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AI model for facial recognition and FSHD diagnosis: The FaceNMD project

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Title:

AI model for facial recognition and FSHD diagnosis: The FaceNMD project

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Abstract

Background: Characteristic facial features can provide important clues for diagnosing neuromuscular disorders and identifying candidate genes. However, their recognition relies heavily on clinicians' expertise, making diagnosis challenging, particularly in rare diseases with heterogeneous presentations. Automated facial and voice analysis offers promising tools to support clinical assessment, improve diagnostic efficiency, and provide objective outcome measures. Despite recent advances, these approaches remain underexplored in neuromuscular disorders. Given the distinctive facial involvement in facioscapulohumeral muscular dystrophy (FSHD), we developed an AI-based model to distinguish patients with FSHD from healthy controls and individuals with other neuromuscular disorders using 2D and 3D facial video analysis.

Results: We enrolled 190 participants (60 with FSHD, 100 with other neuromuscular disorders, and 30 healthy controls), providing 380 video recordings of 14 standardized facial exercises. Due to technical issues, 284 videos were included in the analysis. Model performance was evaluated using the complete dataset and individual exercises. Among all exercises, exercise 14 ("blowing out the cheeks") achieved the highest accuracy (79.1%) in distinguishing FSHD patients from the entire comparison cohort, including healthy controls and individuals with other neuromuscular disorders. The complete dataset was processed in less than one minute. Conclusion: This study highlights the potential of deep learning-based facial movement analysis to distinguish FSHD from other neuromuscular disorders and healthy controls. These findings support the use of facial dynamics as quantifiable clinical features and provide a basis for developing automated AI-assisted tools to support FSHD diagnosis and clinical assessment.

Keywords: FSHD, Facioscapulohumeral dystrophy, neuromuscular disorders, facial recognition system analysis, rare diseases, deepl learning

BACKGROUND

Clinical and genetic diagnosis of neuromuscular diseases (NMDs) traditionally requires in-person evaluation at specialized clinics, involving time-consuming assessments conducted by multidisciplinary teams with expertise in rare diseases. This process is often costly and logistically challenging, particularly for patients living in rural environments or in underdeveloped countries, as well as for individuals with reduced mobility or loss of autonomy. Furthermore, patients with mild phenotypes or those in the early stages of disease

frequently avoid or delay this complex diagnostic pathway (the diagnostic odyssey) (1), and remain undiagnosed for a long time without access to appropriate care or interventions that could mitigate and prevent the evolution of disease burden.

Facial impairment can sometimes be very distinctive for a specific neuromuscular condition, and this can guide the search for a candidate gene (2). However, the recognition of such characteristic facial phenotypes relies heavily on the expertise of trained clinicians, and in the absence of such specialized knowledge, the diagnostic process remains challenging, particularly in the context of rare diseases with heterogeneous presentations (3).

Accurate identification of disease-specific facial features carries important clinical implications. It can streamline and accelerate the diagnostic process, reduce associated costs and delays, thanks to the earlier diagnosis. In turn, this enables timely initiation of appropriate management strategies, access to targeted therapies and clinical trials, and ultimately may contribute to improved patient outcomes and prognosis (2).

Automated computer-assisted facial and voice analysis is a rapidly evolving field that offers clinicians innovative tools to improve diagnostic efficiency while reducing patient burden and addressing unmet medical and patient needs. In particular, such technologies might support the clinician throughout the diagnostic process, by facilitating accurate disease identification and potentially providing outcome measures (4,5). Recent studies have demonstrated that automated computer-assisted 2D or 3D facial analysis can achieve variable effective performance in the diagnosis of rare and ultra-rare genetic syndromes including Cornelia de Lange, Angelman, Noonan, Emanuel and Pallister-Killian syndromes (5). In addition, their role as a potential tool for the detection of novel syndromes is emerging (6). These methodologies enable the extraction of quantitative features at both the individual and group levels, including craniofacial morphology (e.g., interocular distance, nasal contours, perioral

structures, auricular positioning, and chin morphology), facial dynamics, vocal biomarkers ("vocal imprint"), and speech or language characteristics (5,7). Despite these advances, such technologies are not yet widely implemented in routine clinical practice. Their adoption remains limited by concerns regarding reliability, including variability in image and audio acquisition, insufficient clinical validation, and challenges related to integration into clinical workflows (8). In addition, ethical and legal considerations, particularly those related to data privacy and consent, further complicate their clinical applicability.

