World Birth Defects Day 2019

Many Birth Defects – One Voice

Join us on March 3, 2019

#WorldBDday; #ManyBirthDefects1Voice.

Birth defects, also known as congenital anomalies, can be structural or functional defects that are present at birth even though they may sometimes be diagnosed later on in infancy. Birth defects vary widely in severity from relatively minor to major, lethal defects. They occur at an overall rate of 2-3% of all births, however the individual types of birth defects occur at far lower rates and many are considered as rare conditions.

The first World Birth Defects Day (WBDD) was held on March 3rd 2015. The focus of this day has been to raise awareness about birth defects, the scale of the worldwide problem and to advocate for more surveillance, prevention, care and research to help babies and children born with these conditions. In 2018, 130 organizations joined the WBDD efforts formally and millions of people are reached with awareness messages through various social media platforms. Further information can be found at: https://www.worldbirthdefectsday.org/.

More than 8 million babies are born each year around the world with serious birth defects. These are an important cause of death and morbidity in the first year of life. Babies who survive may suffer permanent physical or mental disabilities, with consequent emotional and financial cost to their families, communities and countries. However, much can be done to prevent certain birth defects, hence the importance of raising awareness.

In Malta, birth defects are monitored through the Malta Congenital Anomalies Register of the Directorate for Health Information and Research (DHIR).

Further information on the Malta Congenital Anomalies Register may be found at:
https://deputyprimeminister.gov.mt/en/dhir/Pages/Registries/birthdefects.aspx

Health professionals may notify new cases through the secure portal found at:
https://deputyprimeminister.gov.mt/en/dhir/Pages/Notifications/nocaf.aspx

Hypospadias

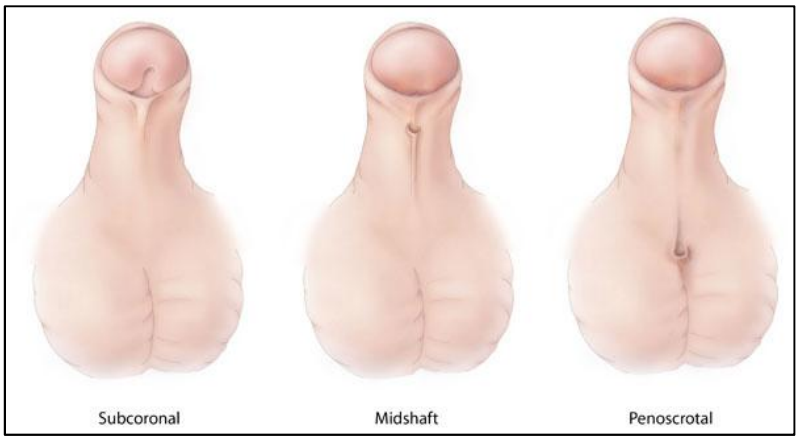
Hypospadias is the most common congenital abnormality of the urethra and second most common genital anomaly after undescended testes, reported to affect 1:300 live births worldwide.[1] Hypospadias is the abnormal location of the urethral opening on the ventral surface of the penis, with the meatus location being anywhere from the glans (head of penis), to penile shaft, scrotum or perineum.[2] Hypospadias varies significantly in severity and classifications are based on the location of the external meatus. The WHO International Classification of Diseases classifies hypospadias into four categories from the least to the most severe types: Balanic (includes coronal and glandular hypospadias), Penile, Penoscrotal and Perineal.[3]

Depending on the location of the urethral opening, the condition can cause misdirected urinary stream and may also lead to problems with sexual function later in life if the penis is also curved (chordee). The frequency of associated genital anomalies increases with the severity of hypospadias, with undescended testis and inguinal hernia being the most common associated anomalies, while curvature of the penis is common in the more severe cases.[1]

There have been a number of studies from different countries showing an increasing trend in the prevalence of hypospadias.[4] It is generally accepted that hypospadias is a highly heterogeneous condition that is subject to multiple genetic, endocrine, placental and environmental factors.[5,6,7]

