Introduction
Many important discoveries have been made in the last five decades in the field of hereditary mechanisms, environmental teratogenesis, prenatal diagnosis and medical/surgical management of affected individuals with congenital malformations. The Maltese group of islands occupies a central position in the Mediterranean, being only 93 km away from Sicily and 290 km from Northern Africa. This location made the Islands an important meeting place for the various Mediterranean cultures throughout the ages. The history of malformations in the Maltese Islands reflects the cultural links with the mainland Mediterranean. The attitude towards congenital malformations can be divided into two periods - the Period of Superstitious Belief which reflected primitive man’s concepts and prevailed to at least the 17th century; and the Period of Modern Thought which began during the 18th century but made the most progress during the twentieth century.

The Period of Superstitious Belief
Congenital malformations causing deformities have been described since early times. Primitive man’s interest in these phenomena has found expression in drawings, carvings and sculptures throughout the world including Australia, the South Pacific Islands, and the Americas. Written records of congenital malformations have come down from Babylon in the form of clay tablets from the Royal Library of Nineveh which was assembled by the Assyrian King Asshurbanipal (c.700 BC). These tablets include a list of sixty-two human malformations with their associated prophetic implications.1-2 It is believed that these records date back to 4000 BC. Direct evidence of congenital malformations in primitive cultures were found in skeletal remains of affected individuals, such as a specimen of an open sacrum and a femur described from Neolithic France with changes suggestive of congenital dislocation of the hip.3 The first treatise on monsters appeared in the sixth century AD by Isidore of Seville who described numerous abnormalities and tried to give natural reasons for their occurrence, although supernatural causes were not discounted. From the mid-16th to the mid-17th centuries, several books on monstrous births were published and a number of authors published a variety of collections of such births and discussions on their cause.4

The Maltese skeletal archaeological record shows evidence of congenital abnormalities which date to the Palaeolithic era, circa 18,000 years ago. Two teeth with taurodontism were found in association with remains of Cervus (deer).5 Taurodontism was a common feature in Homo neanderthalis, but in a minor form is also found in modern man, particularly in association with genetic anomalies such as the presence of an extra X or chromosome 21 and Prader Willi syndrome. Neolithic skeletal remains excavated from Brochtorff Circle at Xaghra (Gozo) have identified other congenital skeletal malformations and non pathological anomalies.

These malformations include a case of a sacral spina bifida in Tarxien Phase material. Other abnormalities include absence of the sagittal suture in a skull from Hal Saflieni Hypogeum (Malta).6 The earliest depicted congenital malformations in Malta date from the Neolithic era and include cases of abnormalities of the hand: one with three digits and one with six digits. These representations were discovered in an incised decoration from Gzibbu Tombs at Zebru (Malta) and a statuette from Hagar Qim. Polydactyly is also evident in a hand print described from the Hal Saflieni Hypogeum.5-8

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Other skeletal remains showing congenital anomalies were excavated from Roman tombs. This included a non-pathological anatomical variation of the sacrum. Here the transverse process of the first sacral vertebra was not fused with the rest of the bone. In addition two adult skulls from St. Agatha Catacombs showed features of non-union of the frontal bones. The archaeological skeletal record also describes a number of late medieval/early modern congenital anomalies from excavations of burials in Maltese churches. These skeletal anomalies included diverse sacral anomalies including a case of spina bifida and other minor anatomical variations.

The written records of the hospitalier period furnish a number of descriptions of congenital malformations which appeared before the Episcopal court, the Inquisitional Tribunal or the Civil Courts. The first congenital malformation described in Malta dates to 1542, a case of severe male genital malformation (hypospadia) which appeared before the Episcopal Court as a case for marriage annulment. Another case of severe hypospadias was described in 1744 in the Law courts for the legal change of sex registration. In 1630 a description of an abnormal infant was submitted by the midwife Olivia Gambino to the Episcopal Court to justify why a stillborn was not buried in consecrated ground. The description suggests that the parturient mother gave birth to two macerated monsters - one with human and the other with avian features.

