



FSHD 101: What every patient needs to know

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FSHD 101

FSHD is a muscular dystrophy

Biology of FSHD

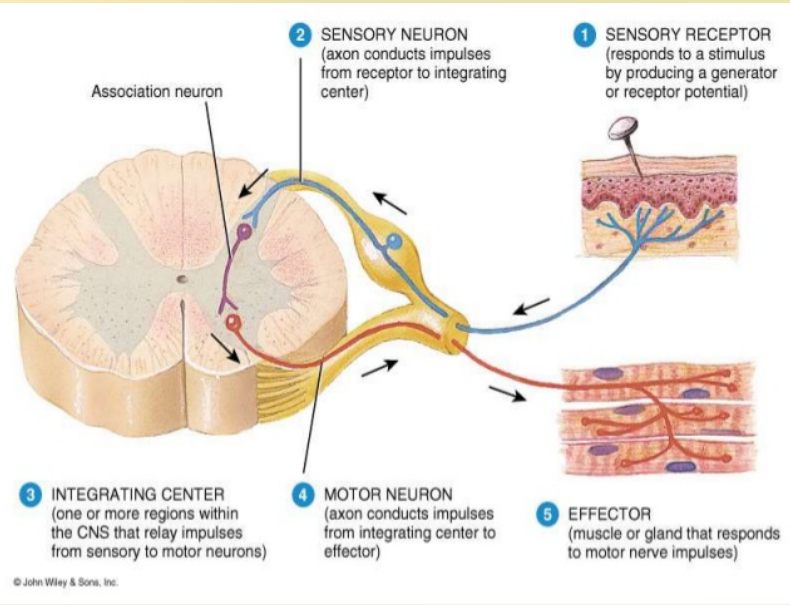
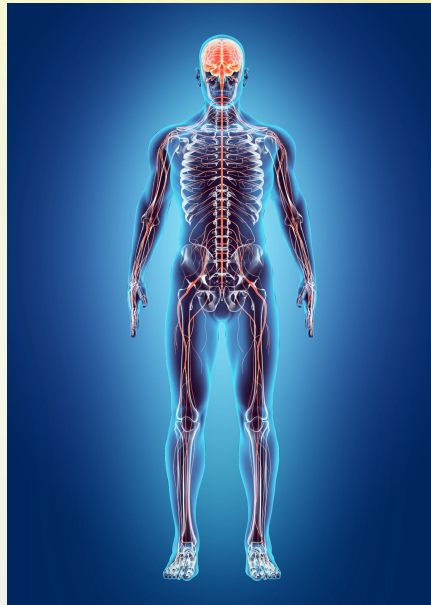
Clinical presentation

