Understanding the Patients' Journey to the Diagnosis of Facioscapulohumeral Muscular Dystrophy (FSHD): a Real-World Retrospective Data Analysis

Chamindra Konersman[1], Kathryn A. Munoz[2], Richard A. Brook[3], Nathan L. Kleinman[3], Kelly DiTrapani[2], Bradley McEvoy[2], Alissa N. Peters[2]; Chao-Yin Chen[2], Mark C. Stahl[2]

[1]University of California San Diego; [2]Avidity Biosciences, Inc.; [3]Better Health Worldwide. All authors have met authorship criteria.

Objectives

Describe the healthcare conditions, costs, and services received by FSHD patients versus matched non-FSHD controls (MCs) 1 year before confirmed FSHD diagnosis

Background

- FSHD is a rare, slowly progressive, genetic skeletal muscle disease. Muscle weakness usually presents in the face and upper extremities, eventually extending to the trunk and lower body[1,2]
- Patients experience significant physical limitations, pain, fatigue, and an overall negative impact on wellbeing[3,4]
- Real-world data characterizing the patients' pre-diagnosis journey is limited
- Currently there is no cure or targeted treatment[5]

Methods

- Retrospective database analysis to compare conditions, costs, and services of FSHD patients versus those of MCs
  - Database: IQVIA US PharMetrics® Plus
  - Time frame: January 2016 through March 2021
- The FSHD cohort is defined as having ≥2 FSHD claims ≥30 days apart
  - Claims identified by International Classification of Disease Tenth Revision (ICD-10) code G71.02
- The first diagnosis date was used for the index date
- FSHD patients were matched to a 5% random sample of eligible non-FSHD controls
  - Matching was done using R's Matchit procedure, with nearest neighbor matching (exact matching on month of index date)
- Cohorts were matched (5-MC:1-FSHD) on index month and baseline age, region, gender, plan, and payer type
- All subjects (patients and matched controls) had a minimum of 12 months of continuous data prior to their index date
  - Because our focus was on the events preceding diagnosis, the index date was not included in the 1-year pre-index evaluation period
- Healthcare conditions, costs, and services used during the 12 months before each subject's index date were compared using 283 US Agency for Healthcare Research & Quality (AHRQ) comorbidity categories
- Comparisons were made using *t*-tests for continuous variables and chi-square tests for discrete variables
- Total per member per year (PMPY) direct medical and prescription (Rx) costs were calculated for both FSHD and MC cohorts
- All costs were inflation adjusted using the US Bureau of Labor Statistics Consumer Price Index (CPI) for December 2020
  - Medical claims used the medical cost CPI and prescription claims used the prescription cost CPI

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This poster is sponsored by Avidity Biosciences.

Abbreviations
AHRQ, Agency for Healthcare Research and Quality; CPI, Consumer Price Index; FSHD, facioscapulohumeral muscular dystrophy; ICD, International Classification of Disease; MC, matched controls; PMPY, per member per year; Rx, prescription; SD, standard deviation.

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[1]Greco A, et al. *Clin Genet.* 2020;97(6):799–814; [2]Statland JM and Tawil R. *Continuum (Minneap Minn)*. 2016;22(6):1916–1931; [3]Hamel J, et al. *Neurology.* 2019;93(12):e1180–e1192; [4]Tawil R and van der Maarel SM. *Muscle Nerve*. 2006;34(1):1–15; [5]Cohen J, et al. *Trends Mol Med*. 2021;27(2):123-137.

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Results

- We identified 290 FSHD patients and 1,450 MCs
  - Descriptive characteristics were similar between cohorts (Table 1)
  - The cohorts had significant (p<0.05) pre-index differences for the Charlson Comorbidity Index:
    - Mean scores were 0.71 (SD 1.29) for FSHD patients versus 0.52 (SD 1.30) for MCs
    - Percent of patients with values >1 was 18.6% for FSHD patients versus 11.4% for MCs
- Overall, compared with MCs:
  - **Conditions:** 84 out of 283 AHRQ categories had significantly higher pre-index use in FSHD patients (top categories by difference shown in Figure 1). Zero categories had significantly greater use in MCs
  - **Cost:** 12 AHRQ categories had significantly different pre-index costs. Those costing more for the FSHD patients versus MCs are shown in Figure 2. Those costing less for FSHD patients versus MCs were "coagulation and hemorrhagic disorders" ($0 vs $2 MCs), "other pregnancy and delivery including normal" ($0 vs $9 MCs), "other complications of pregnancy" ($0 vs $9 MCs), "cataract" ($5 vs $38 MCs), and "sprains and strains" ($26 vs $103 MCs)
  - **Services:** The 23 AHRQ categories with significant pre-index differences in the number of services are shown in Figure 3
- FSHD patients' annual PMPY total cost, days of service, and PMPY services requiring healthcare are shown in Table 2