The theory of supernatural causes for the development of congenital malformations held sway from prehistoric time well into the seventeenth century. Congenital malformations in the various cultures and religions were often regarded as a punishment from the gods. With the advent of Christianity to the Islands, fetal anomalies were believed to originate along biblical beliefs by copulation between a woman and a male beast or between a woman and the devil. The birth and death of a grossly malformed infant or monster posed an intriguing quandary to the canonists. The officiating priest was enjoined to examine the malformed infant or monster and ascertain that its principal parts, namely the head of an animal and the limbs those of a human, the creature was baptised sub conditione si es homo ego et baptism. As late as the mid-nineteenth century canonists were still debating the religious issues raised by the birth of monsters. In their conclusions they were swayed by the conviction that these deformed beings were the result of copulation between a woman and a male beast or devil. The belief was prevalent in 17th century Malta that consort with the devil in an assumed form was possible. In 1647, 17-year old Gertrude Navarre accused herself before the Inquisitional Tribunal of having over the previous six years had carnal relations with men and animals brought to her by the devil. She became pregnant and procured abortion on several occasions. In 1676, a Maltese physician’s wife accused herself of having invoked the devil and invited him to have sexual relations with her if he destroyed her husband.

Another aetiological theory was that of maternal impressions i.e. states of mind, affecting fetal development. However, John Hunter presented scientific evidence against maternal impressions in the late 18th century. In Malta maternal impressions were also considered important aetiological factors. In the 1749, the Maltese physician Dr. Salvatore Bernard adhered to the theory that the fantasy organ of the pregnant woman communicated by means of animal spirits with the fantasy organ of the foetus, so that any perception aroused in the mother’s mind produced a similar impression in the fetal brain, which impression in turn reacted upon and moulded the form of its body. Dr. Bernard held that monsters having the shape of animals and devils were born to women who, during pregnancy, had been exposed to the sight of these creatures. These beliefs remained ingrained in Maltese mentality until relatively recent times, and many congenitally malformed individuals were kept hidden away from view by their families. A special hazard of pregnancy was the emergence of longings or desires which could not be satisfied. According to popular belief, the new-born will manifest a birth-mark resembling in form and colour the object of the unfulfilled desire. If a person neglects to satisfy the wishes of a pregnant woman, he/she would be punished by suffering from a stye. This view was not universal. In 1804, Dr. Francisco Butigieg did not share the notion that “in an alteration of imagination which affected the foetus in such a way as to produce a defective baby or a monstrosity”.

The Period of Modern Thought

Modern science was born in the 15th and 16th centuries during the Italian renaissance. The Renaissance initiated a process which during the eighteenth century, saw the beginnings of modern attitudes towards the aetiology of malformations. The developmental arrest theory of teratogenesis was first developed by William Harvey (1651) who reasoned that cleft lip in human infants was very similar to the normal situation found in early embryos. This concept was extended to explain ectopia cordis and gastroschisis. Although not totally understood, inherited anomalies had been recognised by Ambroise Pare (1649) and John Hunter (1775). However the genetic theory as a cause of malformations gained momentum when the inheritance laws described by Gregor Johann Mendel (1865) were rediscovered in 1900 by Carl Correns, Hugo de Vries, and Erich von Tschermak.

In Malta, superstitious concepts regarding the aetiology of congenital malformations apparently persisted well into the eighteenth century, though an academic interest in the condition was being taken by medical practitioners. A case of severe malformation was described in 1788 by Dr Saverio Fenech. The report records the birth of a monster born to a woman at Nadur. The child had a head and ears which resembled those of a cat. The genital parts were also similar to those of a female cat. The attention given to the description, together with the added comment that the attending priest considered these characters as conforming more to a human form requiring Christian burial suggests that the religious concepts regarding malformations were still prevalent.
This was not surprising since these concepts remained prevalent until the early decades of the twentieth century. Medical publications in Malta during the nineteenth century do not shed light on the beliefs prevalent among Maltese practitioners regarding the aetiology of congenital malformations. However, the description of a Maltese family in 1891 with a genetic predisposition to having twenty-four digits suggests that the hereditary theory was being entertained for at least some anomalies.

Other cases of malformation were described for academic purposes by medical practitioners during the nineteenth century. A case describing the internal and external features of conjoined twins with one head was described in 1816, while further cases of malformed babies born in Malta were published in the 1840s. A case of obstructed labour resulting from fetal gross enlargement of the liver due to malformation was described in 1891. The infant also had a cervical spina bifida and extra digits. Prof Giuseppi Schembri in 1896 referred to malformations which may occur, including conjoined twinning with head fusion and union by the chests, or by the pelvis giving rise to two complete trunks with two or four legs. He also listed a number of malformations that could be met by midwives, including atresia of the rectum, ears, and urethra; hypospadias; harelip and cleft palate; congenital protrusion of bowels within the nasal string; conjoined or supernumerary fingers or toes; club foot; spina bifida situated at the neck and the lumbar region; hydrocele; port-wine stains, mother’s marks, moles, and tongue tie.

