



World Birth Defects Day 2018 – Orofacial Clefts

World Birth Defects Day (WBDD) was first marked on March 3rd 2015 and this year, March 3rd 2018, will mark the fourth WBDD. Organisations from around the world, including the Malta Congenital Anomalies Registry, are marking this day.¹ The vision for WBDD is to raise awareness on birth defects and the message being disseminated this year is that Birth Defects impact millions of families and that a sustained focus is needed to support research, prevention, treatment and services. Further information can be found at: <http://www.worldbirthdefectsday.org/>.



Birth defects, also known as congenital anomalies, affect millions of babies world wide, occurring at around 1 in every 33 babies born. Some defects are more severe than others and it is estimated that, globally, 303,000 new-borns die within the first 4 weeks of life due to congenital anomalies.² Those babies who survive may suffer long term disability with significant impact on the individual, family, society and health care systems. While many birth defects have, as yet, no known cause or prevention, some can be prevented by taking appropriate measures before and during pregnancy. Preventive measures include adopting healthy lifestyles, achieving healthy weight, avoiding smoking and alcohol in pregnancy, vaccination and adequate intake of folic acid and other vitamins and minerals amongst others.³ Continuing research is essential to further identify the causes of birth defects, for prevention programs and for improving the care and support of those individuals affected and their families.

In Malta, the occurrence of birth defects is monitored through the Malta Congenital Anomalies Register of the Directorate for Health Information and Research. Further information on this Register may be found at <https://deputyprimeminister.gov.mt/en/dhir/Pages/Registries/birthdefects.aspx> . Health professionals are encouraged to notify cases of birth defects through the secure web portal found at <https://deputyprimeminister.gov.mt/en/dhir/Pages/Notifications/nocaf.aspx> .

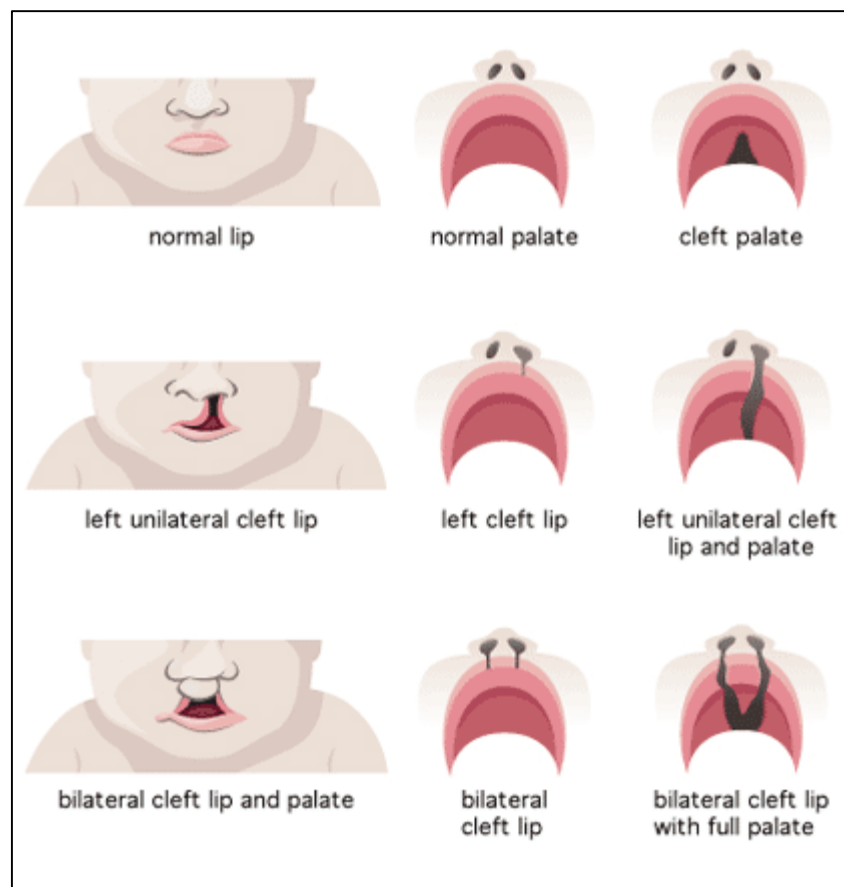
OROFACIAL CLEFTS – CLEFT PALATE AND CLEFT LIP

Orofacial clefts include Cleft lip and Cleft palate; these are defects that occur when the fetal mouth does not develop properly during pregnancy.

