Ascertainment of consanguinity, neonatal outcome and morbidity in the population of Haripur district

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بسم الله الرحمن الرحيم

DECLARATION

I hereby declare that the work accomplished in this thesis is the result of my own research work carried out in Human Genetics lab, Department of Animal Sciences, Quaid-i-Azam University Islamabad. The epidemiological data were collected from Haripur district of KPK. This thesis has neither published previously, nor does it contain any material from the published resources that can be considered as the violation of the international copyright law.

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Anisa Bibi

Dedication

*I dedicated this effort & work to Almighty **ALLAH** and*

*Holy Prophet **HAZRAT MUHAMMAD (PBUH).***

The greatest educationist of mankind

My Mother

For her endless support, encouragement and love. Her prayers always paved the way of success for me

My Father

Who always encourages me to move forward

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Abbreviations	Full form
IC-F	Inbreeding coefficient
FC	First cousin
DFC	Double first cousin
FCOR	First-cousin-once-removed
SC	Second cousin
FBD	Father-brother-daughter
FSD	Father-sister-daughter
MBD	Mother-brother-daughter
MSD	Mother-sister-daughter
DHSs	Demographic and health survey
HLA	Human leukocyte antigen
CM	Congenital malformation
CU	Consanguineous unions
NCU	Non-consanguineous unions
MS	Medical superintendent
OPD	Outpatient department
SCOR	Second-cousin-once-removed
DR	Distantly related
NR	Non-related
DHQ	District headquarter hospital
EPW	Ever pregnant women
NGO	Non-governmental organization
KPK	Khyber pakhtunkhwa

Abstract

Consanguinity is a complex, controversial social and health care issue which is culturally favored in North Africa, West Asia and the Middle East. In addition to migrants from these populations now residing in Europe, Australia and North America also favor cousin marriages. Consanguinity is widespread in Pakistan but its prevalence is highly variable across different populations. Haripur is an important city of Hazara division of Khyber Pukhtunkhwa (KPK) and is the fourth populous city after Mansehra, Abbottabad and Batagram districts. In order to establish various biological aspects of consanguinity and inbreeding coefficient (IC-F), the present epidemiological study was carried out in Haripur district. A total of 1,500 random married females were recruited and information on marriage types, morbidity, congenital disorders, fertility and birth outcome was gathered. The present study revealed that consanguineous unions were 56% of the total marriages, yielding an inbreeding coefficient (IC-F) of 0.0295. The first cousin unions (FC) were the most predominant type of marriages (38.4%). Among the first cousin unions, parallel cousin unions and patrilineal unions were in the majority (57.5% and 51.4%, respectively), and father-brother-daughter (FBD) type had the highest representation (34.5%). The consanguineous unions were significantly associated with certain socio-demographic variables like spouse's literacy, caste-system, family type, exchange or reciprocal marriages and subject's age at marriage. However, no association of consanguineous unions was observed with subject's origin, language, current age, literacy, occupational status, household system, marriage arrangement, and marriage year. These analyses further demonstrated that fertility and mortality were significantly higher in women who had consanguineous unions (CU) compared to the non-consanguineous (NCU) group ($p=0.0137$ and $p=0.025$, respectively). Significantly higher number of post-natal mortalities were calculated for the mothers who had CU ($p=0.001$) compared with the NCU sample. However, there were no differences in the distribution of CU and NCU samples with respect to live-birth sons and live-birth daughters. Fertility was also assessed with respect to various socio-demographic parameters. The total fertility and mortality rates were significantly associated with subject's literacy, family type and subjects' age at marriage. Furthermore, fertility, live-births, number of sons and daughters per women, mortality rate, pre-natal and post-natal mortalities were highest among consanguineous unions. The present epidemiological study also reports the congenital/hereditary disorders prevalence among the subjects. Among a variety of anomalies, neurological disorders were more prevalent (29%). The present study revealed the marked differences between the Haripur population and other populations of Pakistan with respect to the distribution of marriage types and inbreeding coefficient (IC-F). Consanguinity in Haripur was not as higher as reported for other regions of Pakistan. Further, consanguinity was significantly associated with fertility and mortality rates. This study would be helpful in determining the biosocial structure of Haripur population.

Chapter 1

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Chapter 2

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1.1 Consanguinity

The word consanguinity has the origin from two Latin words, "con" means common and “sanguineous” means blood (Joseph *et al.,* 2014). Consanguinity can be described as a marital union between male and female who come from a common ancestor (Tadmouri *et al.,* 2009; Warsy *et al.,* 2014). In medical genetics, consanguineous union (CU) means a marital union between couples related as a second cousin (SC) or closure (Bittles, 2011). An inbred marriage more frequently defined in clinical genetics is a combination of a couple having inbreeding coefficient IC-F ≥0.0156 in their progeny (Bittles, 2001b).

1.2 Coefficient of inbreeding (IC-F) and types of cousin marriages

A measure of the ratio of loci at which inbred union is anticipated to receive similar gene copy from both parents is specified by the coefficient of inbreeding or IC-F. This comprises union designated as first cousin (FC), first-cousins-once-removed (FCOR) and second cousins. In several populations such as Arabs who practice marital union between double first cousins (DFC) and the people of South India who practice uncle-niece marriage having a maximum level of inbreeding coefficient where IC-F reaches 0.125 (Hamamy *et al.,* 2011). Pedigrees with complex consanguinity loops due to inbred unions in successive generations come across in several inbred communities leading to higher inbreeding coefficient. The mean coefficient of inbreeding IC-F and the rates of first cousins marriages are two best variables for the comparison of consanguinity rates among different populations (Hamamy, 2012).

Five types of cousin marriages exist throughout the world named as incest, uncle-niece or aunt-nephew or double first cousins (DFC), first cousin (FC), first-cousin-once-removed (FCOR) and the second cousin (SC). Any sexual association between father-daughter, mother-son, and brother-sister are called incest. It is also deliberated as a first-degree genetic relationship with an inbreeding coefficient IC-F = 0.25. Uncle-niece or aunt-nephew or double first cousins are 2nd-degree genetic relationship with IC-F = 0.125. Islam does not permit the uncle-niece or aunt-nephew marriages. 3rd-degree relationship with IC-F = 0.0625 or first cousin marriage are not only highly predominant in the world but also are highly prevalent in Pakistan. First-cousin-once-removed is known as the 4th degree of genetic relationship with IC-F = 0.0313. FCOR can be defined as the marriage with the child of a real first cousin. The 5th and last degree of genetic relationship is the second cousin with IC-F = 0.0156. SC can be defined as the contract of marriage between the offspring of first cousins (Bittles, 2010).

1.2.1 Sub-types of the first cousin

The first cousin marriages are further subdivided into four types on the basis of the relationship of the parents. These types are classified according to the sex of couple's parents who are sibs.

The four types of 1st cousin marriages named as father-brother-daughter, father-sister-daughter, mother-brother-daughter, and mother-sister-daughter. Father-brother-daughter (FBD) and father-sister-daughter (FSD) are considered as the patriarchal marriages while mother-sister-daughter (MSD) and mother-brother-daughter (MBD) are admitted as matriarchal marriages. In contrast to this, father-

brother-daughter (FBD) and mother-sister-daughter (MSD) are known as parallel cousin marriages while father-sister-daughter (FSD) and mother-brother-daughter (MBD) are called as cross cousin marriages (Al-Gazali *et al.*, 1997).

1.3 Consanguinity through the history

Consanguinity has been observed legally at different periods of times in history by different societies. The members of the ruling empire of prehistoric Egypt and Incas practiced marriage between brother and sister who considered 'regal or royal blood' merely virtuous of mingling with each other. In Biblical heritage, Abraham's first wife, Sarah, was his step sister. Charles Darwin was married to his first cousin Emma Wedgwood. In the end of 1940s, Schull and Neel studied the consequences of consanguineous marriages/inbreeding in the two major cities of Japan named Nagasaki and Hiroshima that initiated the first wide-ranging research into the effects of consanguinity in the human populations (Schull and Neel, 1972).

1.4 Worldwide prevalence of consanguinity

Cousin marriage is a popular social custom. In 20% of the total world population mostly inhabiting the North Africa, West Asia, and the Middle East, consanguineous marriages are a common trend. In addition to migrants from these populations now reside in Europe, Australia and North America also favor the close marriages. It is roughly calculated that one billion of the existing worldwide people live in societies with a fondness for cousin marriages (Hamamy, 2012). Four major global areas have been defined depending upon the geographical distribution of consanguineous union:

1: Regions where cousin marriages are <1% of total marriages, (although inbred kinship away from second cousins, IC-F <0.0156, may perhaps occur), occupied by most of Australasia, North America, and Europe.

2: Regions such as the South America, Iberian Peninsula and Japan where the consanguineous union is 1% to 10% of all marriages.

3: Regions where consanguineous unions are 20% to over 50% of all current marriages, included much of Central, West and South Asia and North Africa.

4: Regions in which the rate of consanguinity or percentage of cousin marriages remains unpredicted or doubtful, either because the data have not been reported or the authenticity of the data is not sufficient to make an estimate with any certainty. According to this classifications, the current numbers in every single group are: 1,061 million; in the regions with less than 1% consanguineous unions, 2,811 million; in the regions having 1% to 10% consanguineous unions, 991 million; in the regions with 20% to 50+% consanguineous unions and 1,064 million are with unknown consanguinity rate (Bittles, 2001a).

The highest level of consanguinity has been reported for the major part of urban Pondicherry's population of South India where 54.9% marriages were consanguineous. The mean coefficient of inbreeding both in urban Pondicherry of South India was 0.0449 Among the army families of Pakistan reported consanguinity was 71% with IC-F = 0.0414. The reported estimate for uncle-niece and first cousin marriages in South India was 20.2% and 31.3%, respectively, (Puri *et al.*, 1978). While in Pakistan the witnessed first cousin unions were 62.5% (Ahmed *et al.*, 1992). In some populated states such as Indonesia, the reported data about consanguinity are incomplete or inadequate, therefore, designated as an unidentified position according

to consanguineous unions. Actually, information is accessible demonstrating that consanguineous marriages are preferred in most of these communities (Christianson *et al.*, 1994; Kromberg and Jenkins, 1982). Information about global consanguinity rate is insufficient and required more detailed study with all parameters and factors which affects the mean value of inbreeding coefficient in different populations, as it may be claimed in Western Europe that in kingdoms like Finland, a growing history of third cousin unions (IC-F = 0.0039) might be clinically important in reserved populations (O'Brien *et al.*, 1988).

Furthermore, the reported rate of overall consanguineous unions in the United Kingdom is less than 1%, while in 0.5 million Pakistani residents in the UK the calculated consanguinity is 50% to 60+%, with the confirmation that their frequency is still growing (Bundey *et al.*, 1991; Darr and Modell, 1988). There are 10.4% people of world's population relates as second-cousin or closure. The actual consanguineous union rate in certain areas is higher than in previous generations. Despite the decline total incidence of inbred/consanguineous unions in most of the countries, possibly imitating an aging adult population that in turn raises the number of unions between relatives (Bittles, 2008; Hussain and Bittles, 2000; Table 1.1).

Table 1.1 Worldwide prevalence of consanguinity

Country	Location	IC-F	CU %age	References
Great Britain	Birmingham	-	0.2	Bundey *et al.,* 1990
U.S.A.	All U.S.A	0.0001	0.2	Freire-Maia, 1968
Mexico	Mexico City	-	0.3	Stevenson *et al.,* 1966
Argentina	All-Argentina	0.0002	0.4	Castilla *et al.,* 1991
Hungary	All Hungary	0.0002	0.5	Czeizel *et al.,* 1976
Norway	All-Norway	0.0002	0.7	Magnus *et al.,* 1985
Chile	All-Chile	0.0007	1.3	Freire-Maia, 1968
Algeria	All-Algeria	-	22.6	Benallegue and Kedji, 1984
Tunisia	North	0.0213	26.9	Riou *et al.,* 1989
All Egypt		0.0101	29.0	Hafez *et al.,* 1983
Spain	All-Spain	0.0014	4.1	Pinto-Cisternas *et al.,* 1979
Nigeria	West (Yoruba)	0.0242	51.2	Scott-Emuakpor, 1974
Sudan	Khartoum	0.0302	52.0	Saha and Sheikh, 1988

1.5 Prevalence of consanguinity in Asia

Cousin marriage is a deeply rooted trend in most of the parts of South and West Asia. Types of cousin marriages differ widely, from uncle–niece marriages mostly preferred in South India to parallel marriages favored mostly in the Middle East (Bittles, 1994). Inadequate information on the topic is available in the main steam bio-demographic literature. The existing bio-demographic data are mostly restricted to Iran fertility survey and bio-demographic and health survey (DHSs) in particular countries including Egypt, Morocco, Tunisia, Pakistan, and India. The data from selected Asian countries varied significantly in terms of the period of assessment, the nature of the studied population, e.g., household survey or antenatal clinic. The country-level data were accessible only for Pakistan and India, with smaller geographically or ethnically-limited datasets for other countries. North Indian Hindus strictly avoid close kin marriages, whereas consanguineous unions are extensively privileged in the majority of Hindu populations of the southern states of Karnataka, Andhra Pradesh and Tamil Nadu (Bittles *et al.*, 1991).

The rates of consanguineous marriages differ from one population to another due to several factors like cultural differences, particular customs and isolation of population. Even though the incidence of cousin marriages is mostly decreasing still most of the Middle Eastern Arabs have a tradition in which intermarriages, mainly between first cousins are favored. Consanguineous unions rates are 57.7% in Saudi Arabia (El-Hazmi *et al.,* 1995), 54.4% in Kuwait (Al-Awadi *et al.*, 1985) , 51% in Jordan (Khoury and Massad, 1992), 33% in Syria (Prothro and Diab, 1974) and 22–33% in Egypt (Hafez *et al.*, 1983; Table 1.2).

Table 1.2 Prevalence of consanguinity in Asia

Country	Location	CU %age	References
China	Zejiang (Han)	2.5	Zhan *et al.*, 1992
Bangladesh	Matlab	6.7	Khan, 1997
India	National	14.0	Hussain and Bittles, 2004
Indonesia	West Timor and W.Flores	17.8	Hussain and Bittles, 2004
Bangladesh	Teknaf	17.9	Khan, 1997
United Arab Emirates	Dubai	32.0	Al-Gazali *et al.*, 1997
Syria		35.4	Othman and Saadat, 2009
United Arab Emirates	Al Ain	37.4	Al-Gazali *et al.*, 1997
Iran	All Iran	38.6	Saadat *et al.*, 2004
Yemen	All Yemen	40	Jurdi and Saxena, 2003
Kuwait	All Kuwait	43.3	Al-Awadi *et al.*, 1986
Afghanistan	All Afghanistan	46.2	Saify and Saadat, 2012
United Arab Emirates	All UAE	50.5	Al-Gazali *et al.*, 1997
Pakistan	National	62.7	Ahmed *et al.*, 1992

1.6 Prevalence of consanguinity in Pakistan

Previous studies showed that overall estimate of the percentage of inbreeding was very high in Pakistan that is 63% (Ahmed *et al.,* 1992). Only a few studies are available about consanguinity rate in different rural and urban communities of Pakistan such as in Punjab (Shami *et al.*, 1989; Yaqoob *et al.*, 1993), Khyber Pakhtoonkhwa (Wahab and Ahmad, 1996), Balochistan (Mian and Mushtaq, 1994), and Kashmir (Jabeen and Malik, 2014). Epidemiological studies showed various proportions of consanguinity in different cities of Pakistan such as it is 38.8% in Lahore city (Shami, 1982), 48.9% in Sheikhupura (Shami and Iqbal, 1983; Table 1.3).

Table 1.3 Prevalence of consanguinity in Pakistan

City	IC-F	CU %age	References
War-affected territory of Bajor	0.0134	22.3	Ahmad *et al.,* 2016
Quetta	0.0217	31.6	Mian and Mushtaq, 1994
Swat (Urban)	0.0163	31.9	Wahab and Ahmad, 1996
Swat (Rural)	0.0166	37.1	Wahab and Ahmad,1996
Mianchannu	0.0236	37.8	Shami, 1983
Muridke	0.0240	41.2	Shami, 1983
Jhelum	0.0262	44.2	Shami, 1983
Lahore	0.0242	46.2	Yaqoob *et al.,* 1993
Lower Dir	0.0249	46.2	Ahmad *et al.,* 2016
Rawalpindi	0.0286	48.1	Shami and Siddiqui, 1984
Gujrat	0.0277	48.5	Shami and Hussain, 1984
Sheikhupura	0.0271	48.9	Shami and Iqbal, 1983
All-Punjab	0.028	50.3	Bittles *et al.,* 1993
Sialkot	0.0261	51.8	Bittles *et al.,* 1993
Faisalabad	0.0293	52.1	Bittles *et al.,* 1993
Sahiwal	0.0295	56.1	Bittles *et al.,* 1993
Sargodha	0.0348	56.7	Hina and Malik, 2015
Rahim Yar Khan	0.0355	58.5	Riaz *et al.,* 2016
Gujranwala	0.0323	58.9	Bittles *et al.,* 1993
Bhimber	0.0348	62.0	Jabeen and Malik, 2014
All-Pakistan	0.0332	62.7	Ahmed *et al.,* 1992

1.7 Bio-demographic aspects of consanguinity

Specific types of cousin marriage differ in various population with ethnic, religious and local or tribal customs which play an important part at local and nationwide levels. Frequently reported reasons for cousin marriages are the custom of cousin marriage in different families, the maintenance of property and family's relationship, the establishment of family bonds and economic benefits associated with wedding gift or bride prosperity expenses. The easiness of matrimonial arrangements, a strong association between the bride and her in-laws and better marriage reliability and resilience make consanguineous unions more popular (Bittles 1994; Hussain 1999).

Cousin marriages were frequently described in land-owing and ruling societies of Western Europe. The worldwide pattern of consanguinity is slightly changed, highest among underprivileged, uneducated and backward societies; whereas several land-owning families also favor cousin marriages for retaining the veracity of their country estate (Bittles, 1994).

1.8 Consanguinity, reproductive behavior, and mortality

Generally, high rates of fertility are reported for cousin marriages. The incompletely explained reasons for these findings are the younger parental age at the first birth who are cousins or close relative couple (Bittles, 1995; Bittles *et al.*, 1993). First pregnancy is delayed in a consanguineous couple, due to consanguinity females are gynecologically immature because they marry at a younger age, consequently, birth intervals are shorter and consanguineous couple have a comparatively larger duration of childbearing (Tuncbilek and Koc, 1994). Use of contraception method is less common in closely related couples (Hussain and Bittles, 1998). The social

variables affect the fertility rates in consanguineous couples, resulting in an increase of maternal reproductive span and to a lesser extent, raise the concentration of child-bearing in the mother's most fertile years.

In consanguineous unions two partners share the human leukocyte antigen (HLA) haplotypes, therefore, couples may have difficulty in inducing pregnancy. As a result, the fertility is lower in such couples (Ober *et al.,* 1992). The second reason for low fertility is the abortion or termination of pregnancy due to the expression of the lethal genes during fetal development in consanguineous couples. On the contrary, as it may be reasoned that greater genetic compatibility between mother and her fetus in consanguineous couples would reduce the unintentional sterility and prenatal deaths. Moreover, there is a strong possibility that greater reproductive outcome or fertility may be ascertained in cousin marriage as a compensatory mechanism for newborn and infant losses (Schull and Neel, 1972).

One of the harmful effects associated with the consanguineous union is the expression of rare, recessive genes inherited due to shared forefather. Higher rates of mortality can be estimated in the population where cousin marriage is a common trend due to the expression of lethal recessive genes. Genetic fitness decreases due to successive cousin marriages. It is probable that even when there is no cousin marriage, alleles which are infrequent in the populations of larger size can rapidly increase in number due to accidental genetic drift and founder effect in a breeding pool of restricted size. Inbreeding and various bio-demographic factors work together in the estimation of deaths during infancy and in early childhood. The major determining factors of premature mortality are the mothers' age greater than 20 years at the time of birth, literacy level of females and greater than 18 months gap between two births. However, even after regulating these issues, odds ratios for postneonatal,

neonatal, and infant mortality which were 1.36, 1.28 and 1.32, respectively, for the offspring's of couples associated as first cousin union had statistically significant values (Grant and Bittles, 1997).

1.9 Impact of consanguinity on morbidity

The rate of cousin marriages has been associated with increased frequency of perinatal mortality and hereditary malformation among different Pakistani communities (Ahmad, 1994). The published literature has shown that relatively high level of newborn deaths was associated with parental consanguinity (Yaqoob *et al.,* 1998). Generally, it has been found that deaths associated with consanguinity were higher during infant, neonatal and childhood periods (Bittles *et al.,* 1993).

Scientific studies show a positive correlation among parental consanguineous union and genetic defects in the progeny. Several studies have been conducted on particular birth defects such as heart defects (OMIM 600001), cleft lip and palate (OMIM 120433) and neural tube defects (OMIM 182940) have a positive relationship with inbreeding. The rate of occurrence of congenital malformations in the offspring's of consanguineous couples is double than the offspring of non-consanguineous couples. In a study conducted in Pakistan, increased rates of inbreeding were described for the patients lying within the range of major adult's diseases such as cardiovascular disorder and some common cancer (Shami *et al.,* 1989).

Morbidity rates reported by experimental studies conducted on the progeny of the first cousin indicate that offspring's of the first cousin have 1% to 4% higher morbidity level than the progeny of non-related couples (Bittles and Makov, 1988). The less common an illness, the greater the effect of the consanguineous union on its prevalence. Due to this reason, various earlier unrecognized hereditary disorders were

identified the first time for highly inbred population, and in a significant proportion of occasions the fundamental change possibly distinctive to the population (Bittles, 2001a). Data on morbidity have been less reproducible, in part reflecting the different diagnostic criteria adopted and the varying socioeconomic and environmental backgrounds of the study populations, with estimates of excess morbidity at first cousin level ranging from 0.7% to 7.5% (Bittles, 2003).

1.10 Consanguinity and hereditary/congenital disorders

Homozygosity for autosomal recessive diseases increases due to consanguinity. The estimated risk for hereditary abnormalities in the descendants of couples related as the first cousin is 2.5 times that of the overall population (Jaber *et al.,* 1998). Hydrocephaly, velo-palatine cleft, post-axial syndactyly, and certain cardiac deformities are the most common hereditary disorder. Inherited deformities have been known and documented for centuries. The study of congenital malformations is the stimulating problem in research due to the high incidence of their occurrence and overwhelming consequences they possibly have on the individual and their families. Previous studies show a significant variation in the frequency of congenital malformations in various populations, from as low such as in Japan (Imaizumi *et al.,* 1991) to as high in Taiwan (1.07% and 3%, respectively; Chen *et al.,* 1992). This wide range of conflict could be due to the different methods used in different studies

Any anomaly which exists at birth either genetic or not is known as congenital malformation (CM) is the major health problem in childhood. Treatment and recovery of children with CM are expensive and complete retrieval is usually difficult (Turnpenny and Ellard, 2011). Previously reported etiology of CM is 5% to 10%

environmental and 30% to 40% genetic causes. Among the genetic etiology, chromosomal anomaly constitutes 6%, single gene illnesses 25% and multifactorial 20-30%; however, for nearly 50% of CM the reason is yet to be identified (Rajangam and Devi, 2007). In one study, the prevalence of CM for single major abnormality and for multiple major abnormalities was 3% and 0.7%, respectively. It has also been reported that 12.3-32% of diseases that have arisen during the perinatal period are associated with congenital abnormalities (Akosy, 2001). A recessive gene gets a chance to be expressed due to cousin marriage after having remained hidden for generations. Genetic effect of cousin marriages can be investigated due to the reason that consanguineous couples have the possibility to carry the two copies of a gene that was present in single copy in the common forefather of his/her consanguineous parents. Due to this reason, consanguineous union effect the frequency of some congenital/hereditary diseases (Khlat and Khoury, 1991). Because of high consanguinity rates within the Muslim population, the incidence of CM in Islamic countries is between 10% to 45% due to high rates of consanguineous unions (Bromiker *et al.,* 2004). In advanced countries such as the United Kingdom, CM account for a significant proportion (26-34%) of perinatal mortality (Young and Clarke, 1987). Also, the prevalence of hereditary abnormalities in Denmark is almost 3% (Søgaard and Vedsted-Jakobsen, 2003).