Nevertheless, these approaches offer several important advantages. They can be non-contact and potentially used without specialized equipment, making them particularly suitable for remote diagnosis and longitudinal follow-up in digital exchanges with patients. This opens the possibility for more patient-centered and decentralized models of care (9).

In neuromuscular diseases, the involvement of facial and bulbar muscles is frequently an early and prominent feature (10), providing valuable diagnostic clues for experienced clinicians. However, the recognition of disease-specific facial and vocal patterns varies considerably depending on disease severity and is highly dependent on clinician expertise and the assessment protocol used. This subjectivity leads to substantial variability in clinical evaluation, limiting reproducibility across examiners, protocols, and specialized centers. These limitations are especially problematic in patients with mild symptoms, early-stage disease, slowly progressive conditions, or significant intra-familial phenotypic variability. However, despite the relatively high prevalence of facial involvement in NMDs, the application of automated facial analysis in this field remains underexplored (11).

In the present study, we aimed to develop and evaluate an algorithm based on a combined analysis of static and dynamic facial features, with the objective of achieving high sensitivity to identify facial patterns suggestive for facioscapulohumeral muscular dystrophy (FSHD) and

discriminate them from those observed in other neuromuscular diseases and healthy individuals. Facial involvement is one of the most characteristic and recognizable features of FSHD, often appearing early in the disease course (12). However, in patients with mild or atypical presentations, or in early disease stages, distinguishing subtle facial weakness from normal inter-individual variability, particularly in aging populations, can be challenging. Moreover, these features are often more apparent during dynamic facial movements, such as eye closure, smiling, blowing, lip pursing, or speech, rather than in static images.

FSHD is also characterized by marked clinical heterogeneity in terms of age at onset, disease progression, and severity (13). Its genetic diagnosis remains complex and time-consuming, often relying on techniques such as Southern blot analysis (14), which may limit access to testing, particularly in resource-limited settings or in rural environments. In this context, improved phenotypic screening tools could facilitate earlier identification and prioritization of patients for genetic testing.

Given the nature of facial involvement in FSHD, static imaging alone is often insufficient to capture subtle manifestations of facial weakness. In contrast, a combined approach integrating both static and dynamic computer-assisted facial analysis may provide greater sensitivity (15), enabling the detection of mild phenotypes and the monitoring of small but clinically meaningful changes over shorter time intervals. Here we present the results of our AI model for detecting FSHD from healthy controls or subjects with other neuromuscular disorders using 2D and 3D video analyses of facial features.

METHODS

Participants

We enrolled subjects aged 18 years or older between 02/09/2020 and 28/08/2024. The study population included individuals with genetically confirmed FSHD1 or 2, presenting with moderate to severe facial involvement. Facial weakness was defined by the involvement of at least two facial muscles, including *orbicularis oculi* and *orbicularis oris*, evaluated with manual muscle testing (MMT). Moderate facial weakness was defined by MMT > 3, whereas severe facial weakness was defined by MMT values ≤ 3 on at least two muscles.

We also included patients with facial involvement due to other neuromuscular disorders, whether of genetic or acquired origin. An age- and sex-matched healthy control group without facial involvement was also enrolled.

Patients affected by other diseases, that could interfere with the assessment of facial function, such as a history of facial trauma or congenital/acquired facial dysmorphism, were excluded.

All participants underwent a standardized neurological examination at enrollment, including MMT assessment. Demographic, genetic, and clinical data were systematically collected for each participant.

Videos and clinical assessments were repeated also at 1 year follow-up in order to evaluate the efficacy of the algorithm in detecting worsening of facial involvement (analyses still in ongoing, not reported in the article).

Clinical protocol and video acquisition

Video data acquisition was performed using a camera system capable of recording both 2D and 3D videos.

During the recording session, participants were comfortably seated in front of a computer in a quiet and silent room (Figure 1). The computer displayed step-by-step instructions and a

short video of the standardized task execution, in order to assist the participants during the registration.

