

Genotype–phenotype associations in Alström syndrome: a systematic review and meta-analysis

Brais Bea-Mascato^{1,2}, Diana Valverde^{1,2*}

¹ CINBIO, Universidad de Vigo, 36310 Vigo, Spain.

² Grupo de Investigación en Enfermedades Raras y Medicina Pediátrica, Instituto de Investigación Sanitaria Galicia Sur (IIS Galicia Sur), SERGAS-UVIGO, Vigo, Spain

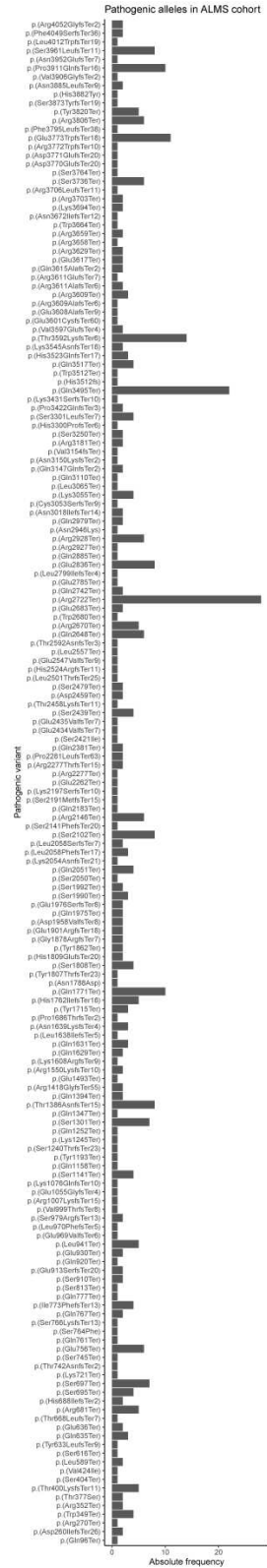
*Correspondence: Diana Valverde

CINBIO Facultad de Biología, Universidad de Vigo, Campus As Lagoas-Marcosende s/n,
36310 Vigo, Spain

