TABLE *Frequency of mothers pregnant with a male with male fetal cells in their peripheral blood. Data on interphase and metaphase counts are presented separately*

	Interphases in uncultured cells	*Interphases after PHA stimulation*	*Metaphases after PHA stimulation*
Walknowska *et al*[4]			19/21 cases
de Grouchy and Trébuchet[5]			8/11
Zimmerman and Schmickel[6]	No good correlation between sex and fluorescent bodies		
Schindler *et al*[7]			4/9
Schröder and de la Chapelle[8]	7/9	7/9	
Grosset *et al*[9]		42/45	
Zilliacus *et al*[10]	7/11		0/10

metaphases, with quinacrine staining, in the peripheral blood of pregnant women after stimulation with PHA, but no previous purification of lymphocytes. Whether these cells are lymphocytes or not has already been discussed earlier by Schröder and Herzenberg.[12]

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Recurrence risk of neural tube defects

SIR,

With reference to Dr Seller's paper (page 245) on recurrence risks for neural tube defects (NTD) derived from a population of women presenting themselves for prenatal diagnostic tests,[1] I would like to confirm some of her conclusions from the experience in South Wales. Among the 4221 amniocenteses carried out between 1974 and 1980, the 927 carried out because the mother had had one or more previous children with an NTD led to the detection of 24 recurrences, giving an apparent recurrence risk of 1 in 39 (2·58%) (table). However, a follow-up of all the pregnancies revealed a further seven cases that were missed (in four because the lesion was closed) giving a total risk of recurrence derived from this population of 1 in 30 (3·34%),

TABLE *Amniocenteses in South Wales 1974–1980*

Amniocenteses	*NTD detected*				*NTD detection rate*	*NTD undetected*	*Total NTD rate*
	No	*An*	*SBC*	*E*			
Indication							
Previous NTD	927	14	10	—	1:39	7	1:30
Family history of NTD	596	3	2	—	1:119	—	1:119
Parent with NTD	40	—	—	—	—	—	—
Raised serum AFP	774	47	45	1*	1:8·3	5	1:7·9
Other CNS malformations	75	1	1	—	1:37	—	1:37
Anxiety	151	1	—	—	1:151	—	1:151
Cytogenetic	1523	1	3	—	1:508	3	1:218
Other	127	2	—	—	1:63	—	1:63
Total	4221	69	61	1	1:32	15	1:30

A, anencephaly; SBC, spina bifida cystica; E, encephalocele. *A case of Meckel-Gruber syndrome.

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a very similar one to that obtained by Dr Seller. In those who had amniocentesis because of a family history of NTD (mostly with an affected second or third degree relative) the overall risk was 1 in 119 (0·83%), while in those where the amniocentesis was performed for cytogenetic indication the incidence of NTD turned out to be 1 in 218 (0·46%), suggesting a population risk of 4·6 per 1000 births. The latter would be in keeping with the fallen population incidence of NTD in the area from 7·67 per 1000 births between 1956 and 1962[2] and 7·0 per 1000 births between 1964 and 1966[3] to just over 4 per 1000 births 12 years later.[4]

The risk of recurrence of 1 in 30 is substantially less than the 1 in 19·2 found in a large family study in South Wales reported in 1968.[5] This lower risk must be a reflection not only of the considerably higher social class distribution of couples coming for amniocentesis than the general population of those who have a child with an NTD,[6] but also of the reduced population incidence in recent years. It is probable that, in addition, some minor cases of spina bifida cystica and encephalocele may not have been reported in the follow up.[7] With some caution this lower risk of recurrence in amniocentesis cases should be reflected in risks that could now be used in genetic counselling.

I wish to thank Dr Seller for letting me see the typescript of her paper.

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