

# Epidemiology of systemic mastocytosis (SM) and indolent systemic mastocytosis (ISM) in Germany

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## Background

- Systemic mastocytosis (SM), a rare disease, is characterized by clonal proliferation of mast cells (MC) that accumulate either in the bone marrow and/or in one or more organ systems. SM is associated with the KIT D816V mutation.<sup>1,2</sup>
- Indolent systemic mastocytosis (ISM), a subform of SM, represents up to 90% of all cases of SM.<sup>3</sup>
- ISM patients are characterized by debilitating skin, gastrointestinal, neurocognitive, musculoskeletal, and systemic manifestations, including anaphylaxis.<sup>4</sup>
- Due to the rarity of the disease, epidemiological data is limited.

## Objectives

- This study aimed to assess the incidence and prevalence of SM. Furthermore, the prevalence of ISM in Germany was assessed.

## Methods

- A retrospective claims data analysis was conducted.
- The study used anonymized German data from more than 50 different Statutory Health Insurances (SHIs) from the Institute for Applied Health Research Berlin (InGef) database.
- The analyzed research sample of the InGef database (~5% of the German population<sup>5</sup>) is adjusted by age, gender, and region to represent the German population. Overall, the study sample shows good correlation with the German population in terms of morbidity, mortality, and use of drugs.<sup>6</sup>
- Incidence and prevalence of adult (≥18 years) SM patients, including advanced SM (AdvSM), smoldering SM (SSM) and ISM patients, were assessed.
- Incident and prevalent SM patients were identified using ICD-10-GM diagnosis codes C94.3 (mast cell leukemia), C96.2 (malignant mast cell neoplasm) or D47.0 (histiocyte or mast cell neoplasm of uncertain or unknown behavior).
- SM patients needed to have at least one recorded diagnosis in the inpatient (primary or secondary discharge diagnosis) and/or outpatient (verified diagnosis) sector in the most recent available year in the database (2021).
- To identify incident SM patients, a diagnosis-free period of two years was chosen. In addition, a sensitivity analysis for incidence determination was performed by extending the diagnosis-free period to five years.
- For the analysis of ISM prevalence for 2021, a subset of adult patients with at least two ICD-10-GM diagnosis codes D47.0 in two different quarters of 2021 in the inpatient and/or outpatient sector (M2Q fulfilled) was selected.
- To omit overlap due to progression of ISM to AdvSM (approximately 5-10%<sup>4</sup>), patients with ISM and AdvSM diagnosis code within the same year were excluded.

