J Med Genet 1992; 29: 140–144

LETTERS TO THE EDITOR

Sociocultural problems in genetic counselling

We have recently encountered two cases which highlight the importance of sociocultural factors in genetic counselling.

Case 1. A Muslim couple was referred to the genetic clinic for counselling regarding the risk of recurrence of thalassaemia. The couple had previously had an abortion, a stillbirth, and two children who had died of thalassaemia major. It was a first cousin, consanguineous marriage. On investigation, both partners were found to be carriers for β thalassaemia. The husband was a graduate and a photographer by profession. During their first visit, he was accompanied by his wife and mother in law, but subsequently only the husband attended, indicating his wife's inability to attend the counselling session because of 'family commitments'. Two weeks later, his wife and mother in law came to the clinic and were anxious to know whether the wife alone was responsible for the occurrence of the disease. The husband had been harassing them, demanding permission for a second marriage or divorce.

Case 2. A similar situation of possible harassment of the wife was anticipated in a Hindu family with a child suffering from Duchenne muscular dystrophy. In this family, both parents were uneducated and of rural background. The parents were accompanied by the paternal grandfather of the child who was educated, with a postgraduate qualification. He requested us not to disclose the carrier state of the wife to the husband, as he anticipated harassment or request for a second marriage by the husband.

Genetic counselling must take into account complex psychological and emotional factors which may affect the consultation. The parents require assistance in dealing with emotions raised by the knowledge that they are both carriers for the same autosomal recessive condition if they are to make an informed decision with regard to management of the pregnancy. This information often leads to feelings of guilt, defectiveness, and loss of self-esteem.

The two cases reported here indicate how a situation can be exploited and misinterpreted in a male dominated society, especially when the wife is uneducated and from a rural background. The problem could be particularly serious in X linked recessive and autosomal dominant disorders where the wife is the carrier of the trait. High values are attached to the marital life of an Indian woman; separation, divorce, second marriage, and the status of the family and individual person in society are regarded differently in the Indian subcontinent compared to western countries. Extra caution is necessary in counselling parents from this background. In such a situation, both the parents and an older member of the family should be included in the counselling session to allow proper interpretation of the information provided to the couple. Written notes and subsequent follow up by a social worker should reinforce the counselling.

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Meckel syndrome and neural tube defects in Kuwait

Meckel syndrome (MS) or dysencephalia splanchnocystica is an autosomal recessive form of neural tube defect (NTD) consisting of posterior encephalocele with polydactyly, cystic dysplasia of the kidneys, and other anomalies.[1] In Kuwait, there is an apparently high frequency of MS, as also observed for some other autosomal recessive malformation syndromes.[2,3] This has only been observed recently, since MS has not been specifically looked for before from among the 'lumped together' NTD, which in general have shown a significant decrease in recent years[4,5] compared to the past.[6]

During 1987 and 1988 at Farwania regional hospital, serving a mixed Arab and non-Arab population of approximately 400 000 (mostly Arabs with 30% Kuwaitis and 15% Bedouin), we have ascertained 21 cases of NTD among 17 652 consecutive births with an overall birth prevalence of 1·19:1000 (table). Among the 21 cases, six had an occipital encephalocele, only one of which was non-syndromic, and five cases (two male and three female) in four sibships of unrelated families had MS. The parents of the MS cases were consanguineous Bedouin or Kuwaitis of Bedouin ancestry. Diagnosis was made by finding, besides occipital encephalocele, associated cystic kidney changes (two sibs) or postaxial polydactyly and polycystic kidneys (three cases). Similar interfamilial variability in MS has been noted previously including a report of a Bedouin family from Kuwait.[7,8]

Incidence of NTD in Farwania Hospital: consecutive births 1987 and 1988.

	1987	1988	Overall	Incidence per 1000 births
Total births	8739	8913	17 652	
Anencephaly	4	6	10	0·57
Spina bifida cystica	3	2	5	0·28
Syndromal and non-syndromal occipital encephalocele	3 (1 non-syndromal, 2 MS*)	3 (All MS)	6 (5 MS)	0·34
Total NTD	10	11	21	1·19

* Meckel syndrome

The birth prevalence of MS in this study is unusually high for a malformation syndrome (1:3530), but is almost comparable to other hospitals in Kuwait serving a significant Bedouin community (unpublished data). It is approximately three times the frequency of MS in Finland.[9] When calculating the birth prevalence among the Bedouin, it appears even higher, a situation similar to the frequency of MS among the Tartars in the Soviet Union[10] and in Gujarati Indians.[11] However, the birth prevalence of anencephaly in Kuwait has shown a progressive decline. In 1968, the estimated birth prevalence was 3·2:1000,[6] which in 1983 became 1·33:1000 with marked geographical variation.[4] For example, at Jahra Hospital, serving an Arab population of 300 000 (80% belonging to the Bedouin community), the birth prevalence was 2·05:1000 which became 1·00:1000 in a recent study[5] and at Farwania Hospital the figure of 0·86:1000 was the lowest in Kuwait. In the present study it is 0·57:1000. This progressive decline, in the absence of prenatal diagnosis and selective abortion on religious grounds, is attributable mainly to the improved quality of food and change in dietary habits, with more fresh vegetables and fruit, in addition to improved health standards as a whole.

Based on the present findings, and as previously suggested,[4] we expect a further decline in the frequency of multifactorial NTD leaving, after a few decades, a baseline prevalence mainly resulting from those with a strong polygenic contribution and mendelian disorders of the neural tube, such as MS, the rare autosomal recessive variant of anencephaly,[12] and hitherto unrecognised disorders.

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