Genital examination of male neonate

The new-born examination includes an overall assessment of the neonate, when hypospadias is diagnosed, its severity is determined and any associated congenital anomalies that could suggest a possible syndromic cause of hypospadias are looked for. It is important to distinguish clinically significant hypospadias that needs urologic referral for possible early surgical correction from the more benign forms which have little or no impact on voiding and sexual function, thus not requiring urgent intervention.[8]



Increasing severity of hypospadias (left to right): distal (sub-coronal); penile midshaft and penoscrotal

(Taken from: Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities)

Risk factors

Several risk factors have been associated with hypospadias, these include: i) advanced maternal age, ii) pre-existing maternal diabetes mellitus, iii) gestational age before 37 weeks, iv) history of paternal hypospadias, v) exposure to smoking and pesticides, vi) placental insufficiency (low placental weight and pathology), vii) prematurity, viii) fetal growth restriction and in-vitro fertilization.[9]

Management

Urologic referral is advised and is most important for patients in whom there is a potential functional issue. Management revolves around surgical correction of the defect, according to Keays & Dave (2017), surgical intervention for hypospadias can be performed at any age, however, most authors recommend operative intervention at 6 to 18 months. The American Academy of Pediatrics suggests this period to limit psychological stress and subsequent behavioural problems which can be seen in toddlers undergoing genital surgery.[10]

Surgery for hypospadias is elective and the decision to operate is based on severity, family preference and surgeon's advice. Patients with mild variations of hypospadias may choose not to have surgical correction especially if the penis is functional in respect to cosmetics, voiding, future sexual function and fertility. The surgical management of hypospadias aims mainly to achieve a straight penis with an adequate meatus at the apex of the glans.[10]

Hypospadias in Malta

In the 10-year period between 2007 and 2016 there were 126 infant boys registered with hypospadias in Malta with a prevalence of 30/10,000 total births (1 in 333 births). The table below shows how many cases there were each year by type of hypospadias:

	2007	2008	2009	2010	2011	2012	2013	2014	2015	2016	Grand Total
Balanic	7	9	4	2	10	5	8	7	4	6	**62**
Penile	4	7	4	4	5	3	4	4	2	5	**42**
Penoscrotal/Perineal	2	2	1	1	2	2	1	0	2	4	**17**
Unspecified	0	1	0	0	0	0	1	0	2	1	**5**
Grand Total	**13**	**19**	**9**	**7**	**17**	**10**	**14**	**11**	**10**	**16**	**126**

Table 1. Number and type of hypospadias registered each year between 2007 and 2016.

Table 1 shows that the most common form of hypospadias encountered is the least severe type - Balanic, which includes both coronal and glandular forms. This is followed by the Penile and then Penoscrotal and Perineal types. In 5 cases (4%) the type of hypospadias was not reported. Cases of isolated congenital chordee were excluded from this analysis.