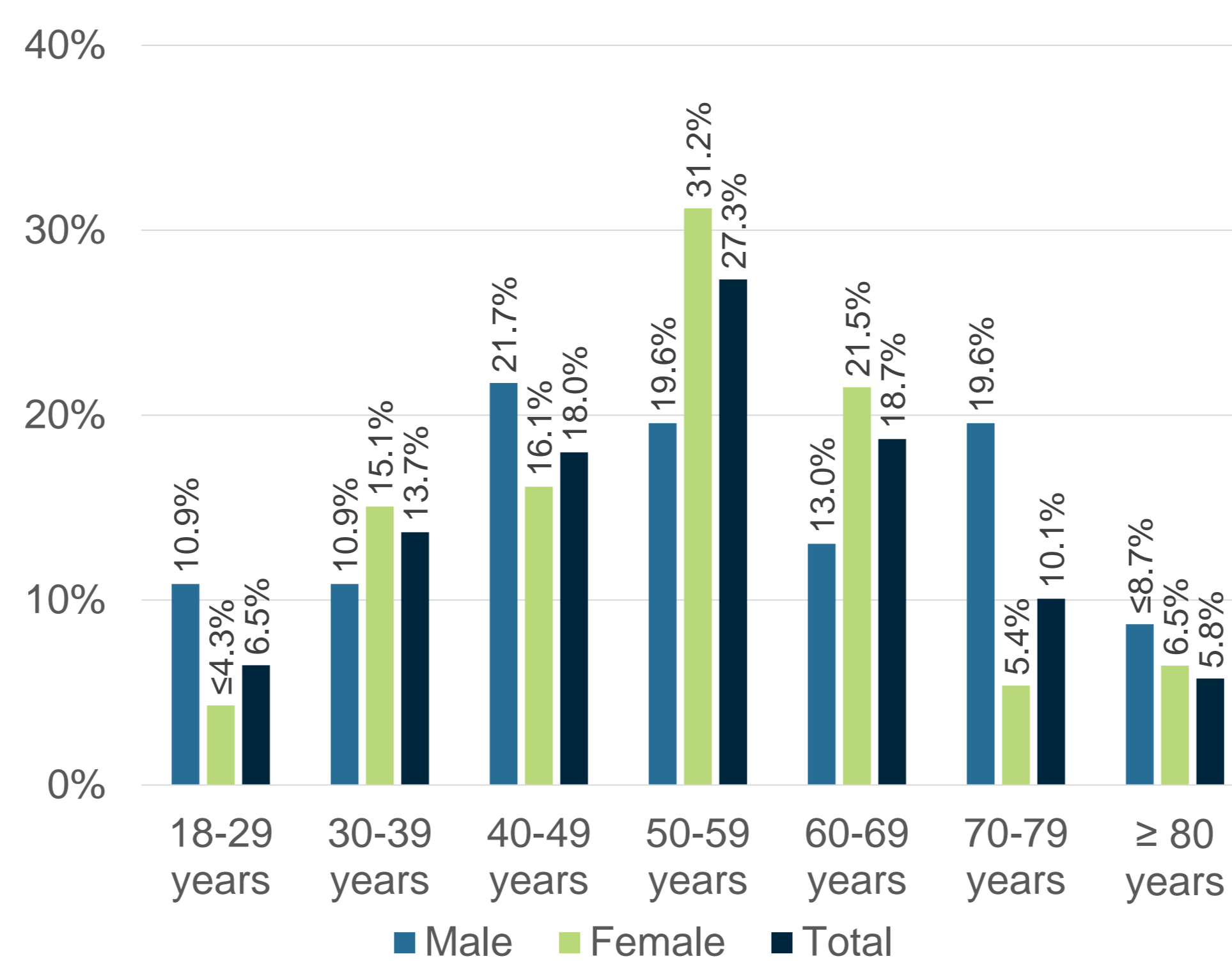
## Results

### Incidence of SM

- In 2021, 139 incident adult SM patients (based on 2-year diagnosis-free pre-observation period) with at least one recorded SM diagnosis in the inpatient and/or outpatient sector were identified, corresponding to 1-year administrative incidence rate of 3.93 per 100,000 individuals.
- Extrapolated to the German population, this resulted in 3,272 incident adults with SM.
- There were 1.5 times more female than male patients (60.1% vs. 39.9%).
- Patients aged 50-59 years accounted for more than a quarter of all incident patients (27.3%), see **Figure 1**.

