

Characterizing Facioscapulohumeral Muscular Dystrophy (FSHD) Longitudinal EHR Activity Using a Large US Electronic Health Records Database Linked to Claims, 2020-2025

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Introduction

- Facioscapulohumeral Muscular Dystrophy (FSHD) is a neuromuscular disorder (NMD) that leads to progressive muscle loss with patients often experiencing significant variations in severity and progression.¹
- Due to the nature of the disease, misdiagnosis and diagnostic delay is common, often requiring patients to consult multiple specialists before receiving a formal diagnosis.²
- Real-world FSHD longitudinal data remains limited, posing a challenge to understanding clinical pathways and assessing outcome measures

Objective

- To examine EHR activity among FSHD patients in a large US real-world database to better understand availability of comprehensive longitudinal data to advance FSHD research.

Methods

Data Source

- Data from Veradigm Network EHR linked to claims from Komodo Health was used to identify patients with an FSHD diagnosis between 1/1/2020 - 12/31/2025. Due to the real-world, observational design of this study, baseline and follow-up periods were not standardized across patients.
- Length of EHR activity and patient demographic characteristics were described as of the earliest diagnosis date (index date). During the variable length follow-up period, conditions were captured using ICD-10 diagnosis codes, medication classes were categorized using NDC codes, and provider types were described based on reported specialty.

