

## 12 CONSANGUINITY

### 12.1 Definition of Consanguinity

Consanguinity is defined as a marriage between relatives and has various degrees. As pointed out by Stern <sup>19</sup> closely related individuals have a higher chance of carrying the same alleles than those less closely related and therefore children from consanguineous marriages are more frequently homozygous for various alleles than those from non-consanguineous unions.

### 12.2 Consanguinity Throughout History

Consanguinity has been afforded various degrees of legitimacy by different societies and at different times in history. This would therefore explain brother/sister marriage as practised between the members of the reigning dynasty of ancient Egypt and the Incas who considered 'royal blood' only worthy of mixing with each other. It is interesting to note that despite consanguinity with sisters among the Pharaohs, no ocular defect was recorded until the 18<sup>th</sup> generation <sup>24</sup>. In Biblical heritage, Abraham's first wife, Sarah, was his step sister.

### 12.3 Invisible Consanguinity

#### 12.3.1 Genetic Non-Relatedness

Consanguinity and genetic non-relatedness cannot be sharply distinguished from one another. There are many people descended from common ancestors who are unaware of the fact that they are related. In most geographical areas, man does not reproduce within pedigrees which are completely isolated from one another but, rather in a network of relationships which joins all, or most strains together in a single reproductive unit. This is the case even where branches of a society seem to be separated. Over generations, prohibitions and barriers to intermarriage break down, particularly as both legitimate and illegitimate unions lead equally to an interchange of genes.

That being the case, tracing of pedigrees of any group of apparently unrelated individuals of similar territorial origin will reveal that many of them possess a common ancestor. Therefore, if two such people, who are apparently unrelated, marry, they do contract a distant consanguineous marriage <sup>19</sup>.

### **12.3.2 Social and Geographical Isolation**

In addition to this form of consanguinity, is that where groups of people live in small isolated pockets which results in social isolation. Under these circumstances, the group from which a mate must be selected is small, therefore the proportion of first cousins in it is higher than in larger populations and random mating will more often result in consanguinity. This has been demonstrated in varying degrees of consanguinity for such isolated groups e.g. in an alpine community in Switzerland (11.5%), a district in Northern Sweden (6.8%) and a Brazilian group (20%)<sup>19</sup>. Another example of genetic isolates are the population of Newfoundland and Labrador <sup>20-22</sup> where inbreeding is common, and also in islands such as Cyprus where the whole island has become practically one large family despite the fact the first cousin marriages are forbidden by the Greek Orthodox Church <sup>23</sup>. In the former, it was suggested that the continuing high prevalence of genetic blindness in this province could be attributed to the genetic structure of the population, which derives from natural increase by settlers who arrived from highly circumscribed areas of Southwest England and Southern Ireland before 1835 and there is very little migration into these communities <sup>22</sup>. Thus, frequencies of specific recessive disorders may be increased owing to inbreeding from mating between distant relatives and the frequency of a recessive disorder is reasonably attributed to 'founder effect'; a finding similar to Fraser's group of sex-linked myopia in South Australia <sup>102</sup>.

### **12.4 Prevalence Of Consanguinity**

Consanguineous marriage remains common in many parts of the world <sup>26</sup> and has been reported in various communities such as the Mormons <sup>27</sup>. It is especially common in most of the Middle-Eastern countries where the custom is considered socially acceptable <sup>28-36,38-39,57</sup>. The same applies to other Muslim countries and regions such as India <sup>41</sup>, Pakistan <sup>37, 547, 548, 549, 552</sup>

and Uzbekistan <sup>43</sup>, and communities in the West such as the Pakistani community in the UK <sup>42</sup>.

## **12.5 Consanguinity And Society**

### **12.5.1 Social Acceptance Of Consanguinity**

The practice of consanguinity is usually cultural rather than religious and this was demonstrated by Tirosh <sup>44</sup> who studied the practice in his survey of infants with visual deficits in Northern Israel. He found consanguinity in three of the ethnic groups studied. The rates recorded were Druze (85%), Christian (77%) and Muslim (72%) thus demonstrating that religious observances in some countries were outweighed by cultural observances in others. Zlotogora <sup>31</sup> found that first cousin marriage has decreased from 75% to 44% when analysed by 20 years periods in a Muslim Village in Israel. Table 22-17 demonstrates the frequency of consanguinity in various parts of the world.