1.11 Consanguinity and sex ratio

Consanguinity disturbs sex ratio (no. of per hundred female births), with the increase in inbreeding coefficient, it relatively decreases the sex ratio (Ansari and Sinha, 1978). On the other hand, any vital consequence of consanguineous union was not found to have an effect on sex ratio (Rao and Inbaraj, 1980). Several causes are

being enlisted for the difference in the sex ratio. Ethnicity, parent's age, the order of birth and sporadically the events which cause strain might affect dissimilarity in the sex ratio with consanguinity.

1.12 Consanguinity and modernization

According to a book "World Revolution and Family Patterns" by William Goode (1963) it has been anticipated that the prevalence of consanguineous marriage will decrease with the progress in modernization. The incidence of consanguineous unions will also be lower as the number of persons begin to select their mates independently. It has been recommended that as the learning ranks of female increases and as they became more involved in the labor force, they demand more independence in the process of selecting a mate (Cherlin, 2012). Many of the researchers have argued second to the explicit/implicit factors described by the Goode's theoretical formulations. Many of the qualified and educated men have gained an awareness to get married by their choice, instead of getting forced to have an arranged marriage with the network of immediate kinship (Khoury and Massad, 1992). Similarly, Hurd (1974) suggested that urbanization exemplifies a new way of life, with two related processes of social change: (1) a continual separation of financial production from the local background and (2) a rising financial independence of women. Current communications and mass media encourage alteration in culture by representative alternate means of living. All these aspects could lead to a disturbance of customary family patterns. Though scholars have tested the worldwide application of Goode's theory (McDonald, 1994; Morgan and Hirosima, 1983), practical support has been established for its various features. Such as, there has been a tendency towards a decrease of intermarriages in several portions

of the emerging world (Tfaily, 2005). Some studies have found that educated females have a higher probability of non-consanguineous marriages than less educated females (Casterline and El-Zeini, 2003; Jurdi and Saxena, 2003). The incidence of cousin marriages is usually lesser in city areas (Givens and Hirschman, 1994). On the other hand, it has been notorious that previous predictions of a quick decline in the total incidence of consanguineous unions have verified to be mainly incorrect (Bittles, 2001a). Though certain countries have practiced a declining trend in the incidence of marriage to biological relatives, in other countries, the prevalence of wedding to genetic relations has either remained persistent or has increased in new generations. For example, Givens and Hirschman studied a rise in the level of intermarriages in Iran between the 1950s and 1970s (Givens and Hirschman, 1994). Also opposing to beliefs based on Goode's theory, a higher or equal level of consanguineous unions has been found in urban areas of Beirut and Yemen as compared with countryside areas (Gunaid *et al.*, 2004). Some scholars have highlighted the importance of cultural factors in studying family change.

1.13 Study objectives

- To determine the prevalence of consanguineous unions and inbreeding coefficient (IC-F) in the population of Haripur district.

- To observe the association of consanguinity and fertility with various bio-demographic variables.

- Determination of neonatal outcome, child mortality and child morbidity in recruited mothers.

- To explore the health effects of consanguinity, in particular, fertility, reproductive wastage, and congenital malformation.

2.1 Sampling area: Haripur district

I belong to the Haripur district, therefore, I selected this district for data collection. Most of the localities and major towns of this district are well known to me hence, I could manage the field work. Furthermore, to the best of my knowledge, no epidemiological study of this nature has ever been conducted in Haripur. Additionally, for Haripur district, there is no published record or biomedical literature available on the parameter which I have explored in my research project.

Haripur is the important city of Hazara division of Khyber Pukhtunkhwa (KPK). After Mansehra, Abbottabad and Batagram, Haripur district is the fourth populous city. According to 2005 census estimate, Haripur district comprised one million individuals. In the west of the city, Swabi and Buner are situated, in the north at a distance of 65km is Islamabad while in south at the distance of 35km Abbottabad city is located. Haripur city is situated in a hilly plain area at an altitude of 520 m. According to 2005 census, the population of Haripur was 803,000. Only 12.0% of the total population lives in urban areas, while 88.0% people in reside rural areas (Ali and Malik, 2014).

According to the historic background, Hari Singh Nalwa (*Sikh* General) set up Haripur in 1822, and Haripur remained headquarter of Hazara until 1853. Mahraja Ranjit (*Sikh King*) had chosen the Hari Singh Nalwa as the second Nazim after the death of Amar Singh Mahjithia who was the first Nazim (Panni, 2006).

Common language spoken by the people of Haripur is Hindko. Hindko (or Hindku) is the ancient language spoken in northern Pakistan. Word “Hindko” exactly translate “Indian Mountains”, or more accurately as “Mountains of Indus country” (Anonymous, 2009).

Haripur district is a very lush valley and is famous for fruits like guavas, while Khanpur area is well known for the vast production of red blood oranges as well as for Khanpur Lake, which supplies drinking water to Islamabad and Rawalpindi. Taxila, a reservoir of ancient heritage is situated at south while, Tarbela Dam the earthwork of Pakistan's power generation, is at North. In close proximity of Haripur lies Hattar that is an industrial area.

The climate of the district Haripur characterized by relatively high temperatures and evenly distributed precipitation throughout the year. The average temperature in Haripur is 69.0°F (20.6°C). June is the warmest month with an average temperature of 89.0°F (31.7°C). January is the coolest month with 50.0°F (10°C) temperature (Hussain *et al.,* 2008).

2.2 Map of Haripur district

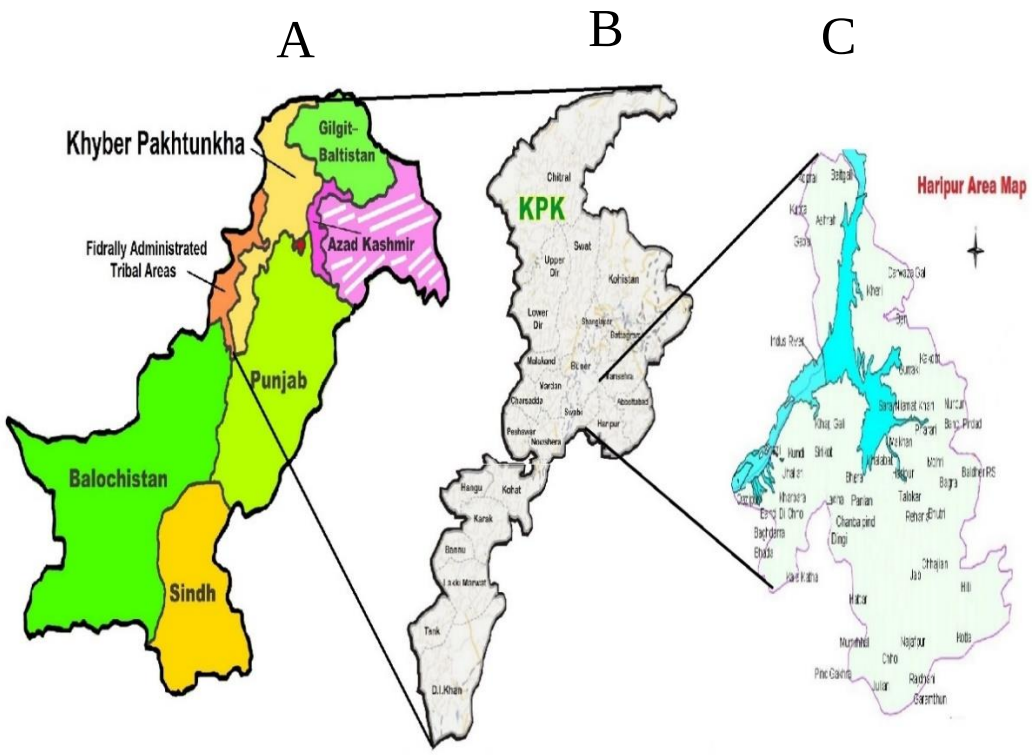

Fig. 2.1. Map of Pakistan (A), with the map of K.P.K (B), and superimposed map of Haripur district (C).

2.3 Proforma design

According to my study objectives, detailed Proforma was designed. During Proforma construction, it was kept in mind that the questionnaire should not be too much complicated and time-consuming. During Proforma designing, different parts of Proforma were modified in later stages after the response of subjects during the field work. Additional questions were added and nonessential parts were excluded. The parameter related to the bio-demographic record, socioeconomic record, marriage record, reproductive record, and disease were included (Annex I, II).

In "bio-demographic records", the parameters like subjects' name, origin, residence, rural-urban differentials, age, mother tongue, caste, (major, minor), education and a number of siblings were included (Annex I, II). Similar information was obtained for the spouse. The bio-demographic record was collected as an integral part of the questionnaire (Proforma).

In "socioeconomic record", the parameters like occupation of subject and her husband, (there were ten occupational categories for spouse namely skilled manual, unskilled manual, services, professionals, sales, agricultural, domestic services, abroad, unemployed and others; while for subject three categories namely housewife, agriculture and others were included), education, family structure (there were five categories of family; single, single-parent-and-children, nuclear, grandparents-and-one-couple and extended family system), and household system (paternal, maternal or mixed) were included.

In "marriage detail" section, the parameters such as a parental relationship, subjects' relationship with her husband were inquired. Because the core objective of the study was to determine the prevalence of consanguineous union (CU) and non-

consanguineous union (NCU), so the spousal relationship like double first cousin (DFC), first cousin (FC) were of four types, father-brothers-daughter (FBD), mother-sisters-daughter (MSD), fathers-sisters-daughter (FSD) and mothers-brothers-daughter (MBD), first-cousin-once-removed (FCOR), second cousin (SC), second-cousin-once-removed (SCOR), distantly related (DR) or *biradari* (brotherhood) and non-related (NR) were observed. Queries about the year of marriage, marriage type, (arrange marriage, self-arrange, reciprocal and forced), the age of subject at marriage, polygamy of both male and female and marriage year were also asked from the subject.

In "reproductive record" section, the information on the ever pregnancy, gap between marriage and first pregnancy, total pregnancies, live total, died total, prenatal and postnatal deaths, male live birth and female live birth, male mortality and female mortality, current pregnancy, and twins were collected.

Finally "medical record" section contained the information on acquired and congenital/hereditary diseases in the subject, husband, children, siblings of both and parents of both subject and spouse.

2.4 Arrangement of the field work

This study was approved by the ethical Review Committee of QAU. I got help from several people, before the proper start of my research work. I obtained a letter from my research supervisor mentioning objectives of my research for the purpose of data collection. I initially approached the head of the institutes and Medical Superintendents (MS) and obtained a written permission for conducting the research work in the respective institute. Sometimes this step was difficult because I was being

asked various questions about my research work. Each time I explained my research work and the study plan to the officials. For their official records, I also gave them the copy of a letter from my research supervisor. Usually, the MS were very kind and humble to accommodate my request to work in their institute for data collection. On several occasions, they provided me a working desk for an interview in general OPD for children and females.

2.5 Travels and field work

Field visits were arranged according to the accessibility of traveling facilities and a resource person. Initially, I arranged door-to-door survey but the people were totally non-supportive and rude. I filled hundred Proforma but that door-to-door survey was very difficult and time-consuming due to the non-supportive behavior of the local people, so I decided to collect data from district headquarter hospital Haripur (DHQ), where the people from all localities of the city visit. I physically visited the (DHG) on daily basis. The hospital was at one hour drive from my home. So hospital-based survey was more convenient for me as compared to the door-to-door survey.

2.6 Subjects' recruitment

Random data were collected through a descriptive epidemiological study carried out during Nov. 15, 2015 to Mar. 5, 2016. Subjects were recruited at their places of residences/work or by visiting public places like community centers and hospitals. A major part of the data was collected from District Head Quarter Hospital (DHQ) Haripur. Generally, a local resource person and lady-health-visitor went along

with the survey team from the door-to-door survey. Only the married females who were a permanent resident of the district and agreed to provide complete information were included. All the data were assimilated through face-to-face contact with the respondent.

2.7 Consent approval from the subject

After approaching each subject, I got her verbal consent for the contribution in the study. For consent approval, firstly I introduced them about myself, my task and objectives of my study. In certain instances, I had to explain them about the purpose of my field work and its potential benefits. Occasionally, I had to make them clear that the data would not be used for any other purpose except research and education, and that the data privacy and subject secrecy would be maintained.

In a number of occasions, the subject and her family raised numerous questions about this survey, before they gave formal consent approval. This was particularly the case for well-educated subjects. Certain educated subjects were very rude, ill-mannered and non-supportive. While the subjects of low socioeconomic status also denied giving information after knowing that they will not be financially supported.

2.8 Proforma filling

After the permission and formal consent approval of subject, I collected the data. I filled the questionnaires after interviewing the subject (Proforma Sheet A; Annex I). I have taken the information about the bio-demographic record,

socioeconomic record, marriage record, family type, reproductive record, twins and finally medical record accordingly. In the case when the subject or her child, sibling, or other family member were found with any genetic/hereditary anomaly then complete phenotypic information and medical record were obtained (Proforma Sheet B). Similarly, in the case of any acquired or non-genetic disease, the symptoms were recorded.

2.9 Ascertainment of subject for congenital/hereditary anomalies

All the respondents were physically examined with the help of medical officer for any congenital/ hereditary disease. Complete medical information was attained in case the subject showed any kind of dysmorphology. In each case, a representative pedigree was drawn, physical measurements were taken and photographs were obtained. Several subjects with hereditary anomalies were re-approached in a second visit in order to acquire necessary medical information.

2.10 Inclusion and exclusion criteria

All the respondents who were residents of Haripur district were included in the study. Additionally, only those subjects were included in the analyses who gave complete information on Proforma A and B. Subjects who were not the resident of Haripur district and subjects not providing complete information were excluded from the study.

2.11 Statistical analysis

Statistical analyses were carried out with the help of MS Excel, SPSS, and Graph Pad Prism software. Results were displayed through MS Excel. Once the CU were classified and tabulated, their types, prevalence, frequency, and associations were statistically examined through the χ^2 test, independent t-test and ANOVA test. Data were presented as 'number', 'types and percentage of each marriage type', 'geographic distribution of collected data', 'total percentage of cousin marriages'. The proportions (i.e., bio-demographic, geographic, etc.) were compared by making contingency tables and the χ^2 test was employed in order to find the significance of variables. The level of significance was $p<0.05$ (Kestenbaum, 2009).

IC-F was calculated by the following method:

$$\text{IC-F of specific CU type} = \frac{\text{No of specific CU type*its IC-F value}}{\text{Total marriage number}}$$

Then IC-F of each CU type was summed up to calculate total inbreeding co-efficient. Likewise, odd ratios were calculated by

$$\text{Ratio of each category} = \frac{\text{CU number}}{\text{Total marriage number}}$$

$$\text{OR} = \frac{\text{Ratio of each category}}{\text{Least ratio}}$$

3.1 Distribution of marital unions in Haripur district

Results were split into nine sections for our convenience.

- Section 3.2 and 3.2.1 described the distribution of CU, NCU and IC-F with respect to various bio-demographic and socio-economic parameters.
- Section 3.3 and 3.3.1 illustrated relative percentage of different marriage types across various bio-demographic and socio-economic parameters respectively.
- Section 3.4 and 3.4.1 described the distribution of four type of first cousin marriages with reference to different bio-demographic and socio-economics parameters correspondingly.
- Section 3.5 and 3.5.1 individually illustrated the distribution of patriarchal and matriarchal marriages among various bio-demographic and socio-economic variables.
- Section 3.6 and 3.6.1 described the distribution of parallel and cross cousin unions with reference to various bio-demographic and socio-economic variables respectively.
- Section 3.7 illustrated the distribution of inter-caste marriages.
- Section 3.8 described the fertility and mortality profile with respect to various bio-demographic and socio-economic factors.
- Section 3.9 described the pattern of genetic/congenital deformities in the Haripur population.

A total of 1,500 subjects were recruited in the present study. Out of those, 840 subjects (56%) had consanguineous unions (CU) while 660 (44%) had non-consanguineous unions (NCU). The first cousin (FC) marriages were the highest in proportion and accounted for 38% (n=576) of the total marriages (Table 3.3).

Accordingly, the proportion of double first cousin (DFC), first-cousin-once-removed (FCOR), and second cousin (SC) marriage types was 1%, 11%, and 6%, respectively. On the other hand, the proportions of second-cousin-once-removed (SCOR), distantly related (DR), and non-related (NR) were 0.4%, 28% and 16%, respectively (Table 3.3). The overall IC-F was estimated to be 0.0295 (Table 3.1).

3.2 Distribution of CU, NCU and IC-F with respect to various bio-demographic parameters

The distribution of CU was observed in various bio-demographic attributes of the subjects and their spouses. With respect to the rural/urban origin, the majority of the subjects originated from rural areas (78%), followed by subjects belonging to urban (15%) and peri-urban (7%) areas. The proposition of CU was slightly higher in the rural sample (58%) compared to the urban (52%) and peri-urban (47%) cohorts (Fig. 3.1). The differences in the distribution of CU and NCU with respect to the location were statistically not significant ($\chi^2 = 5.52$; df.2; p = 0.063; Table 3.1).

With respect to mother tongue, the majority of the subjects were Hindko speaking (92%) while there were 6% and 2% subjects speaking Pashto and other languages (Brushishki, Sindhi, Punjabi, Saraiki, Urdu, and Kohistani), respectively. The incidence of CU was higher in subjects speaking Hindko language compared to the subjects speaking Pashto and other languages (56%, 53%, and 55%, respectively; Fig. 3.2). The differences in the distribution of CU and NCU with respect to mother tongue were statistically not significant ($\chi^2 = 0.43$; df.2, p = 0.807; Table 3.1).

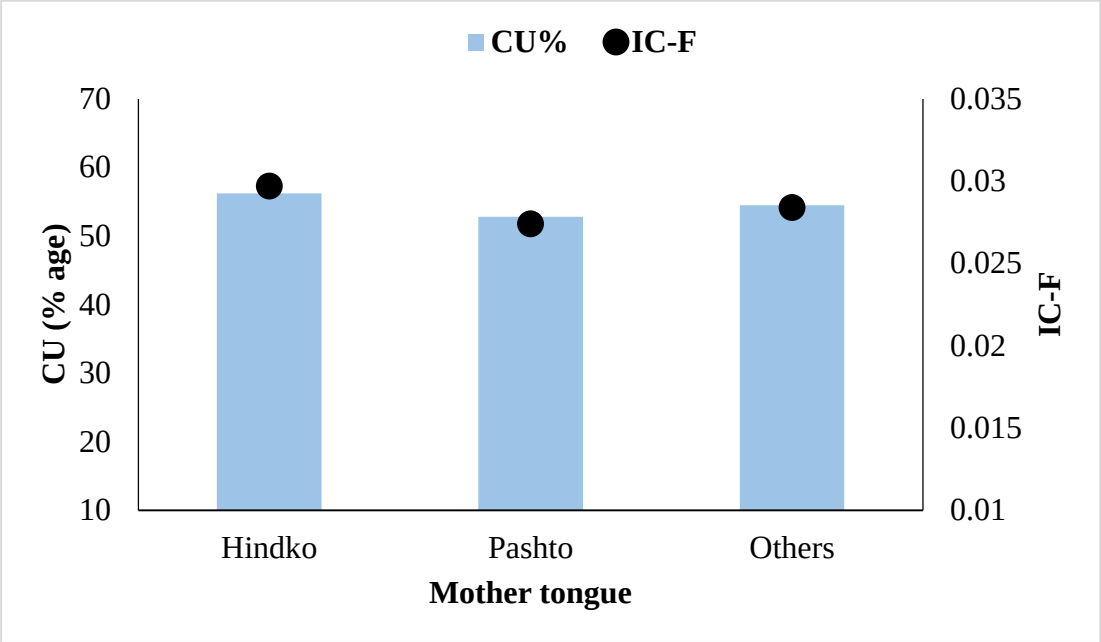

Fig. 3.1. Distribution of CU (%age) and IC-F with respect to subjects' mother tongue

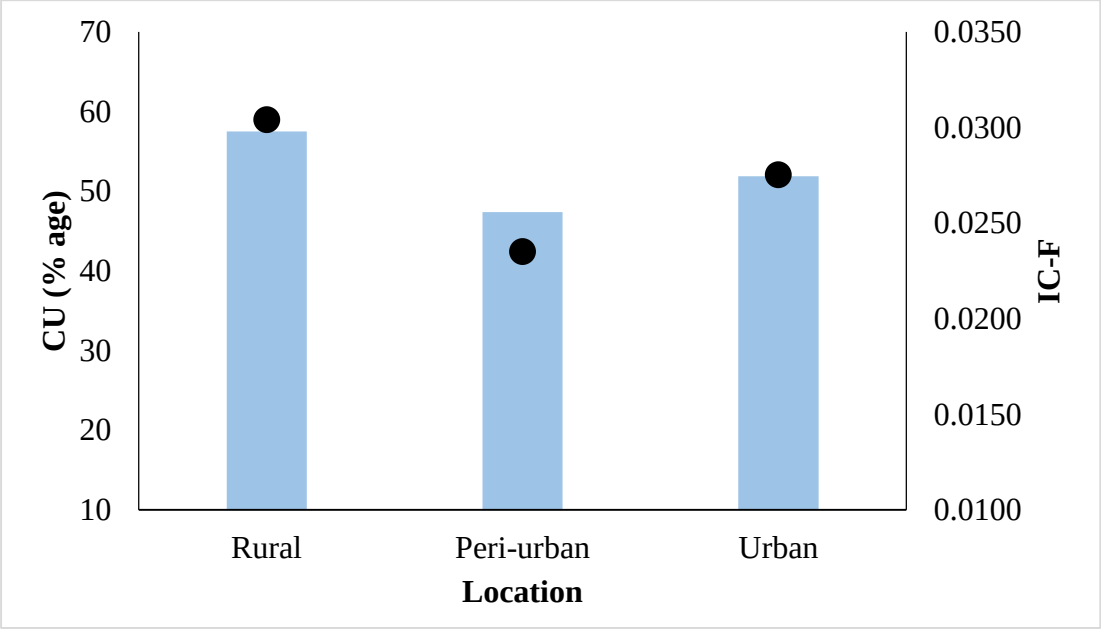

Fig. 3.2. Distribution of CU (%age) and IC-F with respect to subjects' origin

According to spouse's age, the highest incidence of CU was observed in the husbands belonging to the age-range of '>15-25' years category (61%)**,** followed by '>25-30' years (59%), '>30-35' years (56%), '>45' years (55%), '>35-40' years (54%) and '>40-45' years (50%), respectively. There was a declining trend in CU %age and IC-F with increasing the husband's age. The differences in the distribution of CU and NCU with respect to spouse's age were not significant (χ^2 = 4.68; df.5, p = 0.456; Table 3.1).

With respect to the subjects' age, the highest incidence of CU was observed in the subjects belonging to the age-range of '>40' years (63%), followed by '>25-30' years (57%), '>30-35' years (56%), '>20-25' and '>35-40' years (54%) and '>15-20' years (53%). No trend in subjects' age was observed because the difference between mean ages of subject and spouse were declining. The differences in the distribution of CU and NCU with respect to subjects' age were not significant (χ^2 = 3.22; df.6, p = 0.781; Table 3.1).