Cleft lip develops between the 4th and 7th weeks of pregnancy and occurs when the tissue forming the lips does not grow and fuse normally at the midline. *Cleft palate* develops slightly later, between the 6th and 9th week of pregnancy and occurs when the tissue forming the palate does not fuse properly. Defects may range from a small notch to extensive splits in the lip and/or palate, they may be unilateral or bilateral (Diagram 1).

Cleft lip and cleft palate may occur separately or together as cleft lip with cleft palate. Furthermore, orofacial clefts may occur in isolation or as part of a recognised genetic syndrome or sequence such as Pierre Robin Sequence, Van der Woude Syndrome and Velocardial Syndrome. The underlying aetiology is different for non-syndromic and syndromic cases.

Diagram 1 Diagrammatic representation of orofacial clefts.



Taken from: <http://novasurgicalarts.com/procedures/reconstructive-surgery/cleft-lip-palate/>

Orofacial clefts, especially cleft lip with or without cleft palate, may be diagnosed during pregnancy by antenatal ultrasound examination, however many are diagnosed after the baby is born, especially cleft palate. Sometimes certain types of minor cleft palate (such as submucous cleft palate and bifid uvula) might not be diagnosed until later in life.

While orofacial clefts occurring as part of a recognised syndrome or genetic condition are, as the name implies, related to the causes of the particular syndrome, the causes of isolated orofacial clefts not forming part of a genetic condition remain generally unknown.

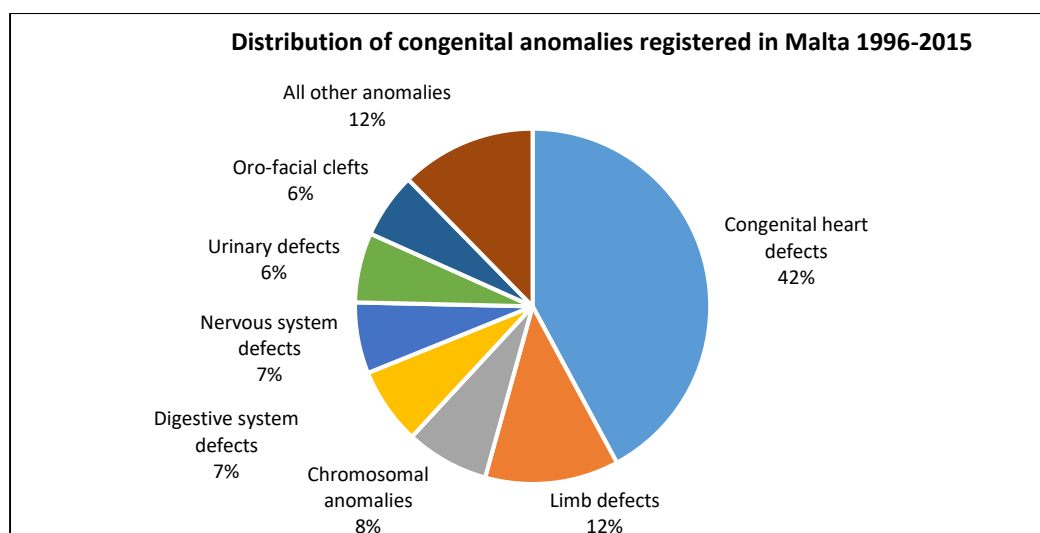
There is increasing evidence that both genetic and environmental factors may be involved in the aetiology of these conditions. Recent studies have shown an association between orofacial clefts and maternal smoking,^{4,5} diabetes⁶ and antiepileptic medication.⁷