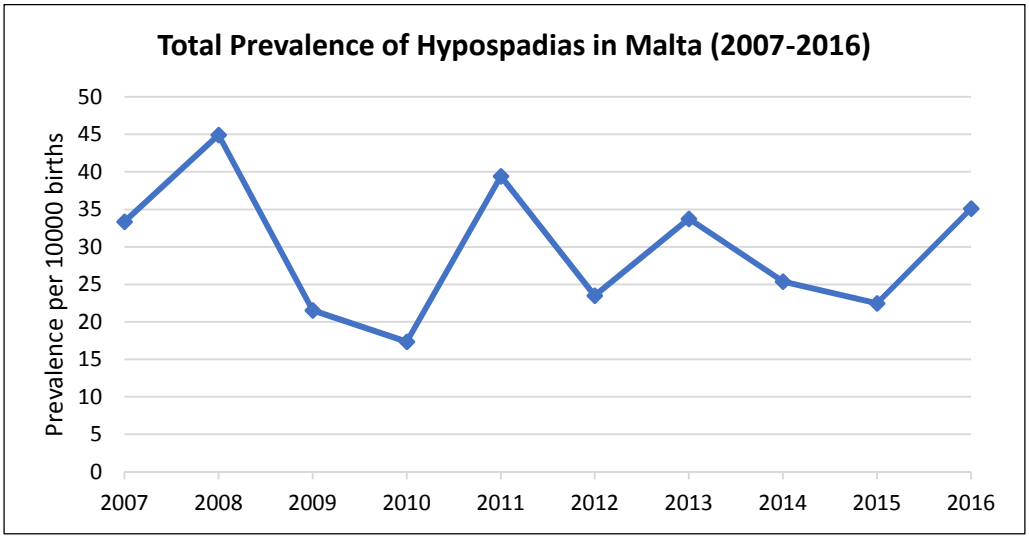


Fig 1. Prevalence of hypospadias between 2007 and 2016.

No significant times trends are seen in this period, with chi square for trend having p=0.59.

The prevalence of the different types of hypospadias over time is presented in Fig. 2. Here as well, no statistically significant time trends are noted for any of the categories of hypospadias.

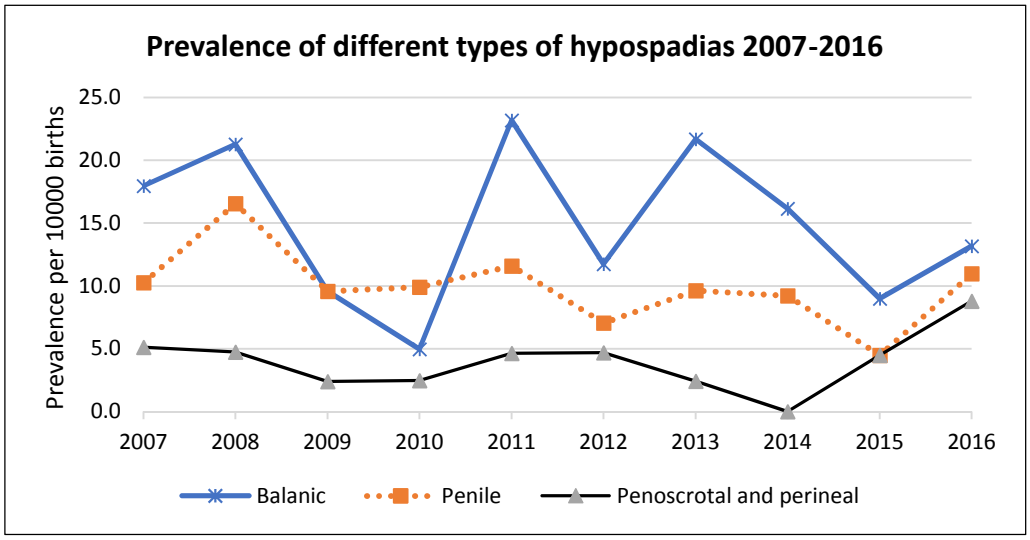


Fig 2. Prevalence of hypospadias by type.

Hypospadias and maternal age

Analysis of the data from Malta shows that overall prevalence of hypospadias is similar in all maternal age groups with p=0.55 (Fig. 3).