In order to check the relationship between consanguinity and education, distribution of marital unions was checked with respect to the literacy levels of the husband and wife. The majority of the husbands were literate (88%). Consanguinity was observed to be significantly higher in the literate group compared to illiterate (57% *vs.* 46%; p = 0.003). The literate group was further categorized into three categories according to years of education, i.e., primary, secondary and graduate/post-graduate groups. The proportion of CU was observed to be highest in spouses with graduate/post-graduate education (59%; Table 3.1).

With reference to subjects' literacy, the literate subjects were higher in proportion compared to illiterate subjects (74% *vs.* 26%). The CU percentage was observed to be highest in literate subjects (57%). The differences in the distribution of

CU with respect to literate and illiterate subjects were not significant (χ^2 = 0.44; df.1, p = 0.505). Furthermore, the percentage of CU was observed to be highest in subjects with secondary education (60%), while in subjects with graduate/post-graduate education highest inbreeding coefficient was noticed (IC-F = 0.033; Table 3.1).

With respect to the spouse caste-system, the largest representatives belonged to Awan community (39%), followed by Pathan, Gujjar, and Tanoli (21%, 10%, and 7%, respectively). The highest rate of CU was witnessed in Abbasi (81%), followed by Gujjar (64%), and Awan (58%) then followed by Syed (56%), and Tanoli caste-system (55%; Fig. 3.3). Within the spouse's caste-system, differences in the distribution of CU and NCU were statistically significant (χ^2 = 24.34; df.7, p = 0.807; Table 3.1).

With reference to subjects' caste-system, the largest representatives belonged to Awan community (38%), followed by Pathan, Gujjar, and Tanoli (21%, 11%, and 7%, respectively). The highest rate of CU was witnessed in Abbasi caste-system (67%), followed by Gujjar (63%), Tanoli (60%), Awan (58%), and Syed (51%; Fig. 3.4). Within the subjects' caste-system, differences in the distribution of CU and NCU were statistically significant (χ^2 = 15.17; df.7, p = 0.034; Table 3.1).

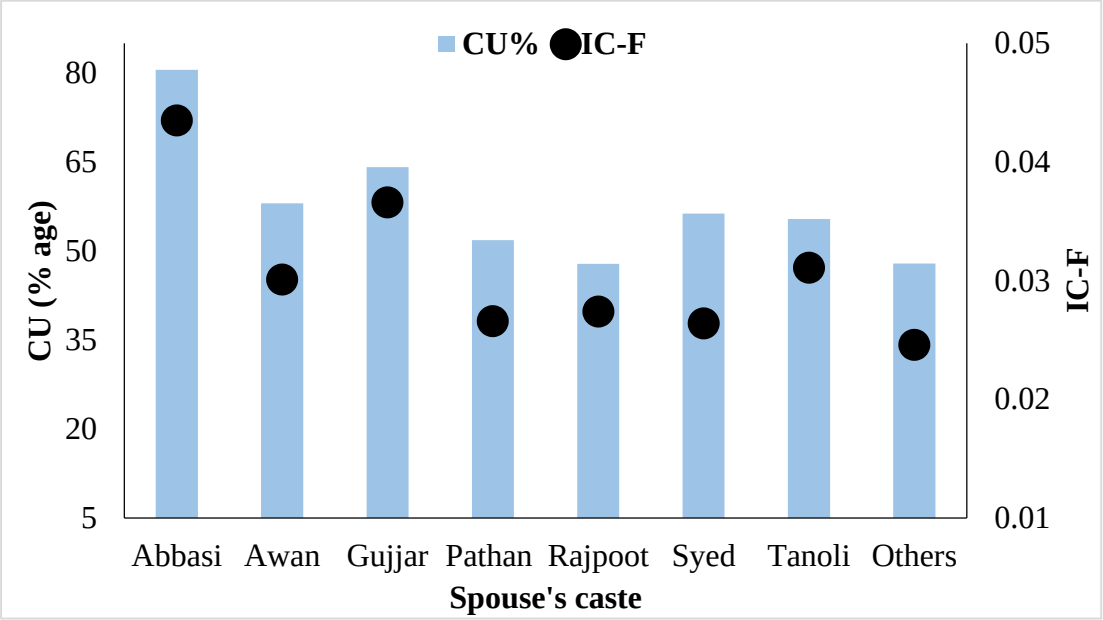

Fig. 3.3. Distribution of CU (%age) and IC-F with respect to spouse's caste-system

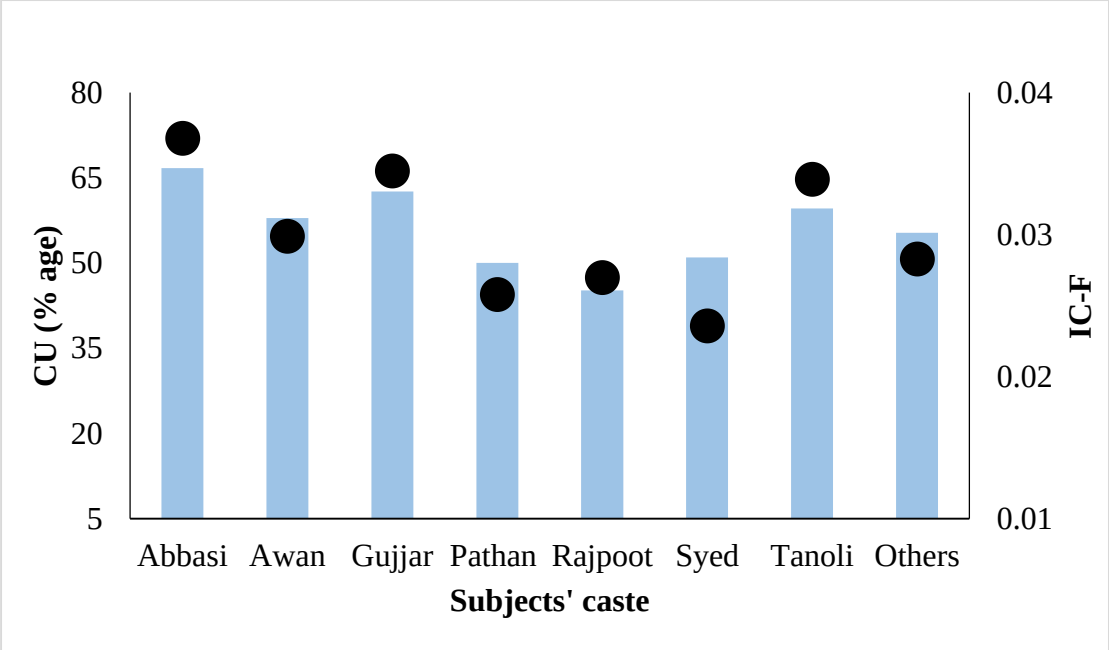

Fig. 3.4. Distribution of CU (%age) and IC-F with respect to subjects' caste-system

Table 3.1 Distribution of CU, NCU, and IC-F with respect to various bio-demographic parameters

Parameters	Consanguineous No. (%)	Non-consanguineous No (%)	Total marriages No (%)	Odds ratio	Inbreeding coefficient (IC-F)
Origin					
Rural	674 (57.5)	498 (42.5)	1,172	1.2	0.0304
Peri-urban	46 (47.4)	51 (52.6)	97	Reference	0.0235
Urban	120 (51.9)	111 (48.1)	231	1.1	0.0275
Total	**840 (56)**	**660 (44)**	**1,500**	**1.2**	**0.0295**
$\chi^2 = 5.52$; df.2, p = 0.063 (Non-Significant)					
Mother tongue					
Hindko	775 (56.2)	603 (43.8)	1,378	1.1	0.0297
Pashto	47 (52.8)	42 (47.2)	89	Reference	0.0274
Others	18 (54.5)	15 (45.5)	33	1.0	0.0284
$\chi^2 = 0.43$; df.2, p = 0.807 (Non-Significant)					
Spouse age (years)					
>15-25	78 (60.9)	50 (39.1)	128	1.2	0.0347
>25-30	222 (58.7)	156 (41.3)	378	1.2	0.0218
>30-35	219 (55.7)	174 (44.3)	393	1.1	0.0212
>35-40	167 (54.0)	142 (46.0)	309	1.1	0.0215
>40-45	65 (50.4)	64 (49.6)	129	Reference	0.0192
>45	89 (54.6)	74 (45.4)	163	1.1	0.0198
$\chi^2 = 4.68$; df.5, p = 0.456 (Non-significant)					
Subjects' age (years)					
>15-20	55 (52.9)	49 (47.1)	104	Reference	0.0204
>20-25	192 (54.4)	161 (45.6)	353	1.0	0.0211
>25-30	271 (56.9)	205 (43.1)	476	1.1	0.0216
>30-35	173 (56.0)	136 (44.0)	309	1.1	0.0215
>35-40	87 (54.4)	73 (45.6)	160	1.0	0.0207
> 40	62 (63.3)	36 (36.7)	98	1.2	0.0222
$\chi^2 = 3.22$; df.6, p = 0.781 (Non-significant)					
Spouse literacy					
Illiterate	83 (45.6)	99 (54.4)	182	Reference	0.0241
Literate	757 (57.4)	561 (42.6)	1,318	1.3	0.0303
$\chi^2 = 9.09$; df.1, p = 0.002 **(significant** between non-literate and literate-all)					
Years of education					
Primary 1-8	197 (55.6)	157 (44.4)	354	Reference	0.0287
Secondary 9-12	478 (57.9)	347 (42.1)	825	1.0	0.0308
Graduate/post-	82 (59.0)	57 (41.1)	139	1.1	0.0310

graduate 13+					
$\chi^2 = 0.69$; df.2, p = 0.710 (non-significant within literate categories)					
Subjects' literacy					
Illiterate	215 (54.6)	179 (54.6)	394	Reference	0.0297
Literate (all)	625 (56.5)	481 (43.5)	1106	1.0	0.0295
$\chi^2 = 0.44$; df.1, p = 0.505 (Non-significant between non-literate and literate group)					
Years of education					
Primary 1-8	269 (52.8)	240 (47.2)	509	Reference	0.0275
Secondary 9-12	283 (60.0)	189 (40.0)	472	1.1	0.0309
Graduate\post-graduate 13+	73 (58.4)	52 (41.6)	125	1.1	0.0325
$\chi^2 = 5.24$; df.2, p = 0.073 (Non-significant within literate categories)					
Caste-system (spouse)					
Abbasi	33 (80.5)	8 (19.5)	41	1.7	0.0435
Awan	339(58.0)	245 (42.0)	584	1.2	0.0301
Gujjar	100 (64.1)	56 (35.9)	156	1.3	0.0366
Pathan	162 (51.8)	151 (48.2)	313	1.1	0.0266
Rajpoot	33 (47.8)	36 (52.2)	69	Reference	0.0274
Syed	27 (56.3)	21 (43.8)	48	1.2	0.0264
Tanoli	56 (55.4)	45 (44.6)	101	1.2	0.0311
Others	90 (47.9)	98 (52.1)	188	1.0	0.0246
$\chi^2 = 24.34$; df.7, p = 0.001 (**Significant**)					
Caste-system(subject)					
Abbasi	32 (66.7)	16 (33.3)	48	1.5	0.0368
Awan	331 (57.9)	241 (42.1)	572	1.3	0.0299
Gujjar	107 (62.6)	64 (37.4)	171	1.4	0.0345
Pathan	158 (50)	158 (50)	316	1.1	0.0258
Rajpoot	33 (45.2)	40 (54.8)	73	Reference	0.0270
Syed	26 (51.0)	25 (49.0)	51	1.1	0.0236
Tanoli	59 (59.6)	40 (40.4)	99	1.3	0.0339
$\chi^2 = 15.17$; df.7, p = 0.034 (**Significant**)					

3.2.1 Distribution of CU, NCU and IC-F with respect to socio-economic variables

The distribution of CU and IC-F was observed in various socio-economic attributes of the subjects and their spouses. With respect to occupational groups of the spouse, ten major categories were established, while minor occupational groups were lumped into 'others' category. The CU was observed to be the highest in spouses who were unemployed (68%), while it was observed to be comparatively low in spouses with other occupational groups, i.e., professional (61%), sales (61%), doing jobs in abroad (61%), others (59%), unskilled manual (54%), skilled manual (53%), agriculture (47 %), and domestic services group (44%). The differences in the distribution of CU and NCU with respect to the occupational status of a spouse were statistically not significant (χ^2 = 15.68; df.9, p = 0.074; Table 3.2).

With respect to the occupational status of the subject, there were only three categories namely housewife, agriculture, and others. The majority of the women were housewives (90%). The proportion of CU was highest in subjects belonging to agriculture category compare to 'housewife' and 'others' categories (64% *vs.* 60% and 49%, respectively). There was statistically non-significant relationship between marriage types and the occupational status of subjects (χ^2 = 3.65; df.2, p = 0.161; Table 3.2).

Regarding the family type, there were 56% respondents having extended family structures while 44% subjects belonged to nuclear family type. The subjects with CU had a higher tendency of belonging to the extended families (61%; Fig. 3.5). The differences in the distribution of CU and NCU with respect to the family type/structure were statistically significant (χ^2 = 16.47; df.1, p<0.0001; Table 3.2).

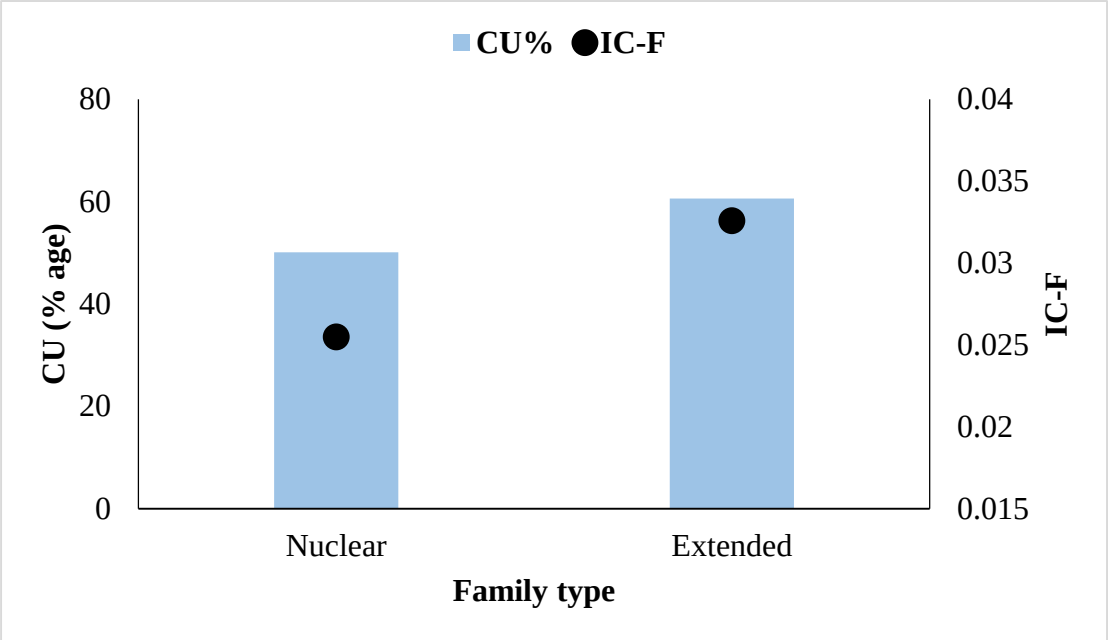

Fig. 3.5. Distribution of CU (%age) and IC-F with respect to family type

Furthermore, the data were also analyzed for the household system. Three types of the household systems were studied during field work namely maternal type, paternal type, and mixed types. The paternal household system was to be observed predominantly with 8% of the total recruited subject, while 15% and 4% of the total subjects were belonging to the mixed and maternal household systems, respectively. The subjects with CU had a higher tendency of belonging to the paternal household system (57%) however, the differences in the distribution of CU with reference to subjects' household system were statistically not significant (χ^2 = 1.51; df.2, p = 0.469; Table 3.2).

Regarding the marriage arrangements, the majority of the subjects had arranged marriages (88%) while there were 13% subjects had self-arranged/love

marriages. The differences in the distribution of CU and NCU with respect to marriage arrangement were not statistically significant (χ^2 = 2.33; df.1, p = 0.127).

The data were also analyzed for exchange/reciprocal marriages, which were observed to be 6% to the total unions. The percentage of CU in reciprocal marriages was higher as compared to non-reciprocal marriages (88% *vs.* 54%, respectively; Fig. 3.6). The distribution of CU and NCU was statistically significant with respect to reciprocal marriages (χ^2 = 38.44; df.1, p <0.0001; Table 3.2).

With respect to the age at marriage, females belonging to '> 9-14' years group had the highest percentage of CU (60%), followed by subjects within the age group of '>14-19' years (58%), '>19-25' years (57%), '>24-30' years (47%) and '>30' years (36%), respectively, (Fig. 3.7). These categories showed that as the age of females at marriage increases the percentage of CU decrease, while consanguinity proportion was higher in the younger females at the time of marriage. The difference between CU and NCU with respect to subjects' age at marriages was statistically significant (χ^2 = 11.49; df.4, p = 0.022; Table 3.2).

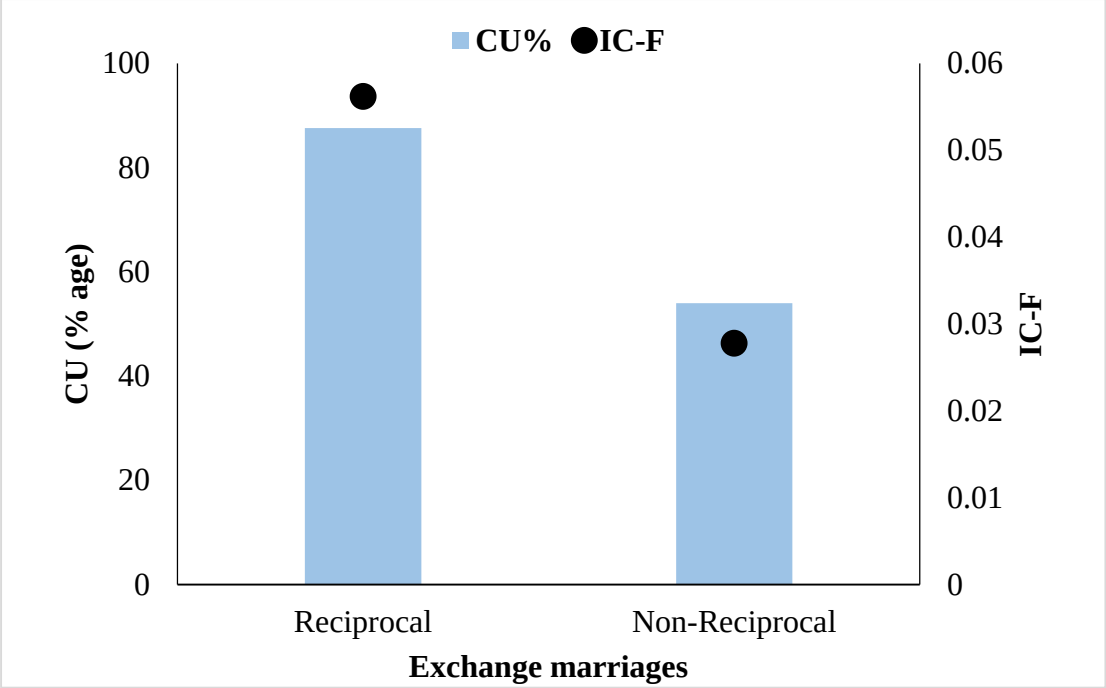

Fig. 3.6. Distribution of CU (%age) and IC-F with respect to Reciprocal marriages

Fig. 3.7. Distribution of CU (%age) and IC-F with respect to subjects' age at marriage

With respect to marriage year, the highest proportion of CU was observed in '>1982-1990' years (70%), followed by '>1998-2006' years (58%), '>1990-1998' years (58%), '>2006-2014' years (54%), '>1974-1982' years (50%) and 'up to 2015' years (49%). There was no increasing or decreasing trend of CU with respect to marriage year of the subject observed. No trend of CU with respect to marriage year was observed because the difference between mean ages of subject and spouse were declining. The differences in the distribution of CU and NCU with respect to marriage year was not significant ($\chi^2 = 8.49$; df.5, p = 0.175; Table 3.2).

3.2.2 Temporal change in age differentials between subjects and spouses

Data were also analyzed to understand the mean age difference between subjects and their spouses with respect to marriage year. Table 3.2.2 revealed the mean age of subjects and their spouses from '>1974-1982' year to 'up to 2015' year. Subject who married in '>2006-2014' were highest in number (776). The age difference between women and their husbands continuously declining from '>1974-1982' (7.4 years) to 'up to 2015' (5.5 years). No trend of increase or decrease in CU rate with respect to marriage year was observed because the mean age difference between subject and their spouse was declining (Table 3.2.1).

Table 3.2 Distribution of CU, NCU and IC-F with respect to socio-economic variables

Parameters	Consanguineous No. (%)	Non-consanguineous No. (%)	Total marriages No. (%)	Odds ratio	Inbreeding coefficient (IC-F)
Occupational status of spouse (husband)					
Abroad/Foreign	84 (60.9)	54 (39.1)	138	1.4	0.0339
Agriculture	20 (46.5)	23 (53.5)	43	1.1	0.0258
Domestic services	14 (43.8)	18 (56.3)	32	Reference	0.0244
Professional	95 (61.3)	60 (38.7)	155	1.4	0.0319
Sales	83 (61.0)	53 (39.0)	136	1.4	0.0314
Services	108 (52.7)	97 (47.3)	205	1.2	0.0277
Skilled manual	78 (52.7)	70 (47.3)	148	1.2	0.0263
Unemployed	52 (68.4)	24 (31.6)	76	1.6	0.0287
Unskilled manual	287 (53.6)	248 (46.4)	535	1.2	0.0347
Others	19 (59.4)	13 (40.6)	32	1.4	0.0303
Total	**840 (56.0)**	**660 (44.0)**	**1,500**	**1.3**	**0.0295**
	χ^2 = 15.68; df.9, p = 0.074 (Non-significant)				
Occupational status of subject (wife)					
Housewife	753 (55.9)	594 (44.1)	1,347	1.2	0.0293
Agriculture	53 (63.9)	30 (36.1)	83	1.3	0.0356
Others	34 (48.6)	36 (51.4)	70	Reference	0.0270
	χ^2 = 3.65; df.2, p = 0.161 (Non-significant)				
Family type					
Nuclear	327 (50.1)	326 (49.9)	653	Reference	0.0255
Extended	513 (60.6)	334 (39.4)	847	1.2	0.0326
	χ^2 = 16.47; df.1, P<0.0001 (**Significant**)				
Household system					
Maternal	35 (53.8)	30 (46.2)	65	1.0	0.0274
Mixed	122 (52.6)	110 (47.4)	232	Reference	0.0279
Paternal	683 (56.8)	520 (43.2)	1,203	1.1	0.0299
	χ^2 = 1.51; df.2, p = 0.469 (Non-significant)				
Marriage arrangement					
Arrange	725 (55.3)	587 (44.7)	1,312	Reference	0.0290
Self-arrange	115 (61.2)	73 (38.8)	188	1.1	0.0330
	χ^2 = 2.33; df.1, p = 0.127 (Non-significant)				
Exchange marriage					
Non-reciprocal	762 (54.0)	449 (46.4)	1,411	Reference	0.0278
Reciprocal	78 (87.6)	11 (12.4)	89	1.6	0.0562

χ^2 = 38.44; df.1, p = P<0.0001 (**Significant)**					
Subjects' age at marriage (years)					
>9-14	20 (60.0)	13 (39.4)	33	1.7	0.0313
>14-19	374 (58.3)	267 (41.7)	641	1.6	0.0310
>19-24	353 (56.7)	270 (43.3)	623	1.6	0.0298
>24-29	84 (47.2)	94 (52.8)	178	1.3	0.0241
>30	9 (36.0)	16 (64.0)	25	Reference	0.0206
χ^2 = 11.49; df.4, p = 0.022 (**Significant**)					
Marriage year					
>1974-1982	7 (50.0)	7 (50.0)	14	1.0	0.0089
>1982-1990	42 (70.0)	18 (30.0)	60	1.4	0.0249
>1990-1998	82 (57.7)	60 (42.3)	142	1.2	0.0213
>1998-2006	245 (58.1)	177 (41.9)	422	1.2	0.0218
>2006-2014	422 (54.4)	354 (45.6)	776	1.1	0.0210
Up to 2015	42 (48.8)	44 (51.2)	86	Reference	0.0194
χ^2 = 8.49; df.5, p = 0.175 (Non-significant)					

Table 3.2.1. Temporal change in age differentials between subjects and spouses

	Subject			**Spouse**			
Marriage year	**Obs**	**Mean (Year)**	**Std. Dev**	**Obs**	**Mean (Year)**	**Std. Dev**	**Difference in age**
>1974-1982	14	56.2	4.6	11	63.6	6.3	7.4
>1982-1990	60	44.8	4.1	58	52.2	6.8	7.4
>1990-1998	142	38.4	3.6	135	45.3	6.2	7.0
>1998-2006	422	32.0	3.6	421	38.1	5.8	6.1
>2006-2014	776	26.1	4.1	776	31.3	5.3	5.2
Up to 2015	86	22.8	3.9	86	28.4	5.3	5.5
Total	**1500**			**1487**			
				13 deceased			

3.3 Relative percentage of different marriage types across various bio-demographic parameters

The distribution of seven types of marriages was observed in various bio-demographic aspects of the subjects and their spouses. The percentage of DFC was observed to be highest in the subjects belonging to urban areas, while the proportion of FC, FCOR, and SCOR was noticed to be highest in females belonging to rural areas, whereas SC, DR and NR were found to be the highest in the subjects from peri-urban areas. Two major languages were studied in district Haripur during the field survey, i.e., Hindko and Pashto while other languages were very less in number so merged them for convenience. The DFC and SCOR marriages were highest in subjects speaking Hindko, while FC and NR unions were more common in subjects speaking other languages, whereas FCOR and DR were highest in Pashto speaking subjects (Table 3.3).