Infants born with an orofacial cleft often require surgical correction, this is usually carried out within the first year of life.⁸ Timely surgical repair not only improves the child's facial appearance, but may also improve breathing, hearing and speech and language development. Infants born with orofacial clefts might need other types of treatments and services, such as specialised dental or orthodontic care or speech therapy and a multidisciplinary approach to management is advisable.⁸

Orofacial Clefts in Malta

The Malta Congenital Anomalies Register within the Directorate for Health Information and Research collects information on all babies born on the Maltese Islands and diagnosed with congenital anomalies until one year of age. This Register follows EUROCAT (European Surveillance of Congenital Anomalies) guidelines in the definition and classification of congenital anomalies.⁹ Over the 20-year period 1996-2015 there were a total of 184 orofacial cleft defects registered in Malta. These defects accounted for around 6% of all registered major birth defects.

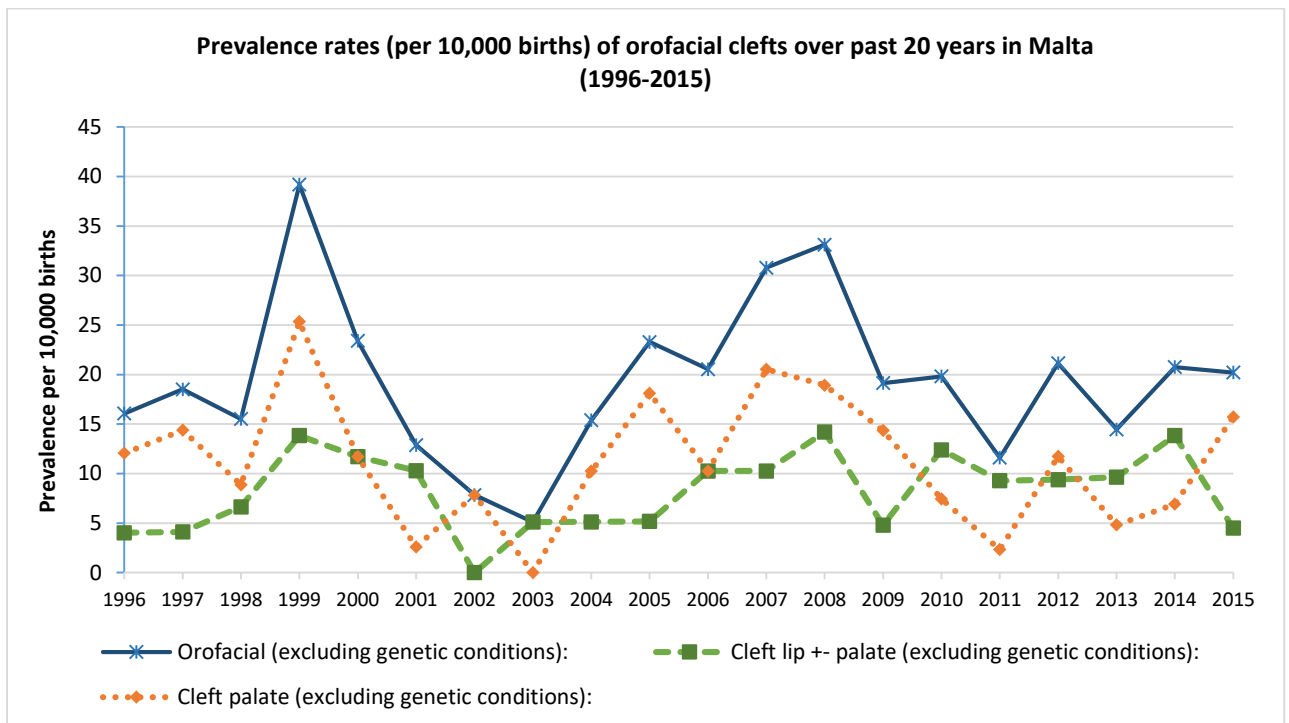
Fig. 1 Occurrence of the various anomaly groups in Malta



Of the total of 184 orofacial clefts registered in Malta and Gozo between 1996-2015, 164 were classified using EUROCAT guidelines as not being part of a genetic syndrome or ‘non-syndromic’, while the remaining 20 cases were classified as forming part of a recognised genetic syndrome or ‘syndromic’. Of these 184 total cases 107 were cases of cleft palate (95 non-syndromic; 12 syndromic) and 77 were cases of cleft lip with or without cleft palate (69 non-syndromic and 8 syndromic).