Figure 1: Patient Selection

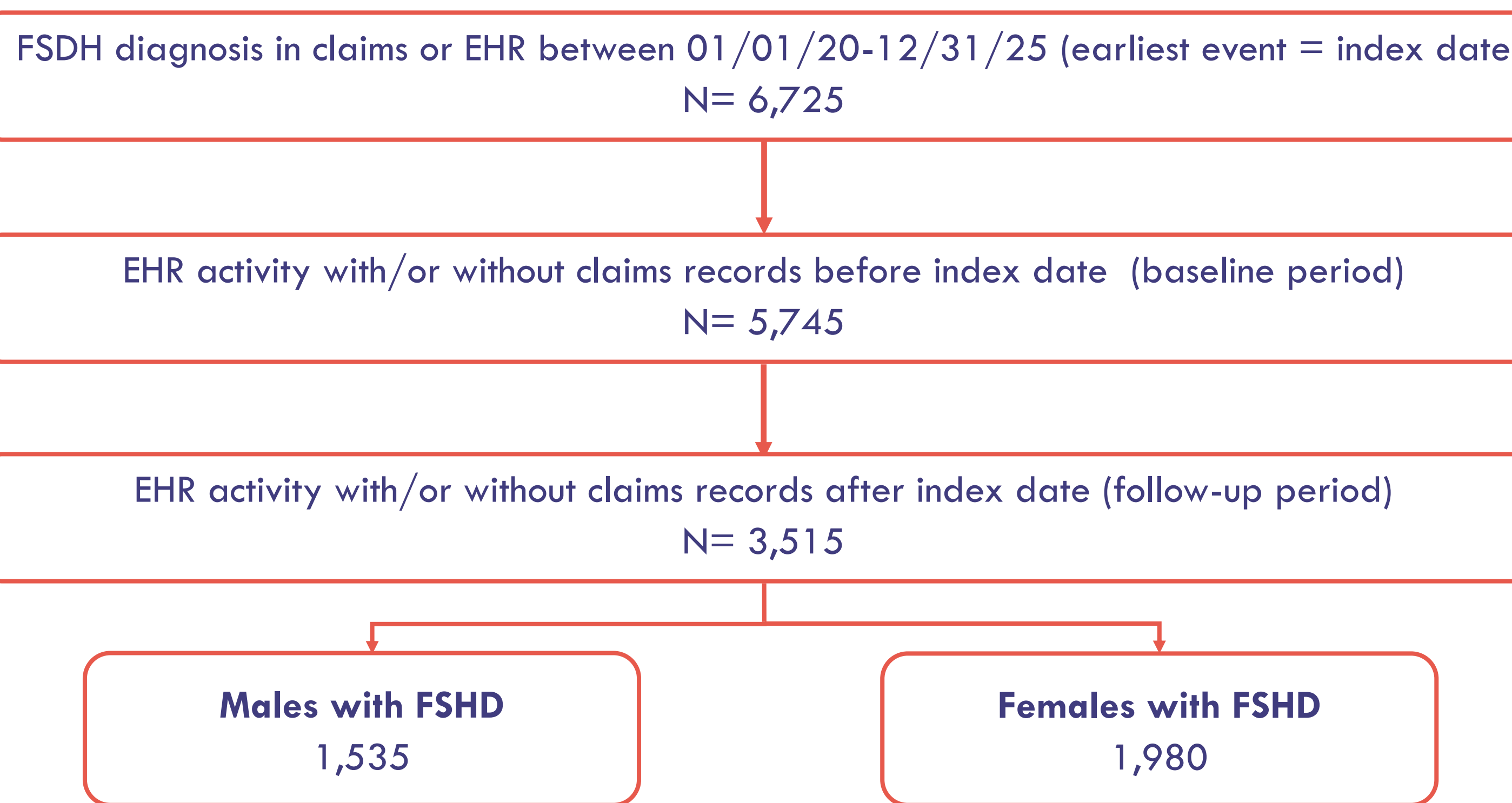


Figure 2: Percent of Patients with EHR Activity

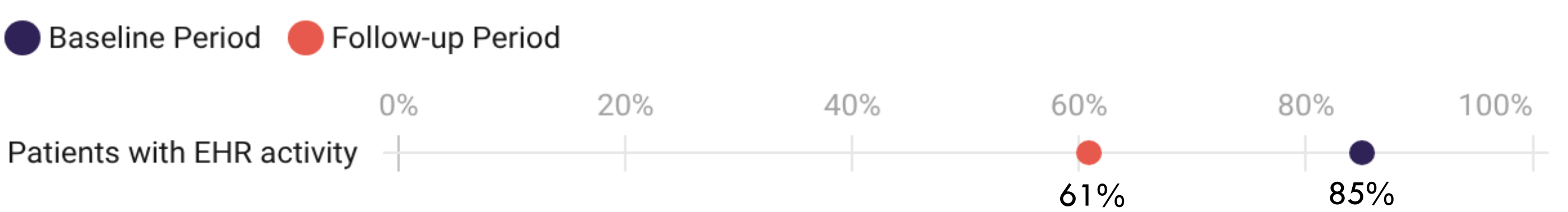


Figure 3: Length of EHR Activity, Overall FSHD Cohort

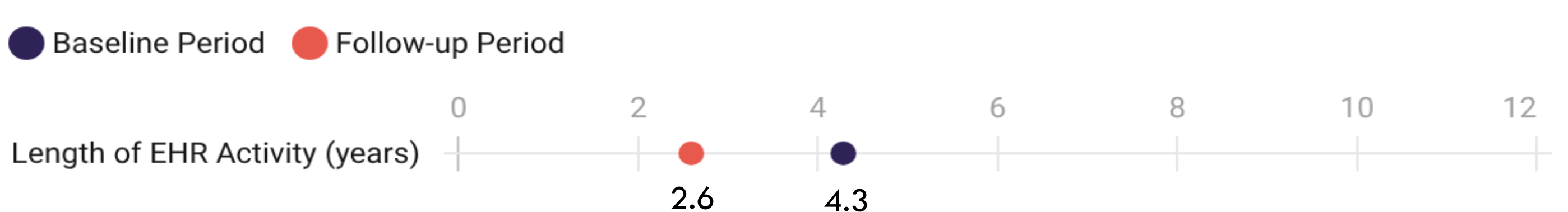


Table 1: Patient Demographics

	Males Patients N = 1,535	Female Patients N = 1,980
Age (Mean, SD)	54.4 (18.6)	55.1 (17.6)
Race (N, %)		
White	1,078 (70.2%)	1,392 (70.3%)
Black	56 (3.6%)	93 (4.7%)
Asian	34 (2.2%)	55 (2.8%)
Other	184 (12.0%)	253 (12.8%)
Unknown/Not Reported	183 (11.9%)	187 (9.4%)
Ethnicity (N, %)		
Hispanic	44 (2.9%)	59 (3.0%)
Non-Hispanic or Unknown	1,491 (97.1%)	1,921 (97.0%)
Geographic Region (N, %)		
Northeast	301 (19.6%)	307 (15.5%)
Midwest	296 (19.3%)	459 (23.2%)
South	512 (33.4%)	730 (36.9%)
West	388 (25.3%)	459 (23.2%)
Other/Unknown	38 (2.5%)	25 (1.3%)