With respect to spouse's age, the percentage of first cousin unions was observed to be decreasing with increasing the spouse age. The proportion of DFC was noticed to be highest and equal in '>15-25' years and in '>35-40' years. FC percentage was highest in '>15-25' years. The proportion of FCOR was highest in '>35-40' years, while SC and DR proportion was noticed to be highest in '>45' years, whereas percentage of SCOR was observed to be highest in two categories '>15-25' years and in '>40-45' years, while NR proportion was highest in '>40-45' years category (Table 3.3).

According to subjects' age, the percentage of DFC and NR was observed to be highest in '>20-25' years category, while FC proportion was noticed to be highest in '>15-20' years category. The FCOR and SC proportion were observed to be highest in '>40' years category, whereas the proportion of SCOR was prevalent in '>35-40'

years category, whereas the percentage of DR was high in '>30-35' years category (Table 3.3).

According to the literacy of the spouse, DFC, FC, FCOR and SC unions were more common in the literate group of a spouse, while SCOR, DR and NR unions were found to be highest in spouses who were illiterate. The difference among the distribution of seven types of marriages with respect to the literate and illiterate groups of a spouse was significant statistically ($\chi^2 = 11.48$; df.5, p = 0.043; Table 3.3). According to the years of education, the highest proportion of DFC and SC were found in the spouse having post-secondary education, whereas FCOR and NR unions were highest in spouse belonging to primary level of education, while the proportion of FC, SCOR and DR unions were noticed to be highest in the spouse belonging to secondary level of education (Table 3.3).

With reference to subjects' literacy, the proportion of DFC, FCOR, SC and NR unions were found to be highest in the subjects who were literate, while FC, SCOR and DR unions were prevalent in the illiterate group of subjects (Table 3.3). According to subjects' years of education the proportion of DFC, FC and SCOR unions was highest in the subjects belonging to graduate/post-graduate level of education, while FCOR and SC unions was observed to be highest in subjects having secondary education, whereas DR and NR unions were found to be highest in subjects having primary education. The difference between the distribution of seven types of marriages with respect to subject's years of education was statistically significant ($\chi^2 = 20.13$; df.10, p = 0.028; Table 3.3).

With reference to caste-system of spouse, the proportion of DFC and FC unions was noticed to be highest in the spouses belonging to Abbasi caste-system, while in Syed caste-system the proportion of FCOR, SC, SCOR and DR unions was

highest, whereas the percentage of NR union was highest in spouses belonging to 'others' category. The difference among the distribution of seven types of marriages with respect to spouse caste-system was statistically significant (χ^2 = 87.69; df.35, p<0.0001; Table 3.3).

Regarding the subjects' caste-system, the highest proportion of DFC and FC unions was observed in subjects of Abbasi caste-system, while in Awan caste-system the percentage of FC union was noticed to be highest, whereas in Syed caste-system SC and SCOR unions were observed to be highest in proportion, while highest percentage of DR union was calculated in subjects belonging to Rajpoot caste-system and the maximum percentage of NR union was studied in Pathan caste-system. The difference among seven types of marriages with respect to subjects' caste-system was statistically significant (χ^2 = 59.04; df.35, p = 0.008; Table 3.3).

Table 3.3 Relative percentage of different marriage types across various bio-demographic parameters

	Consanguineous %				Non-Consanguineous %			
Parameter	**DFC**	**FC**	**FCOR**	**SC**	**SCOR**	**DR**	**NR**	**Total no**
Origin								
Rural	1.0	39.7	10.9	5.9	0.5	27.0	14.9	1,172
Peri-urban	0.0	30.9	10.3	6.2	0.0	29.9	22.7	97
Urban	1.3	35.1	10.0	5.6	0.0	28.6	19.5	231
Total	**1.0**	**38.4**	**10.7**	**5.9**	**0.4**	**27.5**	**16.1**	**1,500**
χ^2 = 11.28; df.12, p = 0.505 (Non-significant)								
Mother tongue								
Hindko	1.1	38.5	10.7	6.0	0.4	27.4	15.9	1378
Pashto	0.0	37.1	11.2	4.5	0.0	32.6	14.6	89
Others	0.0	39.4	9.1	6.1	0.0	15.2	30.3	33
χ^2 = 9.27; df.12, p = 0.679 (Non-significant)								
Spouse age (years)								
>15-25	1.6	46.9	9.4	3.1	0.8	21.1	17.2	128
>25-30	0.8	40.2	10.8	6.9	0.3	23.0	18.0	378
>30-35	1.0	38.2	10.2	6.4	0.5	30.5	13.2	393
>35-40	1.6	37.5	11.3	3.6	0.3	28.8	16.8	309
>40-45	0.0	35.7	9.3	5.4	0.8	27.9	20.9	129
>45	0.6	31.9	12.9	9.2	0.0	32.5	12.9	163
χ^2 = 33.44; df.30, p = 0.304 (Non-Significant)								
Subjects' age (years)								
>15-20	0.0	41.3	7.7	3.8	0.0	22.1	25.0	104
>20-25	1.4	36.3	11.3	5.4	0.6	28.9	16.1	353
>25-30	1.1	38.9	11.1	5.9	0.4	26.1	16.6	476
>30-35	1.0	39.5	10.4	5.2	0.0	30.7	13.3	309
>35-40	0.6	39.4	8.1	6.3	1.3	25.6	18.8	160
> 40	1.0	35.7	15.3	11.2	0.0	27.6	9.2	98
χ^2 = 31.49; df.30, p = 0.392 (Non-significant)								
Spouse literacy								
Illiterate	0.5	31.9	9.3	3.8	1.1	35.2	18.1	182
Literate	1.1	39.3	10.9	6.1	0.3	26.4	15.9	1318
χ^2 = 12.05; df.6, p = 0.061 (Non-significant)								
Years of education								
Primary 5-8 years	0.8	37.0	11.3	6.5	0.0	26.3	18.1	354
Secondary 9-12	1.1	40.2	10.8	5.8	0.5	27.0	14.5	825

years								
Graduate/post-graduate 13+	1.4	39.6	10.8	7.2	0.0	23.0	18.0	139
	χ^2 = 7.05; df.12, p = 0.855 (Non-significant)							
Subjects' literacy								
Illiterate	0.3	41.9	8.1	4.3	0.8	28.9	15.7	394
Literate	1.3	37.2	11.7	6.4	0.3	26.9	16.3	1106
	χ^2 = 12.46; df.6, p = 0.052 (Non-significant)							
Years of education								
Primary 5-8 years	1.2	34.8	10.8	6.1	0.4	27.9	18.9	509
Secondary 9-12 years	0.9	39.0	14.3	6.6	0.0	25.2	14.0	698
Graduate/post-graduate 13+	2.7	41.3	8.0	5.3	1.3	24.0	17.3	225
	χ^2 = 28.15; df.12, p = 0.005 (**Significant**)							
Caste-system (spouse)								
Abbasi	2.4	56.1	12.2	9.8	0.0	7.3	12.2	41
Awan	1.2	37.8	12.3	6.7	0.5	28.4	13.0	584
Gujjar	1.3	50.6	9.0	3.2	0.6	23.7	11.5	156
Pathan	0.6	34.2	11.5	5.4	0.3	34.5	13.4	313
Rajpoot	1.4	37.7	4.3	4.3	0.0	29.0	23.2	69
Syed	0.0	33.3	12.5	10.4	2.1	35.4	6.3	48
Tanoli	1.0	42.6	8.9	3.0	0.0	21.8	22.8	101
Others	0.5	32.4	8.5	6.4	0.0	20.7	31.4	188
	χ^2 = 95.20; df.42, P<0.0001 (**Significant**)							
Caste-system (subject)								
Abbasi	2.1	47.9	10.4	6.3	0.0	8.3	25.0	48
Awan	1.2	37.4	12.8	6.5	0.5	29.0	12.6	572
Gujjar	1.2	46.8	9.9	4.7	0.6	22.2	14.6	171
Pathan	0.6	33.2	11.1	5.1	0.3	34.5	15.2	316
Rajpoot	1.4	38.4	2.7	2.7	0.0	27.4	27.4	73
Syed	0.0	29.4	11.8	9.8	2.0	33.3	13.7	51
Tanoli	1.0	47.5	8.1	3.0	0.0	21.2	19.2	99
Others	0.6	37.6	8.8	8.2	0.0	21.8	22.9	170
	χ^2 = 68.72; df.42, p = 0.006 (**Significant**)							

3.3.1 Relative percentage of different marriage types across various socio-economic variables

In this section, the distribution of seven types of marriage was observed in various socio-economic aspects of the subjects and their spouses. With respect to the occupational status of spouse, the highest proportion of DFC and NR unions was observed in the spouses who were engaged in domestic services, while FC and SC unions were found to be highest in spouses with unskilled manual jobs, whereas highest proportion of FCOR was observed in spouses having different professions and NR unions were found to be highest in spouses belonging to sales group of occupation. Regarding the subjects' occupational status, DFC and FC unions were prevalent in subjects doing agricultural work, while FCOR, SC, SCOR and DR unions were more prevalent in housewife subjects (Table 3.4).

These data were also analyzed for family type. The DFC, FC, and SC unions were prevalent in subjects belonging to the extended family type, while FCOR, SCOR, DR and NR unions were found to be highest in subjects living in the nuclear family type. The distribution of seven types of marriages with respect to family type was statistically significant ($\chi^2 = 25.45$; df.5, $p = 0.0001$; Table 3.4).

According to household system, the proportion of FC union was observed to be the highest among subjects living with paternal household system, while DFC, FCOR, SCOR and DR types were prevalent in the respondents belonging to mix household system, whereas SC and NR unions were more prevalent among subjects with preferred maternal household system (Table 3.4).

With respect to marriage arrangement, the proportion of DFC, SC, SCOR and DR unions were found to be highest in subjects with arrange marriages while the percentage of FC, FCOR and NR unions were observed to be highest in subjects with self-arrange marriages (Table 3.4). Regarding the reciprocal marriages, the highest

percentage of DFC and FC unions was calculated in subjects with reciprocal marriages, while FCOR, SC, SCOR, DR and NR unions were prevalent in respondents with non-reciprocal marriages. The difference between the distributions of seven types of marriages with respect to reciprocal marriages was highly significant ($\chi^2 = 74.43$; dff.5, $p<0.0001$; Table 3.4).

The data were also analyzed for subjects' age at marriage, the highest proportion of DFC union was studied in '>14-19' years and in '>24-29' years categories, while FC and FCOR unions were prevalent in '>9-14' years category, whereas the SC union was prevalent in '>9-14' years and '>19-24' years categories, while SCOR was noticed to be highest in '>24-29' years category. In '>30' years category the highest proportion of DR and NR unions were studied (Table 3.4).

Regarding the marriage year, the highest proportion of DFC and FC was observed in subject married in the decade of '>1982-1990', while FCOR, SC and DR proportion was prevalent in subjects belonging to '>1974-1982' years category, whereas the proportion of SCOR was noticed to be highest in the subject who married in the decade of '>1990-1998', while NR was observed to be highest in subjects' who were married in 'up to 2015' (Table 3.4).

Table 3.4 Relative percentage of different marriage types across socio-economic variables

	Consanguineous (%)				Non-Consanguineous (%)			
Parameter	**DFC**	**FC**	**FCOR**	**SC**	**SCOR**	**DR**	**NR**	**Total no**
Occupational status of spouse (husband)								
Abroad/Foreign	0.7	47.1	9.4	3.6	0.7	24.6	13.8	138
Agriculture	2.3	30.2	11.6	2.3	0.0	37.2	16.3	43
Domestic Services	3.1	28.1	6.3	6.3	0.0	40.6	15.6	32
Professional	1.3	40.0	13.5	6.5	0.0	29.0	9.7	155
Sales	0.7	40.4	13.2	6.6	0.0	18.4	20.6	136
Services	0.0	38.5	8.8	5.4	0.0	28.3	19.0	205
Skilled Manual	0.0	34.5	12.2	6.1	0.7	29.1	17.6	148
Unemployed	1.7	36.1	9.9	6.0	0.7	28.8	16.8	76
Unskilled Manual	0.0	47.4	11.8	9.2	0.0	21.1	10.5	535
Others	0.0	40.6	12.5	6.3	0.0	25.0	15.6	32
Total	**1.0**	**38.4**	**10.7**	**5.9**	**0.4**	**27.5**	**16.1**	**1,500**
$\chi^2 = 49.33$; df.54, p = 0.655 (Non-significant)								
Occupational status of subject (wife)								
Housewife	0.9	38.2	10.8	6.1	0.4	27.8	15.9	1,347
Agriculture	2.4	45.8	9.6	6.0	0.0	24.1	12.0	83
Others	1.4	34.3	11.4	1.4	0.0	25.7	25.7	70
$\chi^2 = 12.08$; df.12, p = 0.440 (Non-significant)								
Family type								
Nuclear	0.8	32.2	11.3	5.8	0.8	30.0	19.1	653
Extended	1.2	43.2	10.3	5.9	0.1	25.5	13.8	847
$\chi^2 = 25.85$; df.6, p = 0.0002 (**Significant)**								
Household system								
Maternal	0.0	38.5	6.2	9.2	0.0	26.2	20.0	65
Mixed	1.3	35.3	11.2	4.7	0.9	29.3	17.2	232
Paternal	1.0	39.0	10.9	5.9	0.3	27.2	15.7	1,203
$\chi^2 = 7.54$; df.12, p = 0.820 (Non-significant)								

Marriage arrangement								
Arrange	1.1	37.3	10.7	6.2	0.5	28.3	16.0	1,312
Self-arrange	0.0	46.3	11.2	3.7	0.0	21.8	17.0	188
χ^2 = 10.81; df.6, p = 0.0943 (Non-significant)								
Exchange marriage								
Non-reciprocal	0.8	35.9	11.1	6.2	0.4	28.5	17.1	1,411
Reciprocal	4.5	78.7	4.5	0.0	0.0	11.2	1.1	89
χ^2 = 83.12; df.6, p<0.0001 (**Significant**)								
Subjects' age at marriage (years)								
>9-14	0.0	42.4	12.1	6.1	0.0	30.3	9.1	33
>14-19	1.1	40.6	10.9	5.8	0.5	24.3	16.8	641
>19-24	1.0	38.8	10.8	6.1	0.3	29.9	13.2	623
>24-29	1.1	29.2	11.2	5.6	0.6	28.7	23.6	178
>30	0.0	32.0	0.0	4.0	0.0	36.0	28.0	25
χ^2 = 26.75; df.24, p = 0.317 (Non-significant)								
Marriage year								
>1974-1982	0.0	14.3	21.4	14.3	0.0	50.0	0.0	14
>1982-1990	1.7	46.7	11.7	10.0	0.0	25.0	5.0	60
>1990-1998	0.7	38.7	10.6	7.7	0.7	24.6	16.9	142
>1998-2006	0.7	40.5	11.8	5.0	0.5	27.3	14.2	422
>2006-2014	1.2	37.4	10.4	5.4	0.4	28.1	17.1	776
Upto 2015	1.2	34.9	5.8	7.0	0.0	25.6	25.6	86
Total	**1.0**	**38.4**	**10.7**	**5.9**	**0.4**	**27.5**	**16.1**	**1500**
χ^2 = 31.91; df.30, p = 0.372 (Non-significant)								

3.4 Distribution of four type of first cousin marriages with reference to bio-demographic variables

In this section, the distribution of four type of first cousin unions was observed in various bio-demographic features of the subjects and their spouses. With respect to the origin of recruited subjects, the trend of FBD and MSD marriages were observed to be highest in the subjects belonging to peri-urban areas (40% and 30%, respectively), while FSD union was common in subjects belonging to urban areas (21%), while MBD union was noticed to be highest in subjects living in rural areas (27%). Regarding the mother tongue of recruited subjects, the highest proportions of FBD and MBD unions were observed in subjects speaking minor ('others') languages (46% and 31%), while FSD unions was prevalent in Pashto speaking subjects (24%), whereas MSD union was observed to be highest in Hindko speaking subjects (24%; Table 3.5)

With reference to subjects', FBD and FSD unions were prevalent in '>40-45' (54%) and in '>30-35' (21%) years categories, respectively, whereas, the proportions of MBD and MSD were observed to be highest in '>15-25' (32%) and '>25-30' (30%) years categories. Whereas among subjects, in '>40' years the percentage of FBD unions was noticed to be highest (54%), while FSD was prevalent in '>25-30' years (23%), on the other hand, the proportion of MBD and MSD was observed to be highest in '>15-20' years (33%) and '>30-35' years (26%), respectively; Table 3.5).

Regarding the literacy level of the spouse, the FBD unions were common in the illiterate group (43%), whereas the proportion of FSD, MBD and MSD was noticed to be highest in the literate group (17%, 26% and 23%, respectively). According to the education level of spouse, the FBD and MBD unions were prevalent in spouse with graduate/post-graduate education (35% and 36%, respectively). While

FSD unions were common in subjects with a secondary level of education (19%), whereas in spouses with primary education MSD union was observed to be highest in proportion (25%; Table 3.5).

With reference to subjects' literacy, the FBD and FSD unions were prevalent in the illiterate group (40% and 19%, respectively). While MBD and MSD unions were found to be highest in the literate group (28% and 24%, respectively). According to subjects' education years, FBD unions were common in subjects with primary education (38%), while FSD unions were noticed to be highest in subjects with secondary level of education (21%), whereas MBD and MSD unions were prevalent in subjects with graduate/post-graduate education (36% and 29%, respectively; Fig. 3.8). The differences among the distribution of four types of first cousin union with respect to subjects' education year were statistically significant ($\chi^2 = 20.31$; df.6, $p = 0.002$; Table 3.5).

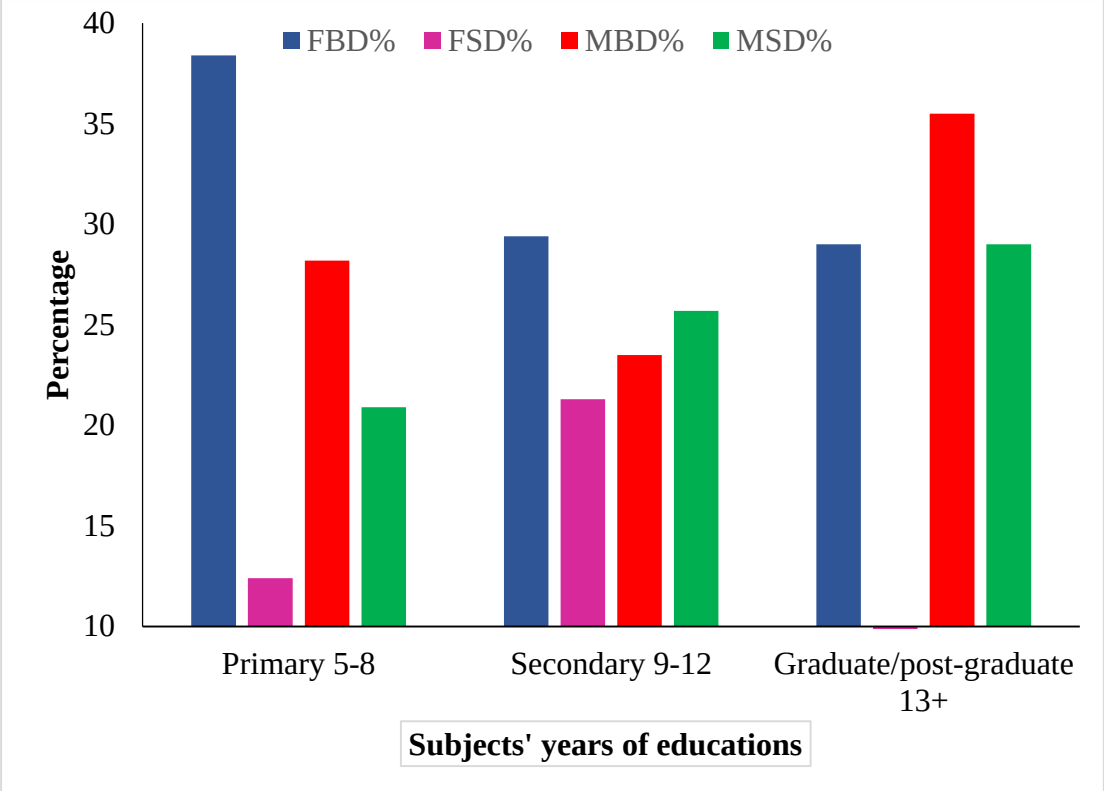

Fig. 3.8. Distribution of FC unions with respect to subjects' years of education

With respect to spouse caste-system, the trend of FBD union was found to be highest in the Abbasi caste-system (44%), while FSD union was more prevalent in Rajpoot caste-system (35%), whereas MBD union was observed to be highest in Syed caste-system (44%), while MSD union was common in Awan caste-system (29%). According to subjects' caste-system, the prevalence of FBD marriages was observed to be highest in Abbasi caste-system (44%), FSD unions were more prevalent in Rajpoot caste-system (36%), whereas MBD unions were found to be highest in Syed caste-system (47%), while MSD unions were common in Awan caste-system (29%; Table 3.5).

