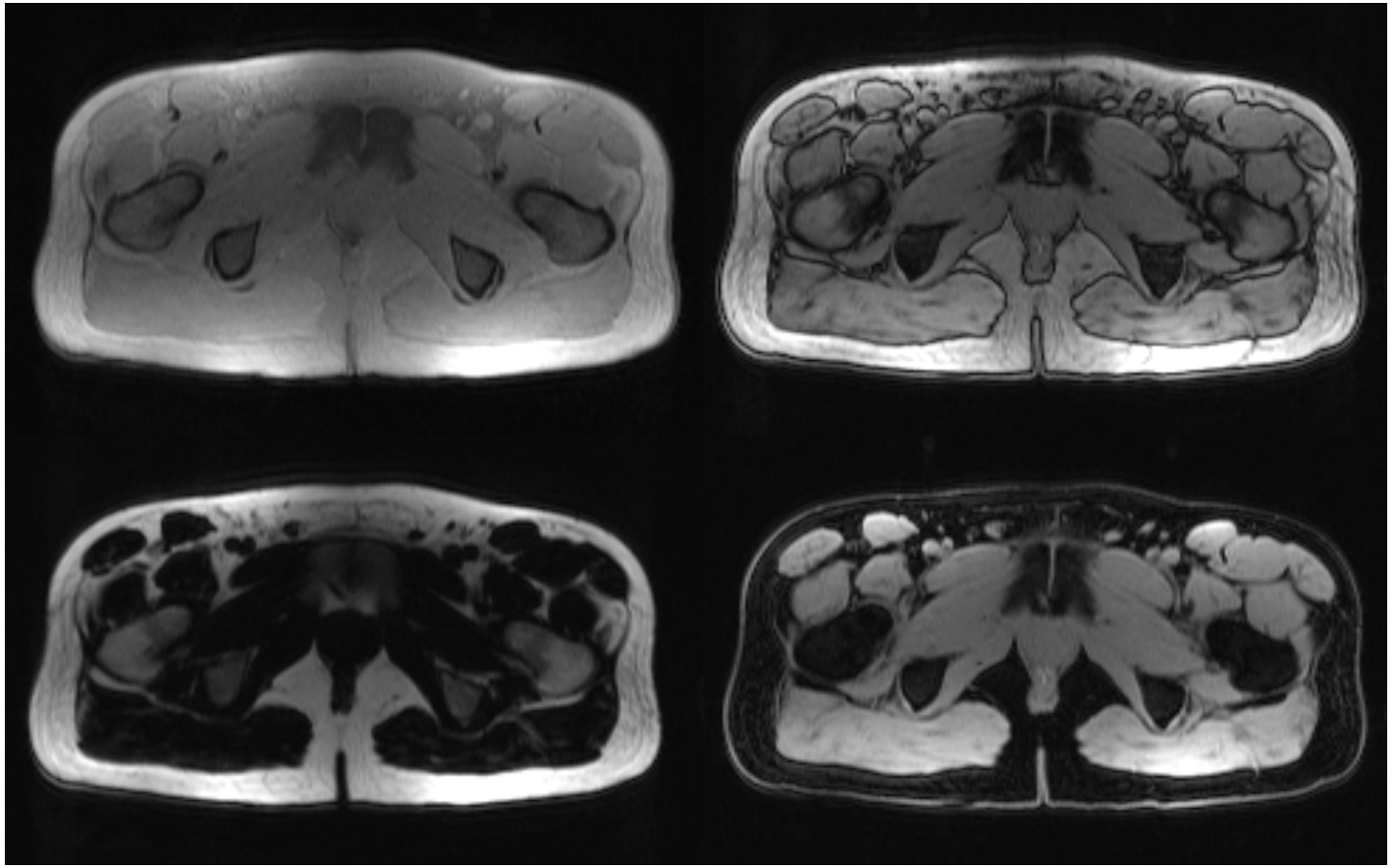


Sub-stages of fat infiltration?



Ferguson, Muscle and Nerve, 2018

Current research on MRI in FSHD



Advantages of imaging

- Large amounts of data
- Repeatable measurements
- Can be used in a wide range of disease
- Interpreters can be blinded
- Images can be stored for future re-analysis
- Ability to study small increments of change