What to monitor

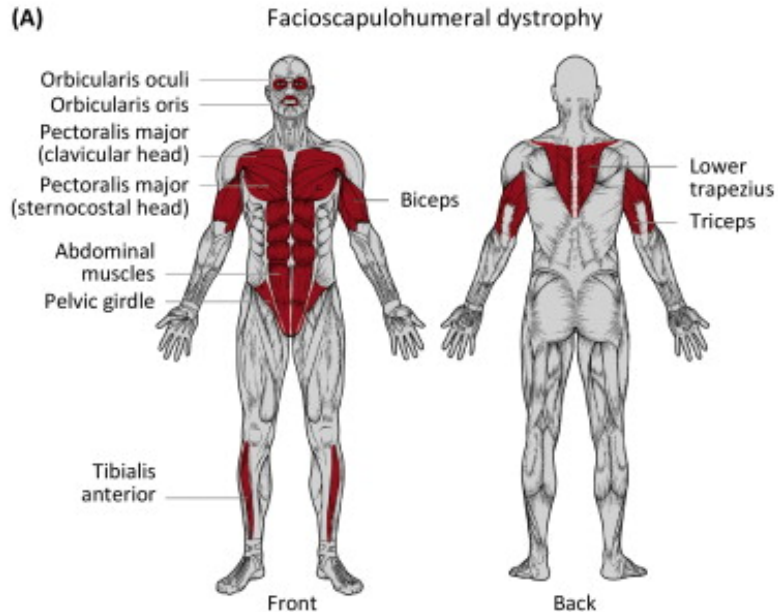
Genetics and Risk estimation

Management

FSHD is a muscular dystrophy



Clinical Presentation



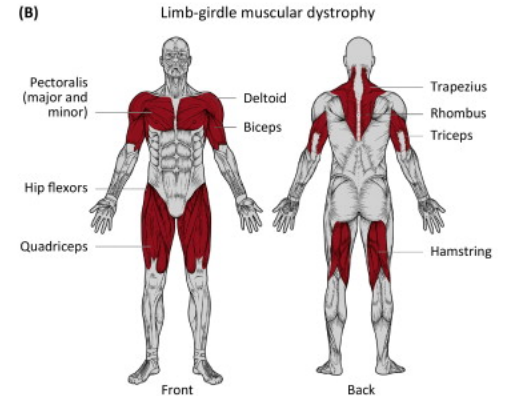
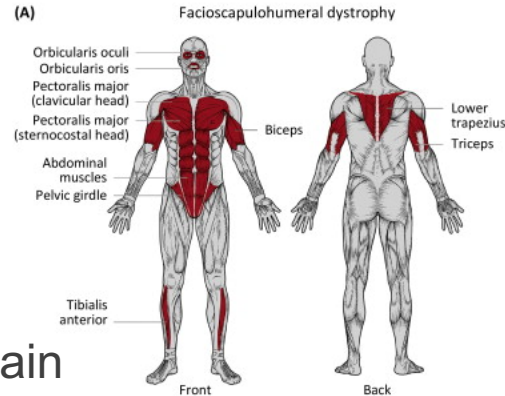
F – Facio - Face

S – Scapulo - Shoulder

H – Humeral - Arm

Clinical Presentation

- One of the most common muscular dystrophies (2 – 7 per 100,000)
- Approximately 21,000 in the USA
- Can be diagnosed at any age
- High degree of variability
- Asymmetry
- Two clinically identical forms
 - FSHD type 1: 95%
 - FSHD type 2: 5%
- Chronic shoulder and neck pain