Table 3.5 Distribution of four type of first cousin marriages with reference to bio-demographic variables

Tehsil	FBD%	FSD%	MBD%	MSD%	Total no
Origin					
Rural	34.0	16.3	27.1	22.6	465
Peri-urban	40.0	13.3	16.7	30.0	30
Urban	35.8	21.0	21.0	22.2	81
Total	**34.5**	**16.8**	**25.7**	**22.9**	**576**
$\chi^2 = 4.16$; df.6, p = 0.656 (Non-significant)					
Mother tongue					
Hindko	33.6	16.6	25.8	24.0	530
Pashto	45.5	24.2	21.2	9.1	33
Others	46.2	7.7	30.8	15.4	13
$\chi^2 = 7.24$; df.6, p = 0.299 (Non-significant)					
Spouse age (Years)					
>15-25	28.3	18.3	31.7	21.7	60
>25-30	29.6	16.4	23.7	30.3	152
>30-35	34.0	21.3	25.3	19.3	150
>35-40	33.6	16.4	27.6	22.4	116
>40-45	54.3	8.7	17.4	19.6	46
>45	42.3	11.5	28.8	17.3	52
$\chi^2 = 20.59$; df.15, p = 0.151 (Non-significant)					
Subjects' age (years)					
>15-20	30.2	14.0	32.6	23.3	43
>20-25	32.8	13.3	28.9	25.0	128
>25-30	29.7	22.7	24.3	23.2	185
>30-35	34.4	17.2	22.1	26.2	122
>35-40	44.4	12.7	28.6	14.3	63
> 40	54.3	8.6	20.0	17.1	35
$\chi^2 = 20.42$; df.15, p = 0.156 (Non-significant)					
Spouse literacy					
Illiterate	43.1	15.5	20.7	20.7	58
Literate	33.6	17.0	26.3	23.2	518
$\chi^2 = 2.20$; df.3, p = 0.532 (Non-significant)					
Years of education					
Primary 5-8	34.4	16.0	24.4	25.2	131
Secondary 9-12	33.1	18.7	25.3	22.9	332
Graduate/post-graduate 13+	34.5	9.1	36.4	20.0	55
$\chi^2 = 5.61$; df.6, p = 0.467 (Non-significant)					

Subjects' literacy					
Illiterate	40.0	19.4	19.4	21.2	165
Literate	32.4	15.8	28.2	23.6	411
χ^2 = 6.75; df.3, p = 0.080 (Non-significant)					
Years of education					
Primary 5-8	38.4	12.4	28.2	20.9	177
Secondary 9-12	29.4	21.3	23.5	25.7	272
Graduate/post-graduate 13+	29.0	6.5	35.5	29.0	93
χ^2 = 20.31; df.6, p = 0.002 (**Significant**)					
Caste-system (spouse)					
Abbasi	43.5	13.0	26.1	17.4	23
Awan	32.1	12.2	26.7	29.0	221
Gujjar	40.5	19.0	24.1	16.5	79
Pathan	31.8	21.5	26.2	20.6	107
Rajpoot	38.5	34.6	15.4	11.5	26
Syed	25.0	18.8	43.8	12.5	16
Tanoli	37.2	18.6	23.3	20.9	43
Others	36.1	14.8	24.6	24.6	61
χ2 = 23.68; df.21, p = 0.309 (Non-significant)					
Caste-system (subject)					
Abbasi	43.5	13.0	30.4	13.0	23
Awan	33.2	10.7	27.6	28.5	214
Gujjar	40.0	20.0	21.3	18.8	80
Pathan	33.3	21.9	25.7	19.0	105
Rajpoot	35.7	35.7	17.9	10.7	28
Syed	20.0	20.0	46.7	13.3	15
Tanoli	36.2	17.0	19.1	27.7	47
Others	32.8	17.2	26.6	23.4	64
Total	**34.5**	**16.8**	**25.7**	**22.9**	**576**
χ^2 = 28.63; df.21, p = 0.123 (Non-significant)					

3.4.1 Distribution of types of first cousin unions by socio-economic variables

The distribution of four types of first cousin unions was observed in various socio-economic aspects of subjects and their spouses. According to occupational status of spouse, the proportion of FBD unions was noticed to be highest in spouses with domestic services (67%), while FSD and MBD unions were more prevalent in spouses with agricultural jobs (31% and 39%, respectively), Whereas MSD unions were observed to be highest in spouses engaged in skilled manual jobs (39%; Fig. 3.9). The difference among the distribution of four types of first cousin union with respect to spouse occupational status was statistically significant (χ^2 = 45.87; df.27, p = 0.013; Table 3.6).

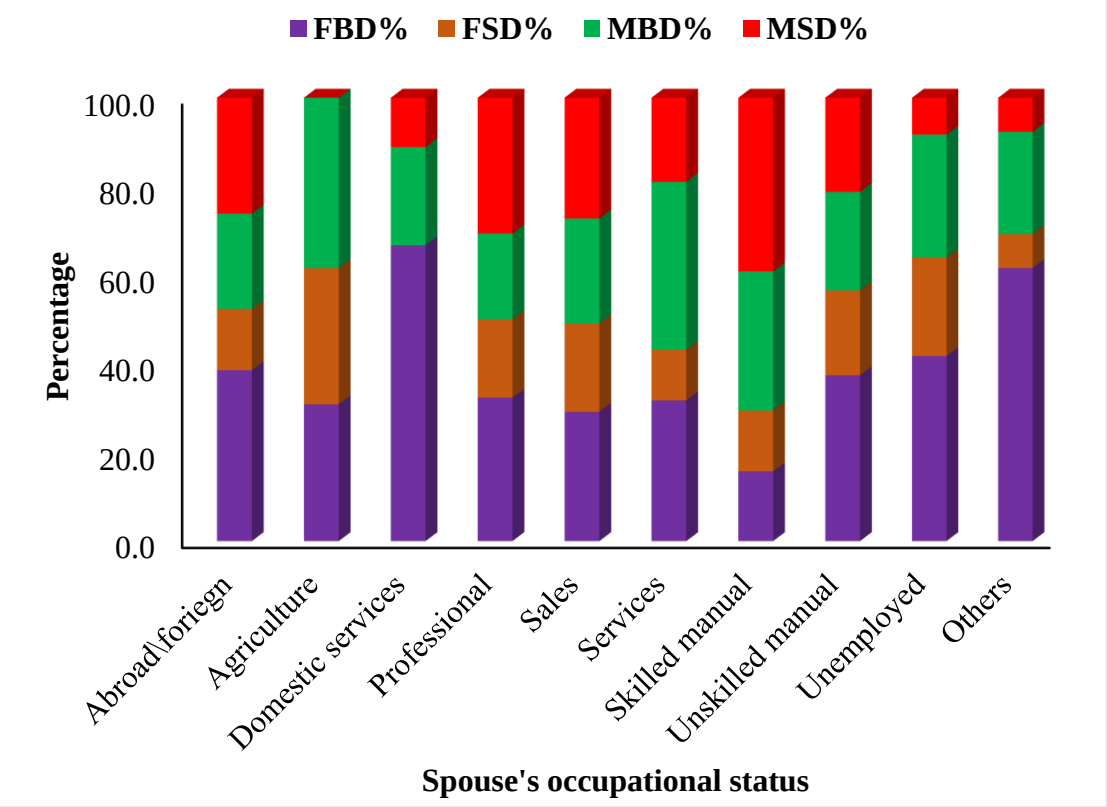

Fig. 3.9. Distribution of FC unions with respect to spouse's occupational status

With respect to subjects' occupation, the FBD and MBD unions were common in 'others' category (38% and 29%, respectively), while FSD was found to be highest in subjects who were engaged in agricultural jobs (29%), whereas MSD union was common in housewife category (23%; Table 6). Similarly, the data were also analyzed for family type, the highest proportion of FBD unions were observed in subject with nuclear family type (38%), while FSD, MBD and MSD unions were highly prevalent in subjects with extended family type (17%, 26% and 25% respectively; Table 3.6).

With respect to household system, the FBD unions were common in subjects with preferred paternal household system (35%), while FSD and MBD unions were more prevalent in subjects living in mixed household system (18% and 28%), whereas MSD unions were common in maternal household system (32%; Table 3.6).

Regarding the marriage arrangement, the FBD and MBD unions were more prevalent in subjects with arrange marriages (36% and 26%, respectively), while FSD and MSD pattern were noticed to be highest in subjects with self-arrange marriages (20% and 29%, respectively). FBD unions were observed to be highest in subjects with reciprocal marriages (49%), whereas the FSD, MBD and MSD marriages were more prevalent in subjects with non-reciprocal marriages (17%, 26%, and 24%, respectively; Table 3.6)

With reference to subjects' age at marriage, the FBD and MBD unions were more prevalent in '>9-14' years category (43%), while FSD unions were common in '>14-19' years category (17%), whereas MSD were found to be highest in '>24-29' years category (39%; Table 3.6).

According to marriage year, the FBD and FSD unions were prevalent in subjects who were married in the decade of '>1990-1998' year (51%) and in '>1998-2006' year (19%), respectively. While MBD and MSD were noticed to be highest in

subjects who were married during ‘upto 2015’ (40%) and during ‘>1974-1982’ year (100%), respectively (Table 3.6).

Table 3.6 Distribution of four types of first cousin marriages with respect to socio-economic variables

Parameters	FBD%	FSD%	MBD%	MSD%	Total no
Occupational status of spouse (husband)					
Abroad/foreign	38.5	13.8	21.5	26.2	65
Agriculture	30.8	30.8	38.5	0.0	13
Domestic services	66.7	0.0	22.2	11.1	9
Professional	32.3	17.7	19.4	30.6	62
Sales	29.1	20.0	23.6	27.3	55
Services	31.6	11.4	38.0	19.0	79
Skilled manual	15.7	13.7	31.4	39.2	51
Unskilled manual	37.3	19.2	22.3	21.2	193
Unemployed	41.7	22.2	27.8	8.3	36
Others	61.5	7.7	23.1	7.7	13
Total	**34.5**	**16.8**	**25.7**	**22.9**	**576**
χ^2 = 45.87; df.27, p = 0.013 (**Significant**)					
Occupational status of subject (wife)					
Housewife	34.2	16.3	25.5	23.9	514
Agriculture	36.8	28.9	26.3	7.9	38
Others	37.5	8.3	29.2	25.0	24
χ^2 = 8.69; df.6, p = 0.192 (Non-significant)					
Family type					
Nuclear	38.1	16.7	25.2	20.0	210
Extended	32.5	16.9	26.0	24.6	366
χ^2 = 2.46; df.3, p = 0.482 (Non-significant)					
Household system					
Maternal	28.0	8.0	32.0	32.0	25
Mixed	32.9	18.3	28.0	20.7	82
Paternal	35.2	17.1	24.9	22.8	469
χ^2 = 3.44; df.6, p = 0.752 (Non-significant)					
Marriage arrangement					
Arrange	35.6	16.4	26.2	21.9	489
Self-arrange	28.7	19.5	23.0	28.7	87
χ^2 = 3.25; df.3, p = 0.354 (Non-significant)					
Exchange marriage					
Non-reciprocal	32.6	17.2	26.1	24.1	506
Reciprocal	48.6	14.3	22.9	14.3	70
χ^2 = 7.68; df.3, p = 0.05 (Non-significant)					

Subjects' age at marriage (years)					
>9-14	42.9	0.0	42.9	14.3	14
>14-19	37.3	17.7	25.4	19.6	260
>19-24	35.1	17.4	24.4	23.1	242
>24-29	15.4	17.3	28.8	38.5	52
>30	37.5	0.0	25.0	37.5	8
$\chi^2 = 20.19$; df.12, p = 0.062(Non-significant)					
Marriage year					
>1974-1982	0.0	0.0	0.0	100.0	2
>1982-1990	50.0	7.1	28.6	14.3	28
>1990-1998	50.9	16.4	20.0	12.7	55
>1998-2006	35.1	18.7	25.7	20.5	171
>2006-2014	31.7	16.9	25.2	26.2	290
Upto 2015	16.7	16.7	40.0	26.7	30
Total	**34.5**	**16.8**	**25.7**	**22.9**	**576**
$\chi^2 = 26.92$; df.20, p = 0.138(Non-significant)					

3.5 Distribution of patriarchal and matriarchal marriages with respect to various bio-demographic variables

In this section, distribution of patriarchal and matriarchal marriages was studied in different bio-demographic aspects of subjects and their spouses. With respect to subjects' origin, the highest proportion of patriarchal marriages was noticed in urban areas (57%), while matriarchal marriages were more prevalent in rural areas (50%). According to the subjects' mother tongue, the patriarchal marriages were highly prevalent in Pashto speaking subjects (70%), while matriarchal unions were common in Hindko speaking subjects (50%; Table 3.7).

With reference to 'spouse's age', the pattern of patriarchal marriages was observed to be highest in '>40-45' years category (63%), while matriarchal unions were prevalent in '>25-30' years category (54%). Whereas according to 'subjects' age', the proportion of patriarchal unions was highest in the subjects belonging to the age category of '>40' years (63%), and matriarchal unions were common in subjects with '>15-20' years age (56%; Table 3.7).

Likewise, the data were also analyzed with respect to 'spouse's literacy'. The patriarchal marriages were observed to be highest in the illiterate group (59%), while matriarchal marriages were more prevalent in the literate group (50%), According to the education years of subject, the pattern of patriarchal marriages was observed to be highest in spouses with secondary education (52%), while matriarchal unions were more prevalent in spouses who attended the school till graduate/post-graduate level (57%).

With respect to 'subject's literacy', the highest proportion of patriarchal marriages was observed in the illiterate group (60%), while matriarchal unions were

more prevalent in the literate group (52%; Fig. 3.10). The difference between the distribution of patriarchal and matriarchal marriages with respect to subject literacy levels was observed to be statistically significant (χ^2 = 5.93; df.1, p = 0.015), according to the education years of subject, the patriarchal marriages were found to be highest in subjects with primary education (51%), while matriarchal unions were highest in proportion in subjects who attended the school till graduate/post-graduate level (65%; Fig. 3.11). There was a significant difference between patriarchal and matriarchal unions with respect to subjects' years of education (χ^2 = 7.22; df.2, p = 0.027; Table 3.7).

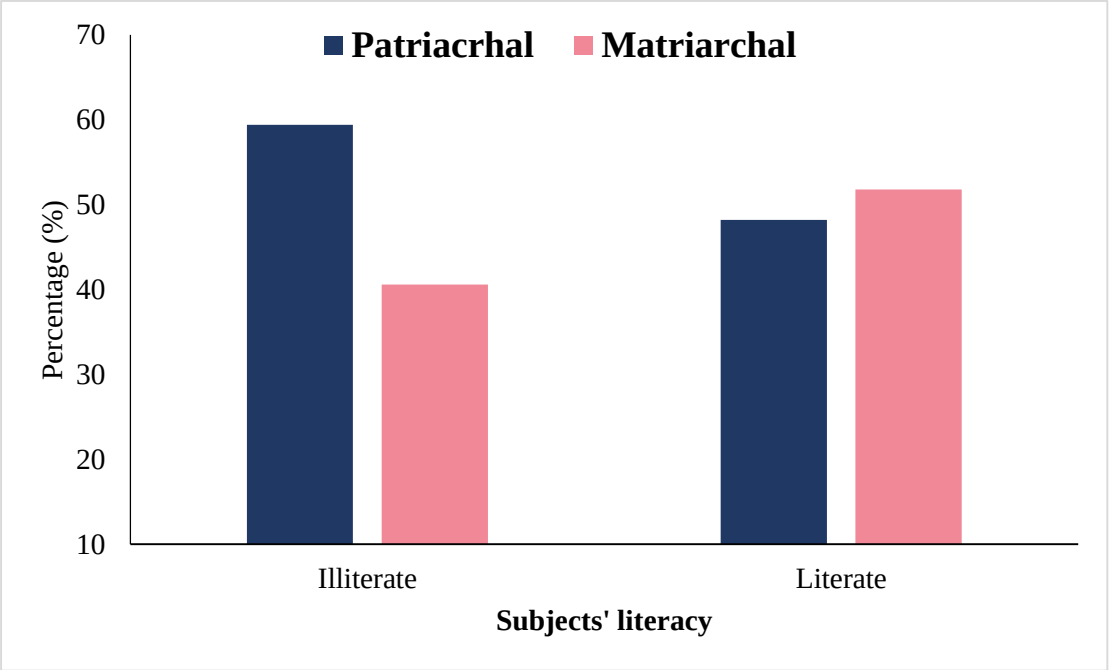

Fig. 3.10. Distribution of patriarchal and matriarchal unions with respect to subjects' literacy

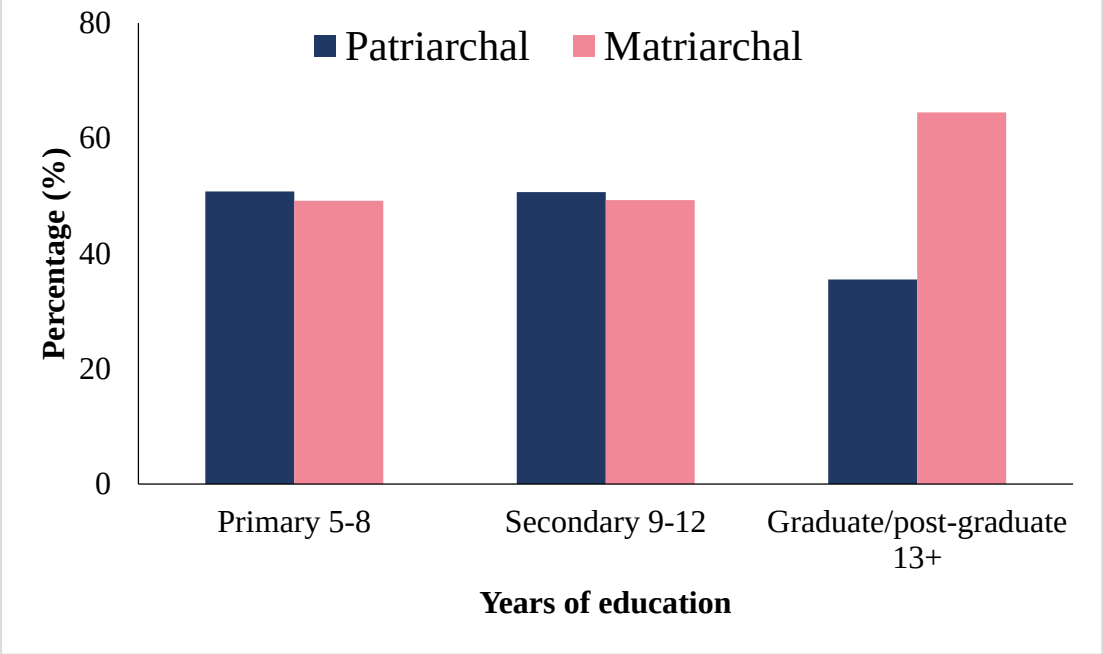

Fig. 3.11. Distribution of patriarchal and matriarchal unions with respect to subjects' years of education

Across the spouse 'caste-system', the highest proportion of patriarchal marriages was noticed in Rajpoot caste-system (73%), whereas matriarchal marriages were more prevalent in Syed caste-system (56%). Likewise, the data were also analyzed for subjects' 'caste-system', patriarchal marriages were found to be highest in subjects belonging to Rajpoot caste-system (71%), while in Syed caste-system the matriarchal marriages were highly prevalent (60%; Table 3.7).

Table 3.7 Distribution of patriarchal and matriarchal unions according to various bio-demographic parameters

Parameters	Patriarchal Total No.	%age	Matriarchal Total No.	%age	Total No.
Origin					
Rural	234	50.3	231	49.7	465
Peri-urban	16	53.3	14	46.7	30
Urban	46	56.8	35	43.2	81
Total	**296**	**51.4**	**280**	**48.6**	**576**
$\chi^2 = 1.20$; df.2, p = 0.548 (Non-significant)					
Mother tongue					
Hindko	266	50.2	264	49.8	530
Pashto	23	69.7	10	30.3	33
Others	7	53.8	6	46.2	13
$\chi^2 = 4.77$; df.2, p = 0.092 (Non-significant)					
Spouse age (Years)					
>15-25	28	46.7	32	53.3	60
>25-30	70	46.1	82	53.9	152
>30-35	83	55.3	67	44.7	150
>35-40	58	50.0	58	50.0	116
>40-45	29	63.0	17	37.0	46
>45	28	53.8	24	46.2	52
$\chi^2 = 5.92$; df.5, p = 0.314 (Non-significant)					
Subjects' age (years)					
>15-20	19	44.2	24	55.8	43
>20-25	59	46.1	69	53.9	128
>25-30	97	52.4	88	47.6	185
>30-35	63	51.6	59	48.4	122
>35-40	36	57.1	27	42.9	63
> 40	22	62.9	13	37.1	35
$\chi^2 = 5.09$; df.5, p = 0.405 (Non-significant)					
Spouse literacy					
Illiterate	34	58.6	24	41.4	58
Literate	262	50.6	256	49.4	518
$\chi^2 = 1.35$; df.1, p = 0.245 (Non-significant)					
Years of education					
Primary 5-8	66	50.4	65	49.6	131
Secondary 9-12	172	51.8	160	48.2	332
Graduate/post-graduate	24	43.6	31	56.4	55

13+					
χ^2 = 1.26; df.2, p = 0.532 (Non-significant)					
Subject literacy					
Illiterate	98	59.4	67	40.6	165
Literate	198	48.2	213	51.8	411
χ^2 = 5.93; df.1, p = 0.015 (**Significant**)					
Years of education					
Primary 5-8	90	50.8	87	49.2	177
Secondary 9-12	138	50.7	134	49.3	272
Graduate/post-graduate 13+	33	35.5	60	64.5	93
χ^2 = 7.22; df.2, p = 0.027 (**Significant**)					
Caste-system (spouse)					
Abbasi	13	56.5	10	43.5	23
Awan	98	44.3	123	55.7	221
Gujjar	47	59.5	32	40.5	79
Pathan	57	53.3	50	46.7	107
Rajpoot	19	73.1	7	26.9	26
Syed	7	43.8	9	56.3	16
Tanoli	24	55.8	19	44.2	43
Others	31	50.8	30	49.2	61
χ^2 = 12.48; df.7, p = 0.086 (Non-significant)					
Caste-system (subject)					
Abbasi	13	56.5	10	43.5	23
Awan	94	43.9	120	56.1	214
Gujjar	48	60.0	32	40.0	80
Pathan	58	55.2	47	44.8	105
Rajpoot	20	71.4	8	28.6	28
Syed	6	40.0	9	60.0	15
Tanoli	25	53.2	22	46.8	47
Others	32	50.0	32	50.0	64
χ^2 = 13.40; df.7, p = 0.063 (Non-significant)					

3.5.1 Distribution of patriarchal and matriarchal marriages among socio-economic parameters

The distributions of patriarchal and matriarchal marriages were observed in various bio-demographic features of subjects and their spouses. With reference to 'spouse occupation', the highest percentage of patriarchal and matriarchal marriages was observed in 'others' category (69%), while the least proportion of patriarchal unions was calculated in spouses engaged in skilled manual jobs (29%), the matriarchal unions were more prevalent in 'skilled manual' category (71%), and least was calculated for 'others' category (31%; Fig. 3.12). Differences in the distribution of patriarchal and matriarchal marriages with reference to spouse's occupation were statistically significant (χ^2 = 19.54; df.9, p = 0.021; Table 3.8).

