A single-centre review of late amniocentesis reliability and satisfaction in carriers of haemophilia

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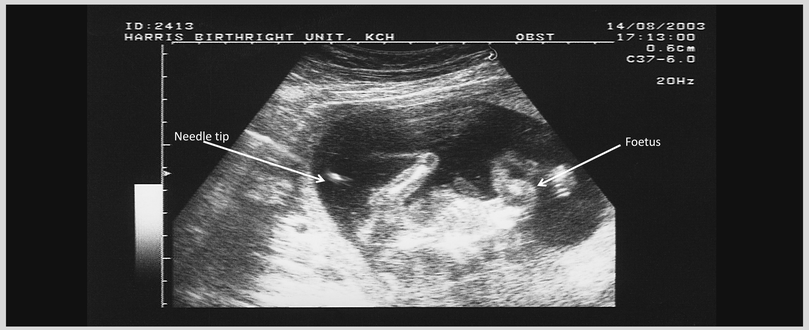
INTRODUCTION

Babies born with haemophilia are at increased risk of intracranial haemorrhage[1]. Late amniocentesis is increasingly offered to inform management of delivery, since unaffected males can be delivered at non-haemophilia centres[2,3].
Few data exist about the reliability of late amniocentesis and no data are published on the experiences of the pregnant woman.

This study reports a single-centre experience of late amniocentesis in haemophilia carrier women during the period January 2013 – January 2016.

AIMS

1. To examine the reliability of late amniocentesis to aid delivery
2. To establish levels of satisfaction with the procedure
3. To determine frequency of side effects experienced.



METHOD

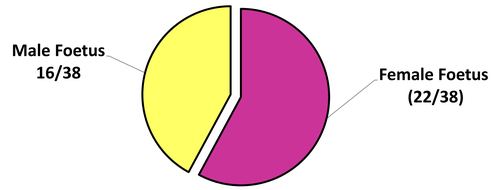
A retrospective review of clinic case notes of all pregnant carriers of haemophilia A or B was undertaken.

Data were collected on amniocentesis result, procedural side effects, and location of delivery. A prospective semi-structured satisfaction telephone survey using a 5 point Likert scale was conducted.

The women were questioned on their satisfaction with the amount of information provided pre-procedure, the procedure itself and whether they would consider a late amniocentesis for future pregnancies.

RESULTS

Total number of Pregnancies - Haemophilia A and B



Amniocentesis

38 pregnancies (16 with males) were reviewed. 9 of 16 (56%) women underwent late amniocentesis.

Six were carriers of severe haemophilia; three haemophilia A (two with intron 22 inversion) and three mild haemophilia (33% haemophilia A).

4 of 9 foetuses were affected with haemophilia (3 severe, 1 mild) – one mother chose an elective Caesarean.

Five women (55%), whose babies were unaffected were able to deliver at a local hospital.

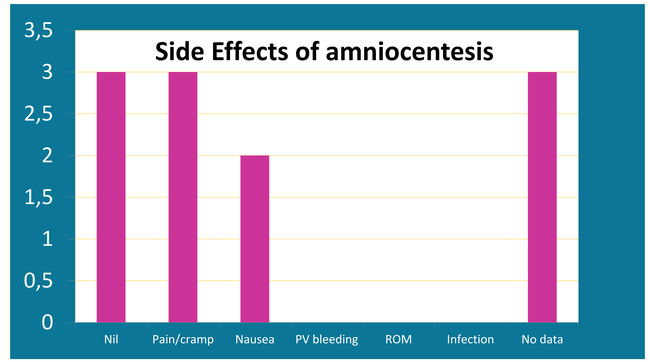
Amniocentesis results with genetic mutation

No. of weeks at procedure	Gene mutation	Result
34	Intron-22 inversion	Not affected
34	F8 gene c.3863dup	Affected
34	c.1220G>A substitution	Affected
35	c.230T>A	Not affected
34	c.11363G>A	Not affected
35	c.230T>A	Not affected
36	c.557A>G, p.D186G	Affected
36	Intron-22 inversion	Failed (affected)
34	F9 gene c.158A>T	Not affected

SURVEY RESULTS

Questions:
Were you satisfied with the level of information and explanations you were given?
Was the procedure as you expected?
Did you experience any side effects and what were they?
Would you consider a late amniocentesis in future pregnancy?

Please rate your overall experience: (5-point Likert scale)
33% women scored 5/5 strongly satisfied
33% women scored 4/5 satisfied
33% women were unable to be contacted



CONCLUSION

Late amniocentesis is a highly informative procedure – our data showed 89% reliability with 66% of delivery plans changed as a result. One case of intron 22 mutation at a later procedure date failed in line with current knowledge.

In addition, high levels of satisfaction were reported with all women contacted stating they would undergo late amniocentesis again. This is an important factor for future counselling.

REFERENCES

1. Huq F. Y. and Kadir R. A. (2011) Management of pregnancy, labour and delivery in women with inherited bleeding disorders. *Haemophilia*. 17(1), 20-30.
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3. Cutler J *et al* (2013) Third trimester amniocentesis for diagnosis of inherited bleeding disorders prior to delivery. *Haemophilia*. 19, 904-907.

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