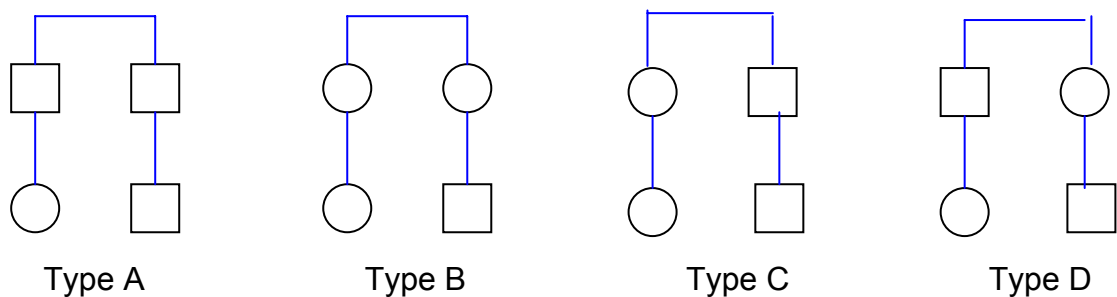
In the rest of the ME, consanguinity has been reported with the highest frequency in Saudi Arabia <sup>38</sup> where it reaches 80% of marriages in certain parts of the Kingdom. The available figures for other countries in the Middle East and the rest of the world are shown in Table 22-17. Average figures ranges between 59% among the Iraqis <sup>34</sup>, 40% among the Palestinians <sup>29</sup>, 44% among the Yemenis in Sana'a <sup>35</sup>, 49-58% among the Jordanian <sup>32-33,114</sup> and 40-54% in the UAE <sup>46</sup>. In Kuwait <sup>108</sup>, high rates of consanguineous marriages within the various Arab communities, with low frequency of intermarriage between them, and also the presence of genetic isolates and semi-isolates in some extended families and Bedouin tribes have been described. Consanguinity is less common in North African Arab countries where it was reported to be 29% in Egypt <sup>36</sup>, however, in another study on the Nubean population <sup>57</sup> in southern Egypt the figures ranged between 41.5-45.5%. The highest rates of such marriages have been reported in rural areas, among individuals with low educational level, and among the poorest.

In Morocco <sup>47</sup>, however, with its contact with the outside world, a marked decrease in consanguineous union is reported and consanguinity is disappearing and does not present a preoccupying problem for public health. However, this cannot be used as a generalisation as the trend has increased in younger generation in other Arab countries such as the UAE where the

rate of consanguinity has risen from 39% in the parent generations to 50.5% in the current generation <sup>46</sup> (Table 22-17).

### 12.5.2 Subtypes Of First Cousin Marriage<sup>19</sup> (Table 12-1)

The first cousin marriage can be subdivided into 4 types according to the relationship of the parents. These are classified according to the sex of the couple's parents who are sibs as follows:



**Figure 12-1: Subtypes of consanguineous marriages**

Adopted from Stern C. Consanguinity. In: Principles of Human Genetics <sup>19</sup>

#### Type A Cousin Marriage

This is the marriage between the siblings of two brothers.

#### Type B Cousin Marriage

This is the marriage of two siblings of two sisters. It is the second most popular form of cousin marriages.

#### Type C Cousin Marriage

This is the marriage of two cousins where the male is the sibling of the brother and the female is the sibling of the sister.

#### Type D Cousin Marriage

This is the marriage of two cousins where the male is the sibling of the sister and the female is the sibling of the brother.

### 12.5.3 Cultural Difference And Consanguinity

In some communities particular emphasis is placed upon certain forms of relationship such a type A of cousin marriage which is the commonest type of cousin marriage, the highest being in Yemen. It is considered

as the duty for the male to marry his cousin and an obligation for the female to accept <sup>19</sup> (Table 22-17). In Palestine, 20% of marriages were between first cousins <sup>31</sup>. The same pattern was reported in Pakistan where 60% of marriages are between first and second cousins <sup>37</sup>. Type B is second commonest and it is believed that such marriages do not constitute a close family marriage as the sisters are from different family from that of the male cousin. In the Jewish society marriage between maternal uncle and niece was also practised <sup>10</sup>. This practice has parallels with a Chinese regulation whereby marriage between the children of a brother and sister or of two sisters is acceptable as a consequence of the social custom which assigned a woman to the family of her husband and thus regarded children as 'not belonging' to the biological family of the mother. However, the children of two brothers were considered to be of the same family and were therefore prohibited from marrying, despite that their genetic endowment is the same <sup>19</sup>. Similar cultural practices and myth regarding the marriage between the children of a brother and sister or of two sisters exist in the Middle Eastern communities, but marriages between the children of two brothers are favoured as mentioned above <sup>19</sup>.

#### **12.5.4 Changing Trends and Non-Acceptance**

Many societies now prohibit marriage between close relatives, perhaps as a result of the observations on the progeny of consanguineous marriages. These prohibitions vary in degree and in some countries include marriages between second cousins. In addition to national laws governing marriage between relatives, there are also religious laws that decree the levels of consanguinity allowed <sup>19</sup>.

In addition to marriage between relatives, it is also custom, or law, for marriages between unrelated persons to be prohibited. These have included marriage between a person and his/her step-parent or between a person and his/her deceased uncle's/aunt's spouse. These prohibitions are based either on biological misconceptions or on non-biological grounds <sup>19</sup>.

Inbreeding is becoming less common world wide except among genetic isolates and regions where consanguinity is practiced. In Europe the change started from the beginning of the last century. In Norway this became

evident from the 1920s onward <sup>25</sup>.