The overall population prevalence rate of non-syndromic orofacial clefts (including livebirths and stillbirths) between 1996-2015 has been 19.5/10,000 births. 11.3/10,000 births for non-syndromic cleft palate and 8.2/10,000 births for non-syndromic cleft lip with or without cleft palate. These rates have not shown any statistically significant change over the time period (Fig. 2).

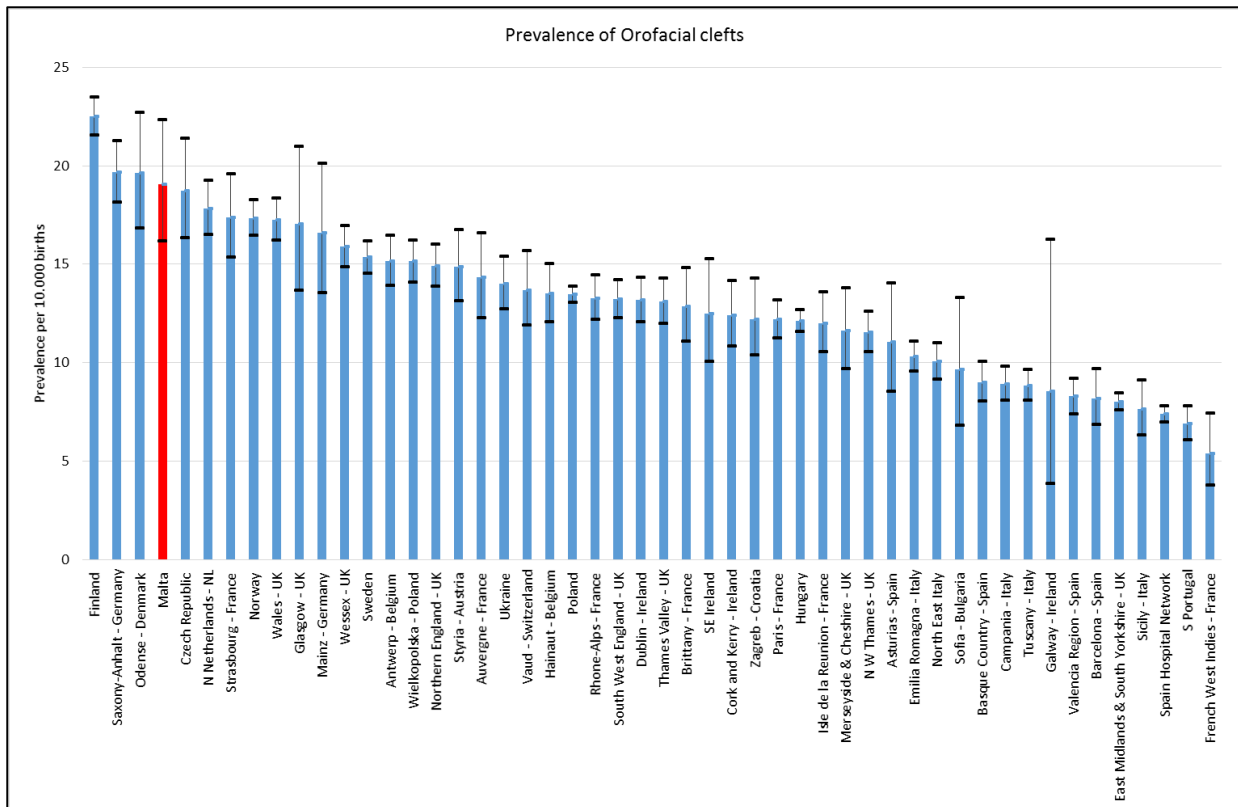
Fig. 2 Prevalence of Orofacial clefts in Malta between 1996-2015.