Table 1. Age, US Region, Insurance, and Payer Type Were Similar for FSHD Patients and MCs

Descriptive Characteristics	FSHD Patients (N=290)
Gender, % female	43.8%
Age, mean (SD) years	47.4 (17.6)
Age, years	
<18	6.6%
≥18 to <35	17.6%
≥35 to <45	11.0%
≥45 to <55	22.4%
≥55 to <65	30.3%
≥65	12.1%
US region	
South	33.1%
Midwest	30.0%
Northeast	20.3%
West	16.6%

Descriptive Characteristics	FSHD Patients (N=290)
Insurance type	
Preferred provider organization	78.6%
Health maintenance organization	13.8%
Point-of-service plan	5.2%
Consumer-directed healthcare	1.4%
Indemnity/traditional plan	1.0%
Payer Type	
Commercial	61.7%
Self-insured	29.3%
Medicaid	1.0%
Medicare Advantage	5.9%
Medicare Supplemental	2.1%

There were no significant differences between FSHD patients and MCs

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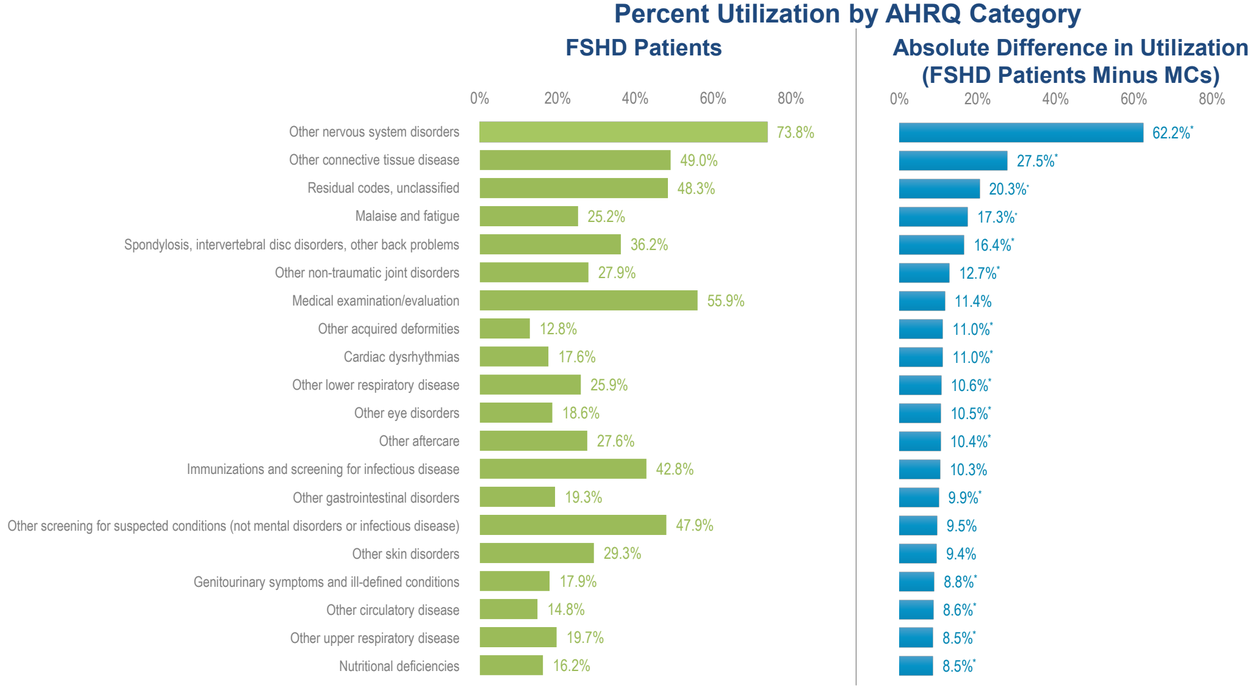
Figure 1. Prior to Diagnosis, FSHD Patients had Higher Absolute Differences[a] in Prevalence for AHRQ Categories of "Other Nervous System Disorders", "Other Connective Tissue Disease", "Residual Codes, Unclassified", "Malaise and Fatigue", and "Spondylosis, Intervertebral Disc Disorders, Other Back Problems" Versus MCs (All $p<0.05$)



