

Age, family history and genetic testing data from myotonic dystrophy patients and carers participating in ENSA survey



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Introduction

- The myotonic dystrophies (DM1 and DM2) are hereditary rare diseases exhibiting heterogeneous symptoms.¹
- People with DM1 and DM2 often experience myotonia, which adversely impacts their quality of life (QoL).^{1,2}
- Many people with myotonic dystrophy (DM) face unacceptable delays between symptom onset and receiving a correct clinical diagnosis.³
- The ENSA (revEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA) survey investigated the impact of myotonia on people living with DM1 or DM2.
- This sub-analysis focused on respondents' data regarding genetic testing for DM1 or DM2, known family history of DM, and ages at symptom onset and DM diagnosis.

Objective

- To identify potential trends relating to the time between symptom onset and diagnosis according to genetic testing and known family history of DM1 or DM2.

Methods

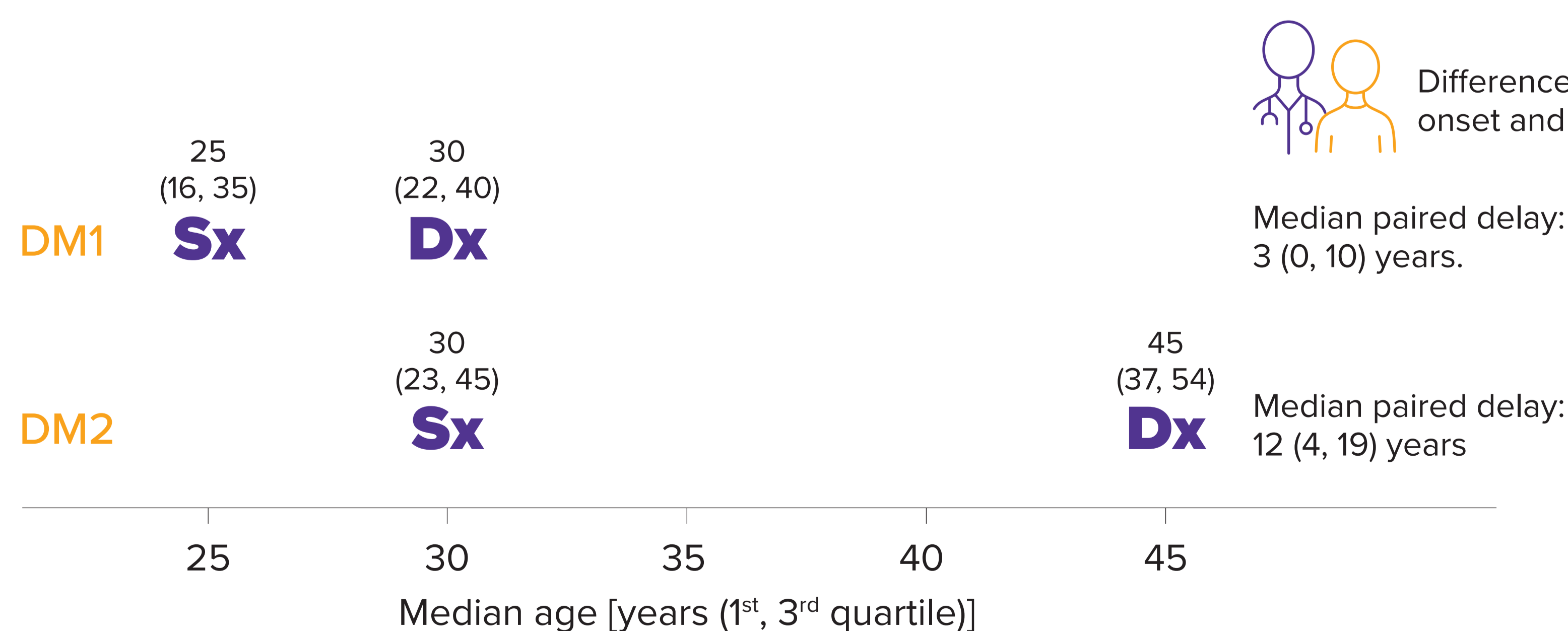
- The anonymised, international, patient-reported, ENSA survey investigated the impact of myotonia in adults (≥ 18 years) living with DM.
- Respondents were people living with DM or caregivers completing the survey on behalf of people with DM.
- Respondents were invited to undertake an online survey via an international outreach campaign.
- Respondents completed up to 32 lay-language questions covering a spectrum of topics:
 - Questions assessed their experiences of the diagnostic process and burden of DM on daily life.
- Descriptive analyses, stratified by age and DM subtype, were undertaken, including paired differences where appropriate.
- Median values are presented with 1st and 3rd quartiles.