## Results (Continued)

Figure 1. Age and gender distribution of incident SM patients



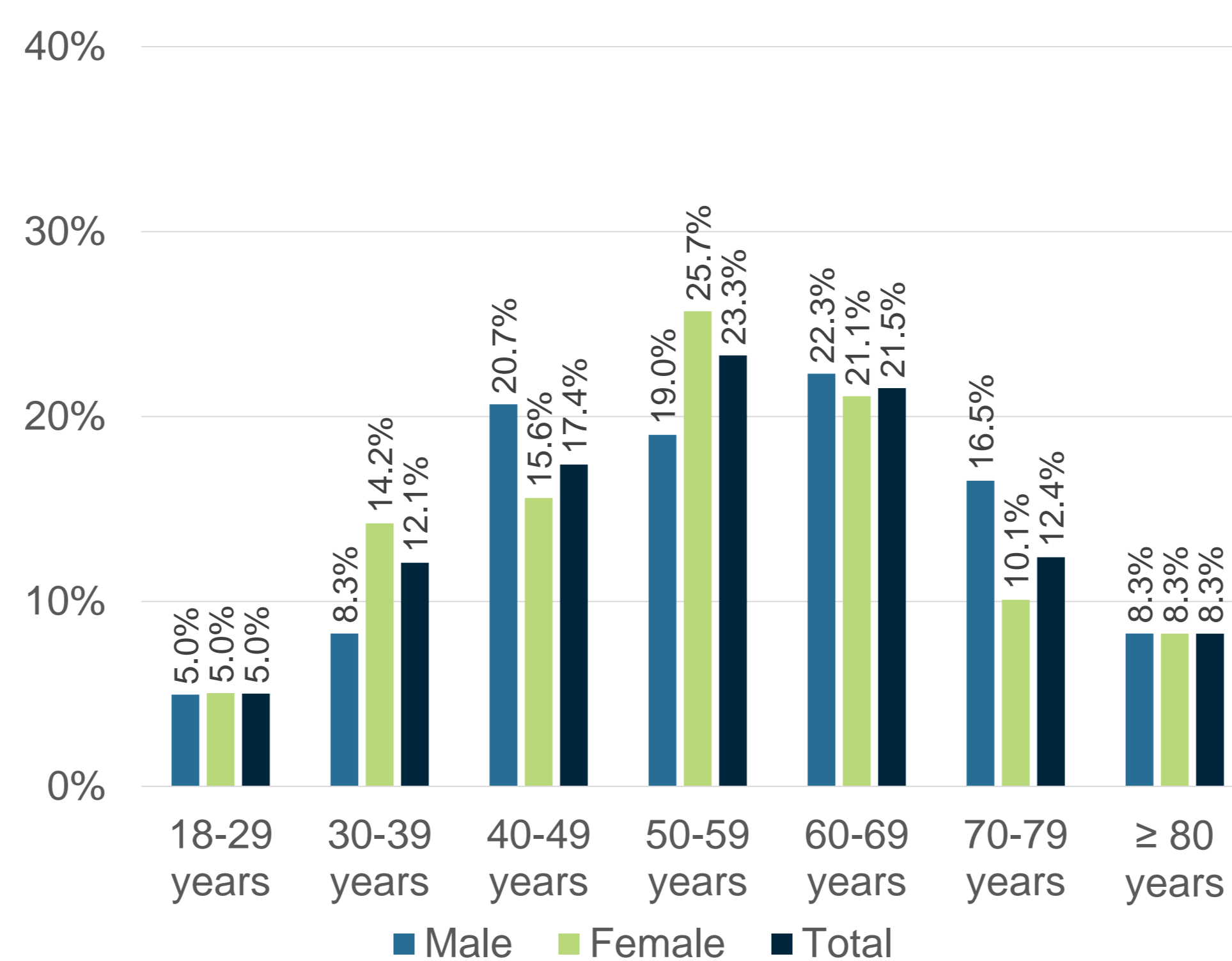
Due to data protection regulations, results for n<5 cannot be presented.

- A sensitivity analysis showed that by extending the diagnosis-free period to five years, 115 incident adult SM patients were identified, which reduced the administrative 1-year incidence rate of adult SM patients to 3.58 per 100,000 individuals.
- Extrapolated to the German population, this resulted in 2,982 incident adults with SM.

### Prevalence of SM

- In 2021, 339 prevalent adult SM patients with at least one recorded diagnosis in the inpatient and/or outpatient sector were identified, corresponding to 1-year prevalence rate of 9.59 per 100,000 individuals.
- Extrapolated to the German population, this resulted in 7,980 prevalent adults with SM.
- There were 1.8 times more female than male patients (64.3% vs. 35.7%).
- Patients aged 50-59 years made up the largest proportion of all prevalent SM patients (23.3%), followed by patients aged 60-69 years (21.5%), see **Figure 2**.