SD, standard deviation

Results

- We identified 6,725 eligible patients with a diagnosis of FSHD during the study period.
- 5,745 (85%) had documented EHR activity prior to FSHD diagnosis (baseline), and 3,515 patients (61%) had activity post-diagnosis (follow-up) (Figure 2).
- Prior to receiving a formal FSHD diagnosis, patients had a mean of 4.3 years (SD 2.2) of EHR activity compared to a mean of 2.6 years (SD 1.8) of EHR activity post diagnosis (Figure 3).
- Mean (SD) age overall was 55 (18.0) years old, and most patients were female (56%) and of White race (70%) (Table 1).
- FSHD patients were predominantly between the ages of 55 and 64, or 65 and older (Figure 4).
- The most common nervous system clinical condition among male patients was chronic pain (28%), whereas females was fatigue (34%) (Figure 5).
- Primary care providers accounted for most visits among FSHD patients (56%), followed by Ophthalmology (15%) and Optometrist (10%) (Figure 6).
- Females showed higher rates of antidepressant use, suggesting a greater burden of mental health conditions (44% vs 32%) (Figure 7).

Figure 4: Age Distribution by Cohort

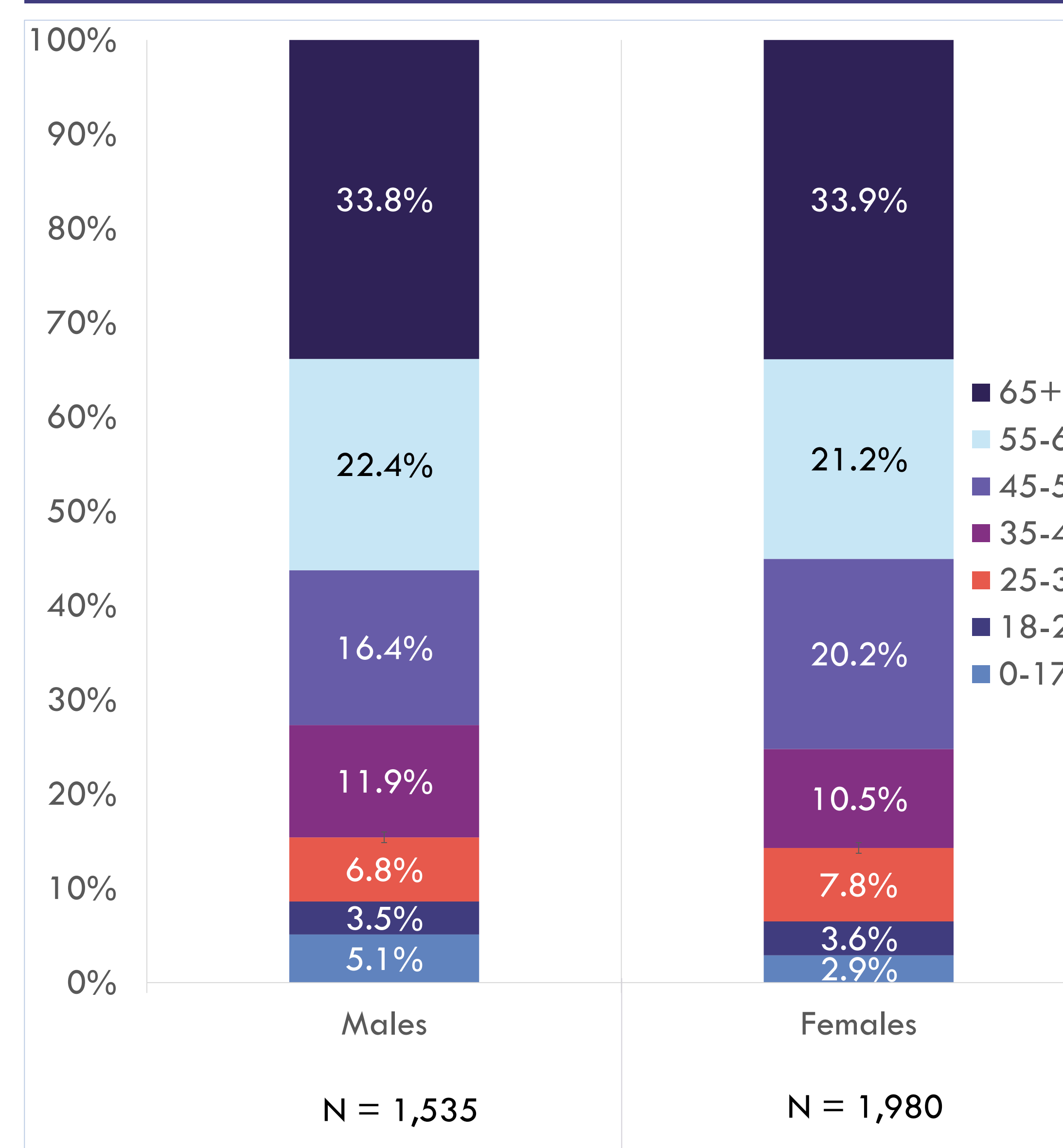


Figure 5: Nervous System Conditions, Follow-up Period

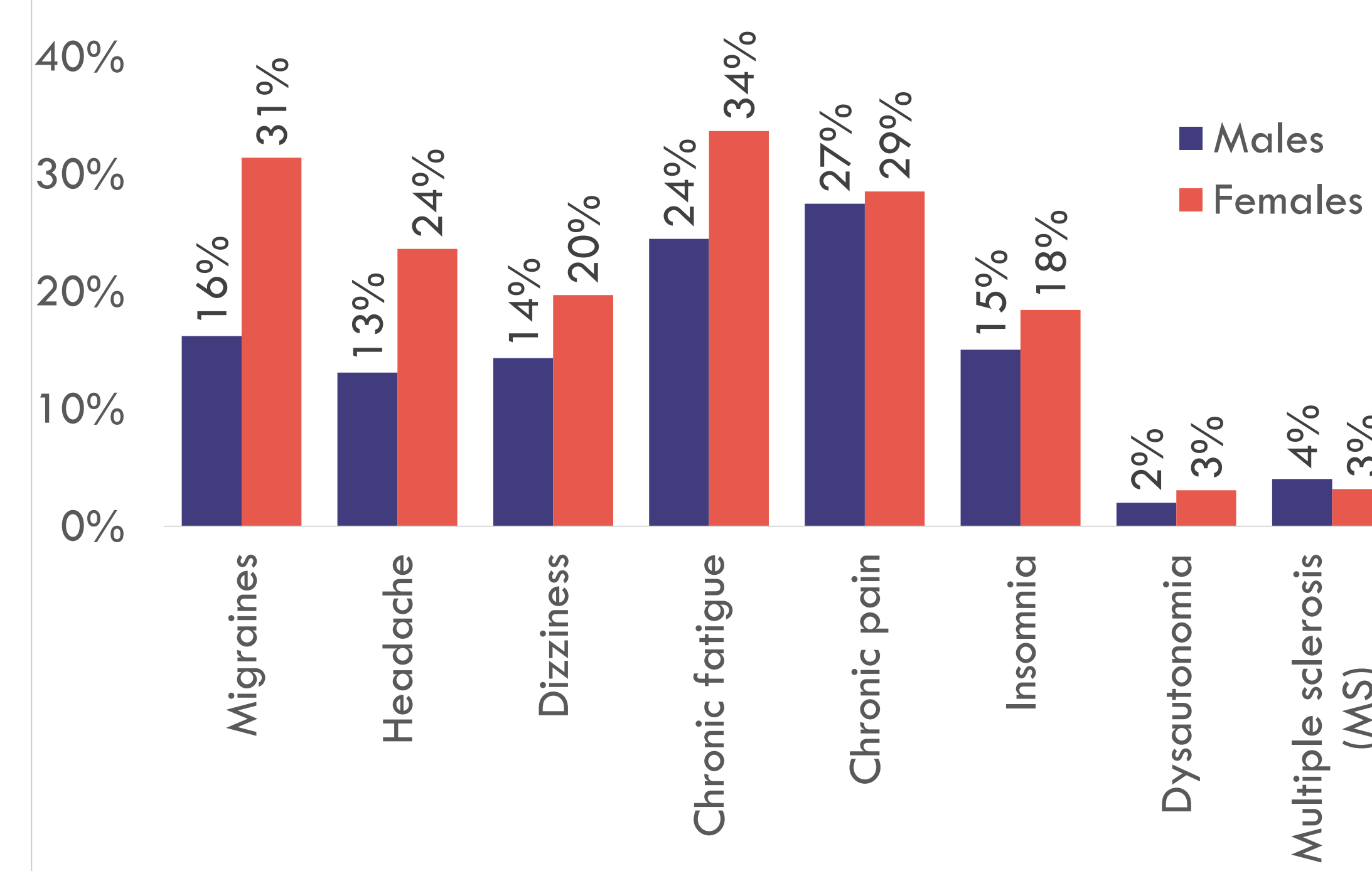
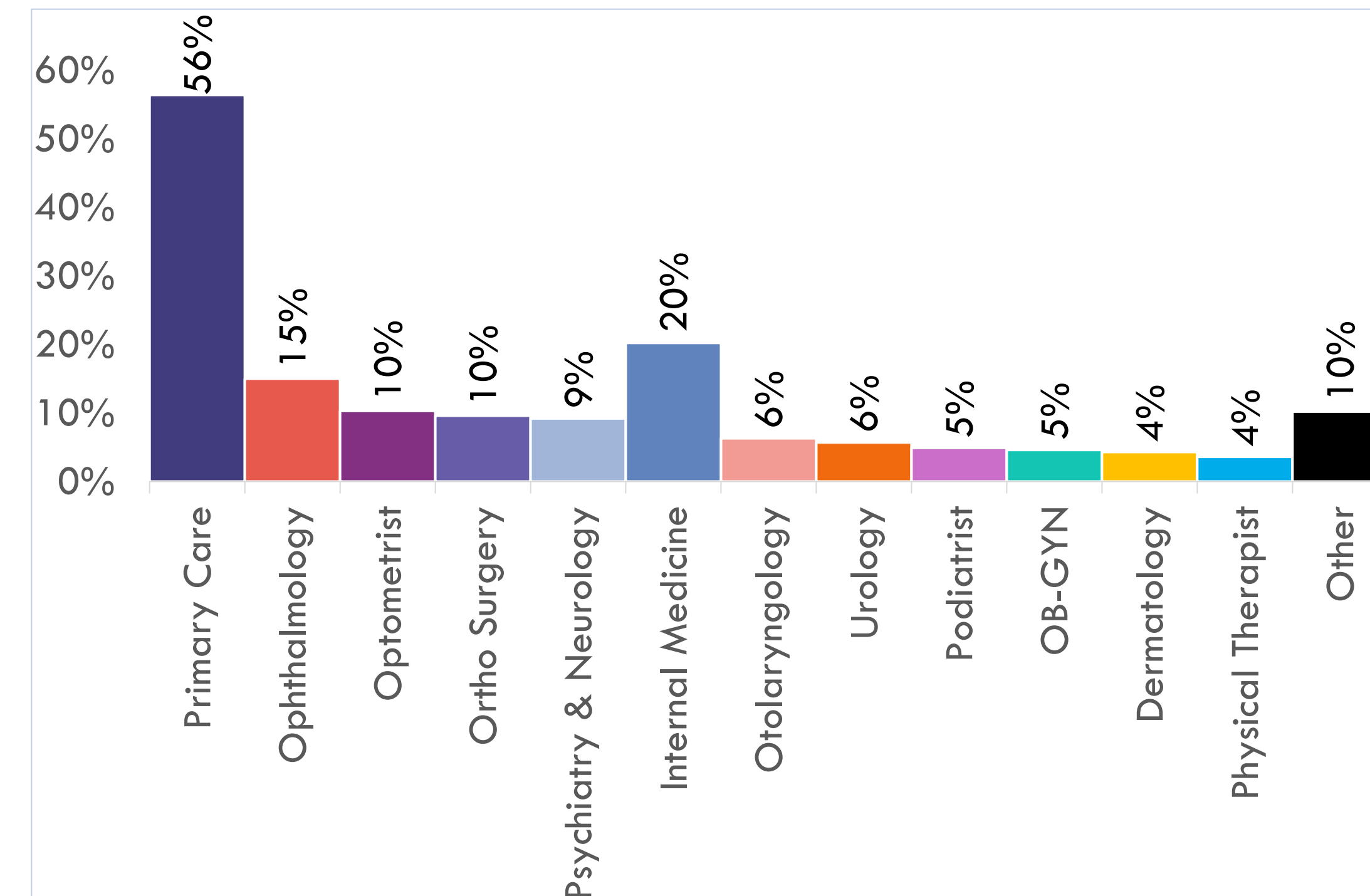