Results

Population

386 (age 18–82 years, 61.7% female) completed the ENSA survey. Of those, **354 (92%)** had, or cared for someone with, genetically confirmed DM.

People with DM face unacceptable delays in diagnosis, particularly those with DM2.



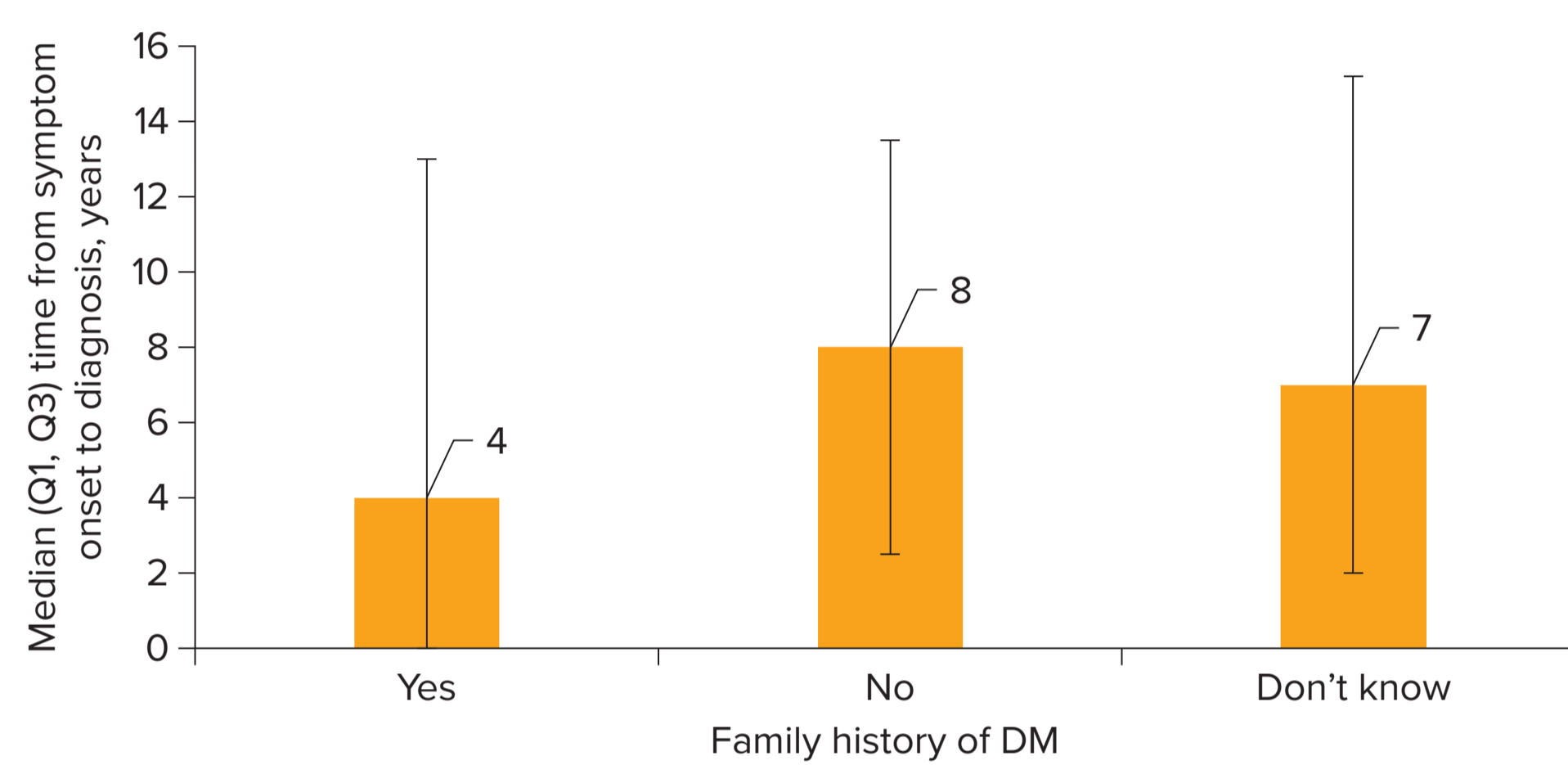
DM, myotonic dystrophy; Q, quartile.