[a]Absolute difference=FSHD cohort minus MC cohort

All $p<0.05$. *$p<0.000177$ (threshold for multiple comparisons [0.05 / 283 comparisons])

Figure 2. Prior to Diagnosis, FSHD Patients had Higher Costs in AHRQ Categories Related to "Other Nervous System Disorders", "Other Connective Tissue Disease", "Headache, Including Migraine", and "Other Lower Respiratory Disease" Versus MCs (All $p<0.05$)

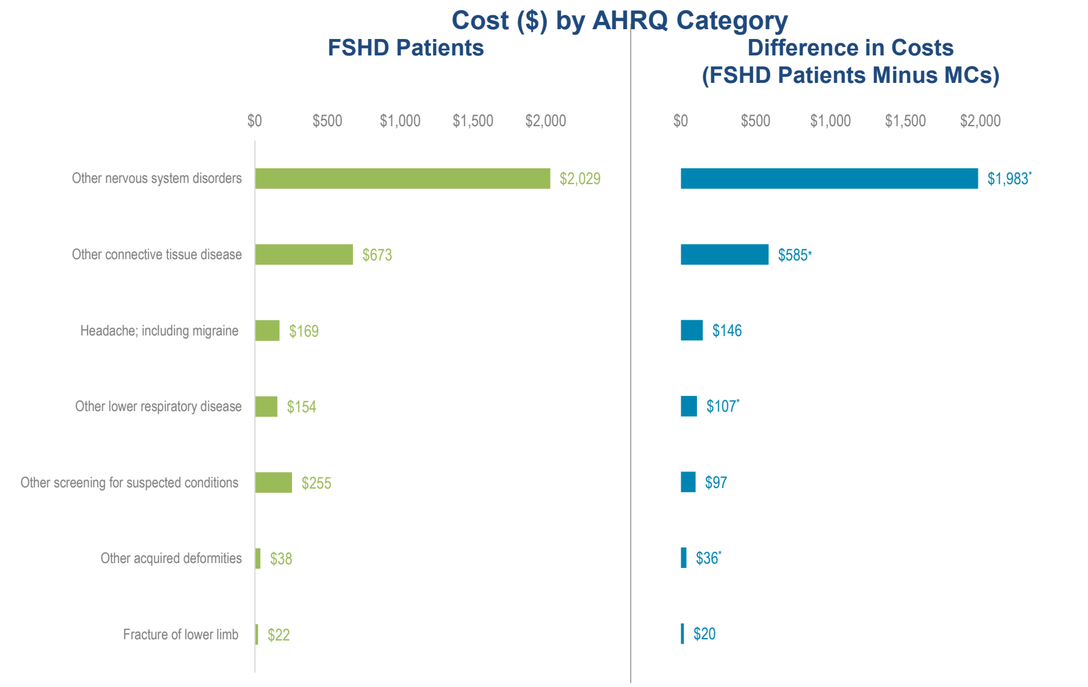


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Figure 3. Prior to Diagnosis, FSHD Patients Had More Services Billed for "Other Nervous System Disorders", "Other Connective Tissue Disease", "Spondylosis, Intervertebral Disc Disorders, Other Back Problems" "Cardiac Dysrhythmias", "Headache, Including Migraines", and "Malaise and Fatigue" Versus MCs