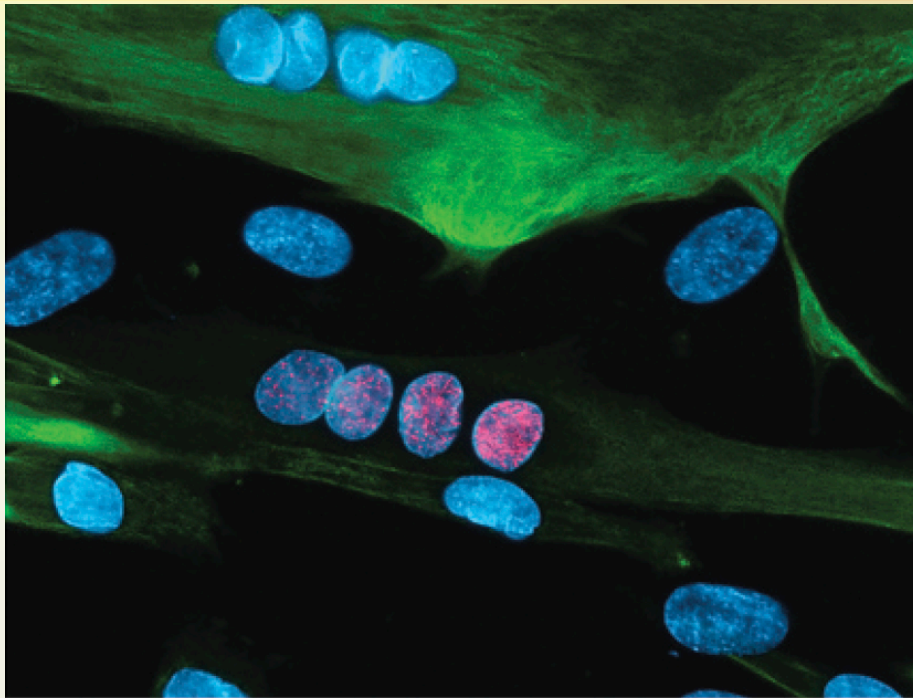


TRENDS in Molecular Medicine

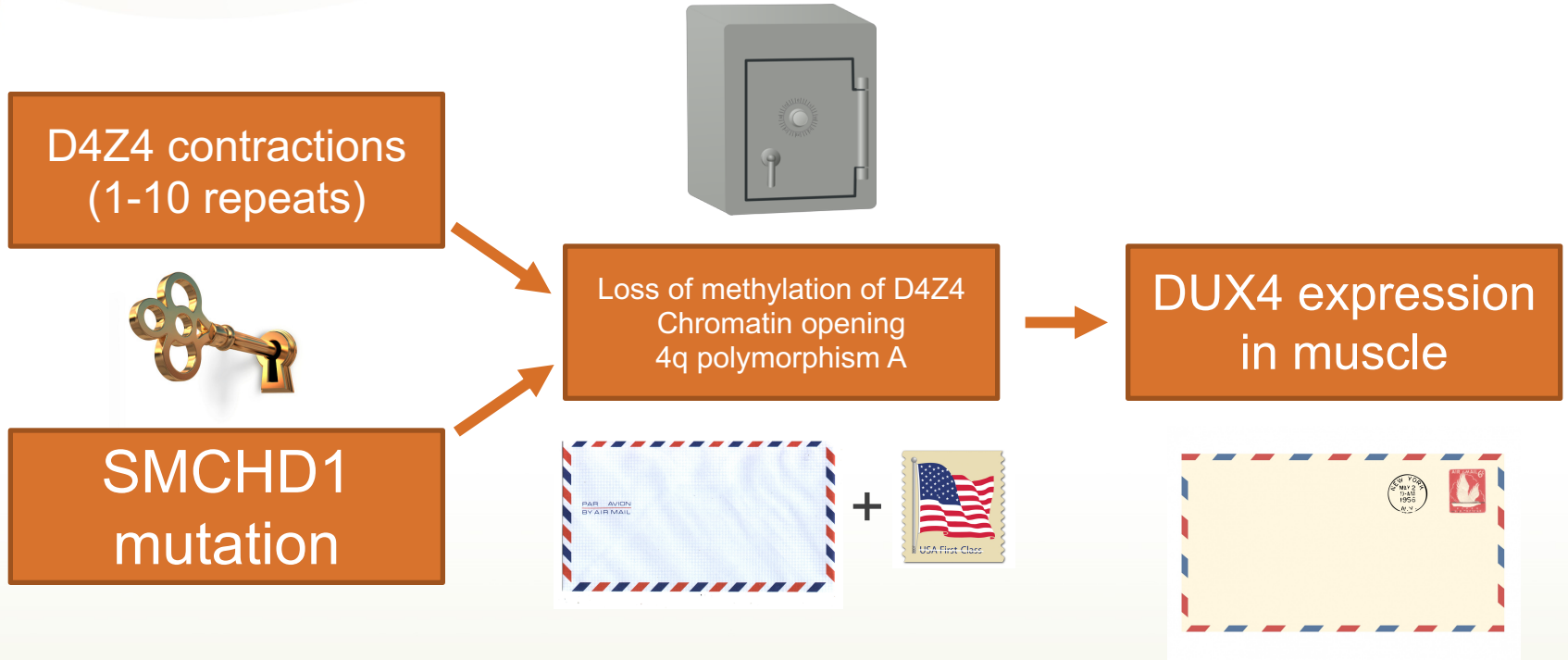
Biology of FSHD



Protein	Gene	Chromosome	Location
MyoD	12O1	2	12p13
Myf5	12O2	2	12p13
Myf6	12O3	2	12p13
Myf2	12O4	2	12p13
Myf3	12O5	2	12p13
Myf4	12O6	2	12p13
Myf1	12O7	2	12p13
Myf7	12O8	2	12p13
Myf8	12O9	2	12p13
Myf9	12O10	2	12p13
Myf10	12O11	2	12p13
Myf11	12O12	2	12p13
Myf12	12O13	2	12p13
Myf13	12O14	2	12p13
Myf14	12O15	2	12p13
Myf15	12O16	2	12p13
Myf16	12O17	2	12p13
Myf17	12O18	2	12p13
Myf18	12O19	2	12p13
Myf19	12O20	2	12p13
Myf20	12O21	2	12p13
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Myf28	12O29	2	12p13
Myf29	12O30	2	12p13
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Myf41	12O42	2	12p13
Myf42	12O43	2	12p13
Myf43	12O44	2	12p13
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Myf75	12O76	2	12p13
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Myf92	12O93	2	12p13
Myf93	12O94	2	12p13
Myf94	12O95	2	12p13
Myf95	12O96	2	12p13
Myf96	12O97	2	12p13
Myf97	12O98	2	12p13
Myf98	12O99	2	12p13
Myf99	12O100	2	12p13