Even in social isolates, the fear of unhealthy offspring may also limit consanguineous marriages as found in the Swiss group above which had a lower frequency of cousin marriage (0.7%) as opposed to the Swedish group (6.8%) although the size of the two groups were similar <sup>19</sup>. This would seem to demonstrate that certain communities show a particular aversion to such marriages even within their isolated communities.

In Japan <sup>48</sup>, the trend of consanguineous marriage has been declining and was found to be 8 times lower in younger groups than the oldest groups. The overall rate of first cousin marriage in 1975 was reported at 2.23% in rural districts and 1.77 in urban areas. No consanguineous marriage was recorded in 32% of the 253 administrative units surveyed (There was also inter-regional variation with the Kyushu district showing the highest figures of inbreeding). Similarly, low consanguinity rates are found in the European countries <sup>25, 60</sup> among the indigenous communities.

In the Arab world, a display of the presence of consanguinity amongst the various categories of visual loss have been demonstrated in a recent retrospective study in Saudi Arabia <sup>715</sup> (Table 22-17).

### 12.6.1 Prevalence of Consanguinity

The available figures on consanguinity rates among the general population in the West Bank and Gaza Strip are those of Pederson <sup>53</sup> and Saunders <sup>14</sup>. The latter studies the rate of consanguinity among Palestinians in all the refugee camps in the Gaza Strip <sup>14</sup>. Saunders analysed data according to the status of the household and professional status. The frequency of consanguinity ranged between 53% to 69% (Table 12-1) and is not related to the professional status of the person as figures were very similar in the various categories ranging from 58% to 63% (Table 22-17). This shows its prevalence in this society.

Status	First Cousin	Not First Cousin	Totals
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<b>By Professional Status</b>			
Professional	<b>63</b>	<b>37</b>	908
Student	<b>60</b>	<b>40</b>	127
Skilled worker	<b>56</b>	<b>44</b>	1023
Labourer	<b>57</b>	<b>43</b>	5317
Not working	<b>58</b>	<b>42</b>	10775
Total	<b>50</b>	<b>41</b>	18150
<b>By Household Status</b>			
Head of the HH	<b>50</b>	<b>41</b>	6207
Wife	<b>59.5</b>	<b>40.5</b>	6409
Child of HH	<b>55</b>	<b>45</b>	2434
Parent of HH	<b>69</b>	<b>31</b>	170
Relation of HH	<b>53</b>	<b>47</b>	3000
Total			

Percentage in bold

**Table 12-2: First cousin marriage in camps**

(Adopted from Saunders CA) <sup>14</sup>

### 12.6.2 Why Consanguinity?

Several factors contribute to consanguinity. These are economic and cultural factors, encounter, social and cultural isolation. The reasons given by Bedouin for favouring cousin marriages were clan solidarity, interpersonal compatibility, preservation of family property, parental authority, and social protection for women <sup>56</sup>. Bener <sup>107</sup> found in Al-Ain, UAE that the only significant variable were the husband's education and parent's consanguinity. The frequency of consanguinity in this region was found to increase significantly with the improvement of the husband's educational level; however, this does not seem the case in other regions such as Jordan <sup>114</sup>.

### 12.7 Deleterious Effects Of Consanguinity

The deleterious effects of consanguineous mating are high and predispose the offspring to the effects of recessively inherited disease. This is

principally due to the frequency with which recessive genes exist over dominant genes within the population<sup>49</sup>. The frequency of malformations was found to parallel the degree of consanguinity<sup>60</sup>.

Higher proportions of first cousin marriage than the proportion in the general population have been reported in schizophrenia<sup>50</sup>, several congenital heart defects such as septal defects (atrial, ventricular and atrio-ventricular). Also reported are pulmonary stenosis and atresia<sup>51</sup>, cystic fibrosis, cystinosis, nephronophthisis, spinal muscular atrophy, albinism, achromatopsia<sup>52</sup>, hearing disorders<sup>110</sup>, and central nervous system anomalies<sup>105</sup>, congenital anomalies, physical handicap, mental retardation and malignancies<sup>106</sup>. In Kuwait, higher incidence of Meckel syndromes, phenylketonuria, and familial Mediterranean fever have been documented<sup>108</sup>.

There is also an added risk of infant and child mortality<sup>53</sup>. Khlal and Khoury<sup>54</sup> found that inbreeding generally increases prereproductive mortality; crude mortality increases with inbreeding in proportion to the mortality rate. Morbidity increases significantly with inbreeding. Tuncbilek & Koc<sup>55</sup> reported that infant and under 5 mortality is higher in first cousin marriage in Turkey. Consanguinity can also influence the age of menopause<sup>109</sup>.

## **12.8 Genetic Counselling**

The option of premarital carrier matching has been found to be acceptable with the Bedouin Muslim community in Israel. This is a form of genetic counselling where two individuals are told, if both are carriers, that they have a 25% risk of each pregnancy of having a child affected by the disease for which they are tested. If one individual is a carrier this information is not disclosed. This method also is supposed to reduce stigmatisation, especially of women<sup>56</sup>.