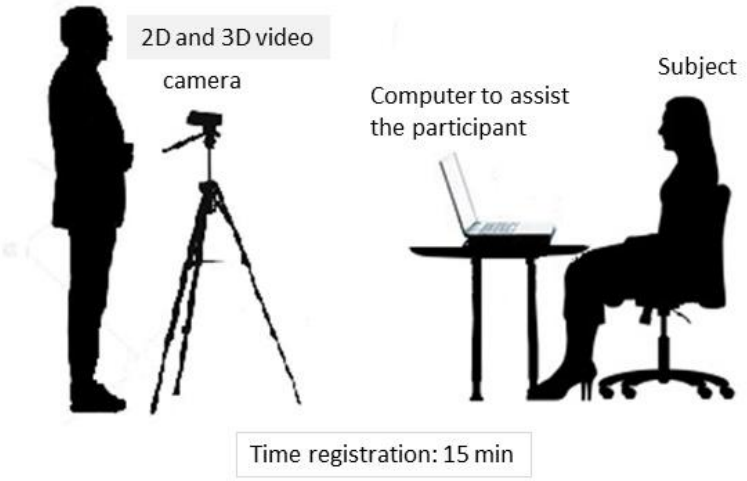

Figure 1: Schematic representation of modalities of video recording

Facial movements and involvement were evaluated using 15 exercises. Participants were instructed to perform the following tasks: [E1] Close your eyes as hard as possible (10s), [E2] Open your eyes as wide as possible (10s), [E3] Look up (30s), [E4] Look down (30s), [E5] Look to the right (30s), [E6] Look to the left (30s), [E7] Look back and forth from right to left (20s), [E8] Look back and forth from bottom to top (20s), [E9] Nod from left to right (10s), [E10] Nod up and down (10s), [E11] Open your mouth as wide as possible (10s), [E12] Show teeth (10s), [E13] Smile big (10s), [E14] Blow out cheeks (10s), [E15] Mimic a kiss (10s).

Each task was performed for a predefined duration. Rest intervals of approximately 20–30 seconds were provided between exercises to minimize fatigue and maintain participant attention throughout the session. The total duration of the recording protocol was approximately 15 minutes.

The model architecture / computational analysis

Pre-training and pre-processing

A subset of our dataset was used to train the algorithm. However, considering the relatively limited size of our study cohort, a pre-training phase was also realized in order to enhance our model's ability to recognize and detect facial features, especially dynamic motion descriptors. For this purpose, the pre-training was made on VoxCeleb, a large-scale facial video dataset available online, composed by millions of videos of human face (16,17). This pre-training allowed the model to recognize facial movement patterns, including micro-facial contractions, asymmetries, intensity of facial expression, eyes-face coordination etc.

Secondly, all videos underwent a standardized pre-processing phase designed to adapt each video to our algorithm, optimize feature extraction and reduce noise and variability. The pre-processing phase consisted of three main steps: (1) reducing the length and size of the video, (2) face cropping, and (3) face alignment.

1. Each video recording, of approximately 15 minutes in duration, was segmented into 15 videos of several seconds. Each video corresponded to the recording of a specific facial exercise. For this procedure, we used the ELAN annotation tool (18).

2. Face cropping was applied to isolate the facial region during the registration and minimize background interference. This technique helped to reduce the influence of background noise, limiting the presence of distractors, and facilitate the model's identification of facial features (19). All videos were also resized to a standardized number of pixels (224x224) to ensure consistency across registrations. Face cropping was performed with MTCNN (Multi-task Cascaded Convolutional Networks)(20) and ELAN annotation tool (18).

3. Face alignment was also realized to standardize the spatial positioning of key facial landmarks across frames. Specifically, alignment was based on the relative positions of the eyes, mouth, and chin, thereby correcting for head pose variations and ensuring consistency in feature extraction.

Finally, the dataset was divided into training and testing sets using a stratified splitting approach, ensuring that the proportion of participants with facioscapulohumeral muscular dystrophy (FSHD) and controls was preserved across subsets (21).

Face detection and feature extraction

The overall architecture of the proposed deep learning framework, from video preprocessing to feature extraction and multi-task classification, is illustrated in Figure 2.