DUX4 expression in adult muscles



What to monitor



Age	Sex	Height	Weight	BP	HR	Temp	SpO2	ECG	ECG
18	M	175	70	120/80	70	37.0	98	Normal	Normal
25	F	160	55	110/70	65	37.0	98	Normal	Normal
35	M	180	80	130/90	75	37.0	98	Normal	Normal
45	F	165	60	120/80	70	37.0	98	Normal	Normal
55	M	175	75	140/100	80	37.0	98	Normal	Normal
65	F	160	55	150/110	85	37.0	98	Normal	Normal
75	M	170	70	160/120	90	37.0	98	Normal	Normal
85	F	155	50	170/130	95	37.0	98	Normal	Normal
95	M	165	60	180/140	100	37.0	98	Normal	Normal





What to monitor

- Respiratory involvement: Present in <10% of patients
 - Pulmonary function testing and sleep studies
- Cardiac involvement: <5% of patients
 - Mostly asymptomatic arrhythmia. ECG
- Retina disease (Coats syndrome): <1% of patients
 - Vascular changes leading to retinal detachment and vision loss. Annual ophthalmology evaluation
- High frequency hearing loss: approx. 16% of patients
 - Mild and usually asymptomatic. Audiometry.
- These complications are usually present only in FSHD type 1 patients with large deletions