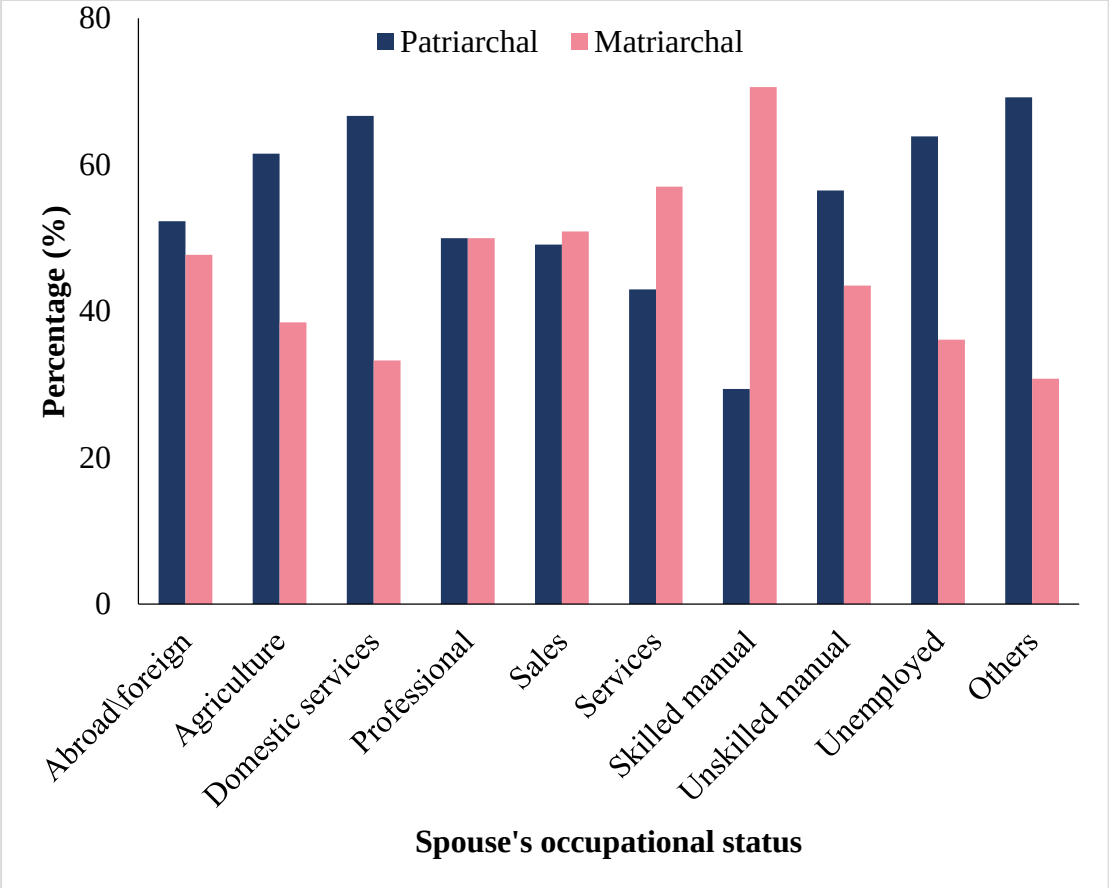

Fig. 3.12. Distribution of patriarchal and matriarchal marriages with respect to spouse's occupational status

While with reference to ‘subjects’ occupation’, the highest proportion of patriarchal unions was calculated for the subjects engaged in agricultural jobs (66%), while least proportion was observed in ‘others’ category (46%).Whereas matriarchal marriages were more prevalent in ‘others’ category (54%) and least prevalent in ‘agriculture’ category (34%; Table 3.8).

In addition, according to ‘family type’ and ‘household system’, the pattern of patriarchal marriages was found to be highest in nuclear family type (55%) and in paternal household system (52%), whereas the matriarchal union was common in extended family type (51%) and in maternal household system (64%; Table 3.8).

Furthermore, the data were also analyzed with reference to ‘marriage arrangement’. The proportion of patriarchal unions was noticed to be highest in the subjects with arranged (52%) and reciprocal marriages (63%), the highest percentage of the matriarchal union was observed in subjects with self-arrange (52%) and non-reciprocal marriage arrangement (50%; Fig. 3.13). There was a statistically significant difference between the distribution of patriarchal and matriarchal unions with reference to reciprocal marriages ($\chi^2 = 4.20$; df.1, p = 0.041; Table 3.8).

Moreover, with respect to ‘subjects’ age at marriage’, the highest rate of patriarchal union was observed in the category in ‘>14-19’ years of age (55%). Whereas the highest percentage of matriarchal marriages was calculated in ‘>24-29’ years category (67%; Fig. 3.14). The difference between patriarchal and matriarchal marriages was significant according to subject’s age at marriage ($\chi^2 = 9.78$; df.4, p = 0.044; Table 3.8).

Furthermore according to ‘marriage year’, the rate of patriarchal unions was observed to be highest during ‘>1990-1998’ (67%; Fig. 3.15) while matriarchal unions were observed to be highest in the subjects who were married during ‘>1974-1982’ (100%; Table 3.8).

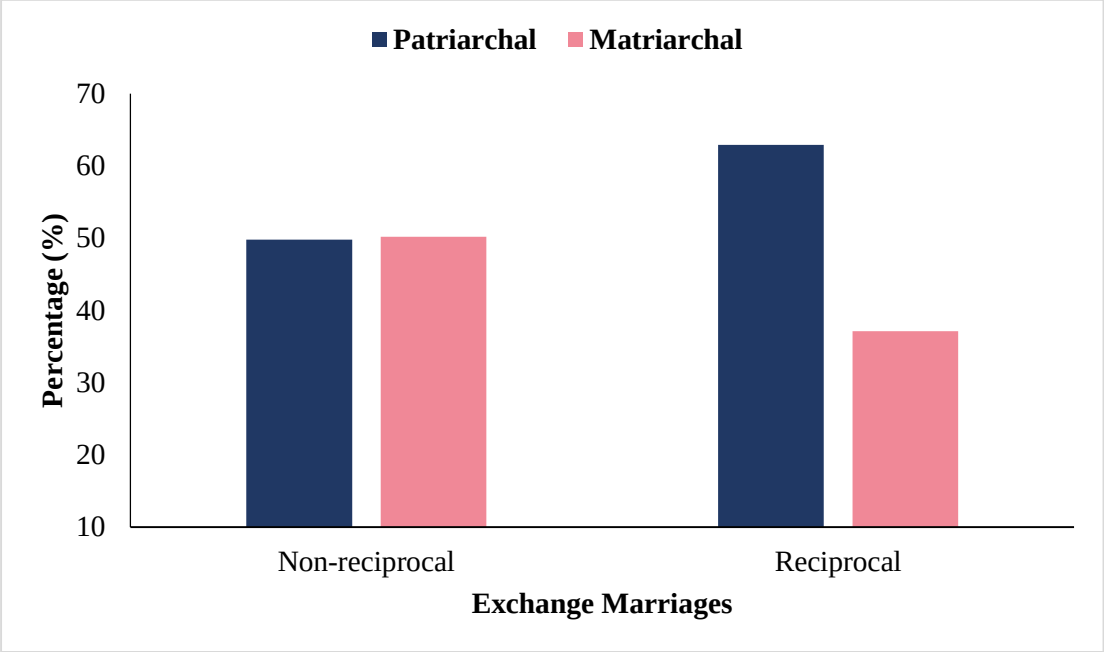

Fig. 3.13. Distribution of patriarchal and matriarchal marriages with respect to reciprocal marriages

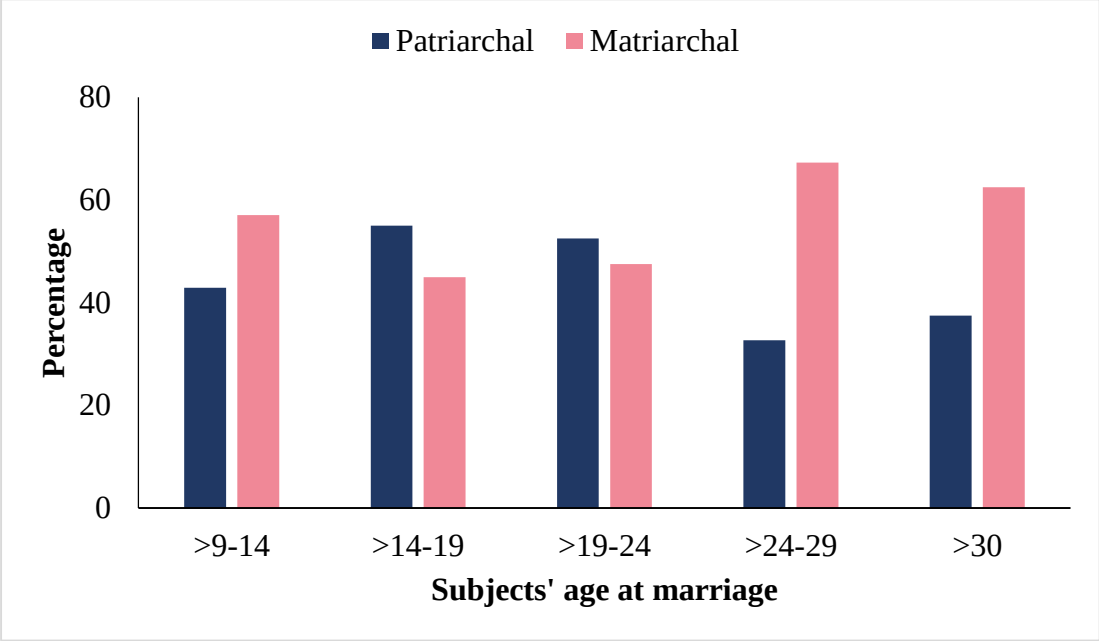

Fig. 3.14. Distribution of patriarchal and matriarchal marriages with respect to subjects' age at marriage

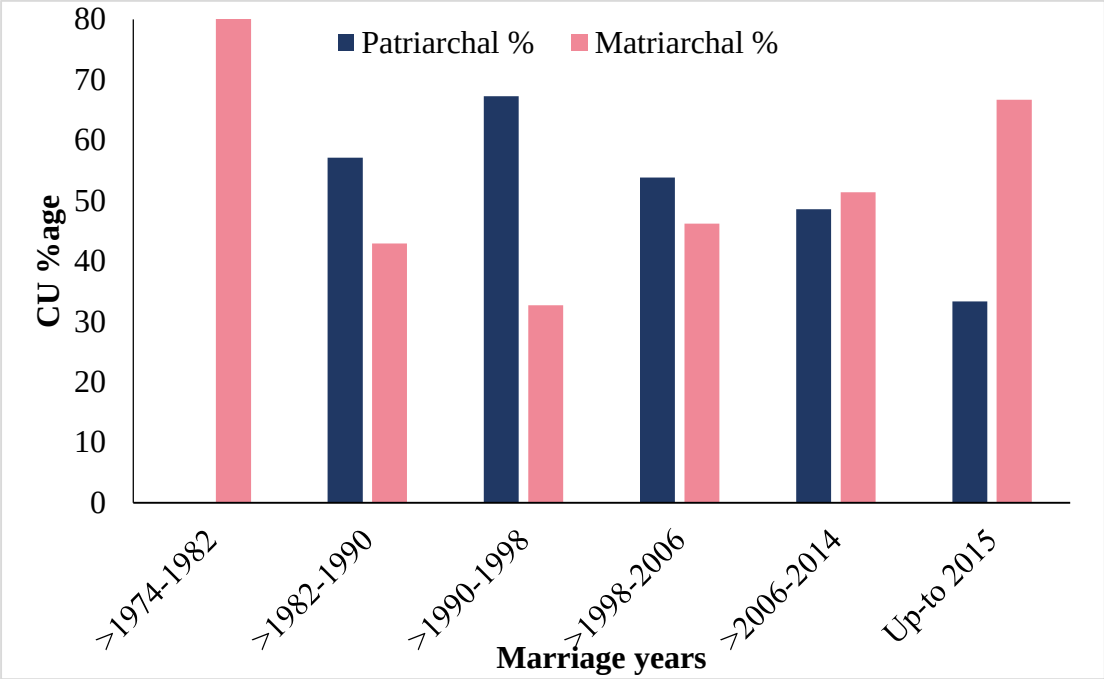

Fig. 3.15. Distribution of patriarchal and matriarchal marriages with respect to marriage year

Table 3.8 Distribution of patriarchal and matriarchal unions according to various socio-economic parameters

	Patriarchal		Matriarchal		
Parameters	**Total no**	**%age**	**Total no**	**%age**	**Total no**
Occupational status of spouse (husband)					
Abroad/Foreign	34	52.3	31	47.7	65
Agriculture	8	61.5	5	38.5	13
Domestic services	6	66.7	3	33.3	9
Professional	31	50.0	31	50.0	62
Sales	27	49.1	28	50.9	55
Services	34	43.0	45	57.0	79
Skilled manual	15	29.4	36	70.6	51
Unskilled manual	109	56.5	84	43.5	193
Unemployed	23	63.9	13	36.1	36
Others	9	69.2	4	30.8	13
Total	**296**	**51.4**	**280**	**48.6**	**576**
$\chi^2 = 19.54$; df.9, p = 0.021 (**Significant**)					
Occupational status of subject (wife)					
Housewife	260	50.6	254	49.4	514
Agriculture	25	65.8	13	34.2	38
Others	11	45.8	13	54.2	24
$\chi^2 = 3.59$; df.2, 0.167(Non-significant)					
Family type					
Nuclear	115	54.8	95	45.2	210
Extended	181	49.5	185	50.5	366
$\chi^2 = 1.51$; df.1, p = 0.220 (Non-significant)					
Household system					
Maternal	9	36.0	16	64.0	25
Mixed	42	51.2	40	48.8	82
Paternal	245	52.2	224	47.8	469
$\chi^2 = 2.51$; df.2, p = 0.286 (Non-significant)					
Marriage arrangement					0
Arrange	254	51.9	235	48.1	489
Self-arrange	42	48.3	45	51.7	87
$\chi^2 = 0.40$; df.1, p = 0.528 (Non-significant)					
Exchange marriage					
Non-reciprocal	252	49.8	254	50.2	506
Reciprocal	44	62.9	26	37.1	70
$\chi^2 = 4.20$; df.1, p = 0.041 (**Significant**)					

Subjects' age at marriage (years)					
>9-14	6	42.9	8	57.1	14
>14-19	143	55.0	117	45.0	260
>19-24	127	52.5	115	47.5	242
>24-29	17	32.7	35	67.3	52
>30	3	37.5	5	62.5	8
χ^2 = 9.78; df.4, p = 0.044 (**Significant**)					
Marriage year					
>1974-1982	0	0.0	2	100.0	2
>1982-1990	16	57.1	12	42.9	28
>1990-1998	37	67.3	18	32.7	55
>1998-2006	92	53.8	79	46.2	171
>2006-2014	141	48.6	149	51.4	290
Upto 2015	10	33.3	20	66.7	30
χ^2 = 13.24; df.5, p = 0.021 (**Significant**)					

3.6 Distribution of parallel and cross marriages with respect to bio-demographic parameters

It was observed that the highest rate of parallel cousin unions was noticed in subjects with the peri-urban origin (70%), while the percentage of cross cousin union was noticed to be highest in subjects with the rural origin (43%). Similarly, according to mother tongue, the highest proportion of parallel unions was calculated in subjects others category (62%), while in Pashto speaking subjects the cross cousin unions were more prevalent (46%; Table 3.9).

With reference to spouse's age, it was witnessed that the parallel marriages were more prevalent in '>25-30' years category (16%), while cross cousin unions were more prevalent in spouses belonging to age category of '>30-35' years (12%). Whereas with reference to subjects' age, the parallel and cross cousin marriages were more frequent in '>25-30' years category (17% and 15%, respectively; Table 3.9).

Contrastingly, with respect to spouse literacy, the highest rate of parallel unions was observed in the illiterate group (64%), while cross cousin unions were prevalent in the literate group (43%). Whereas with respect to years of education, the highest percentage of parallel unions was observed in the spouses who attended school till primary level (60%), while cross cousin unions were noticed to be highest in spouses with graduate/post-graduate education (46%; Table 3.9).

With reference to subjects' literacy, parallel cousin unions were prevalent in the illiterate group (61%), while cross cousin unions were found to be highest in the literate group (44%). Contrastingly with respect to education years, the highest proportion of parallel cousin unions was o in subjects with primary education (59%), while the highest percentage of cross cousin unions was calculated for the subjects who attended the school till graduate/post-graduate level (50%; Table 3.9).

With reference to spouse's caste-system, the highest proportion of parallel cousin unions was observed in Abbasi caste-system (61%), whereas highest proportion of cross cousin unions was observed in Syed caste-system (63%).

According to subjects' caste-system, the highest proportion of parallel cousin unions was noticed in Tanoli caste-system (64%), while the highest percentage of cross-cousin unions was observed in Syed caste-system (67%; Table 3.9).

Table 3.9 Distribution of parallel and cross marriages with respect to bio--bio-demographic parameters

	Parallel unions		Cross unions		
Parameters	**Total No**	**%age**	**Total No**	**%age**	**Total no**
Location					
Rural	263	56.6	202	43.4	465
Peri-urban	21	70.0	9	30.0	30
Urban	47	58.0	34	42.0	81
Total	**331**	**57.5**	**245**	**42.5**	**576**
$\chi^2 = 2.10$; df.2, p = 0.351 (Non-significant)					
Mother tongue					
Hindko	305	57.5	225	42.5	530
Pashto	18	54.5	15	45.5	33
Others	8	61.5	5	38.5	13
$\chi^2 = 0.20$; df.2, p = 0.903 (Non-significant)					
Spouse age (years)					
>15-25	30	5.2	30	5.2	60
>25-30	91	15.8	61	10.6	152
>30-35	80	13.9	70	12.2	150
>35-40	65	11.3	51	8.9	116
>40-45	34	5.9	12	2.1	46
>45	31	5.4	21	3.6	52
$\chi^2 = 8.06$; df.5, p = 0.153 (Non-significant)					
Subjects' age (years)					
>15-20	23	4.0	20	3.5	43
>20-25	74	12.8	54	9.4	128
>25-30	98	17.0	87	15.1	185
>30-35	74	12.8	48	8.3	122
>35-40	37	6.4	26	4.5	63
> 40	25	4.3	10	1.7	35
$\chi^2 = 5.15$; df.5, p = 0.398 (Non-significant)					
Spouse literacy					
Illiterate	37	63.8	21	36.2	58
Literate	294	56.8	224	43.2	518
$\chi^2 = 1.06$; df.1, p = 0.304 (Non-significant)					
Years of education					
Primary 5-8	78	59.5	53	40.5	131
Secondary 9-12	186	56.0	146	44.0	332
Graduate/post-graduate	30	54.5	25	45.5	55

13+					
	χ^2 = 0.60; df.2, p = 0.742 (Non-significant)				
Subject literacy					
Illiterate	101	61.2	64	38.8	165
Literate	230	56.0	181	44.0	411
	χ^2 = 1.33; df.1, p = 0.249 (Non-significant)				
Years of education					
Primary 5-8	105	59.3	72	40.7	177
Secondary 9-12	150	55.1	122	44.9	272
Graduate/post-graduate 13+	54	58.1	39	41.9	93
	χ^2 = 0.81; df.2, p = 0.666 (Non-significant)				
Caste-system (spouse)					
Abbasi	14	60.9	9	39.1	23
Awan	135	61.1	86	38.9	221
Gujjar	45	57.0	34	43.0	79
Pathan	56	52.3	51	47.7	107
Rajpoot	13	50.0	13	50.0	26
Syed	6	37.5	10	62.5	16
Tanoli	25	58.1	18	41.9	43
Others	37	60.7	24	39.3	61
	χ^2 = 5.92; df.7, p = 0.549 (Non-significant)				
Caste-system (subjects)					
Abbasi	13	56.5	10	43.5	23
Awan	132	61.7	82	38.3	214
Gujjar	47	58.8	33	41.3	80
Pathan	55	52.4	50	47.6	105
Rajpoot	13	46.4	15	53.6	28
Syed	5	33.3	10	66.7	15
Tanoli	30	63.8	17	36.2	47
Others	36	56.3	28	43.8	64
Total	**331**	**57.5**	**245**	**42.5**	**576**
	χ^2 = 8.52; df.7, p = 0.289 (Non-significant)				

3.6.1 Distribution of parallel and cross cousin marriages with respect to socio-economic parameters

The distribution of parallel and cross cousin unions were studied in different bio-demographic aspects of subjects and their spouses. Regarding the occupational status of spouse, the highest rate of parallel cousin union was observed in spouses who were engaged in domestic services (79%), while least proportion was noticed in spouses engaged in agricultural jobs (31%). On the other hand, the highest percentage for cross cousin union was found in spouses who were engaged in agricultural jobs (69%) and least proportion was observed in spouses who were engaged in domestic services (22%; Table 3.10).

With reference to subjects' occupation, the highest proportion for parallel cousin union was observed in 'others' category of subjects' occupational status (63%), while highest proportion of cross cousin unions was calculated for the subjects who were engaged in agricultural jobs (55%; Table 3.10).

The data were also analyzed for family type and household system, parallel cousin unions were more prevalent in the nuclear family type and in maternal household system (58% and 60%, respectively), whereas cross cousin unions were common in extended family type and in mixed household system (43% and 46%, respectively; Table 3.10).

With reference to marriage arrangement, the rate of parallel and cross cousin unions was observed to be almost equal. While parallel cousin unions were more prevalent in subjects with reciprocal marriages (61%), whereas cross cousin unions were common in subjects with non-reciprocal marriages (43%; Table 3.10)

Accordingly, with respect to age at marriage of subjects, the proportion of parallel cousin union was high in '>30' years category (75%), whereas the proportion of cross cousin was noticed to be highest in'>24-29' years category (46%). Moreover,

with reference to marriage years, it was observed that the percentage of parallel and cross cousin unions was observed to be highest during '>2006-2014' (29% and 21%, respectively; Table 3.10).

Table 3.10 Distribution of parallel and cross marriages with respect to socio-economic parameters

Parameters	Parallel unions Total no	%age	Cross-unions Total no	%age	Total no
Occupation status of spouse (husband)					
Abroad/Foreign	42	64.6	23	35.4	65
Agriculture	4	30.8	9	69.2	13
Domestic services	7	77.8	2	22.2	9
Professional	39	62.9	23	37.1	62
Sales	31	56.4	24	43.6	55
Services	40	50.6	39	49.4	79
Skilled manual	28	54.9	23	45.1	51
Unskilled manual	113	58.5	80	41.5	193
Unemployed	18	50.0	18	50.0	36
Others	9	69.2	4	30.8	13
Total	**331**	**57.5**	**245**	**42.5**	**576**
$\chi^2 = 10.74$; df.9, p = 0.294 (Non-significant)					
Occupational status of subject (subject)					
Housewife	299	58.2	215	41.8	514
Agriculture	17	44.7	21	55.3	38
Others	15	62.5	9	37.5	24
$\chi^2 = 2.87$; df.2, p = 0.238 (Non-significant)					
Family type					
Nuclear	122	58.1	88	41.9	210
Extended	209	57.1	157	42.9	366
$\chi^2 = 0.05$; df.1 p = 0.817 (Non-significant)					
Household					
Maternal	15	60.0	10	40.0	25
Mixed	44	53.7	38	46.3	82
Paternal	272	58.0	197	42.0	469
$\chi^2 = 0.61$; df.2, p = 0.739 (Non-significant)					
Marriage arrangement					
Arrange	281	57.5	208	42.5	489
Self-arrange	50	57.5	37	42.5	87
$\chi^2 = 0.0001$; df.1, p = 0.999 (Non-significant)					
Exchange marriage					
Non-reciprocal	287	56.7	219	43.3	506
Reciprocal	44	62.9	26	37.1	70
$\chi^2 = 0.95$; df.1, p = 0.330 (Non-significant)					
Age at marriage (years)					

>9-14	8	57.1	6	42.9	14
>14-19	148	56.9	112	43.1	260
>19-24	141	58.3	101	41.7	242
>24-29	28	53.8	24	46.2	52
>30	6	75.0	2	25.0	8
	χ^2 = 1.38; df.4, p = 0.848 (Non-significant)				
Marriage year					
>1974-1982	2	0.3	0	0.0	2
>1982-1990	18	3.1	10	1.7	28
>1990-1998	35	6.1	20	3.5	55
>1998-2006	95	16.5	76	13.2	171
>2006-2014	168	29.2	122	21.2	290
Upto 2015	13	2.3	17	3.0	30
	χ^2 = 5.60; df.5, p = 0.347 (Non-significant)				

3.7 Distribution of inter-caste marriages

The data were also analyzed to study the trend of inter-caste marriages, the proportion of marriage of Syed with Syed was calculated to be highest (88%), followed by Pathan (84%), Awan (85%), Gujjar (81%), and Tanoli caste-systems (74%). The least proportion of intra-caste marriage was noticed in Rajpoot caste-system (66%; Fig. 1.16).

