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

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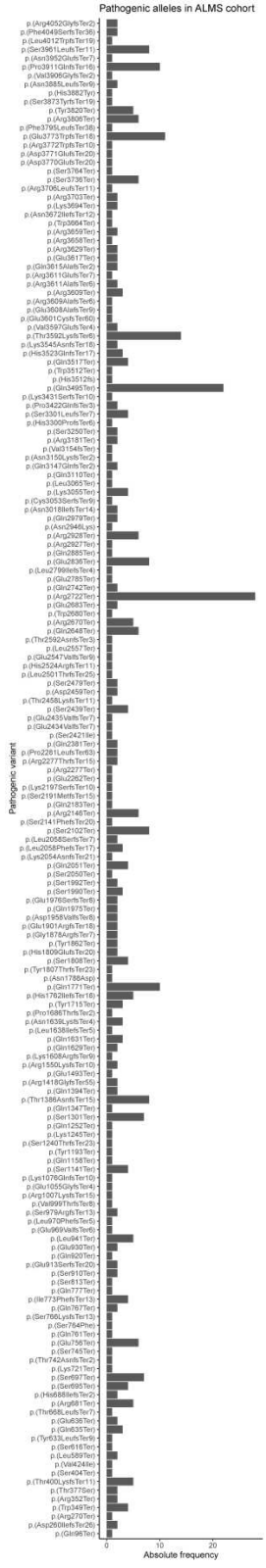
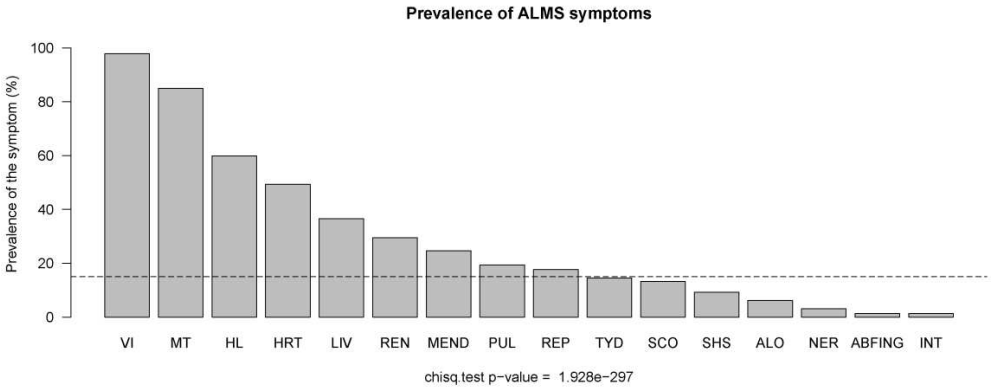
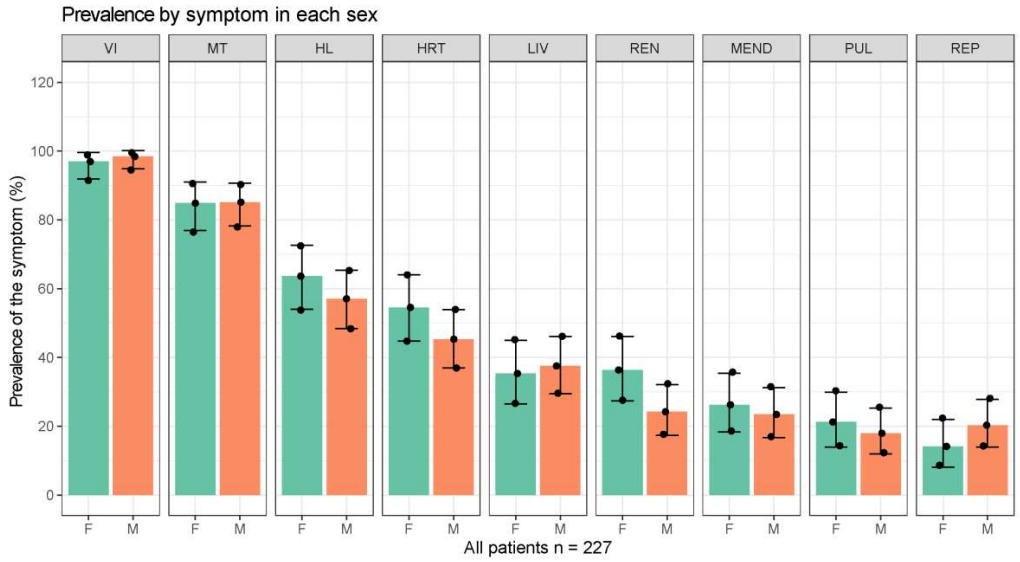
Supplementary Material:



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

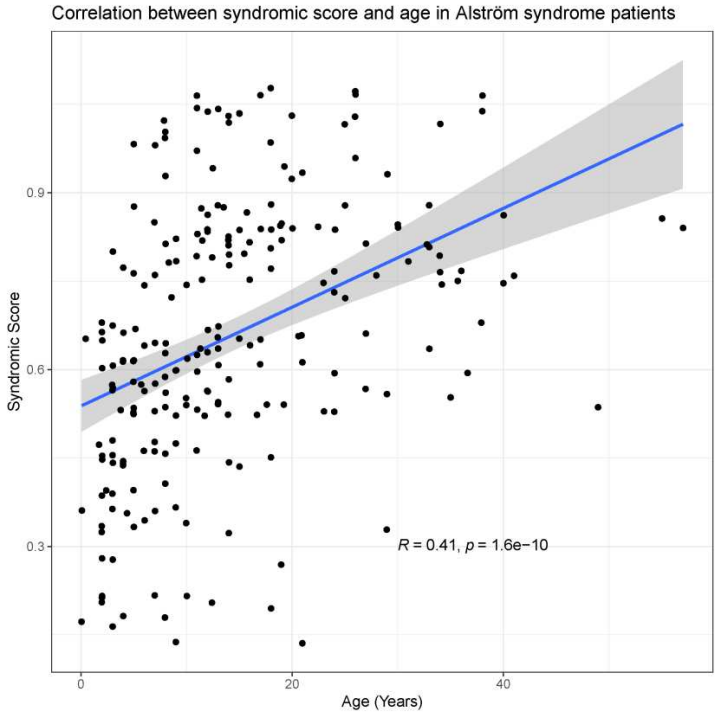


Supplementary Figure S2. Prevalence in the 16 syndromic groups initially collected from the literature and the established minimum prevalence threshold (15%; n=33) to be included in the study. **VI:** Vision impairments; **MT:** Metabolic anomalies; **HL:** Hearing anomalies; **HRT:** Heart anomalies; **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).