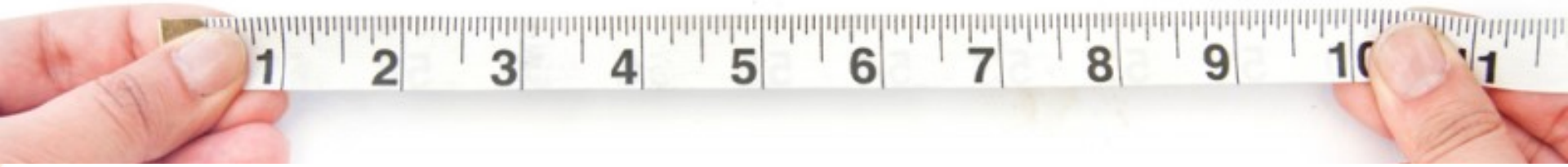
Genetics and Risk Estimation



chr	pos	ref	alt	freq
1	100000000	A	G	0.0001
1	100000001	T	C	0.0002
1	100000002	C	T	0.0003
1	100000003	G	A	0.0004
1	100000004	A	G	0.0005
1	100000005	T	C	0.0006
1	100000006	C	T	0.0007
1	100000007	G	A	0.0008
1	100000008	A	G	0.0009
1	100000009	T	C	0.0010
1	100000010	C	T	0.0011
1	100000011	G	A	0.0012
1	100000012	A	G	0.0013
1	100000013	T	C	0.0014
1	100000014	C	T	0.0015
1	100000015	G	A	0.0016
1	100000016	A	G	0.0017
1	100000017	T	C	0.0018
1	100000018	C	T	0.0019
1	100000019	G	A	0.0020
1	100000020	A	G	0.0021
1	100000021	T	C	0.0022
1	100000022	C	T	0.0023
1	100000023	G	A	0.0024
1	100000024	A	G	0.0025
1	100000025	T	C	0.0026
1	100000026	C	T	0.0027
1	100000027	G	A	0.0028
1	100000028	A	G	0.0029
1	100000029	T	C	0.0030
1	100000030	C	T	0.0031
1	100000031	G	A	0.0032
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1	100000033	T	C	0.0034
1	100000034	C	T	0.0035
1	100000035	G	A	0.0036
1	100000036	A	G	0.0037
1	100000037	T	C	0.0038
1	100000038	C	T	0.0039
1	100000039	G	A	0.0040
1	100000040	A	G	0.0041
1	100000041	T	C	0.0042
1	100000042	C	T	0.0043
1	100000043	G	A	0.0044
1	100000044	A	G	0.0045
1	100000045	T	C	0.0046
1	100000046	C	T	0.0047
1	100000047	G	A	0.0048
1	100000048	A	G	0.0049
1	100000049	T	C	0.0050
1	100000050	C	T	0.0051
1	100000051	G	A	0.0052
1	100000052	A	G	0.0053
1	100000053	T	C	0.0054
1	100000054	C	T	0.0055
1	100000055	G	A	0.0056
1	100000056	A	G	0.0057
1	100000057	T	C	0.0058
1	100000058	C	T	0.0059
1	100000059	G	A	0.0060
1	100000060	A	G	0.0061
1	100000061	T	C	0.0062
1	100000062	C	T	0.0063
1	100000063	G	A	0.0064
1	100000064	A	G	0.0065
1	100000065	T	C	0.0066
1	100000066	C	T	0.0067
1	100000067	G	A	0.0068
1	100000068	A	G	0.0069
1	100000069	T	C	0.0070
1	100000070	C	T	0.0071
1	100000071	G	A	0.0072
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1	100000082	C	T	0.0083
1	100000083	G	A	0.0084
1	100000084	A	G	0.0085
1	100000085	T	C	0.0086
1	100000086	C	T	0.0087
1	100000087	G	A	0.0088
1	100000088	A	G	0.0089
1	100000089	T	C	0.0090
1	100000090	C	T	0.0091
1	100000091	G	A	0.0092
1	100000092	A	G	0.0093
1	100000093	T	C	0.0094
1	100000094	C	T	0.0095
1	100000095	G	A	0.0096
1	100000096	A	G	0.0097
1	100000097	T	C	0.0098
1	100000098	C	T	0.0099
1	100000099	G	A	0.0100