Conclusions

- Most ENSA survey respondents reported a genetically-confirmed diagnosis, but those with a family history had a shorter diagnostic odyssey than those without.
- The 52 respondents diagnosed with DM while asymptomatic were likely identified early as a result of family screening.
- Unacceptable diagnostic delays of 3 and 12 years between symptom onset/first seeking medical help and diagnosis were reported for DM1 and DM2, respectively.
 - These data show there has been a limited improvement in diagnostic delays over the past decade (7 and 14 years for DM1 and 2 respectively), particularly for DM2.³
 - There is a need for anyone affected by DM (even asymptotically) to obtain prompt access to monitoring, support and clinical management.
- These data reinforce the importance of family screening and genetic testing in DM.

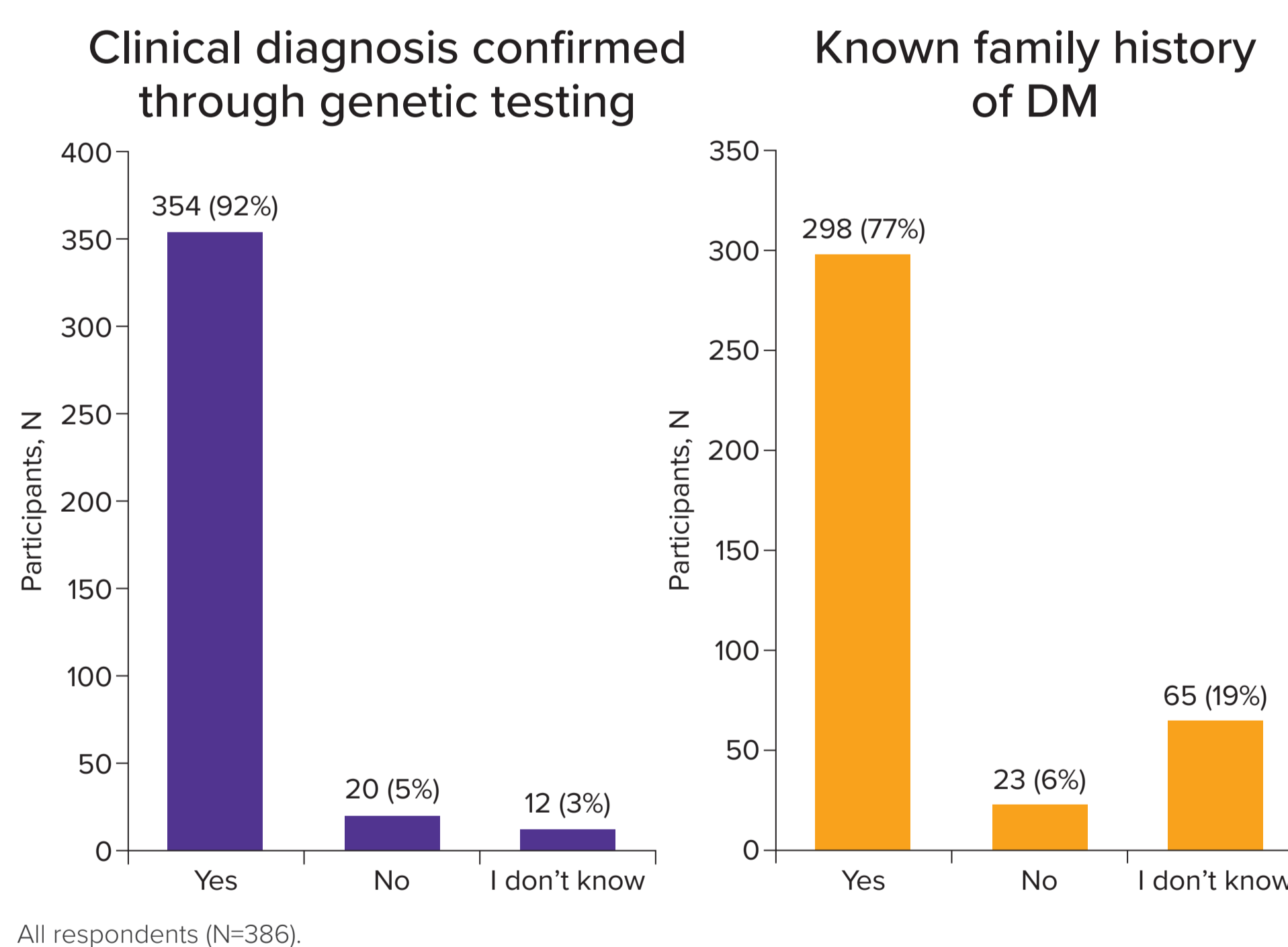
Impact of family history on speed of diagnosis

Having a known family history of DM cut diagnostic delays in our dataset from 8 to 4 years.