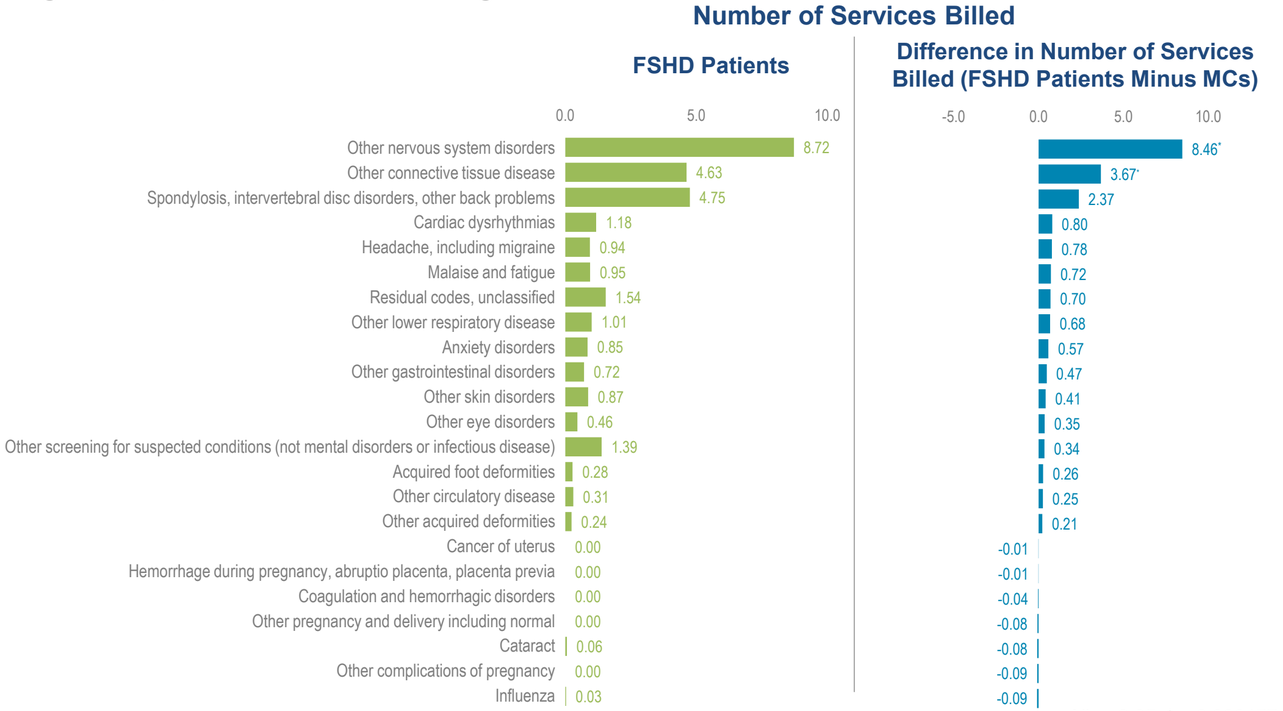

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Table 2. FSHD Patients' Annual PMPY Total Cost, Days of Service and PMPY Services Requiring Healthcare

Category		Mean (SD)		Difference (FSHD minus MCs)
		FSHD patients	MCs	
PMPY Total Cost	Medical	$11,082 ($24,879)	$5,525 ($22,990)	$5,556*
	Rx	$2,768 ($9,759)	$1,589 ($5,845)	$1,179+
Days of Service Requiring Healthcare	Medical	20.51 (20.86)	9.67 (15.36)	10.84*
	Rx	14.31 (16.04)	9.40 (12.13)	4.92*
PMPY Services Requiring Healthcare	Medical	59.97 (67.58)	29.80 (49.66)	30.17*
	Rx	21.70 (28.71)	13.54 (18.93)	8.16*

*$p<0.0001$; +$p<0.05$

Conclusions

- The journey to an FSHD diagnosis encounters higher use, cost, and services in many AHRQ categories prior to the FSHD diagnosis, including nervous system disorders, connective tissue disease, headache, and lower respiratory disease
  - This corroborates the premise that the FSHD diagnosis process is complex and often delayed, and that symptom management on the journey to diagnosis contributes to the cost and burden of FSHD to patients and society
  - Identifying patterns of care prior to diagnosis of FSHD may support earlier diagnosis
- Based on the high unmet need, Avidity Biosciences is planning clinical trials with a first-in-class antibody oligonucleotide conjugate targeting DUX4, the underlying cause of FSHD, in 2022
- Future research should investigate changes in healthcare condition diagnoses and incurred costs post diagnosis

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