Whole body MRI

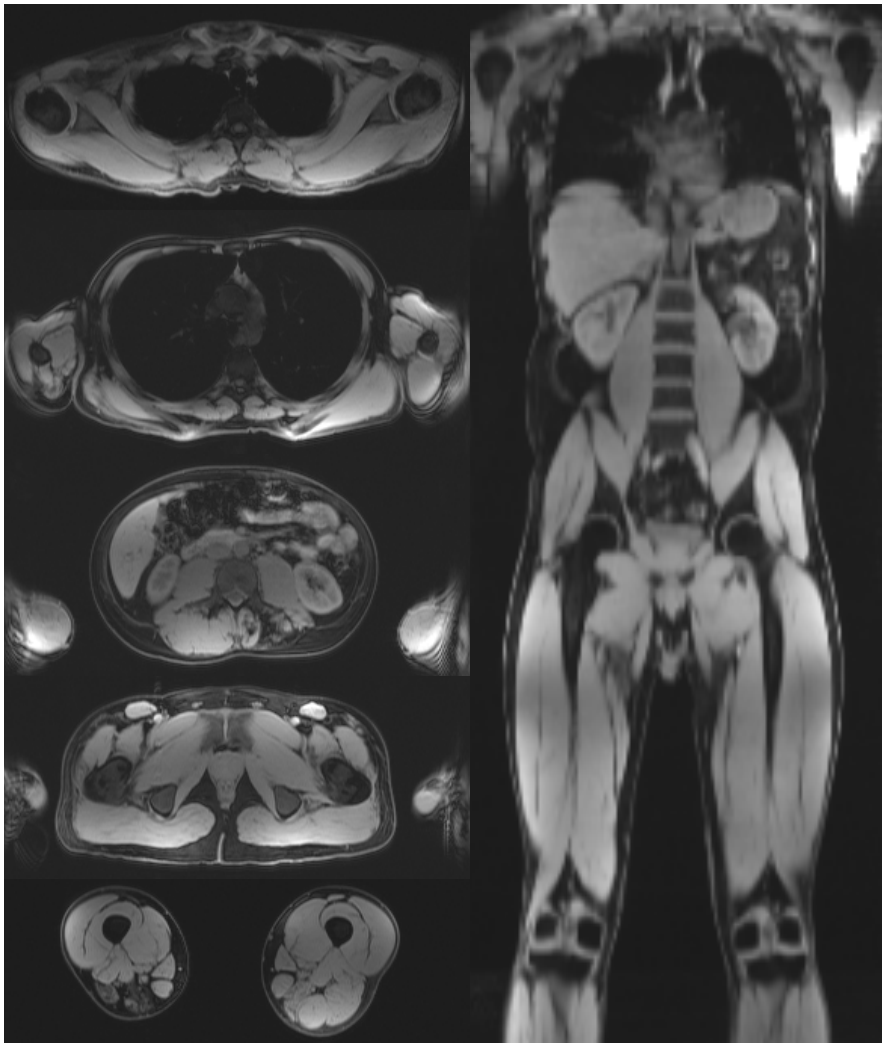
Shoulder

Chest

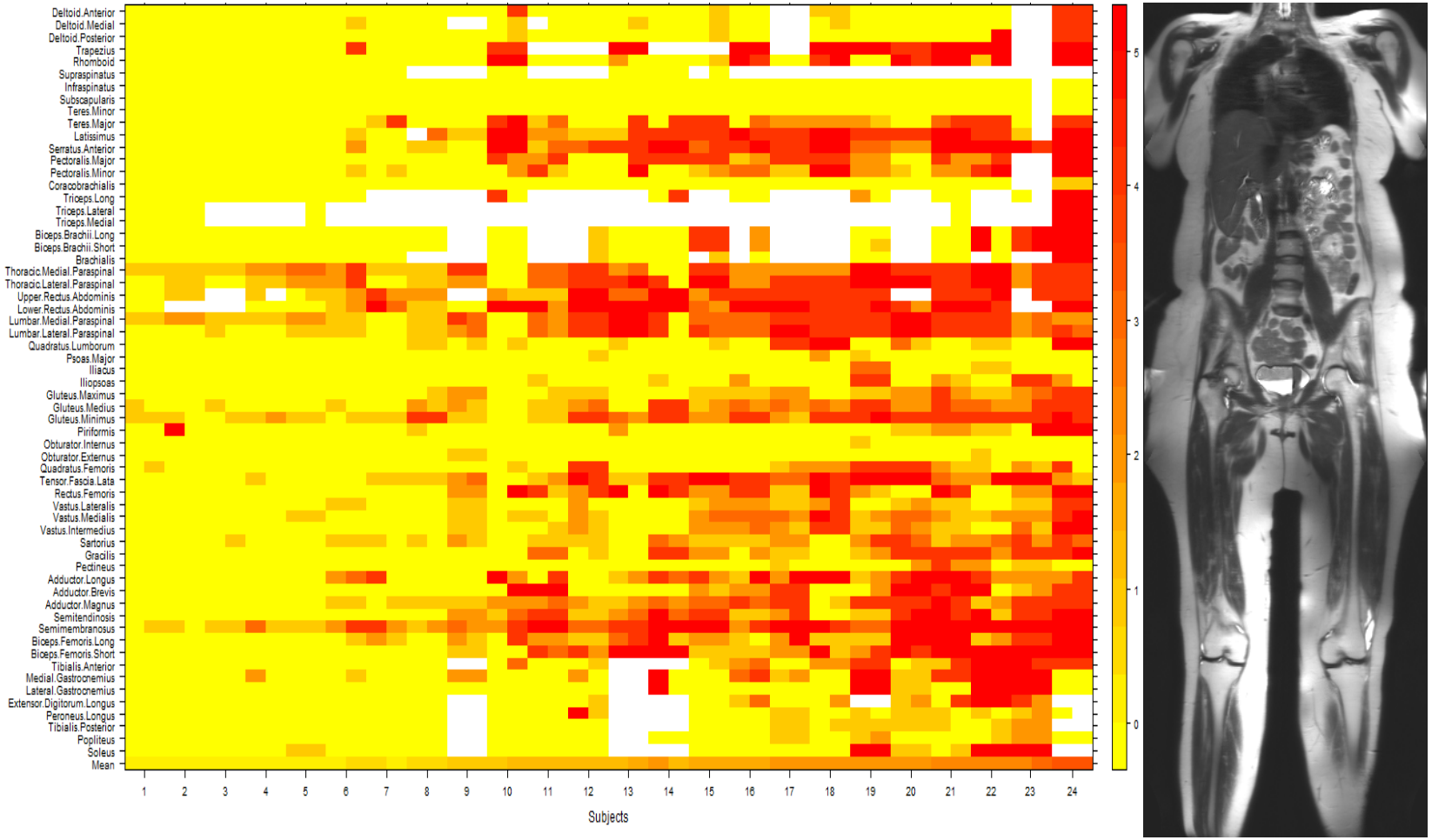
Abdomen

Pelvis

Legs



Fat infiltration scores



Rationale for longitudinal cohort

- Imaging does not necessarily follow neurological exam
  - Imaging changes can precede measurable loss of strength
- Cross-sectional findings may not reflect longitudinal changes in an individual
- Clinical trial preparedness
  - Variability in rate of change across population
  - Relationship with strength and function
  - Imaging targets that are clinically meaningful
  - Modifiers of disease progression
    - Gender, deletion size

Study visits

Baseline → 3m → 9m → 15m → 21m

Whole-body MRI scan

- Qualitative – T1 and STIR
- Quantitative – Dixon, diffusion weighted images