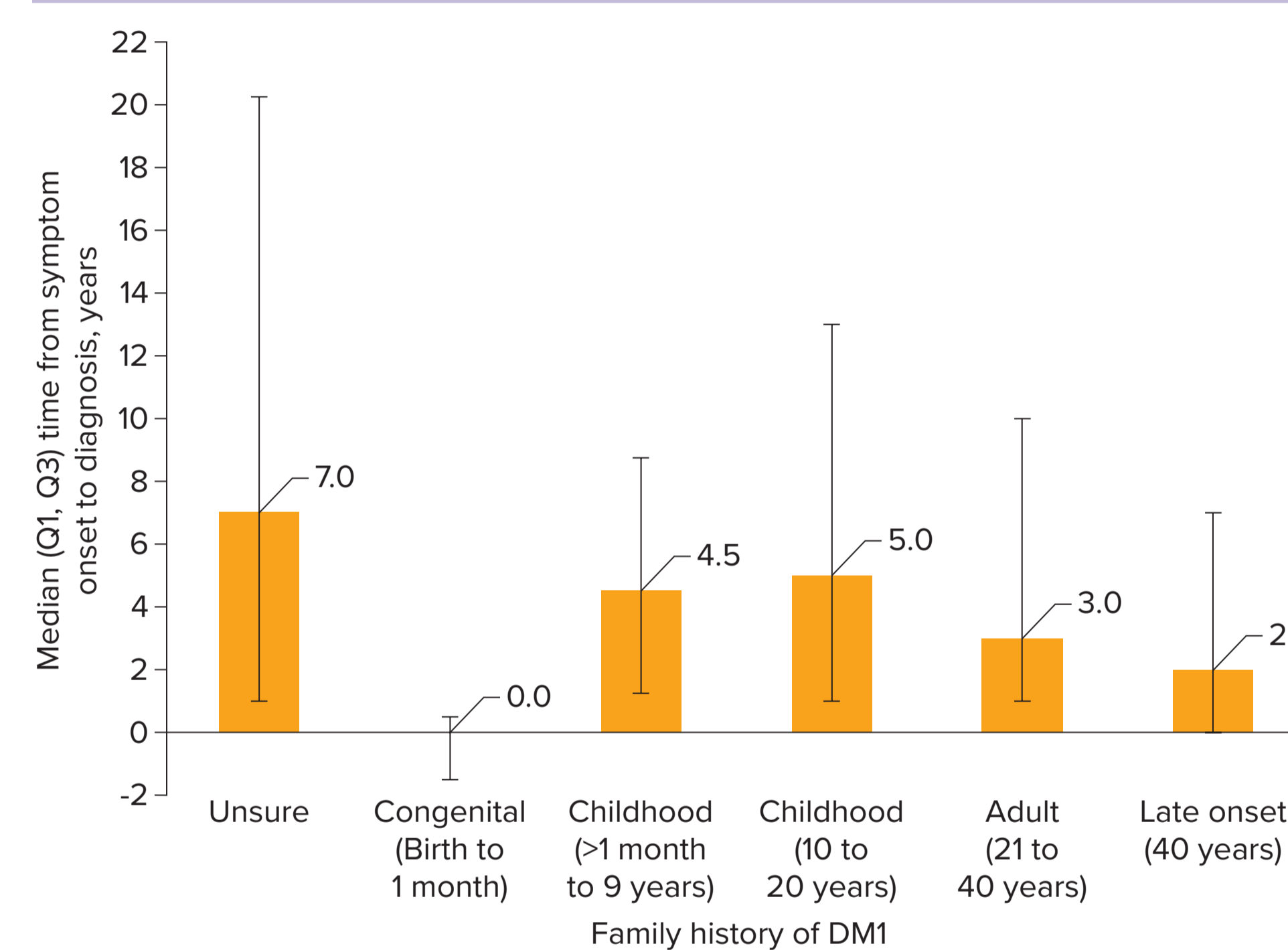
Availability of genetic testing

Most people with DM received genetic testing and reported a known family history. These results suggest that individuals with a confirmed genetic diagnosis of DM may share their status with family members, which encourages them to undergo genetic testing.