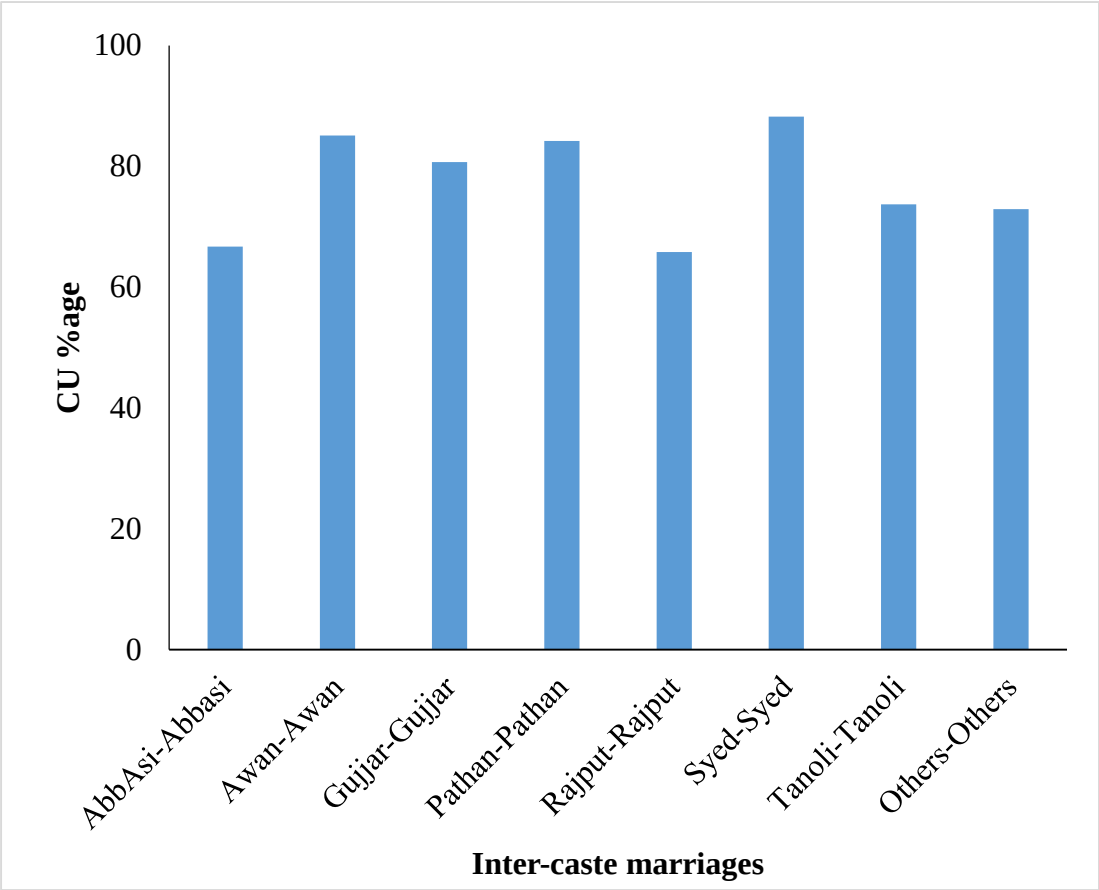

Fig. 1.16. Distribution of CU with respect to inter-caste marriages

3.8 Fertility and mortality profile with respect to various socio-bio-demographic variables

3.8.1 Fertility and mortality with respect to origin

There were a total of 1,453 (97%) ever pregnant women (EPW) in the sample. With respect to their origin, the highest representation of EPW was observed in subjects originating from peri-urban areas (99%) and was the least in the subjects from urban areas (97%). Fertility was estimated by calculating the total pregnancies per woman. The highest fertility rate was observed in subjects from urban (3.30±1.84) areas (3.30±1.84; p=0.755). The total live-births per women were also calculated. The highest rate was observed in subjects originating from urban areas (2.64±1.80) and was lowest in peri-urban areas (2.27±1.58) the differences were not significant (p=0.075). However, with respect to male live-births, the differences were statistically significant (p=0.019). The data were also analyzed to estimate the mortality. The highest mortality rate was observed in the peri-urban areas (0.64±1.21) the differences were statistically not significant (p=0.112). The prenatal mortality rate was also calculated. Prenatal mortalities were more prevalent in the peri-urban areas (0.51±1.17) than rural and urban areas (0.41±0.86 and 0.35±0.72, respectively; Table 3.11).

Table 3.11 Subjects' fertility, live-births and mortality with respect to their origin

Parameters	Rural	Peri-urban	Urban	Total	P value
Variable (No.)	1,172	97	231	1,500	
Fertility					
Ever pregnant women (No., %)	1,134 (96.75)	96 (98.96)	223 (96.53)	1,453 (96.86)	
Total pregnancies (No.)	3741	307	762	4810	
Fertility: preg./women (mean± SD)	3.19±2.09	3.16±2.09	3.30±1.84	3.21±2.06	t: 0.755
Currently pregnant (No.)	254	25	41	320	
No pregnancy (No)	38	1	8	47	
Live-births					
Total live-births(No.)	2,786	220	610	3,616	
Live-births/women (mean ± SD)	2.38±1.73	2.27±1.58	2.64±1.80	2.41±1.73	t: 0.0757
Live-birth: sons (No.)	1380	113	322	1815	
Live-birth: sons (mean± SD)	1.18±1.05	1.16±1.19	1.39±1.17	1.21±1.08	t: 0.0186 *
Live-birth: daughters (No.)	1406	107	288	1801	
Live-birth: daughters (mean± SD)	1.20±1.22	1.10±0.91	1.25±1.24	1.20±1.21	t: 0.6145
Mortalities					
Total mortalities (No.)	707	62	105	874	
Mortality/women (mean± SD)	0.60±1.05	0.64±1.21	0.45±0.78	0.58±1.02	0.1118
Prenatal mortality (No.)	476	49	80	605	
Prenatal mortality (mean± SD)	0.41±0.86	0.51±1.17	0.35±0.72	0.40±0.86	t: 0.3066
Postnatal mortality (No.)	229	13	27	269	
Postnatal mortality (mean± SD)	0.20±0.59	0.13±0.34	0.12±0.41	0.18±0.56	t: 0.1031

* Statistically significant; ** highly significant

3.8.2 Effect of subject's literacy on fertility and mortality rates

The data were also examined to find out the relationship between subjects' literacy and fertility. EPW were more prevalent in the illiterate sample (97%). Fertility was assessed by calculating pregnancy per women. The highest rate of fertility per women was observed in the illiterate group (4.13±2.36; $p>0.001$). The total live-births per women were also calculated. The highest rate was noticed in the illiterate subjects (3.16±2.04; $p>0.001$). In the case of male live-births, the highest rate was calculated in the illiterate subjects (1.95±1.05), and the differences were statistically significant ($p>0.001$; Table 3.12).

The data were further studied to check the relationship between mortality and subjects' literacy. The highest rate of mortality was calculated in the illiterate females (0.80±1.21; $p<0.001$). Even though prenatal mortality rate was high in the illiterate subjects (0.51±0.98; $p=0.007$). According to postnatal mortality, the differences were statistically highly significant ($p<0.001$; Table 3.12).

Table 3.12 Subjects' fertility, live-births and mortality with respect to their education

Subjects' literacy	Literate	Illiterate	Total	p-value
Variable (No.)	1,106	394	1,500	
Fertility				
Ever pregnant women (No., %)	1,071 (96.83)	382 (96.95)	1,453 (96.86)	
Total pregnancies (No.)	3,181	1,629	4,810	
Fertility: preg./women (mean± SD)	2.88±1.83	4.13±2.36	3.21±2.06	<0.001 **
Currently pregnant (No.)	249	71	320	
No pregnancy (No)	35	12	47	
Live-births				
Total live-births(No.)	2,369	1,247	3,616	
Live-births/women (mean± SD)	2.14±1.52	3.16±2.04	2.41±1.73	<0.001 **
Live-birth: sons (No.)	1202	613	1815	
Live-birth: sons (mean± SD)	1.53±0.85	1.95±1.05	1.65±0.93	<0.001 **
Live-birth: daughters (No.)	1167	634	1801	
Live-birth: daughters (mean± SD)	1.06±1.07	1.61±1.46	1.20±1.21	<0.001**
Mortalities				
Total mortalities (No.)	558	316	874	
Mortality/women (mean± SD)	0.50±0.94	0.80±1.21	0.58±1.02	<0.001 **
Prenatal mortality (No.)	403	202	605	
Prenatal mortality (mean± SD)	0.36±0.82	0.51±0.98	0.40±0.86	t: 0.0072 *
Postnatal mortality (No.)	155	114	269	
Postnatal mortality (mean± SD)	0.14±0.44	0.29±0.78	0.18±0.56	t: 0.0004 **

*Statistically significant; **highly significant

3.8.3 Association of fertility and mortality with consanguinity

Furthermore, the data were also analyzed to assess the correlation of consanguineous unions and non-consanguineous unions with fertility, live birth and mortality rates. The highest proportion of ever pregnant women (EPW) was calculated in subjects with consanguineous unions (97%). The fertility was estimated by calculating the pregnancy per women. The fertility rate was higher in CU (3.32±2.12), and the differences were significant (p=0.014). Further live-births per women were also evaluated. The highest rate was observed in CU (2.48±1.74; p=0.081). Whereas both male live-birth and female live-birth were higher in CU, and the differences were not statistically significant (p=0.095 and p=0.031, respectively). The mortality rate with respect to CU and NCU were also studied. The highest mortality was observed in subjects with CU (0.63±1.06), the differences were significant (p=0.025). The prenatal mortality was prevalent in CU (0.42±0.87; p=0.427). However, with respect to postnatal mortality, the differences were significant (p=0.001; Table 3.13).

Table 3.13 Subjects' fertility, live-births and mortality according to CU and NCU

Parameters	**CU**	**NCU**	**Total**	**p-value**
Variable (No.)	840	660	1,500	
Fertility				
Ever pregnant women (No., %)	815 (97.02)	638 (96.67)	1,453 (96.87)	
Total pregnancies (No.)	2790	2020	4810	
Fertility: preg./women (mean± SD)	3.32±2.12	3.06±1.96	3.21±2.06	t: 0.0137 *
Currently pregnant (No.)	171	149	320	
No pregnancy (No.)	25	22	47	
Live-births				
Total live-births(No.)	2,083	1,533	3,616	
Live-births/women (mean± SD)	2.48±1.74	2.32±1.72	2.41±1.73	t: 0.0811
Live-birth: sons (No.)	1051	764	1815	
Live-birth: sons (mean± SD)	1.25±1.08	1.16±1.08	1.21±1.08	t :0.0952
Live-birth: daughters (No.)	1032	769	1801	
Live-birth: daughters (mean± SD)	1.23±1.20	1.17±1.21	1.20±1.21	t: 0.3122
Mortalities				
Total mortalities (No.)	533	341	874	
Mortality/women (mean± SD)	0.63±1.06	0.52±0.97	0.58±1.02	t: 0.0252 *
Prenatal mortality (No.)	352	253	605	
Prenatal mortality (mean± SD)	0.42±0.87	0.38±0.86	0.40±0.86	t: 0.4266
Postnatal mortality (No.)	183	86	269	
Postnatal mortality (mean± SD)	0.22±0.65	0.13±0.41	0.18±0.56	t= 0.0014 *

*Statistically significant; **highly significant

3.8.4 Association of family type on fertility and mortality

Data were also analyzed to check the association of fertility with family type. In the extended family system, the highest percentage of EPW was noticed (97%). The fertility rate was assessed to be highest in the nuclear family system (3.96±2.24) and differences were highly significant ($p<0.001$). The live-births per women were also calculated. The highest rate was observed in the nuclear family system (3.03±1.83; $p<0.001$). The male live-births were also higher in the nuclear family system (1.48±1.15), the differences were highly significant ($p<0.001$). Regarding the mortality per women, the highest rate was observed in the nuclear family (0.77±1.16) and the differences were significant ($p<0.001$). While prenatal mortality rate was highest in nuclear family type (0.54±0.98; Table 3.14).

Table 3.14 Subjects' fertility, live-births and mortality with respect to family type

Parameters	Nuclear family	Extended family	Total	p-value
Variable (No.)	653	847	1,500	
Fertility				
Ever pregnant women (No., %)	632 (96.78)	821 (96.93)	1,453 (96.86)	
Total pregnancies (No.)	2587	2223	4810	
Fertility: preg./women (mean± SD)	3.96±2.24	2.62±1.69	3.21±2.06	<0.0001 **
Currently pregnant (No.)	88	232	320	
No pregnancy (No.)	21	26	47	
Live-births				
Total live-births(No.)	1980	1636	3616	
Live-births/women (mean± SD)	3.03±1.83	1.93±1.49	2.41±1.73	<0.0001 **
Live-birth: sons (No.)	969	846	1815	
Live-birth: sons (mean± SD)	1.48±1.15	1.00±0.97	1.21±1.08	<0.0001 **
Live-birth: daughters (No.)	1011	790	1801	
Live-birth: daughters (mean± SD)	1.55±1.34	0.93±1.01	1.20±1.21	<0.0001 **
Mortalities				
Total mortalities (No.)	506	368	874	
Mortality/women (mean± SD)	0.77±1.16	0.43±0.88	0.58±1.02	<0.0001 **
Prenatal mortality (No.)	354	251	605	
Prenatal mortality (mean± SD)	0.54±0.98	0.30±0.74	0.40±0.86	<0.0001 **
Postnatal mortality (No.)	152	117	269	
Postnatal mortality (mean± SD)	0.23±0.67	0.14±0.45	0.18±0.56	t: 0.0018 *

*Statistically significant; **highly significant

3.8.5 Subjects' age at marriage and fertility

The data were further analyzed to check the effect of subjects' age at marriage on fertility. Ever pregnant women were higher in lowest age group, i.e., ">914" (100%). Fertility was also highest in ">9-14" years age category (5.09±2.36; p<0.001). Other parameters like live-births/women, male live-birth, female live-birth were prevalent also prevalent in ">9-14" years age category. Mortality was also analyzed with respect to subjects' age at marriage. The highest mortality was calculated for ">14" years age category (0.85±1.12). The differences were statistically significant (p= 0.014). Prenatal mortality rates were higher among subjects with ">14-19" years age category (0.48±0.92) and the differences were statistically not significant (p= 0.086), while postnatal mortalities were highest among ">9-14" years age group (0.48±0.67). The differences were significant (p=0.004; Table 3.15).

Table 3.15 Subjects' fertility, live-births and mortality with respect to subject's age at marriage

Subject's age at marriage	>9-14	>14-19	>19-24	>24-29	>30	Total	p-value
Variable (No.)	33	641	623	178	25	1,500	
Fertility							
Ever pregnant women (No., %)	33(100)	623 (97.19)	604 (96.95)	169 (94.94)	24 (96)	1,453 96.86)	
Total pregnancies (No.)	168	2345	1797	450	50	4810	
Fertility: preg./women (mean± SD)	5.09±2.36	3.66±2.20	2.88±1.83	2.53±1.71	2.00±1.19	3.21±2.06	<0.001 **
Currently pregnant (No.)	2	131	141	39	7	320	
No pregnancy (No.)	0	18	19	9	1	47	
Live-births							
Total live-births(No.)	138	1,783	1,339	327	29	3,616	
Live-births/women (mean± SD)	4.18±1.93	2.78±1.85	2.15±1.53	1.84±1.48	1.16±0.99	2.41±1.73	<0.001 **
Live-birth: sons (No.)	63	911	671	156	14	1,815	
Live-birth: sons (mean± SD)	2.17±1.04	1.80±0.98	1.51±0.86	1.43±0.79	1.17±0.72	1.65±0.93	<0.001 **
Live-birth: daughters (No.)	75	872	668	171	15	1,801	
Live-birth: daughters (mean± SD)	2.27±1.77	1.36±1.29	1.07±1.08	0.96±1.07	0.60±0.65	1.20±1.21	<0.001 **
Mortalities							
Total mortalities (No.)	28	431	317	84	14	874	
Mortality/women (mean± SD)	0.85±1.12	0.67±1.09	0.51±1.00	0.47±0.80	0.56±0.96	0.58±1.02	0.0140
Prenatal mortality (No.)	12	305	212	66	10	605	
Prenatal mortality (mean± SD)	0.36±1.03	0.48±0.92	0.34±0.83	0.37±0.72	0.4±0.87	0.4±0.86	0.0860
Postnatal mortality (No.)	16	130	101	18	4	269	
Postnatal mortality (mean± SD)	0.48±0.67	0.20±0.59	0.16±0.56	0.10±0.38	0.16±0.47	0.18±0.56	0.0041 *

*Statistically significant; **highly significant

3.9 Pattern of genetic/congenital deformities in Haripur population

The present epidemiological study was carried out in the Haripur district KPK. A total of 1,500 married females were recruited. The clinical information about the health status of their children gathered. Total 58 children with congenital/hereditary anomalies were observed. Required information related to socio-bio-demographic variables as well as the clinical detail of diseases was also obtained from each participant. Here distribution of anomalies was established across the bio-demographic variable like the gender of the subject and family/sporadic nature and isolated/syndromic nature of anomaly. The studied congenital/hereditary disorder were categorized into ten major groups.

Neurological disorder had the highest representation (n=17; 29%), followed by musculoskeletal defects (n=8; 14%) and ear/auditory defects (n=6; 10.3; Fig. 3.17). While studying male to female ratio, it was observed that the affected males were higher in number (n=32; 55%) compared to affected females (n=26; 45%). It was observed that significantly higher number of the congenital/hereditary cases were of sporadic nature (n=49; 84%), while 16% cases had familial presentations and distribution of anomalies among familial/sporadic was statistically significant ($p= 0.003$). Likewise, it was observed that the most of the disorders were of isolated presentation (n=41; 71%) while 29% subjects had a syndromic presentation of diseases and differences in the distribution of anomalies among isolated/syndromic nature were highly significant ($p<0.0001$; Table 3.16).

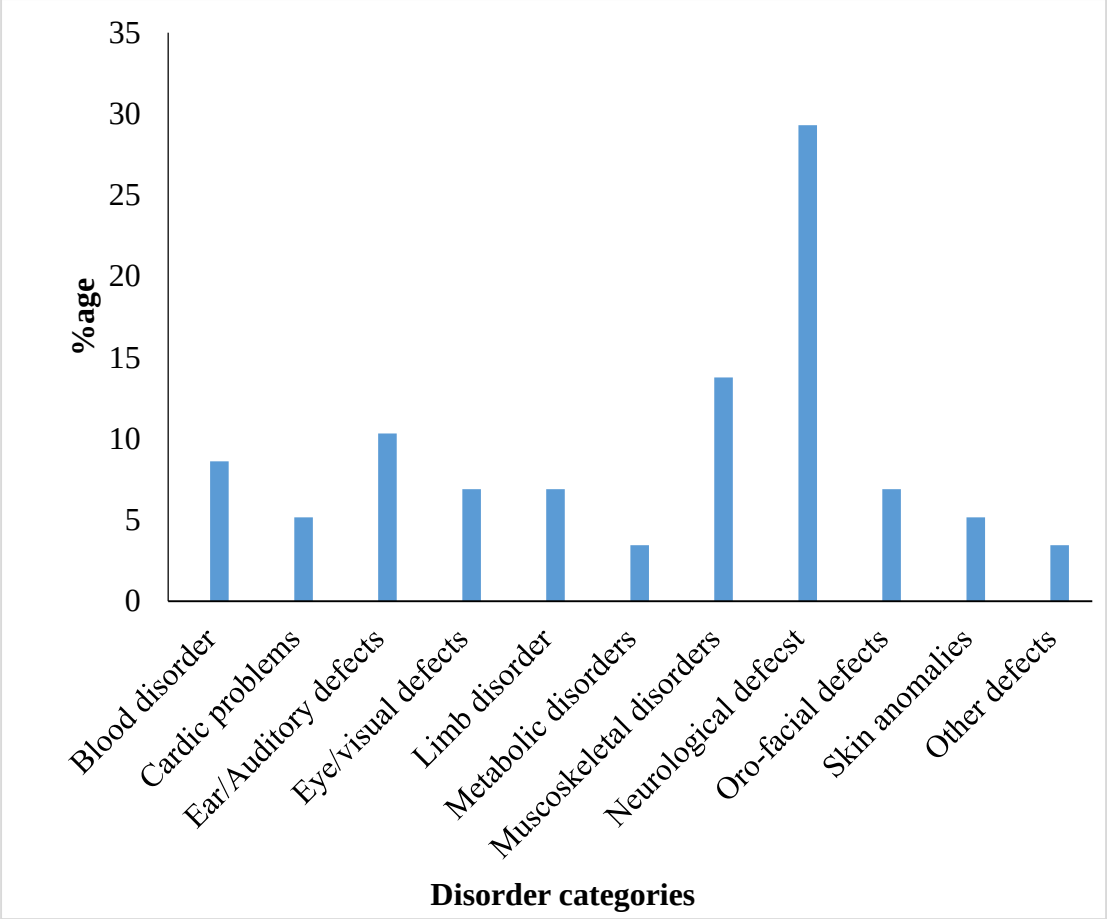

Fig. 3.17. Distribution of hereditary/congenital anomalies in the population of Haripur district

Table 3.16 Major categories of the hereditary/congenital anomalies: distribution with respect to gender, familial/sporadic nature, and isolated/syndromic presentation

	Gender		Familial/sporadic		Isolated/syndromic		
Disorders category	**Male**	**Female**	**Familial**	**Sporadic**	**Isolated**	**Syndromic**	**Sum**
Blood disorder	3	2	0	5	5	0	5
Cardiac problems	1	2	0	3	3	0	3
Ear/Auditory defects	4	2	1	5	6	0	6
Eye/visual defects	2	2	2	2	4	0	4
Limb disorder	2	2	0	4	4	0	4
Metabolic disorder	1	1	1	1	2	0	2
Muscoskeletal disorder	5	3	1	7	7	1	8
Neurological defects	9	8	1	16	1	16	17
Oro-facial defects	3	1	0	4	4	0	4
Skin anomalies	2	1	3	0	3	0	3
Other defects	0	2	0	2	2	0	2
Total	32	26	9	49	41	17	58
	$\chi^2 = 26.35$; df.10, p = 0.920 (Non-significant)		$\chi^2 = 26.35$; df.10, p = 0.003 **(Significant)**		$\chi^2 = 49.23$; df.10, p<0.0001 **(Highly significant)**		

Consanguinity is a deeply rooted socio-cultural trend in several societies including Pakistan. The highest rates of cousin marriages were described in the Middle East, where consanguinity reaching >80% in specific regions. Cousin marriages have some social and economic benefits in these populations (Bosdou *et al.,* 2016). Previous studies showed that the overall percentage of inbreeding is very high in Pakistan that is 63% (Ahmed *et al.,* 1992). Only few studies are available about consanguinity rate in different rural and urban communities of Pakistan, such as in Punjab (Shami *et al.,* 1989; Shami *et al.,* 1990; Yaqoob *et al.,* 1993), Balochistan (Mian and Mushtaq, 1994), Khyber Pakhtoonkhaw (Wahab and Ahmad, 1996), Kashmir (Jabeen and Malik, 2014), Sargodha (Hina and Malik, 2015), Dir Lower (Ahmad *et al.,* 2016) and Rahim Yar Khan (Riaz *et al.,* 2016).