There is evidence of a three-legged conjoined twin who lived in Malta for some time in the early twentieth century. Photographs taken in Malta of a seven to nine year old child with this anomaly have been found among the belongings of Professor Ruggiero Busuttil. The origins of this child remain unknown, and it has been suggested that the child may have been the same three-legged Francisco Lentini who was born in Sicily in 1889 and who later emigrated to America where he lived in Malta for some time in the early twentieth century. In 1891, with a genetic predisposition to having twenty-four digits. Other cases of malformation were described for academic purposes by medical practitioners during the nineteenth century. A case describing the internal and external features of conjoined twins with one head was described in 1816, while further cases of malformed babies born in Malta were published in the 1840s. A case of obstructed labour resulting from fetal gross enlargement of the liver due to malformation was described in 1891. The infant also had a cervical spina bifida and extra digits. Prof Giuseppi Schembri in 1896 referred to malformations which may occur, including conjoined twinning with head fusion and union by the chests, or by the pelvis giving rise to two complete trunks with two or four legs. He also listed a number of malformations that could be met by midwives, including atresia of the rectum, ears, and urethra; hypospadias; harelip and cleft palate; congenital protrusion of bowels within the nasal string; conjoined or supernumerary fingers or toes; club foot; spina bifida situated at the neck and the lumbar region; hydrocele; port-wine stains, mother’s marks, moles, and tongue tie.

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Further to the list given by Prof Schembri in his lecture notes to midwives in 1896, there is to date no further identifiable historical material which deals with the epidemiology of abnormalities. Published mortality data suggests that in 1896, malformations accounted for a total of 76 deaths resulting in a specific mortality rate of about 43.1 per 100,000 population. All of these deaths occurred in children under five years of age. The number of deaths from malformations apparently decreased in the first decade of the twentieth century to eventually maintain an annual fluctuation around 8.5 per 100,000 population in the 1920s. The specific mortality rate from congenital malformations started to rise just prior to the Second World War to peak to a value of about 18 per 100,000 population in the 1950s. After the 1950s, the specific mortality rate fell progressively to reach mean values of about 7 per 100,000 population in the 1980s. The majority of these deaths occurred in childhood so that the annual childhood mortality rate from congenital malformations during 1981-85 was 54.2 per 10,000 total births. The larger proportion of these deaths throughout the years was caused by congenital heart lesions, with the second commoner group of malformations being abnormalities of the central nervous system including hydrocephalus and spina bifida.

While a number of studies had been undertaken by individual clinicians in Malta on specific congenital disorders, a comprehensive epidemiological study of congenital malformations in Malta was not seriously initiated in the early 1980s. This lack of data is not surprising in view of the fact that before the First International Conference on Congenital Malformations in 1960, there were very few studies in the world literature on incidence of congenital malformations.

A small preliminary survey of congenital anomalies in live-born Maltese children was performed in 1972 reporting 20 anomalies in a total of 1016 consecutive births. In 1983, a computerised database was initiated by the Department of Obstetrics and Gynaecology in order to allow the registration of all births at Karen Grech Hospital. This database allowed a number of epidemiological studies to be initiated. Malformations were shown during 1983-1986 to occur in about 3.5% of all births, with minor malformations accounting for about 77% of all malformations. While minor malformations were shown to be approximately the same in all age groups, the major malformation rate increased with increasing maternal age.

A further database was initiated after 1986 by the Departments of Genetics and Obstetrics-Gynaecology in conjunction with the EUROCAT Project. This project is a concerted action of the European Union in the epidemiology of congenital anomalies. The EUROCAT data base initiated in 1978 has indicated that the malformation rates in Europe should range from a minimum of 0.9% to a maximum of 4.6%. The EUROCAT project has further shown that the incidence of anomalies caused by chromosomal problems in Europe varied in incidence from 9.4 - 22.3 per 10,000 total births. Of all the registered cases, approximately 74% were Down Syndrome. In Malta during the period 1983-87, the prevalence of chromosomal abnormalities was estimated at 22.0 per 10,000 births or a frequency at 1:434. The prevalence of Down Syndrome in Malta was estimated at 18.8 per 10,000 births (frequency 1:532) or 85.5% of chromosomal anomalies diagnosed. The frequency of Down Syndrome was markedly higher in older mothers. The lowest risk of occurrence appears to be in the 25-34 year age group, and it is approximately three times higher in mothers below 20 years and 25-30 times higher in the 40-45 years age group.

The last decade has seen further efforts at epidemiological studies with the continuation of the EUROCAT Project and the establishment of the Malta Congenital Anomalies Register, and the initiation of several databases to assess the incidence and prevalence of specific congenital malformations particularly thalassaemia (Laboratory of Molecular Genetics) and congenital heart disease (Maltese Paediatric Cardiology Database).
References

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