

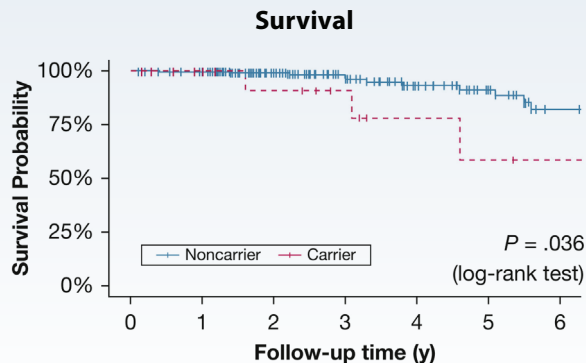
## How Are Rare Gene Variants in Patients With SLE-Associated PAH Associated With Clinical Phenotypes and Outcomes?

### STUDY DESIGN

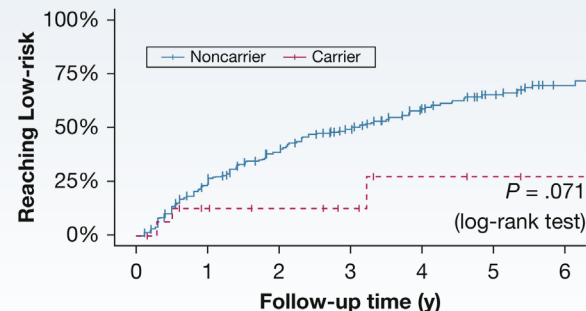
- 241 patients with systemic lupus erythematosus (SLE)-associated pulmonary arterial hypertension (PAH) were screened for rare deleterious variants in 12 definitive and 9 candidate PAH-related genes by whole exome sequencing
- Variants were classified as pathogenic, likely pathogenic, uncertain significance, likely benign, or benign
- Another 87 patients with SLE-associated PAH were included as a genetic replication cohort

### RESULTS

- 17 patients (7.2%) carried rare variants
  - 27.8% of variants (n = 5) were likely pathogenic
  - 72.2% of variants (n = 13) had uncertain significance



### Cumulative Rates of Reaching European Society of Cardiology/European Respiratory Society Low-Risk Strata of PAH



The findings of this study show that rare variants in PAH-related genes are associated with a distinct vasculopathy phenotype and worse outcomes in patients with SLE-associated PAH.