World Birth Defects Day 2020

Many Birth Defects – One Voice

Spina Bifida

World Birth Defects Day (WBDD) is an annual global event, initially held in 2015. Its purpose is to raise awareness and provide information about birth defects to help with their prevention, management and thus improve care. It is estimated that 3% of babies are born with a congenital anomaly (birth defect). Although some of the defects are minor, the major ones contribute to a high morbidity and mortality.

Some birth defects are known to be preventable by proper measures before and during pregnancy, thus making it necessary to strengthen and support research and policy programs to find the causes and potential avoidance of such defects. Educating the public is essential as it is one of the most effective interventions for the primary prevention of these conditions.

A major birth defect that is potentially preventable is Spina bifida.

What is Spina bifida?

Spina bifida is a birth defect caused by a flaw in the development of the nervous system namely the neural tube. The neural tube is a developmental structure within the embryo from which the brain and the spinal cord are formed. The neural tube starts developing from a special group of cells soon after the conception of the embryo. It is fully formed by around the 28th day from conception, following which further development continues to occur leading to the formation of the brain at the head, with the rest of the tube developing into the spinal cord. In Spina bifida, part of the neural tube fails to develop properly and close completely into a tube resulting in abnormal development of spinal cord. The development of the meninges (protective coverings of the spinal cord) may also be affected.

Different types of Spina Bifida

There are different types of Spina Bifida which vary both in the way they present and in their severity.

Spina bifida Occulta is the least severe type of defect and is also the most common. It manifests as a malformation of one or more vertebrae, however the gap in the vertebrae is very small, and a layer of skin covers the malformation (hence the name “occulta”). It rarely causes any symptoms and in fact most affected individuals would be asymptomatic.

Meningocoele is a more serious type of Spina bifida. In this type, the protective membranes around the spinal cord (meninges), along with spinal fluid, protrude through an abnormal opening in the bony vertebral column

which houses the spinal cord. This protrusion however does not contain any neural elements and therefore the spinal cord usually develops normally. The protrusion may or may not be covered by a layer of skin. While some individuals have no symptoms due to sparing of neural elements, others may be more severely affected, suffering from bladder and bowel problems.

Myelomeningocoele is the most severe form of Spina bifida. In this type, the baby's spinal cord and neural elements are exposed through an abnormal opening along several vertebrae in the back. Individuals are generally severely affected with partial to complete paralysis of the body below the defect. Bladder and bowel function are also commonly affected.

Causes and Risk Factors for Spina bifida

The exact cause of Spina bifida remains unknown, however there are multiple factors which may increase the risk of a baby developing the condition.[1]

A low intake of folic acid by the mother prior to conception, and throughout the first four weeks following conception, is strongly associated with the development of Spina bifida and other neural tube defects. While it is still unclear how folic acid helps prevent Spina bifida, it most likely plays an important role in biochemical reactions which drive neural tube development.

Furthermore, the risk of having a baby suffering from a neural tube defect is higher if a family member already suffers from the condition. When a mother already has a child with Spina bifida, the chances of having other children suffering from the condition is significantly increased - from less than 1 in 1500, to around 1 in 25.[2]

Diagnosis of Spina bifida

Often the diagnosis of Spina bifida is made before the baby is born, during the antenatal anomaly ultrasound scan - which is generally offered to all pregnant mothers between 18 to 21 weeks of pregnancy.[3] In some cases, such as in the case of the milder forms of the disease, the diagnosis may go unnoticed until after birth.

Apart from ultrasound scans, Spina bifida may also be suspected when testing the maternal blood serum alpha-fetoprotein (AFP). Although this test is not specific for Spina bifida, when higher than expected, it can indicate that the baby may have a neural tube defect. Normal AFP levels vary according to the fetal age therefore the gestational age needs to be known accurately when interpreting this blood test. On its own, AFP levels cannot definitively determine the presence of Spina bifida and in cases where there is a high level of AFP in maternal serum, further testing is required.[4]

Once the baby is born, tests may be carried out to assess the severity of the condition and help direct treatment. These tests may include magnetic resonance imaging (MRI) or a computed tomography (CT) scan to get a clearer view of the spinal cord and vertebrae. Hydrocephalus (excess fluid inside the brain) commonly accompanies Spina bifida.