Orofacial Clefts reported from European Registries

The reported total prevalence of orofacial clefts in European Registries varies. Figs. 3-5 show the reported total prevalence (including livebirths, still births and terminations of pregnancy) for orofacial clefts, cleft palate and cleft lip with or without cleft palate and the 95% confidence intervals in EUROCAT Registries.⁴ The presented rates include those defects that do not form part of a genetic syndrome.

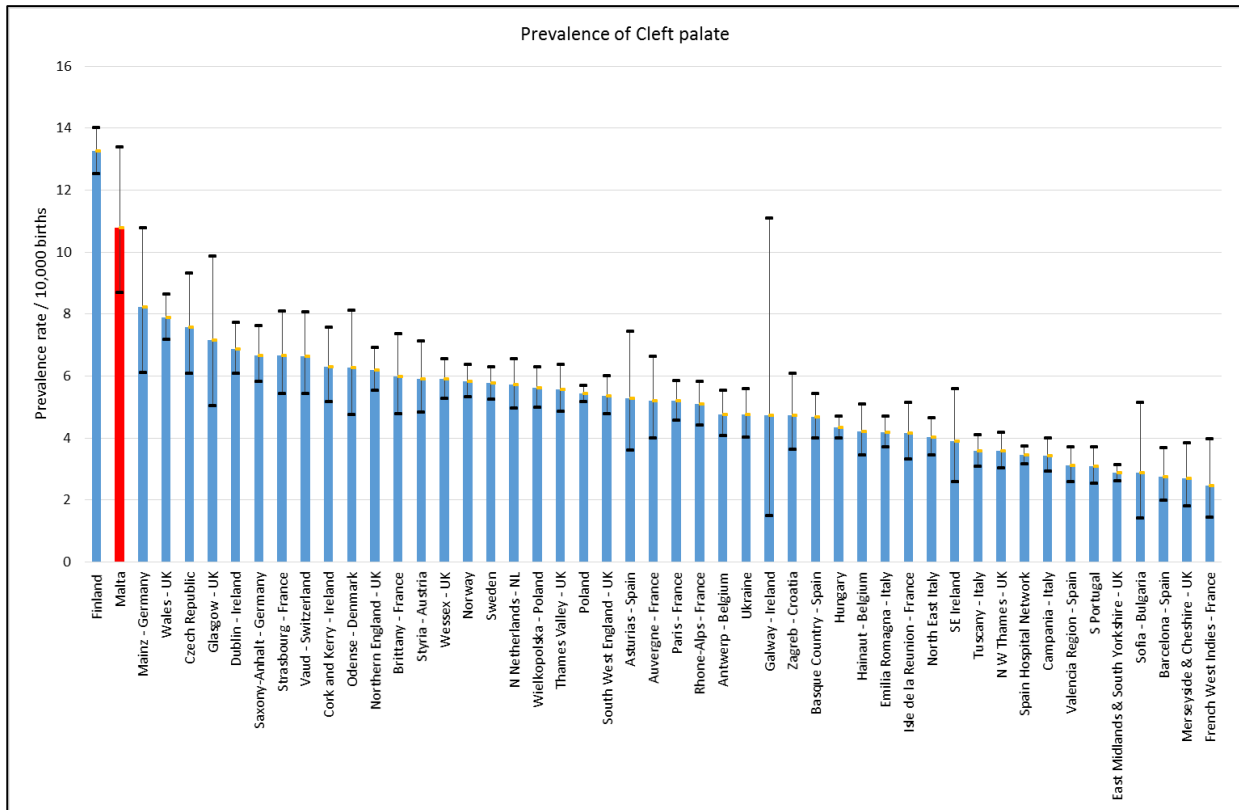
Fig.3 Prevalence of non-syndromic total Orofacial clefts reported by European Registers 1996-2015