Figure 2. Age and gender distribution of prevalent SM patients



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## References

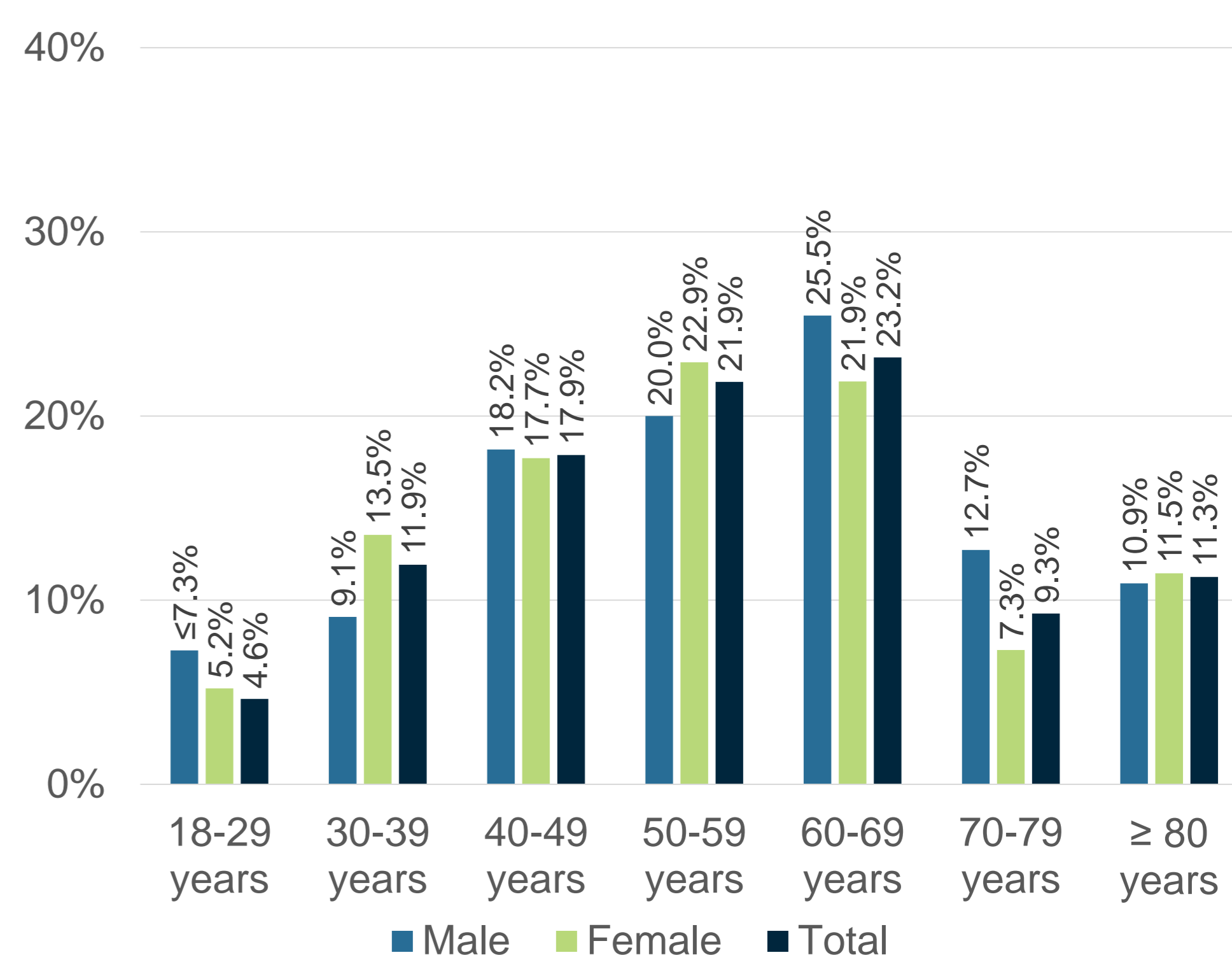
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## Results (Continued)

### Prevalence of ISM

- There were 164 adult ISM patients who fulfilled the M2Q criteria in 2021.
- After exclusion of 13 patients with AdvSM, 151 adult ISM patients were identified, resulting in 1-year prevalence of ISM of 3.89 per 100,000 individuals.
- Extrapolated to the German population, this resulted in 3,242 prevalent adults with ISM.
- There were 1.7 times more female than male patients (63.6% vs. 36.4%).
- Patients aged 50-69 years made up 45.0% of all prevalent ISM patients, see **Figure 3**.

Figure 3. Age and gender distribution of prevalent ISM patients



Due to data protection regulations, results for n<5 cannot be presented.

## Conclusions

- The estimated SM prevalence of 7,980 individuals, corresponding to a 1-year prevalence rate of 9.59 per 100,000 individuals, is consistent with published literature and with a previous study conducted with 2020 as a reference year.
- The prevalence of ISM, when using commonly established M2Q criteria for epidemiological studies, is estimated to be 3,242 patients, corresponding to a rate of 3.89 per 100,000 inhabitants.
- To our knowledge this is the first time ISM prevalence was described with a claims database in Germany.

## Disclosures

Study sponsored by Blueprint Medicines (Germany) GmbH. MH and HS are full-time employees of Blueprint Medicines (Germany) GmbH. SAS and CJ are full-time employees of Xcenda GmbH, part of Cencora, acting as contractors of Blueprint Medicines (Germany) GmbH for the execution of this study.