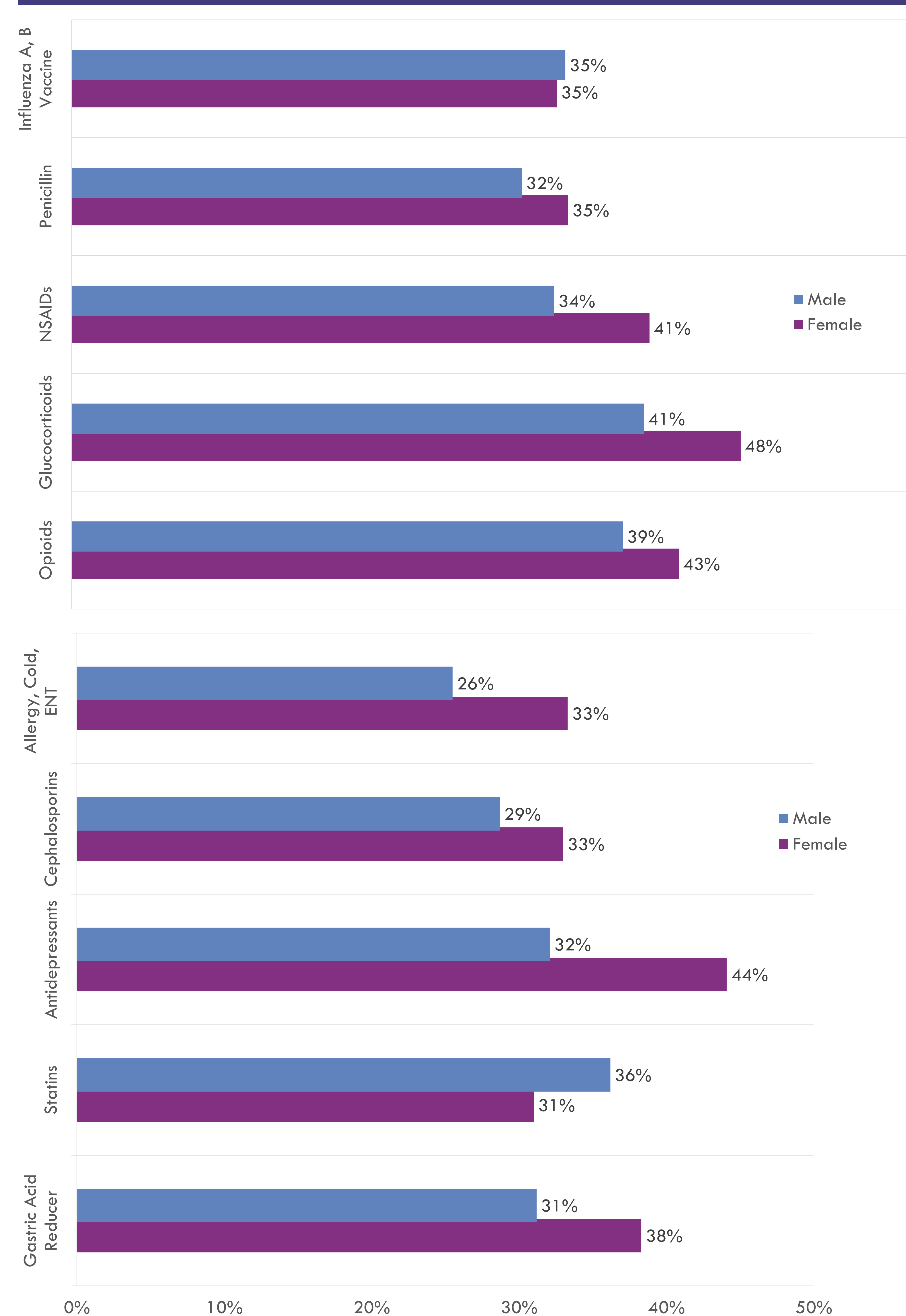
Figure 6: Distribution of Provider Specialty, Overall FSHD Cohort, Follow-Up Period



Results (cont'd)

- Alternately, males had notably higher statin use compared to females (36% vs 31%), suggesting a greater focus on cardiovascular management in males (Figure 7).

Figure 7: Medication Used Over Follow-up Period



Conclusions

- Early disease detection may contribute to delays in timely care in FSHD, as EHR activity suggests some diagnoses may have occurred prior to the study index date. Our findings highlight the value of longitudinal real-world EHR data for understanding care patterns to further advance FSHD research.
- Gender-based differences should be considered in the clinical management and treatment of FSHD patients.

References

- Mul K. Continuum. 2022; 28(6):1735-1751.
- Facioscapulohumeral Muscular Dystrophy Society. Understanding FSHD? <https://www.fshdsociety.org/living-with-fshd/understanding-fshd/>

Disclosures

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