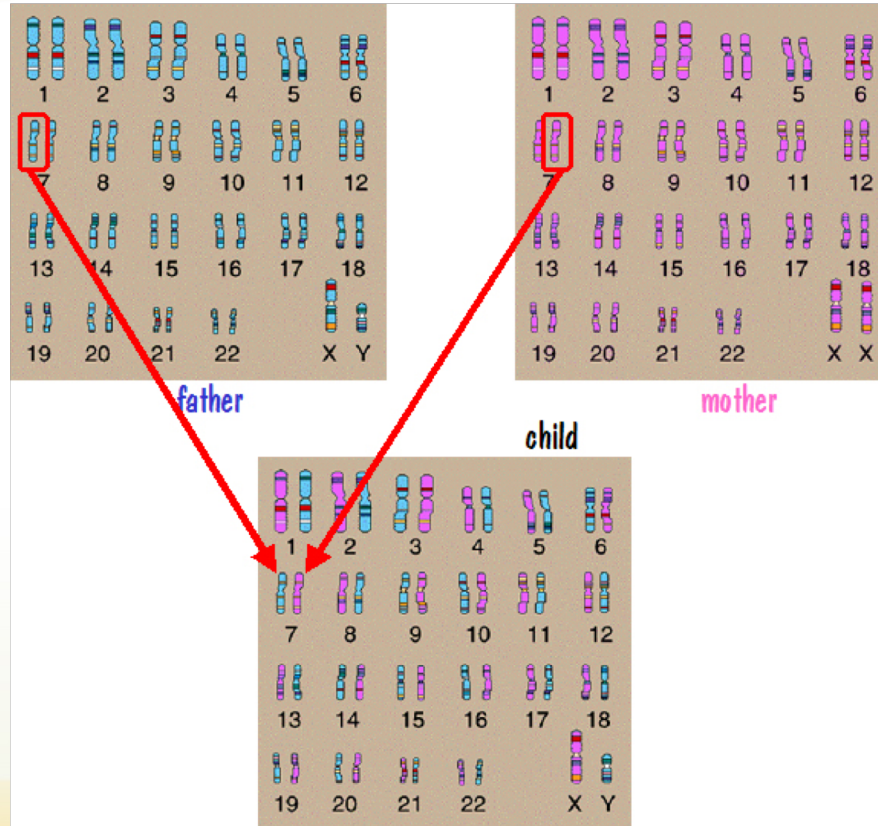


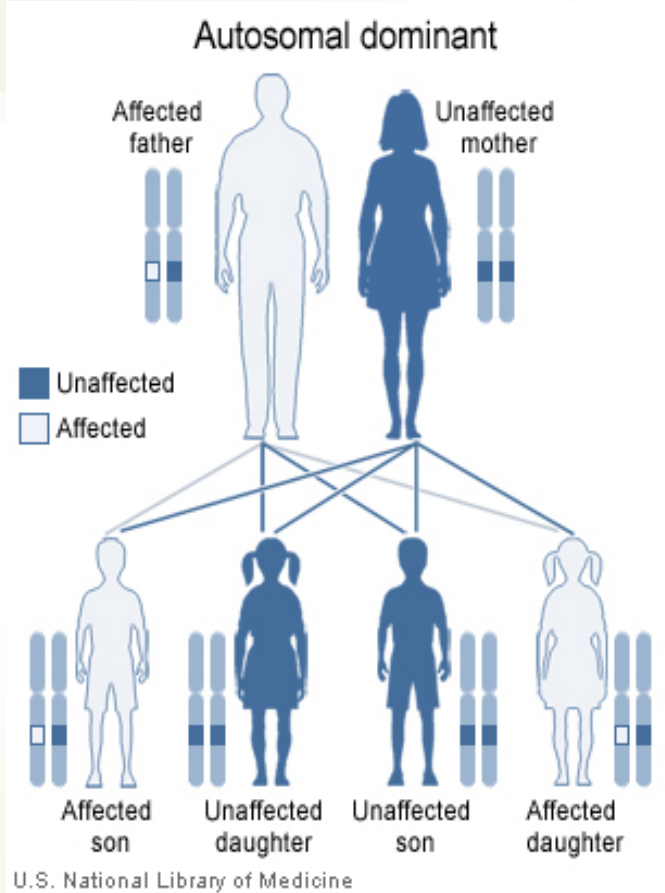
RISK



ESTIMATING RISK IN GENETIC CONDITIONS

HOW DO WE INHERIT OUR LIBRARY OF GENES?





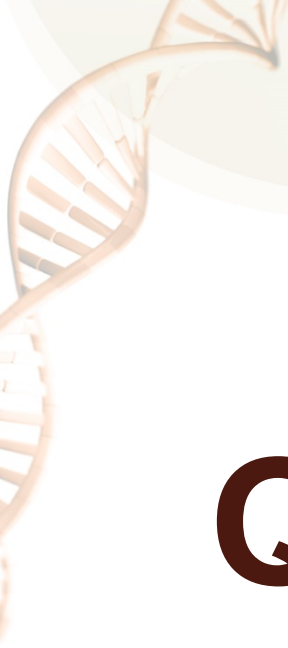
Autosomal Dominant

- FSHD type 1
- Mutation in one copy is enough to cause disease
- Risk for passing disease on is 50%
- Risk does not depend on gender
- Up to 30% of cases are new, spontaneous mutations



Guidelines for the treatment of FSHD

- Multidisciplinary care
 - Stretching and range of motion exercises
 - Assistive devices (braces, mobility)
- Respiratory evaluation
- Dilated eye exam
- Pain management
- Scapular fixation?



QUESTIONS?