Prenatal diagnosis of haemophilia – how is it experienced today?

Charlotta Perers

Supervisor: Rolf Ljung, Department of Clinical Sciences, Lund

It is known from earlier studies that carriers of haemophilia face many psychological difficulties when going through prenatal diagnosis (PND). However, no studies have been made on this subject in Sweden for the last ten years. We therefore evaluated, by semi-structured interviews, how carriers of haemophilia (n=17) experienced PND, by either chorionic villus sampling or amniocentesis, in Sweden during the interim 1997-2008. The aim was to improve the care for carriers going through PND in the future. We found that for 65% the carriershhip had affected the decision to get pregnant mainly negatively. More than half of the women, 58%, choose PND with the aim to interrupt pregnancy with affected foetus, 24% to be psychologically prepared for having a child with haemophilia and 18% primarily to exclude other chromosomal aberrations. The majority, 76%, received genetic counselling prior to the PND but almost half of these women did not feel that the information fully corresponded to what they later experienced. The procedure was unexpectedly troublesome for 65%, 47% described it as very painful and the majority expressed concern about the risk to the foetus. In total 28 PND:s were performed. Ten of these were male foetuses with haemophilia, of which five pregnancies were interrupted and five continued to birth. Among the five women interrupting a pregnancy due to haemophilia, three experienced the following months as troublesome and one woman still regretted the decision. Two of these women later gave birth to a child with haemophilia. In conclusion the women accepted PND relatively well but the care can be improved by a more detailed description of the procedure before the PND and recommendation of psychological support for couples who decide to terminate a pregnancy following PND.