LGMD1D DNAJB6 Foundation and Autosomal Dominant Registry
Poster Presentation
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Title: LGMD and Access to Genetic Diagnoses, A Novel Approach.

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Abstract: Limb girdle muscular dystrophies are among a subset of rare genetic disorders, usually caused by a monogenetic variant. There are many barriers to arriving at a genetic diagnosis including patient and physician knowledge and comfort with new technology, availability of sponsored testing, geographic availability of testing and referral to appropriate specialists upon genetic testing.

The foundation has been able to surmount these issues with the help of sponsored testing, social media, telemedicine and up to date medical education focused on the individual's journey.

The results are as follows:

Methods: From July 2020 to July 2021 we were able to get referrals from a variety of LGMD social media sites, foundations and national meetings (via ZOOM) for assistance in undiagnosed LGMD persons from around the US and Canada who were eligible for sponsored testing (the majority from underserved areas). In many cases our educational quarterly newsletter generated referrals.

Inclusion into the study: Patients' clinical features had to include confirmed neuromuscular signs: Gower's sign,or some degree of proximal weakness which was true in all cases. Neuropathic features were included for completeness in a differential diagnosis and further genetic testing where needed. Suspected mitochondrial disorders could not be tested due to lab limitations but were addressed with individuals as they arose.

The process: In conjunction with their local medical team, DNA kits for neuromuscular and in some cases neuropathic panels were mailed to persons and returned to a central lab.

The results: A total of 134 patients received genetic panel testing, 89 females and 45 males and the average age was 45 years (ages 4- 80). Ten patients did not return tests. Of the 124 who did 38 (28%) had a positive pathogenic diagnosis not known before . Fifty five (44%) had 1-8 variants of uncertain significance but the lab felt these variants were suspicious enough for additional family testing which was followed up upon as circumstances allowed for segregation analysis. Twenty nine patients (23%) had variants of uncertain significance for which the lab declined any further family testing. Two patients had entirely negative results. The average resulted test had 3 variants. Genetic counselling was available in all cases.

In conclusion, with access to expanded sponsored testing, social media and telemedicine capabilities, patients suspected of rare genetic conditions can acquire access to up to date genetic testing and appropriate referrals as our model demonstrates.

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