Management of Spina bifida

There is no cure for Spina bifida since damaged neural tissue cannot be repaired. Treatment is focused at supporting the child's health and the management of complications. Surgery to close any open defects, and therefore minimize the risk of infection or further trauma to the spinal cord, is generally carried out within the first few days of the baby's life. When hydrocephalus is diagnosed, a ventriculoperitoneal shunt may be inserted to help drain the fluid from around the brain into the abdomen.

Physiotherapy and occupational therapy are also important in the child's care. These therapies aim to maximize the child's (and future adult's) functionality and improve independence. Furthermore, assistive devices, such as crutches and wheelchairs, where indicated, can also help in achieving these aims.

As people with Spina bifida may have bowel and urinary problems, care and treatments may also be offered to help manage these issues. Bladder catheterization may be used in cases where problems to empty the bladder exist. This will in turn help to avoid the development of urinary tract infections and kidney malfunction. Bowel management regimens may also be taught to the child where bowel continence is a problem.

In cases where there is paralysis of the lower body, and where sensation of the lower part of the body is impaired, care is needed to make sure that the child's skin stays healthy. This is important as the child may not be able to feel pressure on their legs and feet, and therefore they would not recognize when they have hurt themselves. This in turn may subsequently lead to further skin complications, such as skin ulceration.

Prevention of Spina Bifida

Research has shown that prevention of certain cases of Spina bifida is possible by taking folic acid supplementation before and during pregnancy. While taking folic acid does not guarantee complete prevention of this condition, there is very strong evidence from studies and experience that it significantly reduces the incidence.

International guidelines[5] recommend that women of childbearing age take 400 micrograms of folic acid daily prior to and until the twelfth week of the pregnancy. Foods high in folic acid include dark green vegetables, egg yolks, and some fruits. In some countries, foods are fortified with folic acid (such as breakfast cereals, flours, rice). Fortification of food stuffs with folic acid in these countries has been associated with a decline in the prevalence of neural tube defects including Spina bifida.[6-8]

Some women may be at a higher risk of having a child a neural tube defect. Women at higher risk are those with a family history, those who have already had a pregnancy affected, women who smoke, are on anti-epileptic drugs or folate antagonists. These women benefit from taking a higher daily dose of folic acid.[9]

Outlook for individuals with Spina bifida

Prognosis varies according to the type and severity of Spina bifida. Although there may be significant challenges to daily life, many children with the condition lead active and fulfilling lives. The spectrum of severity of the abnormalities is wide, most will have normal intelligence and will be able to walk, sometimes with assistive devices. These children can attend school, make friends and contribute to society.

Epidemiology of Spina Bifida in Malta

During a 25-year period (1993-2017), 65 infants were born with spina bifida in Malta and Gozo, giving an overall prevalence rate of 6 per 10,000 total births (total births include both live and still births).

Gender distribution

More male infants (55.4%) were born with spina bifida (36 males vs 29 females), this pattern has also been reported in other studies.[10] A total of 6 (9.23%) cases of Spina bifida occurred as part of a chromosomal syndrome (3 females, 3 males).

Maternal Age

Maternal ages at both ends of the age spectrum (i.e. young and older mothers) may be associated with an increased risk of neural tube defects. In this cohort, the mean maternal age of mothers was 29.82 (±5.89) years. In Malta, spina bifida was more prevalent in the 25-36 age group. Of all 65 mothers, 26 were primiparas (first pregnancy).

