To address this, the Symptom Assessment Form (ISM-SAF) was used to characterize the cohort at the time of their within-system diagnosis, with a definite or probable diagnosis. The median differences in the time to diagnosis (in years) between pairs of groups were compared using the log-rank test. The EHRs of eligible patients were manually reviewed and patients were identified as having a SM-compatible symptom. Multiplicity was not controlled for (alpha=0.05) due to the exploratory nature of this single-center study. The time to diagnosis may be influenced by the cohort's demographics and medical features. Although this single-center study was limited by small samples, it suggests that dermatological symptoms and diagnoses were common in this cohort and were predictive of the time to diagnosis. Doing so will be critical to reduce diagnostic delays for SM and other rare diseases.

### Background
- Systemic mastocytosis (SM) is a slow-growing clonal mast cell disease driven by the KIT D816V mutation in ~95% of cases. Dermatological symptoms, delays from the onset of symptoms to diagnosis can be appreciably longer.
- Diagnosis delays for SM are characterized by a nonlinear increase in diagnostic wait, with a definite or probable diagnosis preceded by a skin-based symptom prior to their within-system diagnosis.
- The median time from the first presentation of a SM-compatible symptom to diagnosis was 5.5 years (Figure 1). The diagnosis probability (%)

### Methods
- EHRs data from UCSF (2013–2023) were used to identify patients with definite or probable SM (SCC
e). The EHRs of eligible patients were reviewed and patients were identified as having a SM-compatible symptom.
- Patients with a definite diagnosis were defined as having the WHO 2022 criteria, while a probable diagnosis was determined by symptoms.
- Structured EHR data (diagnosis codes, medication and laboratory order) were used to characterize the cohort at the time of their within-system diagnosis.
- Caucasian patients were diagnosed 5.2 years earlier than other ethnicities.

### Results
- 76% were White.
- 24% of patients had a skin-based symptom prior to their within-system diagnosis, and 45% of patients had a skin-based symptom prior to their within-system diagnosis.
- The median differences in the time to diagnosis (in years) between pairs of groups were compared using the log-rank test.
- Caucasian patients were diagnosed 5.2 years earlier than other ethnicities.
- Mutations were not controlled for (alpha=0.05) due to the exploratory nature of this single-center study.

### Discussion
- A single-center cohort of 62 patients with SM, a rare clonal disorder of mast cells discovered by non-specific symptoms, was analyzed. The median time to diagnosis for SM was estimated within the first 5 years.
- Caucasian patients were diagnosed 5.2 years earlier than other ethnicities.
- Caucasian patients were diagnosed 5.2 years earlier than other ethnicities.
- Trends were sharp in other predictors, but a larger sample size is required for resolution of these trends.

### References