The present epidemiological study was conducted in Haripur district of Khyber Pakhtunkhwa (KPK) Pakistan. A total of 1,500 subjects were recruited to assess consanguinity, neonatal outcome, and morbidity. In this study, consanguineous unions (CU) were observed to be 56%. Formerly, the highest rate of CU (62%), was observed in the Bhimber city of Kashmir. To date, several studies have been conducted in Pakistan, and around the globe to observe the prevalence of consanguinity and its socio-bio-demographic differentials. Consanguinity in various cities/populations of Pakistan was as follows: Rahim Yar Khan (58.5%; Riaz *et al.,* 2016), Bhimber (62.2%; Jabeen and Malik, 2014), Bajaur Agency (22.3%; Ahmad *et al.,* 2016), Sargodha (56.7%; Hina and Malik, 2015), Lahore (38.8%; Shami, 1982), Muridke (41.2%; Shami, 1983), Sheikhupura (48.9%; Shami and Iqbal, 1983), Rawalpindi (48.1%; Shami and Siddiqui, 1984), Jhelum (44.2%; Shami and Minhas, 1984) Mianchannu (37.8%; Shami *et al.,* 1989), Sialkot (51.8%), Faisalabad (52.1%), Sahiwal (56.1%), Gujranwala (58.9%), All Punjab (50.3%); (Bittles *et al.,* 1993),

Sawt (rural) (37.1%) and Swat (urban; 31.1%; Wahab and Ahmad, 1996), Quetta (31.6%; Mian and Mushtaq, 1994), Lahore (46.2%; Yaqoob *et al.*, 1993), Rahim Yar Khan (58.4%; Riaz *et al.*, 2016), and Dir Lower (46.2%; Ahmad *et al.*, 2016).

In the present study, the most prevalent type of marriage was observed to be FC type (38%). Our findings are consistent with the studies conducted around the world, i.e., Arab and Muslim communities in North Africa, in the West and almost all parts of South Asia, and also within Pakistan. In all these studies, FC marriages were observed to be the commonest type of union (Bittles 2010; Bittles *et al.*, 1993; Hina and Malik 2015; Hussain 1999; Hussain and Bittles 1998; Jabeen and Malik 2014; Rajab and Patton 2000; Riaz *et al.*, 2016). A suggestive description can be made for the higher rate of FC marriages as people do not want the intermixing of blood marrying in other caste-systems, strictly yearn to strengthen the family bonds and do not want to share the family property by marrying non-related. In Pakistan, the similar reasons have been previously highlighted in the population of Sargodha district (Hina and Malik, 2015).

With respect to the rural/ urban origin, the percentage of CU in the subjects from rural origin was the highest (58%). Previously reported studies including Pakistan Bio-demographic and Health Survey (PDHS) from 1990 to 1991 were consistent with this finding (Bio-demographic, 1992). This may be explained by the fact that people of the rural origin have low socio-economic status, poor literacy level and generally living in an extended family system where the older person in the family control decision of all family matters including marriage decisions. Our observations are consistent with the findings of studies conducted in Rahim Yar Khan (Riaz *et al.*, 2016), Swat (Wahab and Ahmad, 1996), Southern Punjab and India (Hussain and Bittles, 1998).

In this epidemiological study, consanguinity was relatively more prevalent in Hindko speaking subjects compared to Pashto and other languages speaking subjects (56.2% *vs.*52.8% and 54.5%, respectively). Hindko is the major language of Hazara division and local people of Haripur speak only Hindko language. Pashto is the language of Afghan refugees who migrated from Afghanistan and living in camps built in Haripur or spoken by migrant Pakistani Pathans who are living in Haripur for their job purposes, while other languages, e.g., Saraiki, Urdu, Sindhi, Kohistani, Brushishki, and Punjabi, are spoken by only a few migrated people from different regions of Pakistan. Haripur is an industrial city having different industries e.g., T.I.P, NRTC, Hattar factory, different oil and ghee factories, cement industry, etc. For that reason, people from other areas of Pakistan with different cultures and languages also live in Haripur.

With respect to subjects' age, no increasing or decreasing trend of consanguinity was observed. The present analysis showed that subject's age was not apparently associated with consanguinity. This is not consistent with many previous studies (Riaz *et al.*, 2016). The possible reason might be that female mostly hide their original age, and do not give correct information. Therefore, there might be some underestimate in the subjects' age. While the study conducted in Bhimber district of Azad Jammu And Kashmir, consanguinity was gradually declining as the age of subjects' increased (Jabeen and Malik, 2014).

With respect to the subjects' literacy, the highest percentage of CU was observed in the literate group (57%). Our findings were inconsistent with other studies (Afzal, 1994; Ahmad *et al.*, 2016). With the increase in education level, the rate of CU was observed to be increasing, i.e. 53% in the subjects with primary education, 60% in the subjects with secondary school education, and 59% in

individuals with graduate/post-graduate level education. Our findings are consistent with the study of Bhimber district of Azad Jammu and Kashmir Pakistan (Jabeen and Malik, 2014) and with the findings of Wahab and Ahmad during the study of Swat (Wahab and Ahmad, 1996). The possible reason behind the association of consanguinity with literacy is the economic status of subjects. The rates of consanguineous unions are higher in economically well-off families such families prefer arranging marriages within close relatives, compared to subjects who are not only poor but also have low literacy level have a tendency to marry outside the family. Subjects with low literacy face the problem of finding a match within the family, therefore, their parents have no choice except to arrange the marriage more often beyond the close kinship. While opposite results were found during an epidemiological study of district Rahim Yar Khan of Punjab Pakistan in which highest proportion of CU was reported for illiterate subjects and as the educational levels of subjects increased the percentage of consanguinity decreased (Riaz *et al.*, 2016).

With respect to spouse's literacy, the highest proportion of CU was observed in the literate group (57%). There was a significant difference in the distribution of CU and NCU unions with respect to literate and illiterate spouses. With the increase in higher education, the rate of CU was observed to be increasing, i.e., 57% in the primary educated group, 58% in the spouses having secondary school education, and 59% in the spouses with graduate/post-graduate education. In contrast to our results, Kerkeni observed that as the subjects' education increased the consanguinity rate also increased significantly, while in males no rise in consanguinity was noticed as their literacy rate increased (Kerkeni *et al.,* 2006). In support of his findings, the author proposed that even in the countries and communities in which cousin marriage are

predominant due to cultural norms, the relationship with occupational status and literacy level is primarily observed among females but not in their spouses. These results give clear indications about the plan of genetic epidemiological studies that inspects the effect of cousin marriages on human health. For males, the association intensely depends upon the cultural framework. According to described examples, extending from negative association between consanguinity and social values to the concept that the highly educated males were more prone to be married to their cousin (Khoury and Massad, 1992; Proctor and Smith, 1992).

According to the caste-system, the highest proportion of consanguinity was estimated in Abbasi caste-system and Gujjar caste-system compared to the others caste-system. The differences between the distribution of CU and NCU with respect to subjects' and spouse's caste-system was statistically significant. The reason behind more consanguinity in some caste-systems than others might be that some castes tend to keep their family customs, traditions, values and practices intact by marrying close relatives (Shami *et al.,* 1990). Moreover, people of some caste-systems consider themselves to be superior to other castes and even to their distant relatives and preferably marry inside the family with their close relatives. In addition, some people consider it very odd to add the non-related and even distantly related person as a part of their family as they think that outsiders might intervene in their peculiar lifestyle they are living for years and consequently force them to give up things they are pursuing since long (Hussain, 1999).

Consanguinity has been shown to be associated with the economic status of individuals (Shami *et al.,* 1994). According to the occupational status of spouse, the highest proportion of CU was observed in the unemployed group (68%; IC-F = 0.0287), it was observed to be comparatively low in other occupational groups. While

least percentage of CU was observed in spouses who were engaged in domestic services (44%; IC-F = 0.0244). Our results were different from reported data about the association between occupational status and consanguinity of spouse, for example in studies which have been conducted in district Rahim Yar Khan Punjab (Riaz *et al.*, 2016), Bajaur Agency (Ahmad *et al.*, 2016), Bhimber district of Azad Jammu and Kashmir (Jabeen and Malik, 2014). The possible explanation could be that the unemployed individuals usually have illiteracy or low educational levels, have no skills, and belong to rural areas and have fewer job opportunities. Consequently, their parents arrange their marriage inside the family because to find a perfect match outside the family for unemployed son is very difficult for them. Family relative compromised the low income or no job of a man for other benefits for their daughter.

According to family type, the highest rate of CU was noticed in subjects who were belonging to extended family type (61%; IC-F = 0.0326), our findings are consistent with the study conducted in Karachi (Hussain and Bittles, 1998). The highest rate of CU may be attributed to the fact that in the extended family system, overlapping generations form consanguineous loops by making marriage arrangements among blood-connected relatives (Hina and Malik, 2015). The possible reason might be that people after marriage prefer to live with their parents and sibs, because to start a new household system independently is difficult due to their low monthly income. In the extended family system, most of the economic problems are shared by parents and brothers. Therefore, for potential benefits elder member of the family arrange marriages within the family. As a result, the proportion of consanguineous unions among extended family system rises.

According to our study, the rate of consanguinity was comparatively higher in the paternal household system (57%). In the patrilineal systems, the families are prone

towards cousin marriages than non-related marriages. The factor for the comparatively lower proportion of consanguinity in the mixed household system can be ascribed as female become more dominant in the absence/death of her husband (Jacoby and Mansuri, 2010). In the absence of the male head of the family, females are independent to decide marriage matters outside the family.

With respect to marriage arrangement, the highest proportion of cousin marriages was studied in "self-arrange marriages", i.e., 61%, these marriages are also arranged by the parents and are mostly among cousins/close relatives. In these unions, the couple has influenced the decision of parents or have triggered the condition almost completely by themselves. Such marriages had been uncommon but increasing with the passage of time (Shaw, 2001). Reciprocal marriages are also organized by parents and are mostly arranged between cousins. Usually, land-owning families arrange reciprocal marriages to protect their land and financial adjustment as well as to secure their daughters (Hina and Malik, 2015). In rural communities, reciprocal marriages have many potential benefits, e.g., preservation of family structure and property, wealth, economic benefits associated with dowry and ease of marriage arrangement (Bittles, 1994). In reciprocal unions large age gap between spouses also allow. In our study, the highest proportion of consanguineous unions was observed in reciprocal marriages (88%). The differences between the distribution of CU and NCU were statistically highly significant ($\chi^2 = 38.44$; df.1, $p<0.0001$).

With reference to subjects' age at marriage, the highest proportion of cousin marriages were observed (60%) in ">9-14" years age category, and the least proportion of CU was noticed in ">30" years age category (36%). There was a declining trend of CU with an increase in subjects' age at marriage. The reason for the highest rate of consanguinity in younger females compared to older subjects can be

illustrated that female with low socio-economic status, education, and occupational status had married at an early age compared to those female with higher socio-economic status (Sheela, 2003). As the age of subjects increasing the perfect match in the family become very difficult to find. In rural areas and in regions with low literacy rate the people mostly favor to marry with a younger girl, they thought that younger females have large reproduction period and are healthy. If bride and groom are cousins their parents arrange early marriage because they compromised the education and earning of males for other insecurities of marriages among non-related couples. Females' parents want to marry their daughter as soon as possible at a younger age, therefore, they prefer closely related males. But as the age of females become increasing marriage arrangement within close relative is difficult, so subjects' parents have no option, therefore, they accept proposals from outside the family.

According to marriage year, the highest rate of cousin marriages was observed in subjects who were married in the decade of ">1982-1990", while least proportion of CU was noticed in subjects who were married in the duration of "up to 2015". The possible reason for this difference in the rate of CU in different years may be that as the time passes people become more educated. Further, the developments in the health system have increased the awareness about genetic disorders and marriage decision making. Awareness about recessive genetic disorders through genetic counseling that initiated at basic hospitals level have changed the consanguinity rate over the years. Not only the genetic counselor but the general physician and medical specialist also provides awareness to the people about genetic diseases due to cousin marriages. This effort is working in the decrease of consanguineous union rate with the passage of time. While these results were inconsistent with the studies which have been conducted in Swat, Pakistan. Where a significant a significant increase in the

incidence of consanguinity over the years has been observed (Wahab and Ahmad, 1996). A recently reported study in Bhimber Kashmir revealed the gradual rise in CU over time. The reason might be the deteriorating law-and-order condition between AJK and Indian-occupied Kashmir, due to which the mate choice reduce and CU rate increased (Jabeen and Malik, 2014).

Although there is an extensive literature on the effects of inbreeding on human health and reproduction, but the results are conflicting. Several studies have revealed that cousin marriages are linked with increasing risk of autosomal recessive disorders, congenital malformation, and mental retardation. Reported studies also demonstrated that consanguinity is associated with higher pre-reproductive deaths, low birth weight and higher postnatal mortality among offspring (Assaf *et al.,* 2009; Bittles and Black, 2010; Gowri *et al.,* 2011; Mumtaz *et al.,* 2007).

The current study was conducted to observe the effect of subjects' location on its fertility. Present results indicate that the fertility (pregnancy/female) and total live-births were highest among subjects who were living in urban areas compare to peri-urban and rural areas. The possible reason can be cited that in urban areas the health facilities are much better than peri-urban and rural area. In rural areas, due to lack of proper health care, the rate of total live-birth is lower compared to the urban areas. The reason might be illustrated as in rural areas the literacy rate is low. Others factors such as female education, economic status, diet conditions, use and disuse of contraceptive can also affect the total fertility rate.

In this study, literacy is negatively associated with fertility. There are several reasons for low fertility in educated women, like educated female lighten the burden of repeated pregnancies by taken different actions. This may perhaps happen for the reason that educated females have some other sources of their prestige and

fulfillments as well as reproductive performance. Literate females have more influence on household system besides their greater participation in reproductive decisions (Cain 1984; Dyson and Moore 1983). Another reason might be that educated women are financially less dependent on their son so, they do not have any old age insecurity this too may lead the small family size. The literate females are very ambitious for their children, combined with lower expectations from them in terms of labor services provided. While mortality rate was highest among illiterate subjects in present study.

Some studies show a negative association between fertility and consanguinity (Ansari and Sinha 1978; Reid 1976), while other showing significantly higher rates of fertility among inbred marriages (Bittles, 2002; Bittles *et al.*, 1991). Inadequate information is available about the bio-demographic profile of consanguinity. Even though the cousin marriages are prevalent in various regions of West and South Asia and in several parts of North Africa (Bittles, 1994), the available data are very limited to Bio-demographic and Health surveys (DHSs) in particular countries such as Tunisia, Egypt, Morocco, India, and Pakistan. Subsequently, there are noteworthy errors according to the most important socio-bio-demographic determinants of fertility, like subjects age at marriage and use of contraceptives in a consanguineous couple (Givens and Hirschman, 1994).

Certain methodological flaws, namely small sample size and poor allowance for potential confounders might be considered the reason for the discrepancy in the outcomes of aforementioned studies. The relationship between consanguinity and fertility still remains inconclusive. Consequently, for a firm conclusion about the association of consanguinity with reproductive behavior, further research is required

(Islam, 2012). The review of literature is failed to show any relationship between fertility and consanguinity.

A lot of studies described the direct relationship between live-births and consanguineous unions. Meta-analysis results of 30 different studies which have been conducted in African and Asian countries revealed a higher average number of live-birth in consanguineous couples compared to non-consanguineous couples (Bittles, 2010). The present data were analyzed to calculate the relationship between fertility and consanguinity, ever pregnant women were almost equal both in CU and NCU categories (97% and 97%, respectively), while fertility (pregnancy/women) was significantly higher among consanguineous couples ($p=0.013$). Our results were consistent with a study conducted in North-East Brazil (Weller and Santos, 2013). Earlier studies which have been conducted in Asian communities revealed that other bio-demographic variables such as subjects' age at marriage and non-contraceptive use can also affect the rate of fertility, hence consanguinity is not the single factor that affects the fertility rate (Bittles, 2002; Hussain and Bittles, 2004). In our study mortality rates were highest among consanguineous couples (0.63 ± 1.06; $p=0.0252$). Our findings are consistent with the study of Shah *et al.,* (1998). The significant result of his study suggests that the rate of mortality or child death is higher in consanguineous families compare to non-consanguineous families in Pakistan. Consanguineous couple's children have 1.1825 times higher risk of death before their fifth birthday, even after controlling the other factors which can affect the child mortality. Generally, mortality and morbidity is higher in inbred population due to the expression of detrimental recessive genes (Bittles, 2001b).

On the basis of total family members, family type is determined. There are two major types of family namely 'nuclear family' and 'extended family' (Smith *et*

al., 1986). The family influence the behaviors of its members from the early years of life and continues to have an effect even after maturity is reached (Sarker, 1993). The reason of a large number of children in the extended family can be illustrated as in extended family type elder member of the family like mother-in-law and father-in-law tend to have a traditional attitude towards fertility. They have a strong belief of benefits related to large family e.g. economic, bio-demographic and socio-cultural benefit (Sarker, 1993; Adongo *et al.,* 1997; Sheykhi, 1995). Similarly, in South Asian countries fertility associated matters are controlled by the senior family member and husbands (Kadir *et al.,* 2003; Karra *et al.,* 1997). The impact of family type on fecundity or fertility could be confused by the sex composition of living children due to the phenomenon of son preference, especially in Bangladesh, India, Nepal, and Pakistan (Karki, 1988; Greenspan 1992; Mwageni *et al.,* 2001). Reported studies revealed that with an increase in the number of living son the desire to have more children rises (Ali, 1989; Farooqui 1990; Hussain *et al.,* 2000). As reported by Pakistan Bio-demographic and Health Survey, Women who favored a baby with a specific sex were ten times more likely to desire that baby to be a male (Studies *et al.,* 1992). In addition, other factors affecting the fertility predilections of husbands within the ecology of family types could be socio-bio-demographic characteristics and knowledge and attitude towards family planning (National Institute of Population Studies); (Piet-Pelon *et al.,* 1999).

The present study was conducted to assess the association of subjects' age at marriage with fertility. The highest rate was observed in women with ">9-14" years age. The fertility was lowest among "upto 30" years age category. The fertility was negatively related to subjects' age at marriage. In many regions of Asia and Africa, present consanguinity rates are accounted for approximately 20-25 %. People from

these areas who have been migrated to Western Europe and North America also contract marriages with close cousins. Consanguinity is linked with increased gross fertility due to the younger maternal age at first live-birth. Earlier marriages and the younger maternal age at first live-birth in the close relative union are the factors which can increase both the pace of fertility and completed family size. Gynecological immaturity due to younger maternal age at first live-birth may have a hostile effect on developing fetus, which may be a contributing factor in greater mortality and morbidity in consanguineous couples (Bittles *et al.*, 1991).

4.1 Problems during field work

Most of the people and especially females of Haripur district are illiterate and due to lack of opportunities/facilities, they do not have an idea of field work conducted for data collection. Therefore, mostly female considered me as lady heath worker, from Benazir income support program, polio team worker, Pakistan census team member or any other financial assistant of NGO. Every woman was briefed and convinced separately for the participation and most of the time females left without completing the Proforma. Subjects showed no interest in the interview, despite the five to ten minute of briefing they still raised question during Proforma filling and gave no importance to the questionnaire

During the door-to-door survey, most of the time people do not allow me to enter in their houses and to interact with their females. People raised the question, such as, why we disclosed our marital information with you, consanguineous or non-consanguineous marriage is our personal matter and we did not need any genetic counseling or help from your team. The majority of the people thought that there were

no genetic anomaly in their family due to successive cousin marriages that is why they did not need any help or information, so they refused to participate in the study.

Due to worsening law-and-order situation in Haripur district, there is a general lack of trust on strangers. Sometimes it was very hurting for me to obtain data when respondent refused to co-operate by considering me a female robber. In such situations, I could not blame them because they are unaware but it was due to people who threatened them with losing their property or family information.

If female agreed and convinced for participation, their husband or other male guardian stopped her, got angry and excused us for their participation without any inquiry and clarification. Very few people were co-operative, most of them felt insecure upon disclosing their marital information. Most of the time, they avoid to give complete information and got irritating after few personal question like family income and household goods. Although I briefed them about my study but people remained in a doubtful condition throughout the process of Proforma filling. One of the most frequently asked question during home to home as well as the hospital-based survey was, “what will be the immediate benefit for us through this research”?

Currently, Pakistan is facing a number of challenges and one of the main problems is economy crises or unemployment that has forced people to rely on social support programs inaugurated by the local government like Zakkat fund and Be-Nazir income support scheme. Hence, considering me a team surveyor of that program they participate warmly but when I declared them about my purpose, suddenly their attitude was changed, they became unresponsive and some of them left without completing their Proforma.

As mentioned before, I collected the major part of my data from DHQ hospital, in general child OPD. Only one doctor was available for hundred to one hundred and fifty children on daily basis. Subject waited for their turn as their number called they instantly left without completing their Proforma. So, within the limited time of five to ten minutes, I had to fill my Proforma with complete and correct information.

During the door-to-door survey, in many cases, I visited for families but could not get data due to the absence of subject at home. Sometimes I visited families and faced problems when female denied giving any information in the absence of male at home, in hospital female first confirm from their male member then agreed to interview. Occasionally, I also face the problems of transport and could not continue my field survey according to my set schedule.

4.1.1 Problems due to perceptions about genetic diseases

Sometimes people disagreed to give information due to the following reasons.

- They considered that all the health problems they have are due to curse of saints to their forefathers.
- So people gave importance to their culture rather than to their health because in their view man is known by a culture so they prefer to follow them and said that problems are from God, and their culture (consanguineous marriage) is not responsible for any disease.
- Many subjects thought that genetic defects were due to some changes or events in nature like moon eclipse and are inherited in the family due to consanguinity.

4.2 Future prospective

The information gained through this epidemiological study would be valuable

- To assess the prevalence of morbidities/non-communicable diseases in the population of Haripur district.
- This study would be beneficial in the etiological study of hereditary/congenital anomalies in the population of Haripur district.
- Percentile breakdown of the Haripur district into bio-demographic and socio-economic variables would be helpful in order to understand the dynamics of consanguinity and IC-F.
- This study would be very beneficial to set the baseline for the different aspects related to consanguinity like mutational load, genetic burden and population stratification in Haripur district.
- This study would help to control different bio-demographic aspects which affect the total consanguinity rate. It would set an initiative for awareness program related to adverse effect and risk of rare recessive disorder due to consanguineous unions.

4.3 Limitations of this study

- The data were collected from females only and male subjects were not recruited. The bulk of the data were collected from housewives while professional ladies were very less in number and may be unrepresentative.
- Due to less awareness about the epidemiological survey, women often hesitated to give complete bio-demographic information. This deficiency was compensated by recruiting a large number of subjects.
- Direct information about the economic status could not be gathered accurately because people did not give the complete information. So data were not analyzed according to the economic status.
- This study does not present the association, if any, between subjects' consanguinity and parental consanguinity because the data about parental consanguinity is missing.
- The major part of these data consist of subjects less than 50 years age, that's why the exact information about the morbidity rate in recruited subjects may be unreported.
- Lastly, the influence of consanguinity on adult mortality/morbidity remains to be explored in Haripur district of KPK. Recently reported study in Kashmir, Pakistan revealed that consanguinity was not associated with mortality/morbidity in adult women.

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