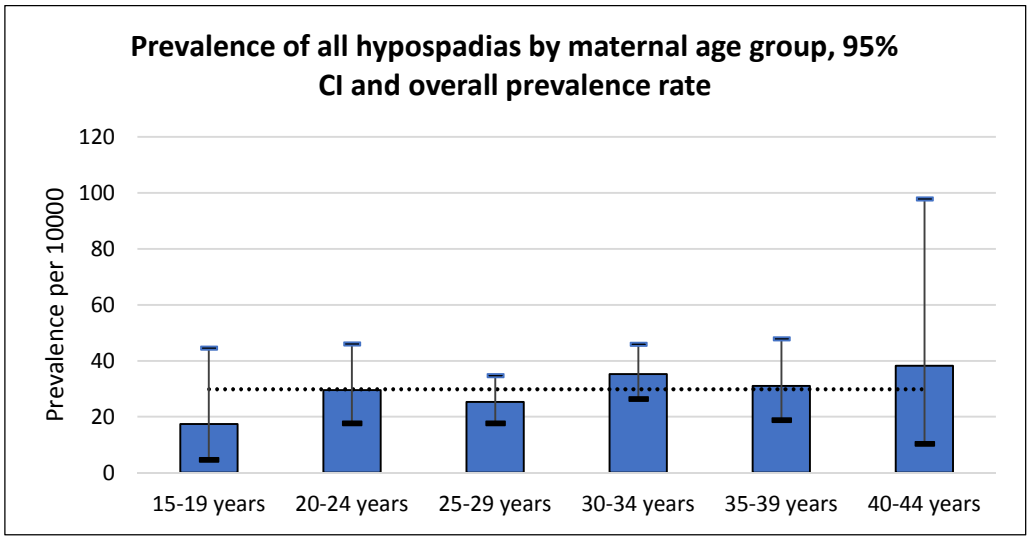


Fig 3. Prevalence of hypospadias per 10,000 births by maternal age group (2007–2016).

Hypospadias and associated anomalies

Fig.4 shows that 86.5% of all the recorded cases were isolated hypospadias, while 10.3% occurred within a picture of multiple comorbidities, 1.6% occurred in babies with chromosomal anomaly and a further 1.6% occurred in babies with a genetic disorder.

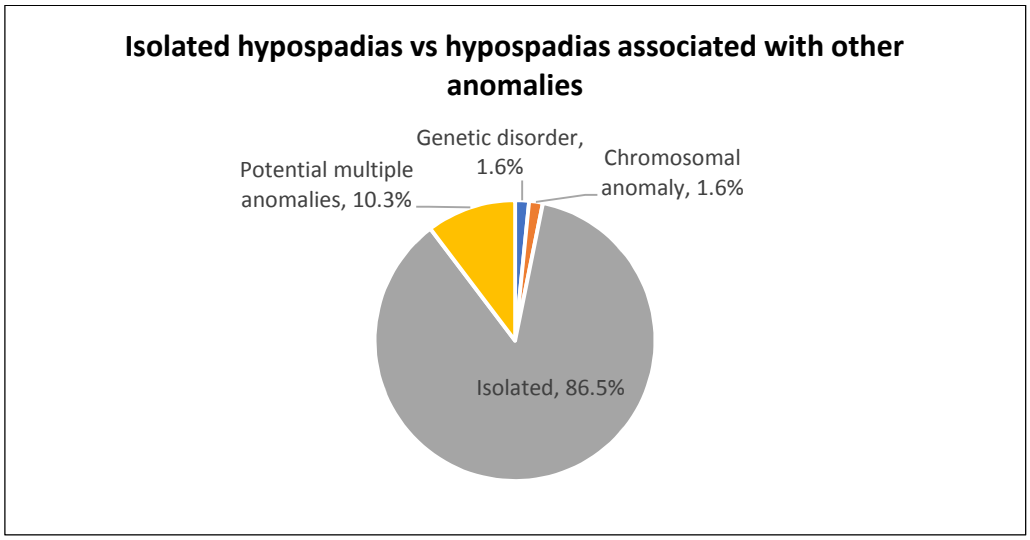


Fig 4. Percentage of cases being isolated vs cases having associated anomalies.

Hypospadias in Europe

EUROCAT is a European network of population-based registries involved in the epidemiologic surveillance of congenital anomalies. The prevalence of hypospadias reported by the Registries within this network varies widely from 31.28/10,000 in Czech Republic to 6.41/10,000 in Southern Portugal for the years 2007-2016. The prevalence reported from Malta for the same period was 30.89/10,000 (Fig. 5).

The wide variation in reported prevalence has been partly attributed to differences in case ascertainment by the different registries, especially for the minor cases.[5,11] Higher rates are often reported from the Registries that have very good ascertainment of even the minor forms of hypospadias.

