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INTRODUCTION

- Haemophilia B is an X-linked recessive bleeding disorder characterized by an absence or deficiency of coagulation factor (F) IX due to genetic mutations in the FIX gene.
- Genetic testing is the only reliable tool to determine carrier status. Yet genetic testing is not always available and clinicians sometimes rely on women's FIX levels to determine carrier status.
- Our clinic seeks to determine the mutation responsible for haemophilia in all our patients and to determine their mothers' carrier status.

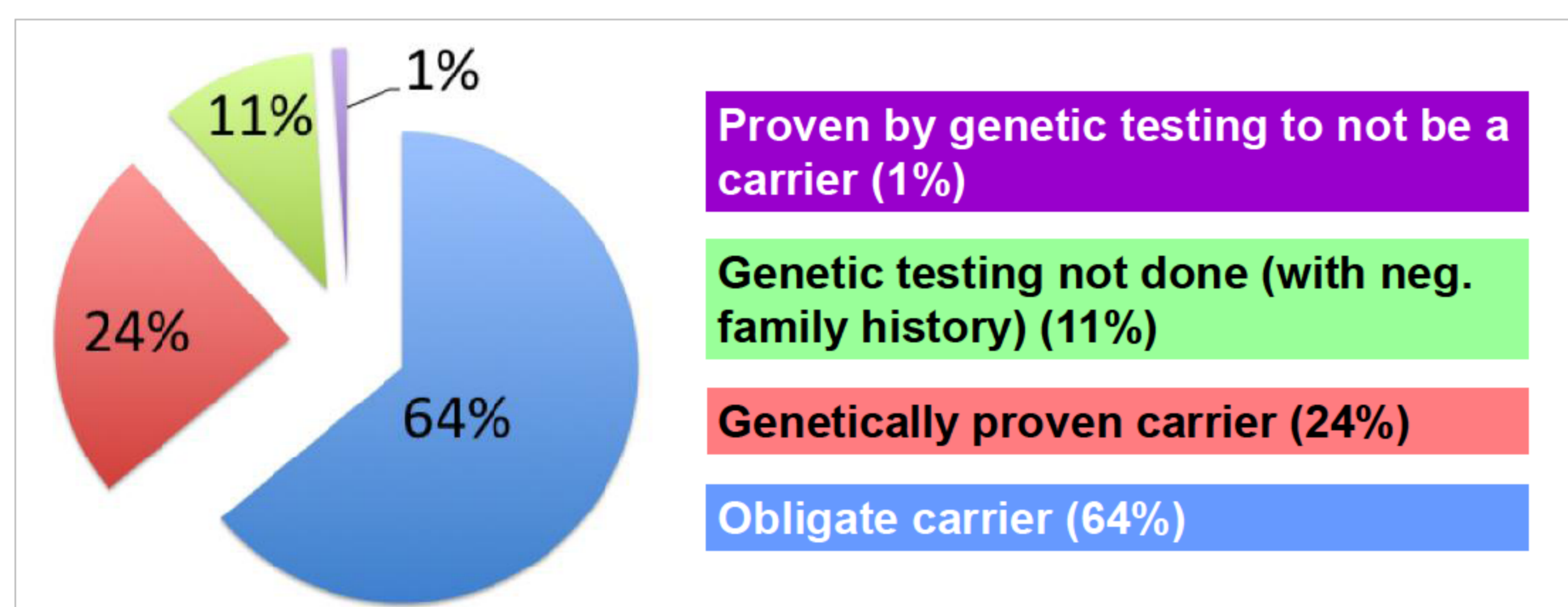
OBJECTIVES & METHODS

- Factor IX (FIX) levels show considerable variability in carriers of Haemophilia B (HB).¹
- Recently, we have been obtaining baseline FVIII/FIX levels on all carrier mothers regardless of whether they do/do not demonstrate clinical bleeding.
- We present the results of an unselected population of carrier mothers of boys with HB.
- Aims:
 - Determine the distribution of FIX levels in carrier mothers
 - Determine if FIX levels in carriers are correlated with the genetic mutation causing haemophilia in the family or with the severity of haemophilia in their children with haemophilia