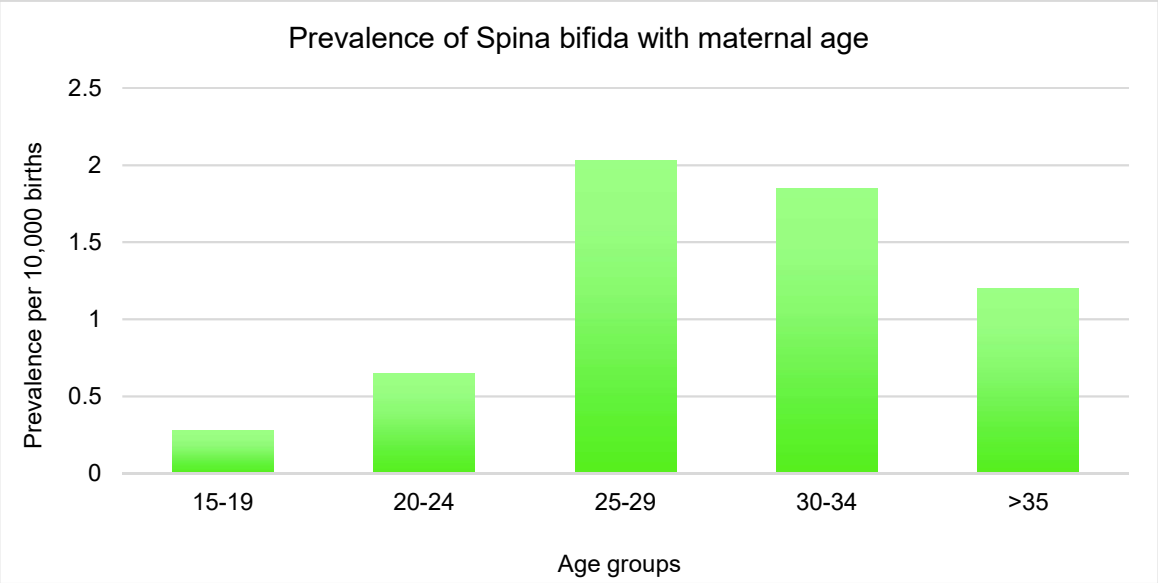

Figure 1: Prevalence of spina bifida in Malta with maternal 5-year interval age groups

Preconception Folic acid

Maternal pre-conception folic acid consumption is known to decrease the risk of neural tube defects, unfortunately not all mothers take folic acid before pregnancy and a recent study in Malta analysing 2015 data showed that only 25.7% of all mothers took folic acid before pregnancy.[11] In Malta, folic acid intake has been recorded since 2006, for the registered spina bifida cases born between 2006-2017, only 5 mothers out of 30 (16.7%) were reported to take pre-conception folic acid.

Antenatal diagnosis

Between 1993 and 2017, 25 (38.5%) cases were diagnosed at antenatal screening, whilst the rest were diagnosed at birth. Of the those diagnosed prenatally, 7 cases were diagnosed at 24 weeks and below whilst 17 cases were diagnosed above 24 weeks. Most of the foetuses were alive at time of discovery (64 alive, 1 dead).

Spina bifida case diagnoses at the beginning of the 25-year time series (1993-2002) showed that only 9 (31%) were diagnosed antenatally whilst 20 diagnosed at birth. However, at the end of the time series (between 2007-2017), 14 (63.6%) were diagnosed antenatally whilst 8 were diagnosed at birth. This shows that antenatal diagnosis of Spina bifida has improved throughout this time period.

Prevalence in Malta and Europe

The overall European prevalence for spina bifida cases (both live and dead) reported by EUROCAT (European Surveillance of Congenital Anomalies)[12] over the period 1993-2017 was 5.14 per 10,000 births. Over this period, there was an increase in trend in prevalence between 1993-2000 and since then there has been a downward trend in Europe (Figure 2).