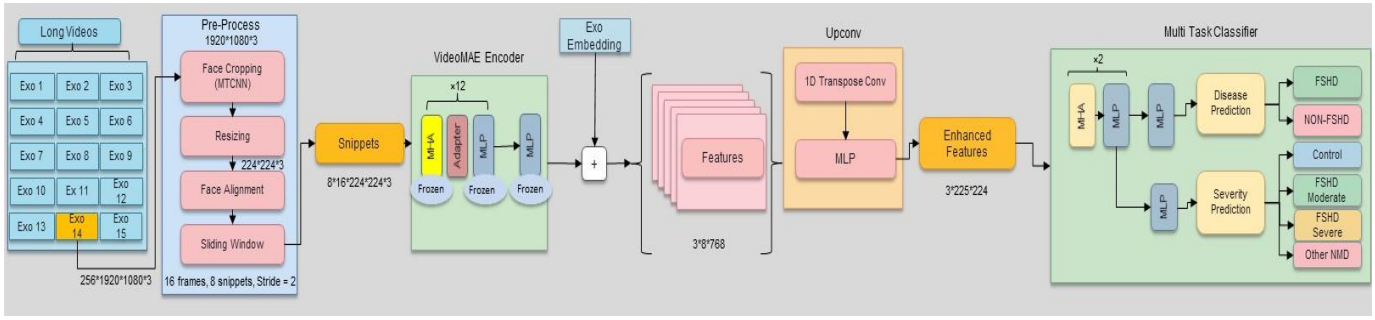

Figure 2: Overview of the deep learning architecture for automated facial movement analysis and FSHD classification.

For feature detection and extraction, we used VideoMAE software (22), a transformer-based encoder-decoder architecture. This model converts each video into numeric descriptors capturing salient spatial and temporal characteristics of facial movements. By at first masking un-necessary pixels, the model is encouraged to focus on identifying the most informative features/descriptors. Then the decoder uses these descriptors to reconstruct a new video.

Thanks to this process, a loss is generated that helps the encoder learn the best descriptors to extract from the video.

To further enhance temporal representation and improve classification performance, additional adapters were integrated into the architecture. Specifically, adapters, as upconv modules, were incorporated to enhance temporal feature representations and refine motion descriptors. These components facilitate the capture of subtle temporal variations in facial activity.

Finally, a multi-task classification was implemented to increase the model's discriminative capacity. This component enables simultaneous feature extraction and classification, allowing the model to categorize first in a binary-way participants into FSHD or not and then to classified them into moderate and severe FSHD, healthy control (HC), or other NMD groups.

Ethics

The study was submitted to the Delegation for the Protection of Persons of the Nice University Hospital (CHU de Nice), which granted authorization for its conduct. The study was registered at ClinicalTrials.gov (NCT04377217) on May 6, 2020 (first posted).

RESULTS

Patients

We enrolled 190 subjects in the study. Sixty of them were diagnosed with FSHD, 100 with other NMDs, and 30 healthy controls (HC).

The FSHD group was divided into two subgroups based on MMT evaluation of facial muscles: 40 were classified as moderate and 20 as severe FSHD (MMT mean of 15.3 ± 2.8 and 10.3 ± 2.6 with $p<0.0001$, respectively).

Patients with moderate FSHD consisted of 15 females, with a mean age at enrolment of 57.5 ± 13.6 years, and a disease duration of 12.5 ± 13.4 years. Similarly, patients with severe FSHD consisted of 11 females, displayed a mean age at enrolment of 55.1 ± 17 years and a disease duration of 19.5 ± 10.5 years, with evidence of a statistical difference for disease duration with the moderate group ($p=0.04$). Subjects belonging to the FSHD moderate group carried a mean D4Z4 length of 7.1 ± 2.1 RU, whereas a mean of 5.2 ± 1.8 RU was found in the severe FSHD group ($p<0.001$).