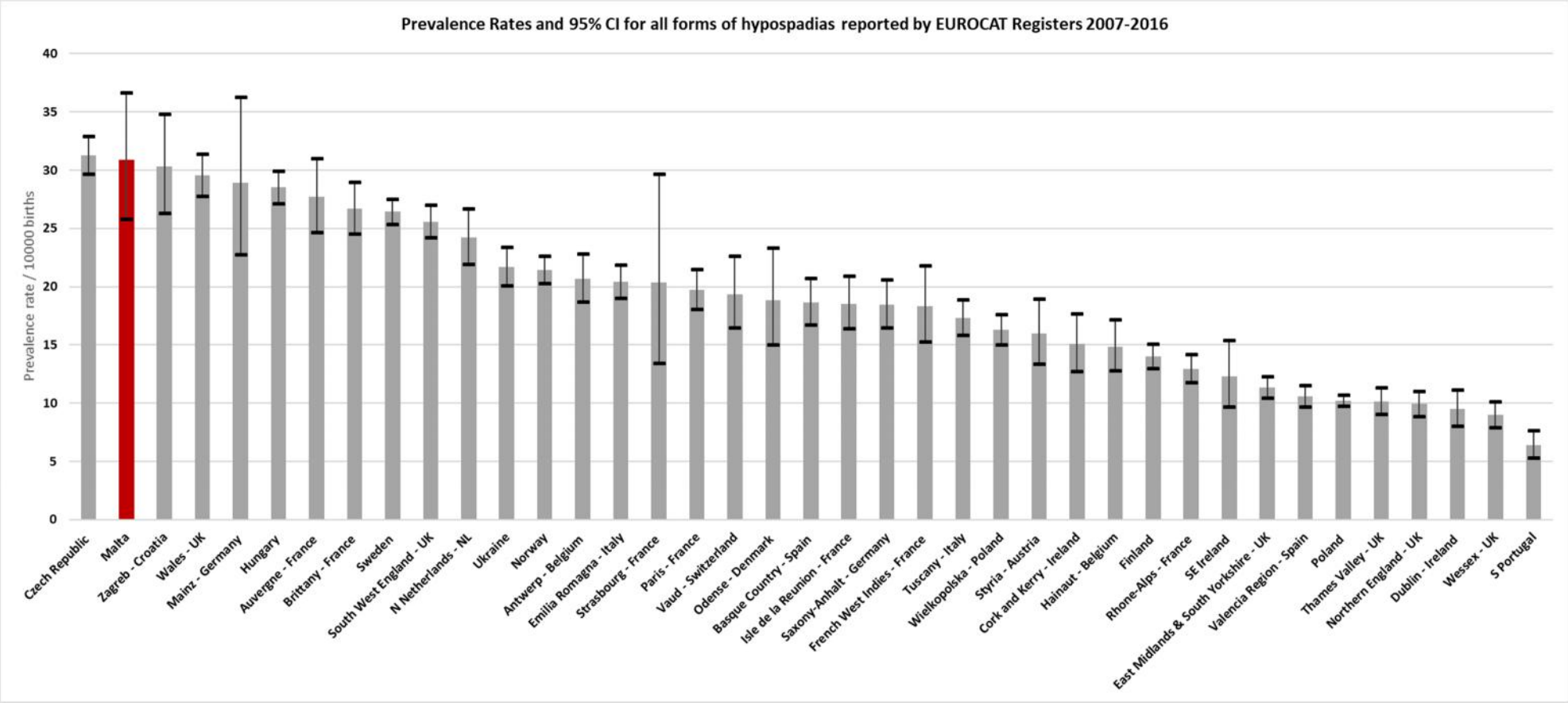


Fig 5. Prevalence of Hypospadias reported by European Registries.

Source: EUROCAT Website Interactive Database: http://www.eurocat-network.eu/ACCESSPREVALENCEDATA/PrevalenceTables

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Compiled by: Dr Norman Galea, Dr Miriam Gatt
Malta Congenital Anomalies Registry, Directorate for Health Information and Research
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