Differences in diagnostic delays by age of DM1 symptom onset

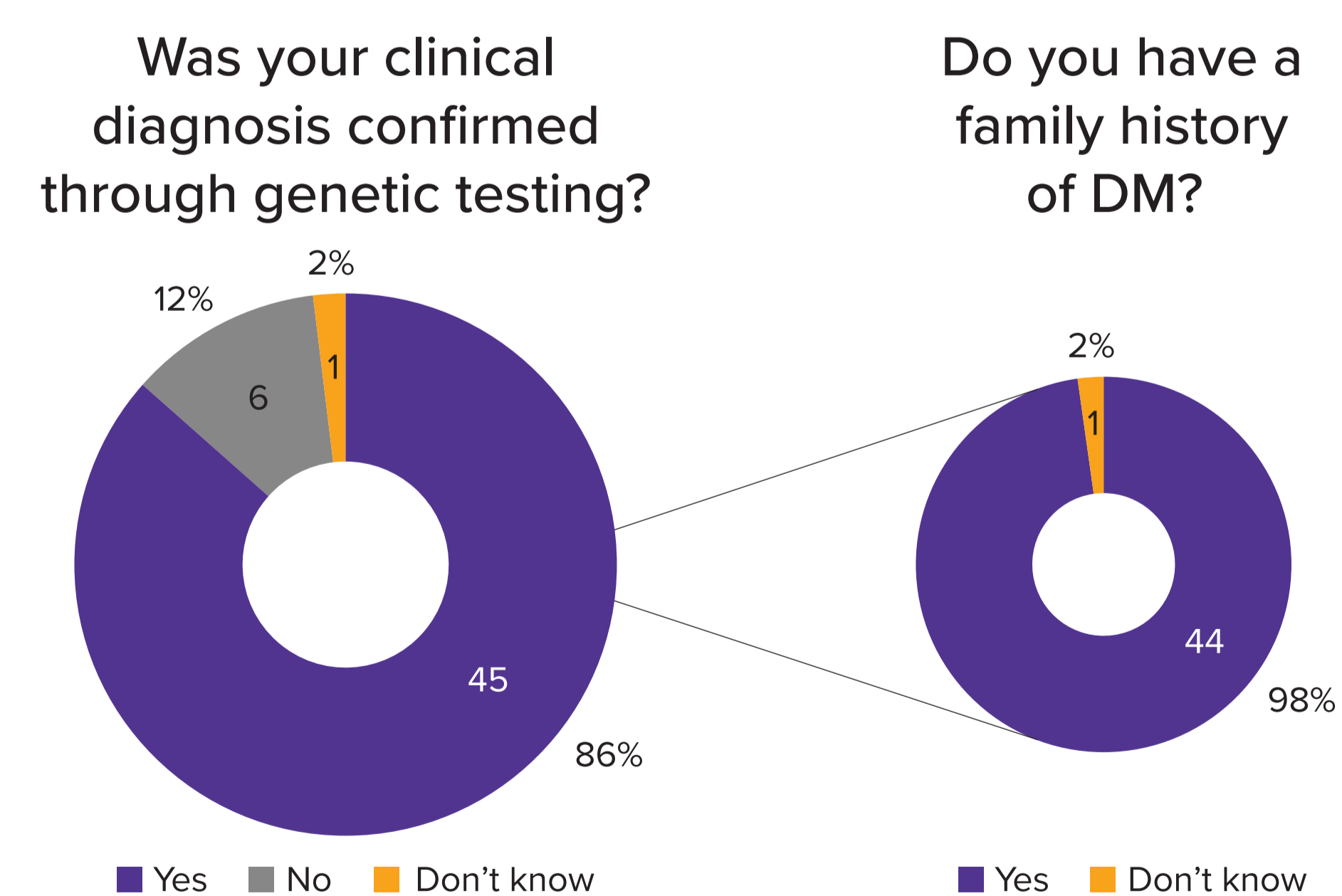
Diagnostic delays are longest in those who were unsure of symptom onset and those with childhood onset; ENSA data show a wide range of delays. The numbers of respondents in the Congenital and Childhood (pre-puberty) DM1 categories were both low.



Genetically confirmed subset (n=354)

Some people with DM received a diagnosis before symptom onset

52 people (13.5%) received their DM diagnosis before symptom onset. Within the subgroup with a confirmed genetic test (n=45/52, 86%), 44/45 (98%) had a family history of DM.



Subset diagnosed before symptom onset (n=52)

References

- Landfeldt E, et al. J Neurol 2019;266:998–1006; 2. Heatwole C, et al. Neurology 2015;85:2136–46; 3. Hilbert JE, et al. J Neurol 2013;260:2497–504.

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Disclosures

Zozulya-Weidenfeller is employed by Lupin. Other authors received honoraria from Lupin Neurosciences as consultants during the ENSA creation.