RESULTS

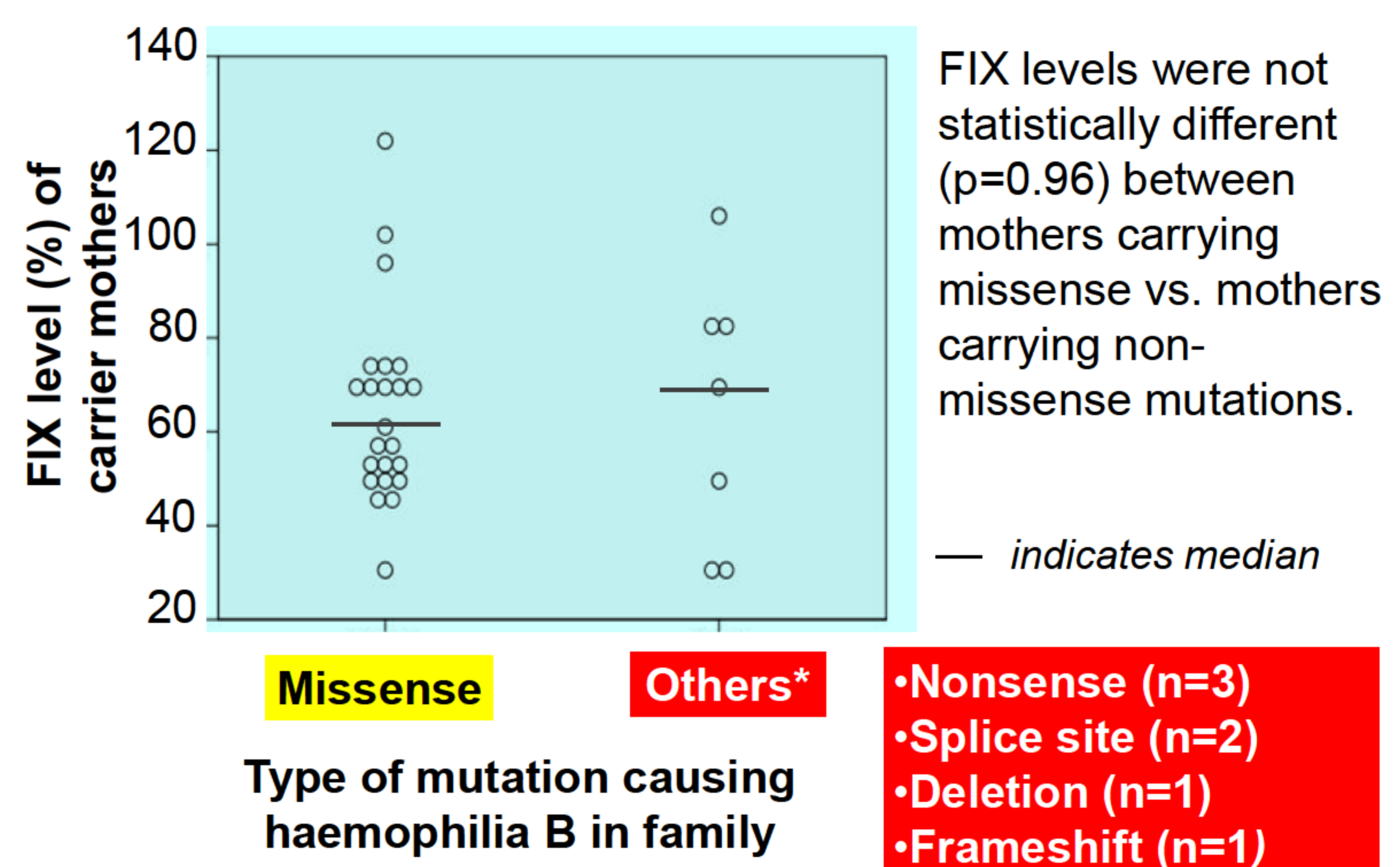
Demographics

- 75 boys with HB are registered in our genetics' database
- Collectively they have 66 mothers (some mothers have ≥1 affected son).
- In at least 88% of children the mothers are known to be carrier.
- Distribution of Carrier status:**