Muscle testing

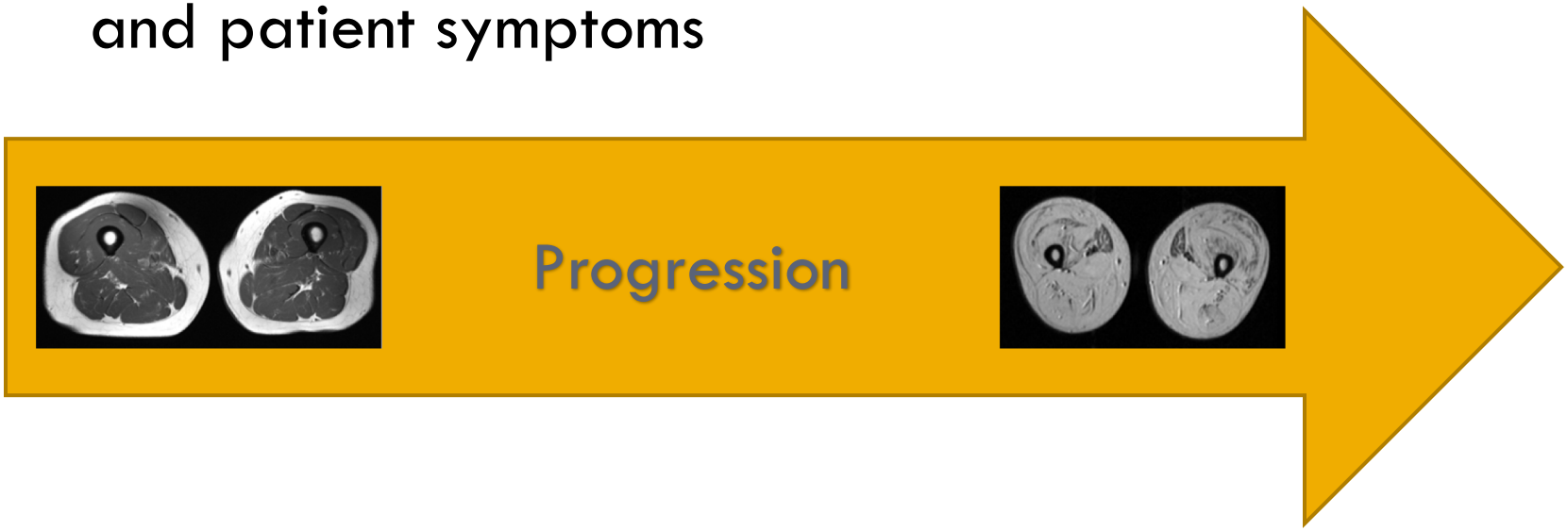
- Strength testing - dynamometry
- Timed function testing

Medical history/symptoms

- History – medications, falls, comorbidities
- Questionnaires – pain, fatigue, function

Study goals

- To evaluate longitudinal changes in muscle over time
- To characterize relationships between imaging and changes in strength/function
- To characterize relationships between imaging and patient symptoms



Future directions in MRI research

- Automated quantification methods
- Standardized techniques across centers
  - Acquisition
  - Post-processing
- Validation in larger studies and clinical trials
- Relationship to treatment

Acknowledgements

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