The group of subjects with other NMDs consisted of 100 patients of both acquired and genetic origin. Among them, the largest subgroups were composed by patients with myotonic dystrophy type 1 (DM1) (44%), myasthenia gravis (21%), and mitochondrial myopathy (13%). A minor proportion of patients was diagnosed with other neuromuscular disorders as Charcot-Marie-Tooth disease (CMT), Oculopharyngeal muscular dystrophy (OPMD), and congenital myopathy. A complete list of all diagnostic subgroups is displayed in Table 1.

Subjects with other NMD	N
Myotonic Dystrophy Type 1 (DM1)	44
Myasthenia Gravis	21
Mitochondrial myopathy	13
Congenital myopathy	4
Charcot-Marie-Tooth disease (CMT)	4
Chronic Inflammatory Demyelinating Polyneuropathy (CIDP)	3
Oculopharyngeal muscular dystrophy (OPMD)	2
Idiopathic Inflammatory Myositis (IIM)	1
Pompe Disease	1
Motoneuron Disease (MND)	1
Myotonic Dystrophy Type 2 (DM2)	1
Duchenne muscular dystrophy (DMD)	1
Myopathy without molecular definition	4
TOTAL	100

Table 1. Composition of the Other NMD group by diagnostic subtype

Subjects diagnosed with other NMD displayed a mean age at enrolment of 54.46 ± 17.8 years and a disease duration of 13.8 ± 13 years. No differences were found in age at enrolment and disease duration when compared to FSHD group. A statistical difference was found regarding the severity of facial involvement between subjects with other NMDs and severe FSHD group for greater involvement in the FSHD group (10.3 ± 2.6 vs 15.3 ± 3.4, $p<0.0001$). Detailed demographic and clinical data of subjects are reported in Table 2.

	moderate FSHD	**severe FSHD**	**Other NMD**	**HC**	**p-values**
Sex (F)	15 (38%)	11 (55%)	56 (56%)	18 (60%)	
Age at enrollment (y)	57.5 ± 13.6	55.2 ± 17.0	54.5 ± 17.8	48.9 ± 16.5	0.57 (moderate vs severe) 0.33 (moderate vs other NMD) 0.87 (severe vs other NMD)
Disease duration (y)	12.5 ± 13.4	19.5 ± 10.5	13.8 ± 12.9	N/A	0.04 (moderate vs severe) 0.6 (moderate vs other NMD) 0.06 (severe vs other NMD)
Facial MMT score	15.3 ± 2.8	10.3 ± 2.6	15.3 ± 3.4	20 ± 0	<0.01 (moderate vs severe) 0.9 (moderate vs other NMD) <0.01 (severe vs other NMD)
D4Z4 RU	7.1 ± 2.1	5.2 ± 1.8	N/A	N/A	0.01 (moderate vs severe)

Table 2. Demographic and clinical characteristics of study participants. For countable variables, mean and standard deviation is displayed.

Videos

Our dataset was initially composed of 380 video recordings. However, due to technical and/or registration problems, as reduced quality of recordings, 96 videos were excluded from the analysis, resulting in a final dataset of 284 long videos derived from 113 subjects.

Considering the diagnostic group classification, the final dataset was composed of 126 videos from FSHD patients, 92 videos from patients with other NMDs and 66 videos from HC subjects.

Within the FSHD group, this stratification into moderate and severe FSHD resulted in 82 and 44 video recordings, respectively.

In order to train the algorithm on the recognition of FSHD patients, 70% of the videos were used for training the algorithm, whereas the remaining 30% was used for testing (Table 3).

Classes	Training test	Testing set
Moderate FSHD	58	24
Severe FSHD	30	14
Other NMD	64	28
HC	46	20

Table 3. Subdivision of videos into training and testing set

The model

The model was tested both on the complete dataset and on each single exercise. Among all the exercises used for discriminating FSHD from healthy controls and patients with other neuromuscular disorders, the exercise 14 (“blow out cheeks”) achieved the highest discriminative performance, reaching an accuracy of 79.1%.

When all the exercises were considered, the overall accuracy of our model in discriminating FSHD reduced to 72%.

Notably, the model performed the complete analysis of the dataset in less than 1 minute.

In addition, we also compared the accuracy of our proposed algorithm with alternative architectures composed of part of our model or different pre-training datasets. Across all possible configurations, our model gained better accuracy among all of them (Table 4).