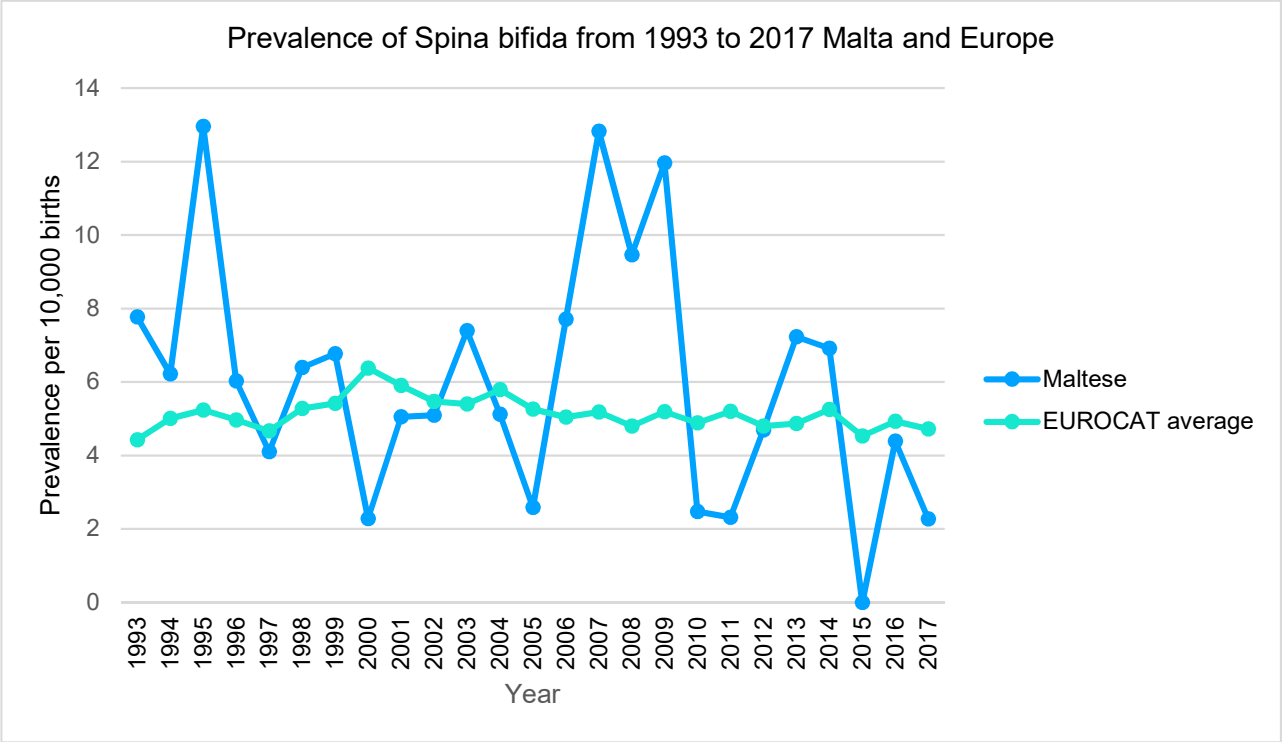

Figure 2: Trends of Malta and EUROCAT average over the 25-year period (EUROCAT data obtained for all full registries).

While EUROCAT average prevalence rates of spina bifida show a downward trend, prevalence rates in Malta over the 25-year period do not show statistically significant trends. Given the small numbers in Malta it is more difficult to elicit statistically significant trends.

Figure 2 shows the ***total*** prevalence rates of Spina bifida reported by different EU countries. Bulgaria had the highest reported total prevalence (12.29 per 10,000 births) whilst Portugal has the lowest (2.41 per 10,000 births). The EUROCAT average total prevalence rate was 4.32 per 10,000 births. Malta reported a total prevalence of 6.01 per 10,000 births which is higher than the EUROCAT average rate and similar to the rate reported by the UK.

The ***live birth*** prevalence for Malta was 5.45 per 10,000 births (1993-2017), being much higher than the average EUROCAT live birth prevalence rate (2.07 per 10,000 births) (Figure 3). This is to be expected as termination of pregnancy is illegal in Malta.