Taken from EUROCAT: Available from: <http://www.eurocat-network.eu/accessprevalencedata/prevalencetables>

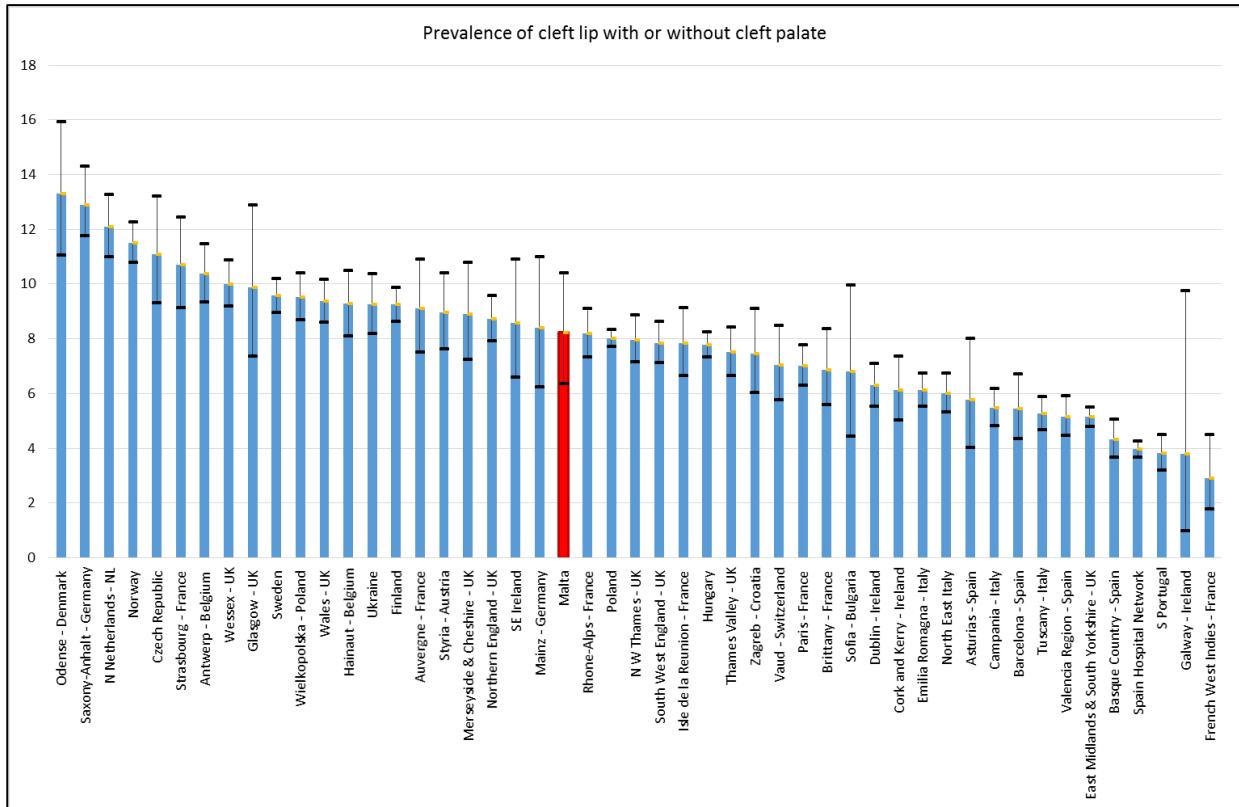
Malta reports a comparatively high rate of orofacial clefts and this is mainly due to a higher rate of reported cleft palate (Figs 4 – 5).

Fig.4 Prevalence of non-syndromic Cleft palate reported by European Registries 1996-2015



Taken from EUROCAT. Available from: <http://www.eurocat-network.eu/accessprevalencedata/prevalencetables>

Fig.5 Prevalence of non-syndromic Cleft lip with or without cleft palate reported by European Registries 1995-2015



Taken from EUROCAT. Available from: <http://www.eurocat-network.eu/accessprevalencedata/prevalencetables>

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