The model showed a sensitivity and specificity in detecting FSHD patients of 63.2 % and 79.2%, respectively.

Architectures	Pre-train Dataset	Accuracy
I3D	Kinetics	48.8%
VideoMAE	Kinetics	55.8%
VideoMAE	VoxCeleb	74.4%
VideoMAE + Upconv + Multi-Task	VoxCeleb	76.7%
OURS (VideoMAE + Adapter + Upconv + Multi-task)	VoxCeleb	79.1%

Table 4. Comparison of accuracy of different model configurations and architectures

DISCUSSION

In this study, we developed a novel dataset, composed of 142 unique patients performing 15 facial exercises resulting in 284 high-quality video recordings. In addition to video recordings, we also collected clinical data of NMDs, creating a unique database of subjects affected by neuromuscular diseases.

The primary aim of the study was to evaluate the ability of the algorithm to distinguish individuals with FSHD from healthy controls and, more importantly, from patients with other neuromuscular diseases. This latter comparison is particularly relevant from a clinical perspective, as a useful screening or diagnostic-support tool should be able to identify the characteristic facial phenotype of FSHD in the presence of other neuromuscular disorders rather than simply distinguish pathological from healthy facial patterns. As a secondary objective, we explored whether the model could discriminate different degrees of facial

involvement, thereby assessing its potential as a quantitative digital marker of disease severity. Despite the rapid expansion of artificial intelligence applications in facial analysis, its application to neuromuscular disorders remains limited. In particular, few studies have addressed facial phenotyping and analyses in rare neuromuscular diseases using automated approaches (4). In this context, FSHD represents a particularly suitable model because facial weakness is a characteristic feature of the disease and may be evident early in its clinical course. However, the recognition of subtle facial abnormalities remains highly dependent on clinical expertise, particularly in patients with mild, atypical, or early-stage disease.

One of the main findings of our study was that the highest classification performance was obtained using exercise 14, consisting of blowing out the cheeks. When this task was considered alone, the algorithm achieved an accuracy of approximately 80% in distinguishing FSHD from the other study groups. This finding suggests that specific facial motor tasks may contain substantially more disease-discriminative information than others. The performance of the cheek-blowing task may be related to the characteristic involvement of perioral and buccal musculature in FSHD, including weakness and asymmetry affecting movements of the mouth (11). Dynamic tasks may therefore accentuate abnormalities that are less apparent during resting facial examination or static image analysis. However, the biological mechanisms underlying the higher discriminative performance of this specific task remain to be established.

Interestingly, model performance was lower when all facial exercises were considered together in the comparison between FSHD and other neuromuscular diseases. This finding may reflect the heterogeneity of facial involvement across neuromuscular disorders. In our cohort, patients with other neuromuscular diseases were predominantly affected by disorders such as myotonic dystrophy type 1, myasthenia gravis, and mitochondrial

myopathies, each characterized by distinct patterns of facial and especially ocular involvement. Therefore, the ability of individual facial tasks to discriminate FSHD may depend on the specific neuromuscular disorders represented in the comparison group. Further studies including larger and more phenotypically diverse cohorts will be important to determine whether the observed performance is maintained across the broader spectrum of neuromuscular diseases encountered in clinical practice.

This work advances the field of automated face phenotyping, in the neuromuscular field, by demonstrating that clinically meaningful information can be extracted from standardized facial movements using deep learning approaches. Importantly, this approach supports the broader goal of enabling earlier, more objective, and more scalable diagnostic pathways, as well as facilitating longitudinal disease monitoring.

Indeed, a significant proportion of repeat-related NMDs remains undiagnosed especially in underdeveloped countries or isolated rural environments (23). This is largely due to limited access to specialized genetic testing, variability in clinical expertise, and the high cost and logistical constraints associated with molecular diagnostics. These patients lose the opportunity to be diagnosed and generate occasional expensive critical care in hospital, contributing to a substantial healthcare burden.