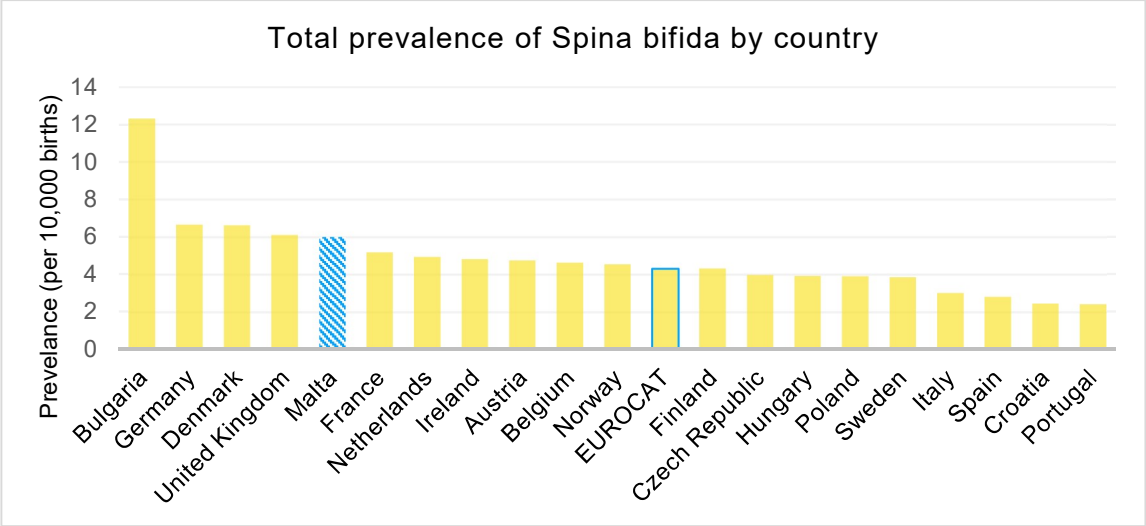

Figure 3: Total prevalence of Spina bifida in EU countries and EUROCAT average over the 25-year period

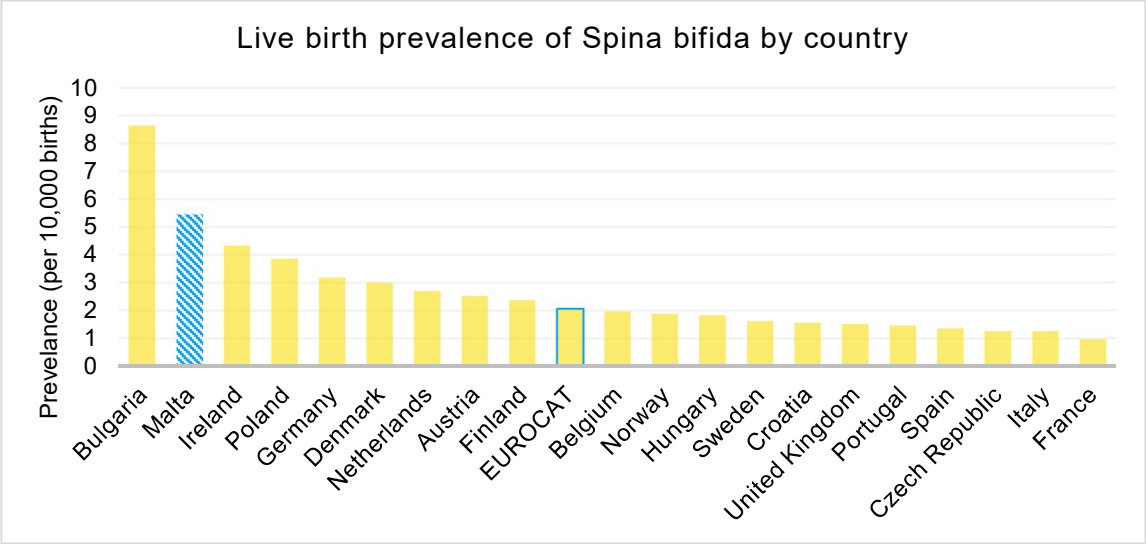

Figure 4: Live birth prevalence of Spina bifida in EU countries and EUROCAT average over 25-year period

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