

## Evaluation of prevalence and predictor factors of respiratory impairment in a cohort of Italian Myotonic Dystrophy type 1 (DM1) patients.

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### Introduction

Respiratory failure due to restrictive syndrome either precipitated by pulmonary infections or following general anesthesia represents the main cause of death in DM1. Studies about the prevalence of restrictive syndrome in DM1 and the predictive role of DM1-related features have been performed on small cohorts, so there are no consensus guidelines on the management of respiratory problems for these patients. Aim of this study was to assess prevalence and any predictor of restrictive syndrome in DM1 analyzing a large cohort of patients, in order to stratify respiratory risk and eventually improve their follow-up.

	FVC<80% vs FVC>80%		Pearson χ <sup>2</sup>
	Mann-Whitney U-test	p	
Gender			0.908
Age	2856.5	0.124	
BMI	2229.5	0.310	
Smoking			0.589
N° of cigarettes	2579.0	0.571	
Onset	2370.500	0.981	
Congenital			0.270
Expansion n(CTG)	1768.0	<b>0.001</b>	
MIRS	1688.5	<b>0.000</b>	
Cardiopathy			0.263
Lung disease			0.757
Rest dyspnea			0.897
Exertion dyspnea			0.067
Sleepiness			0.693
Snoring			0.904
OSAS			<b>0.035</b>
Dysphagia			0.159
FEV1%	4568.0	<b>0.000</b>	
FEV1/FVC	2351.5	0.465	
TLC%	1875.0	<b>0.000</b>	
FVC%	6636.0	<b>0.000</b>	
VC%	4595.0	<b>0.000</b>	
MIP%	2880.5	<b>0.042</b>	
MEP%	2813.5	0.080	
NIV indication			<b>0.000</b>
NIV in use			<b>0.004</b>
Use of mexiletine	Not applicable		

Tab.1 Univariate analysis

### Methods

We designed a retrospective cross-sectional study on 164 adult DM1 patients in follow-up in one of the participating centers.

#### Inclusion criteria:

- Molecular diagnosis of DM1
- Performing at least one pulmonary function test (in case of more tests the last one was evaluated)

**Spirometric parameters analyzed:** FEV1, FVC, FEV1/FVC, TLC, VC, MIP and MEP.

**Parameters analyzed** are listed in table 1.

For statistical analysis patients were divided in 2 groups based on the presence or lack of pulmonary restriction (FVC<80% of predicted). Comparison between the two groups by univariate analysis (Tab. 1, Fig.1) was performed using Mann-Whitney U-test for numeric variables and Pearson's Chi-squared test for categorical variables. Also multivariate analysis was carried out (Fig.2). Significance level was set at p<0.05.