Earlier and more accessible diagnosis has the potential to significantly reduce the social, economic, and clinical burden of these diseases by enabling timely therapeutic interventions, appropriate surveillance of complications, and improved care planning (24). In this context, automated computer-assisted facial analysis represents a promising complementary tool that may help bridge diagnostic gaps in selected rare diseases, including neuromuscular disorders (4). Such systems offer the advantages of scalability, low cost, minimal training requirements, and compatibility with widely available devices (25). In our study, the full analysis of all facial

exercises required less than one minute per subject, demonstrating the feasibility of a rapid and potentially usable clinical tool suitable for both diagnostic support and remote patient monitoring.

From a broader perspective, the diagnostic algorithm can be used for a remote assessment of FSHD patient during follow up and, if confirmed accuracy in the longitudinal assessments, it can be potentially used for detecting subtle changes of facial involvement during time.

The standardized nature of facial video acquisition also raises the possibility of longitudinal applications. Repeated assessments could potentially be used to quantify changes in facial motor performance over time and complement established clinical outcome measures. Longitudinal evaluations are still ongoing, and further analyses are required to determine whether the proposed approach is sufficiently sensitive to detect clinically meaningful changes in disease severity or progression.

CONCLUSION

In conclusion, this study demonstrates the potential of deep learning approaches to distinguish FSHD from other neuromuscular diseases and from healthy controls using standardized facial movement analysis. Among the evaluated exercises, the “blow out cheeks” task showed the highest discriminative power in separating FSHD from other groups, suggesting that specific facial motor tasks may serve as sensitive disease-specific biomarkers (26). Overall, these findings support the utility of facial dynamics as quantifiable clinical features and contribute to the development of automated, AI-assisted supportive diagnostic tools for FSHD and related neuromuscular disorders.

However, these results should be interpreted in light of some limitations. Our cohort was characterized by a marked imbalance between the small healthy control group (n = 30) and

the larger groups of patients with FSHD (n = 60) and other neuromuscular disorders (n = 100), which may have influenced model performance and generalizability; validation in larger, balanced cohorts is therefore warranted.

Finally, this tool should be regarded as complementary to, rather than a replacement for, established clinical methods: it is intended to support clinicians in the diagnostic workup, for instance, by facilitating earlier identification of patients who may benefit from genetic testing, while routine diagnosis and care remain anchored in clinical evaluation and molecular confirmation, whose role in supporting clinical assessment is well established.

LIST OF ABBREVIATIONS

2D: Two-dimensional

3D: Three-dimensional

AI: Artificial intelligence

CMT: Charcot–Marie–Tooth disease

CNRS: French National Centre for Scientific Research

DM1: Myotonic dystrophy type 1

ELAN: EUDICO Linguistic Annotator

FSHD: Facioscapulohumeral muscular dystrophy

HC: Healthy controls

NSERM: French National Institute of Health and Medical Research

MMT: Manual muscle testing

MTCNN: Multi-task cascaded convolutional neural network

NMD: Neuromuscular disease

NMDs: Neuromuscular diseases

OPMD: Oculopharyngeal muscular dystrophy

RU: Repeat units

DECLARATIONS

Ethics approval and consent to participate

The study was approved by the Sud Méditerranée II Committee for the Protection of Persons (Comité de Protection des Personnes Sud Méditerranée II) on June 7, 2019. The Nice University Hospital (CHU de Nice) was authorized to implement data processing for a study investigating computer-based analysis of facial movements for the automated diagnosis of facioscapulohumeral muscular dystrophy (FSHD), under authorization decision DR-2020-069 (authorization application No. 919314). All the participants signed an informed consent form prior any study related procedures. The study was conducted in accordance with the principles of the Declaration of Helsinki.

Consent for publication

Not applicable

Availability of data and materials

Data available on request

Competing interests

SS has received consulting from UCB pharma, Alexion, argenx, Sanofi and research grant from UCB pharma. The other authors (GT, LV, JP, CS, AS, MC, AE, IB, AP and FB) have declared that no conflict of interest exists.

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Authors' contributions

Giulia Tammam, Luisa Villa, Jonathan Pini, and Abderhmane Slioui contributed to the initial drafting of the manuscript. Michele Cavalli, Luisa Villa, Ikram Bekkhoucha, and Angela Puma contributed to patient recruitment and data collection. François Bremond was responsible for video processing and methodology. Sabrina Sacconi critically reviewed and revised the manuscript. All authors reviewed and approved the final version of the manuscript

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