Applying Supervised classification to children with Fragile X Syndrome gait: introducing Dynamic Time Warping

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Summary

This study investigates the use of supervised machine learning algorithms to classify children with Fragile X Syndrome (FXS), the leading inherited cause of intellectual disability and autism spectrum disorder (ASD). Since children with FXS often show musculoskeletal disorders, gait analysis can be adopted to quantify their motor impairments. As gait analysis features have been successfully used to classify individuals with motor disturbances, this study aimed to apply Support Vector Machine as a supervised learning algorithm driven by Dynamic Time Warping features to classify children with FXS from healthy subjects. Additionally, the model was used to classify two different FXS phenotypes.

Introduction

Fragile X Syndrome (FXS) is the leading inherited cause of intellectual disability and autism spectrum disorder (ASD)[1] but it is often misdiagnosed due to a reduced referral to genetic screenings. In addition to neurodevelopmental and neuropsychiatric disorders, children with FXS often present musculoskeletal disorders with flat feet, ligamentous laxity, and hypotonia, warranting a referral for gait analysis (GA). Recent studies involving kinematics and surface electromyographic (sEMG) of children with FXS have identified significant differences compared to typically developing children (healthy subjects, HS). Thus, this study investigates the possibility to classify FXS children using sEMG signals and by applying Dynamic Time Warping (DTW) [3]. DTW-features extracted from sEMG signals were then used in machine learning (ML) models to distinguish FXS from HS and to further differentiate between the two major FXS phenotypes: FXS with only Full Mutation (FXSFull) and FXS with Full Mutation and somatic mosaicism (FXSMos).

Methods

This study included 35 children with FXSFull (BMI: 19.1 ± 6.2 kg/m²; age: 10.2 ± 3.6 years), 24 with FXSMos (BMI: 17.15 ± 7.6 kg/m²; age: 10.1 ± 2.9 years), and 14 HS (BMI: 19 ± 3.19 kg/m²; age: 9.3 ± 2.4 years). GA was performed at a self-selected walking speed using four synchronized GoPro Hero7 cameras (60 fps) and a Cometa sEMG system (2000 Hz, 8 channels) to record muscle activity bilaterally from Gastrocnemius Lateralis (GL), Tibialis Anterior (TA), Rectus Femoris (RF), and Biceps Femoris (BF). Six trials per subject were analysed, and muscle envelopes extracted following the method described in [2]. The mean value of the envelope from all HS trials served as a reference for the DTW algorithm. DTW distance (DTWdist) and the energy of the optimal path (DTWpath) were extracted. A Support Vector Machine (SVM) model with a radial basis function was trained with 5-fold cross validation, assigning subjects to training, test (Tset) or validation (Vset) sets. Model performance was assessed based on accuracy, precision, and recall. Three comparisons were analysed: HS vs. FXS; HS vs. FXSFull vs. FXSMos; FXSFull vs. FXSMos.

Results and Discussion

The classification accuracy for both Tset and Vset is shown in Figure 1, across all comparisons.

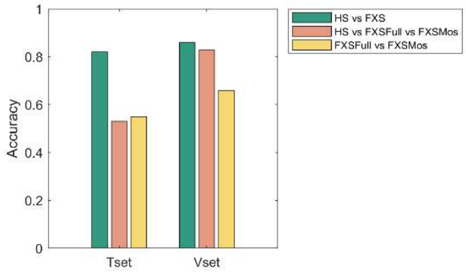

Figure 1: Accuracy of the SVM model for Tset (left) and Vset (right) for all the comparisons.

The SVM model demonstrated acceptable performance in distinguishing between HS and FXS subjects. However, classification between the two FXS phenotypes (FXSFull vs. FXSMos) yielded lower accuracy, indicating greater difficulty in differentiating FXS phenotypes.

Conclusions

These study findings suggest that DTW-related features extracted from sEMG can effectively classify FXS individuals from HS. However, to further improve classification accuracy, future research will explore alternative supervised ML models and integrate additional sEMG-derived features to enhance differentiation within FXS phenotypes.

Acknowledgments

In memory of Prof. Alessandra Murgia. This work was supported by FRAXA Foundation and PRIN project (2022, Prot. 20227JA8R3).

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