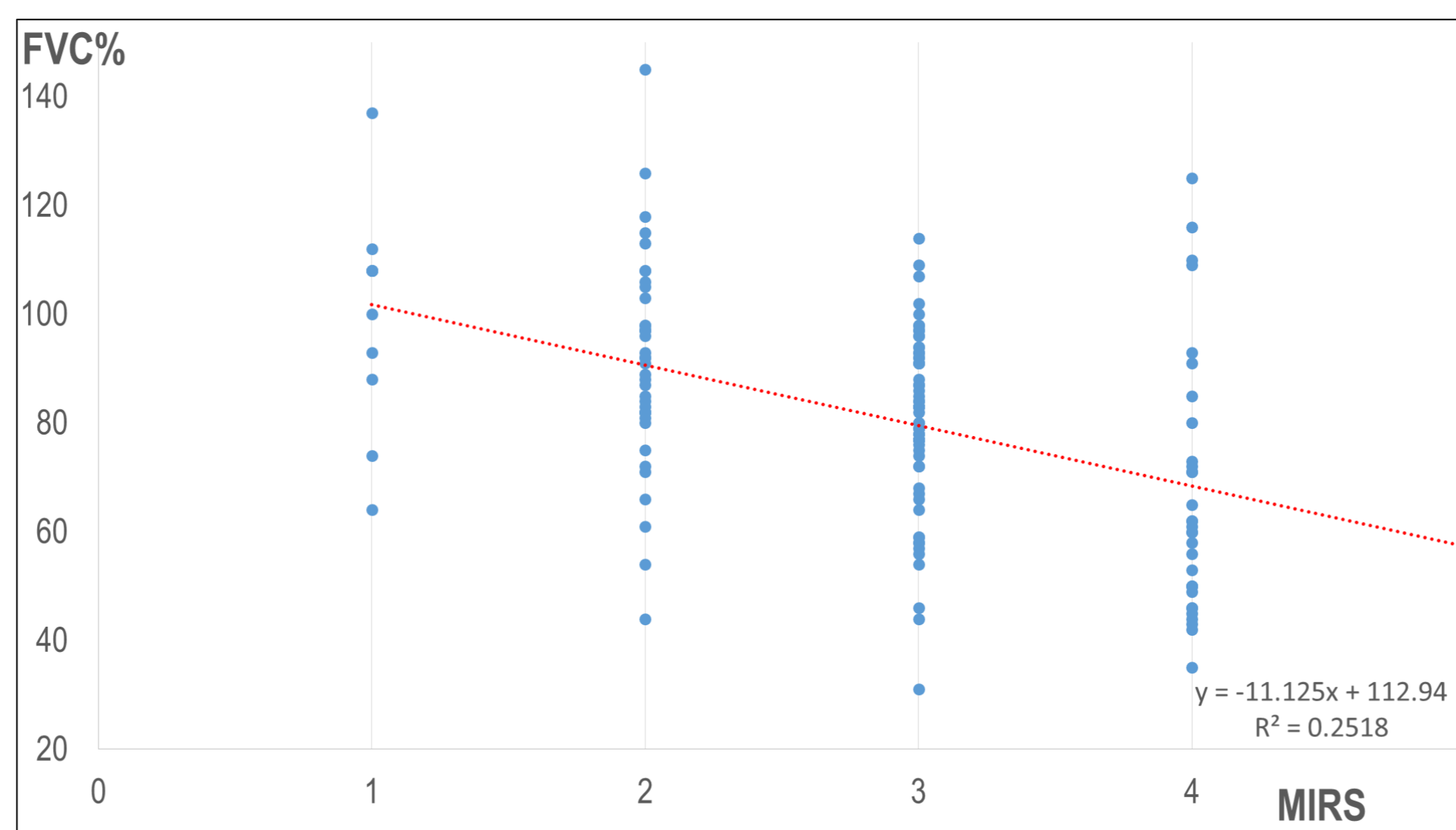


Fig.1 Linear regression between MIRS score and percentage of FVC

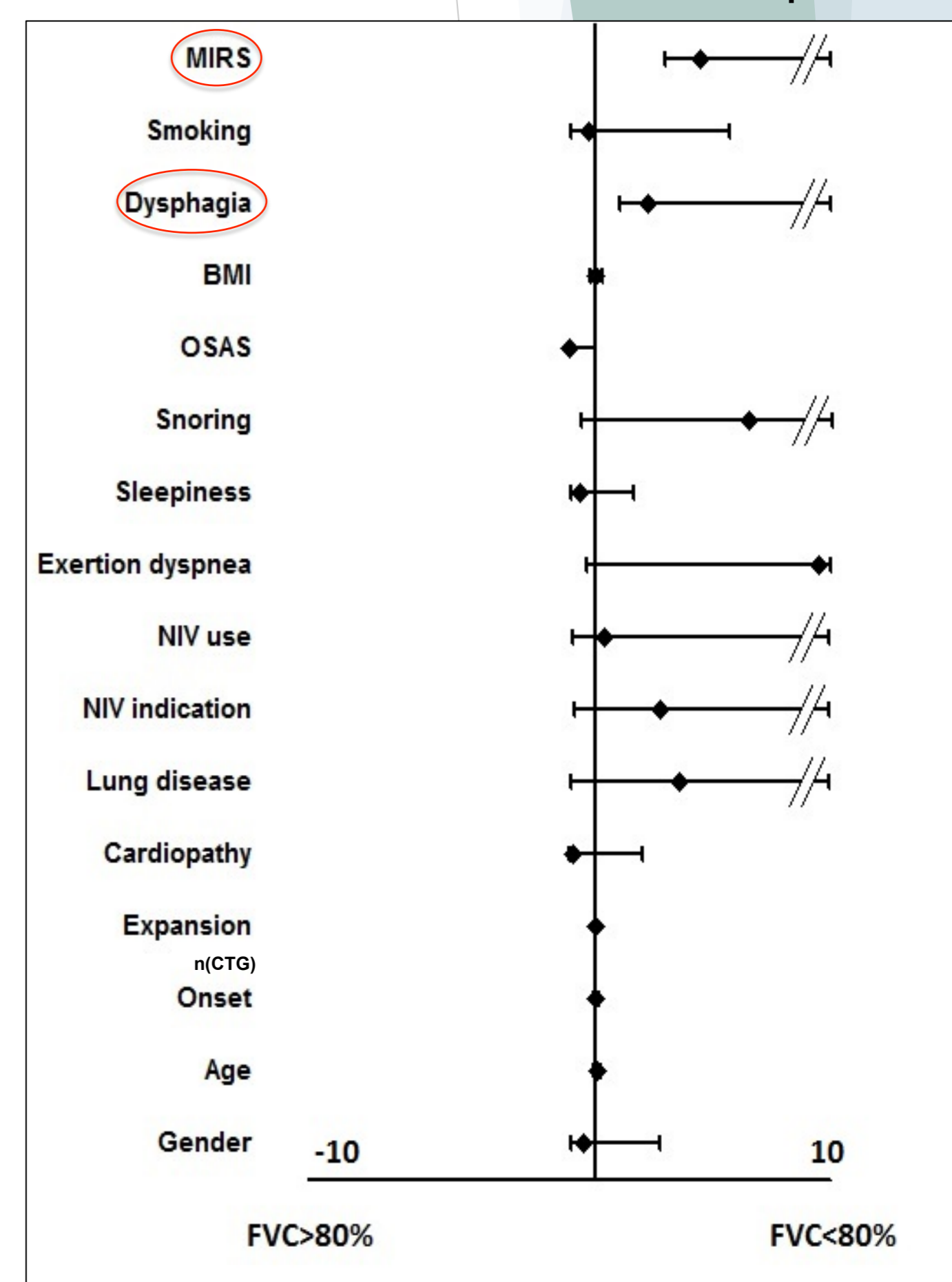


Fig.2 Forrest plot of the multivariate analysis

### Results

48.78% DM1 patients showed a restrictive syndrome (FVC<80% of predicted): of those about 50% had indication to NIV, but only half of them performed it adequately. All pulmonary measures of respiratory muscle involvement, n(CTG), MIRS, OSAS, indication and use of NIV were predictive of restrictive syndrome. In the multivariate model, only MIRS and dysphagia were independently associated with restrictive syndrome (p=0.003 and p=0.015 respectively).

### Conclusions

Our findings obtained on a large cohort of DM1 patients indicate, according to literature, that in DM1 MIRS severity and the n(CTG) are significantly associated with restrictive syndrome. Moreover, our data indicate that dysphagia and OSAS, but not BMI, are also significantly associated.

**Higher MIRS score** and **dysphagia** are the only independent predictors of restrictive syndrome. Therefore a strict respiratory follow-up should be recommended in DM1 patients showing these features.

Our study also confirms poor compliance to NIV being the major concern, likely responsible of the high frequency of respiratory-related mortality in DM1.

### References

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