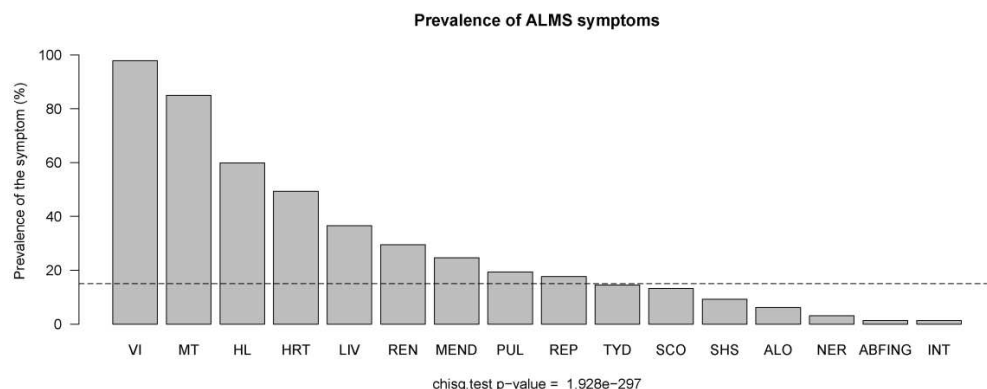
Tel +34 986 811 953

Email: dianaval@uvigo.es

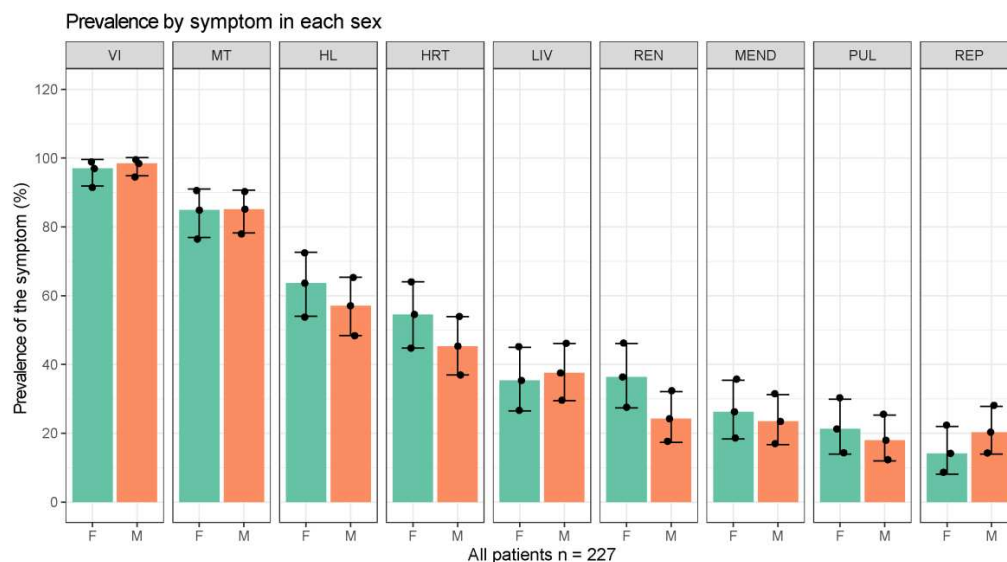
Supplementary Material:



Supplementary Figure S1. The 176 pathogenic variants and the number of alleles of each variant in the cohort.

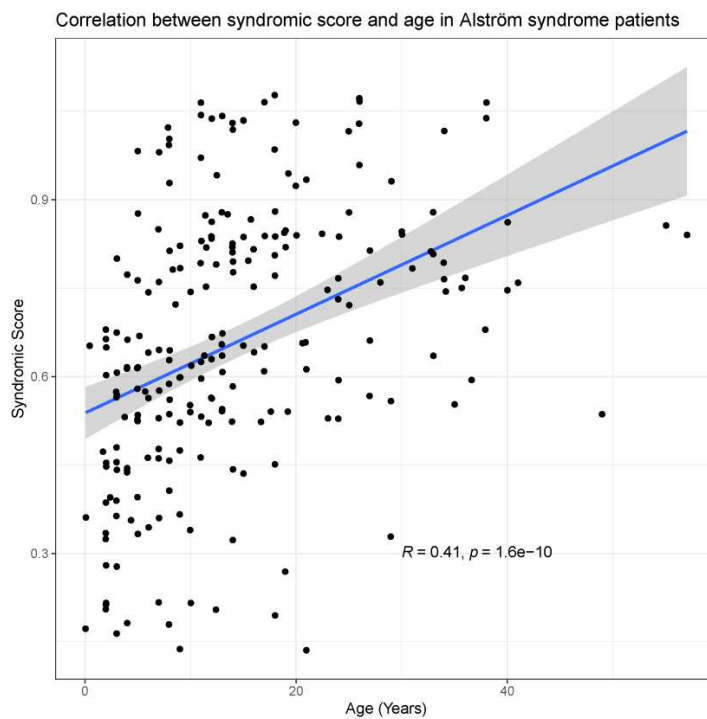


Supplementary Figure S2. Prevalence in the 16 syndromic groups initially collected from the literature and the established minimum prevalence threshold (15%; n=33) to be included in the study. **VI:** Vision impairments; **MT:** Metabolic anomalies; **HL:** Hearing anomalies; **HRT:** Heart anomalies; **LIV:** Liver anomalies; **REN:** Renal anomalies; **MEND:** Mental anomalies; **PUL:** Pulmonary anomalies; **REP:** Reproductive system anomalies; **TYD:** Thyroid metabolism anomalies; **SCO:** spine/feet anomalies; **SHS:** Stature anomalies; **ALO:** Alopecia; **NER:** Nervous system anomalies; **ABFING:** Finger anomalies; **INT:** Intestinal anomalies.



Supplementary Figure S3. Prevalence by sex in the 9 most prevalent syndromic groups initially collected from the literature. **F:** Female; **M:** Male; **VI:** Vision impairments; **MT:** Metabolic anomalies; **HL:** Hearing anomalies; **HRT:** Heart anomalies; **LIV:** Liver anomalies; **REN:** Renal

anomalies; **MEND**: Mental anomalies; **PUL**: Pulmonary anomalies; **REP**: Reproductive system anomalies.



Supplementary Figure S4. Correlation between syndromic score and age of patients included in the meta-analysis (n=227).