**Appendix**

**Genetic defects may lead to recurrent miscarriage or foetal mal-development**

Couples carrying genetic diseases may experience recurrent miscarriage (two or more miscarriages) or foetal mal-development in their babies (e.g. structural malformation or intellectual disabilities). For example, parents may have a balanced chromosomal translocation, which may then result in unbalanced translocation (deletion and duplication) in their offspring, leading to miscarriages. On the other hand, an asymptomatic couple carrying the same autosomal recessive gene may have a one-quarter chance of having a child with a major genetic disorder. The chance of the couple carrying the same genetic defect is much higher when they are consanguineously related. Sometimes, if a mother carries an X-linked genetic defect, half of her male babies will suffer from the disease caused by the abnormal gene (e.g., mental disability in Fragile X disease, or coagulation problems in haemophilia), even if her partner has no genetic defects.

In the above three scenarios, the carrier parents are healthy and asymptomatic. They are unaware of their underlying conditions until they experience repeated pregnancy loss, malformation or disability in their babies. In the past, due to limited awareness of the genetic defects related to adverse pregnancy outcomes, as well as limited resources, affected couples suffered immense physical and psychological distress. Their family planning was also delayed.