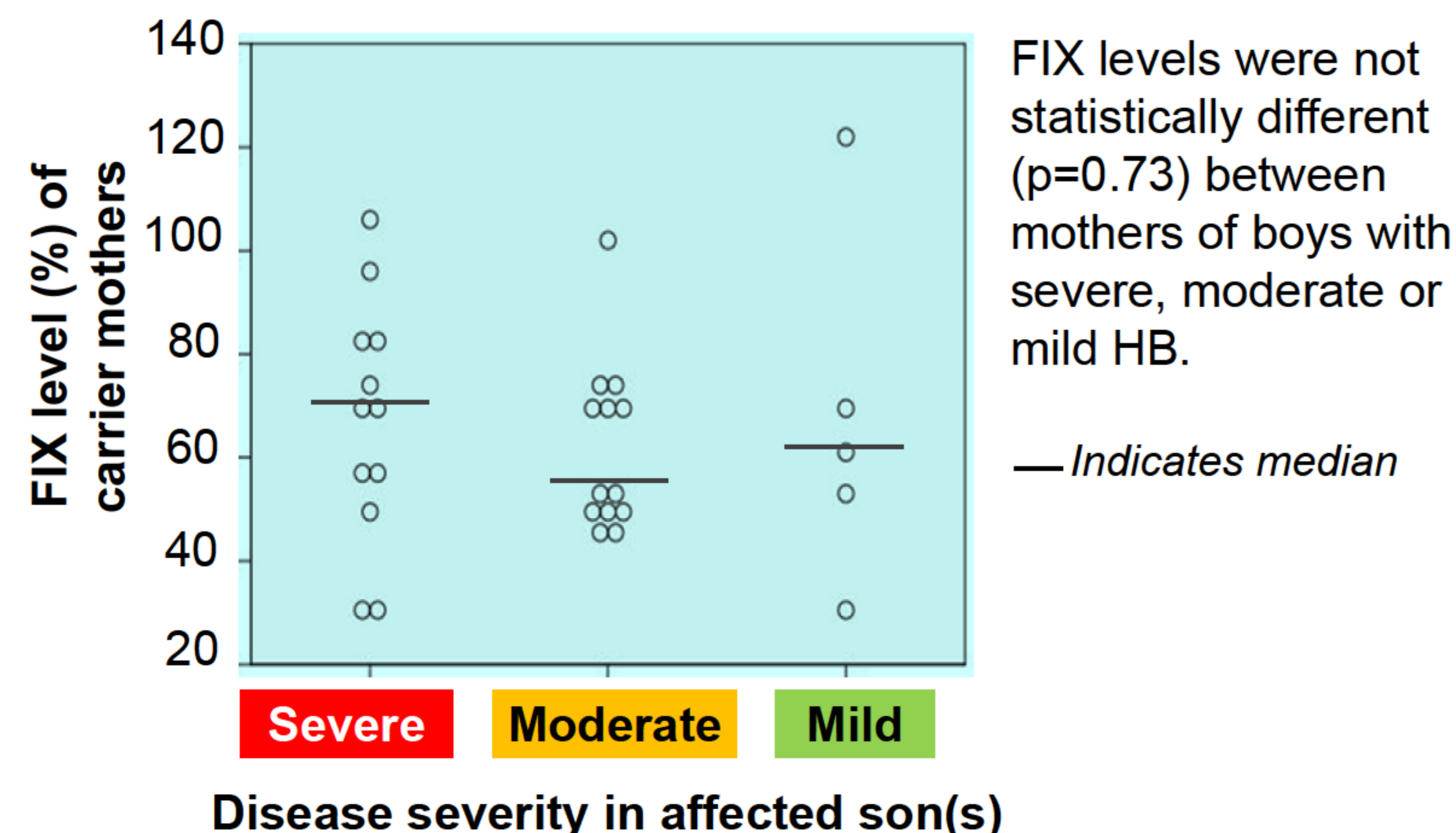


- Of the 66 carrier mothers in 30 (45.5%) both genetic results and FIX levels were available:
 - Mother's Median age (at the time of FIX testing) 29yr (IQR 26-33)
 - FIX levels in carrier mothers varied between 29% and 122%
 - Median FIX levels in all carrier mothers was 68%
 - % of carrier mothers with FIX level > 50%: 73%

Relation between Mutation type and Carrier FIX level



Relation between Severity of Haemophilia B and Carrier FIX level



SUMMARY & CONCLUSIONS

- Our data indicate that when a boy is diagnosed with HB, the likelihood of his mother being a carrier is between 88% (58/66) and 99% (65/66).
- Most carrier mothers (73%) have baseline FIX levels that are >50% proving that FIX levels are unreliable at predicting carrier status in mothers of boys with haemophilia.
- FIX levels in carriers do not correlate with the severity of HB in their sons or the type of mutation.

REFERENCES

¹Knobe K.E. and Ljung R.C.R.. Haemophilia (1999), 5, 238-242

