

**European Registries for Rare Endocrine Conditions (EuRECa):
Results from the Platform for e-Reporting of Rare Conditions (e-REC)**

Introduction

e-REC (e-Reporting of Rare Endocrine Conditions) is an electronic reporting system which captures activity and allows for a better understanding of the occurrence of a wide range of rare endocrine and bone conditions. For clinical networks such as Endo-ERN and ERN-BOND, the e-REC platform allows continuous reporting of core indicators of activity and enables these clinical networks to objectively map the conditions and related activity. However, the platform is open to all centres that look after people with such conditions and not just limited to reference centres within ERNs. More recently, the e-REC platform has also been assisting the Rare Disease Committee of the ESE in mapping the occurrence of COVID-19 infection in people with rare conditions.

Aim

This report has been developed to describe the current activity in e-REC over a 2-year period from July 2018 to June 2020 inclusive.

Methods

The e-REC platform issues invitations to clinicians who had registered to participate in e-REC from July 2018 to June 2020 to ask them to complete a monthly return. Participants can create a bespoke reporting set up and can report any newly encountered cases of any of the conditions that have been included in Endo-ERN, ERN-BOND and more recently, COVID-19 infections.

Results

A total of 60 centres from 26 countries have registered to participate in e-REC between July 2018 and June 2020. Of these 60 centres, 46 from 20 countries have used the platform and 40 of these centres are part of Endo-ERN and/or ERN-BOND.

Country	Centres	Country	Centres
 Austria	2 (2)	 Luxembourg	1 (1)
 Belgium	6 (6)	 Netherlands	4 (5)
 Croatia	1 (1)	 Norway	0 (1)
 Cyprus	0 (1)	 Poland	1 (1)
 Czech Republic	1 (1)	 Portugal	1 (1)
 Estonia	1 (1)	 Romania	0 (1)
 France	3 (4)	 Slovakia	0 (1)
 Georgia	0 (1)	 Slovenia	1 (1)
 Germany	8 (9)	 Spain	1 (3)
 Hungary	1 (1)	 Sweden	1 (1)
 Italy	6 (7)	 Switzerland	1 (1)
 Israel	1 (1)	 United Kingdom	4 (6)
 Lithuania	1 (1)	 Ukraine	1 (1)

Table 1 – Number of centres in each country that are active and those who have registered (in parentheses)

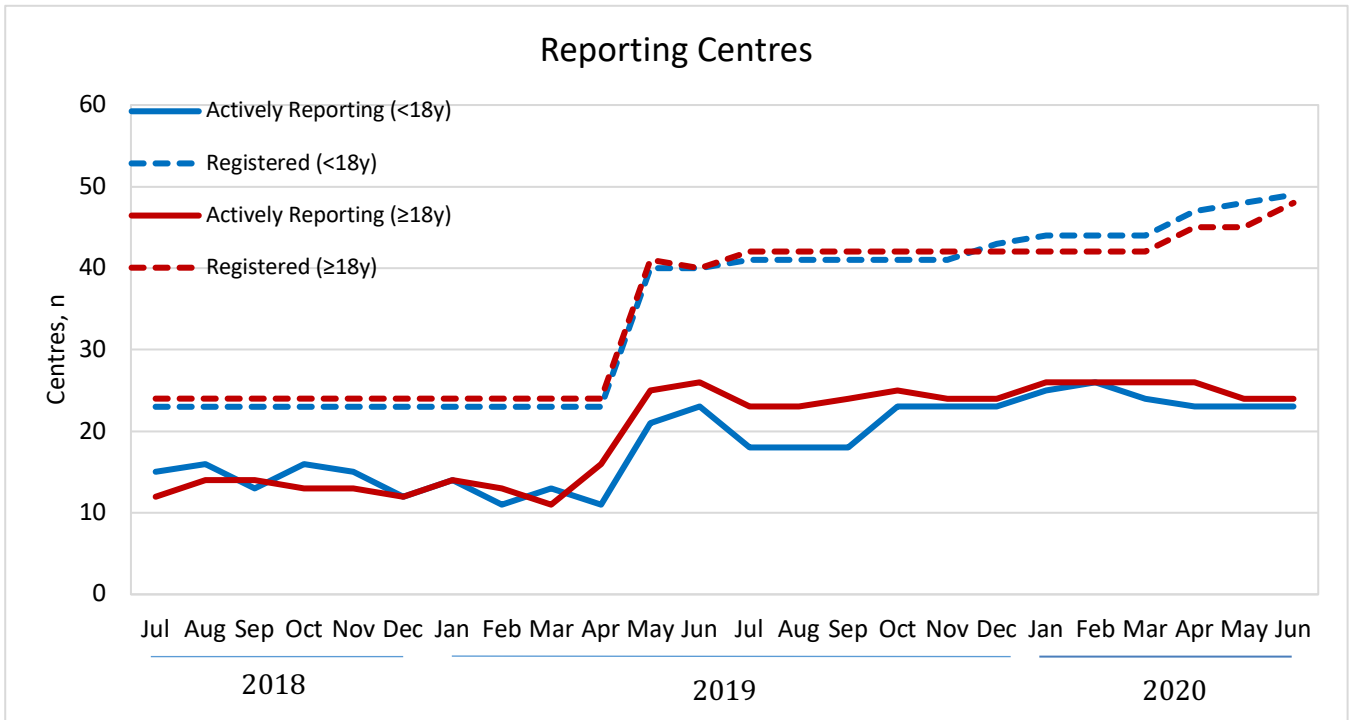


Fig.1 – Change in the number of centres that registered and that were actively reporting between July 2018 and June 2020. Currently, 49 paediatric centres and 48 adult centres have registered to participate in e-REC.

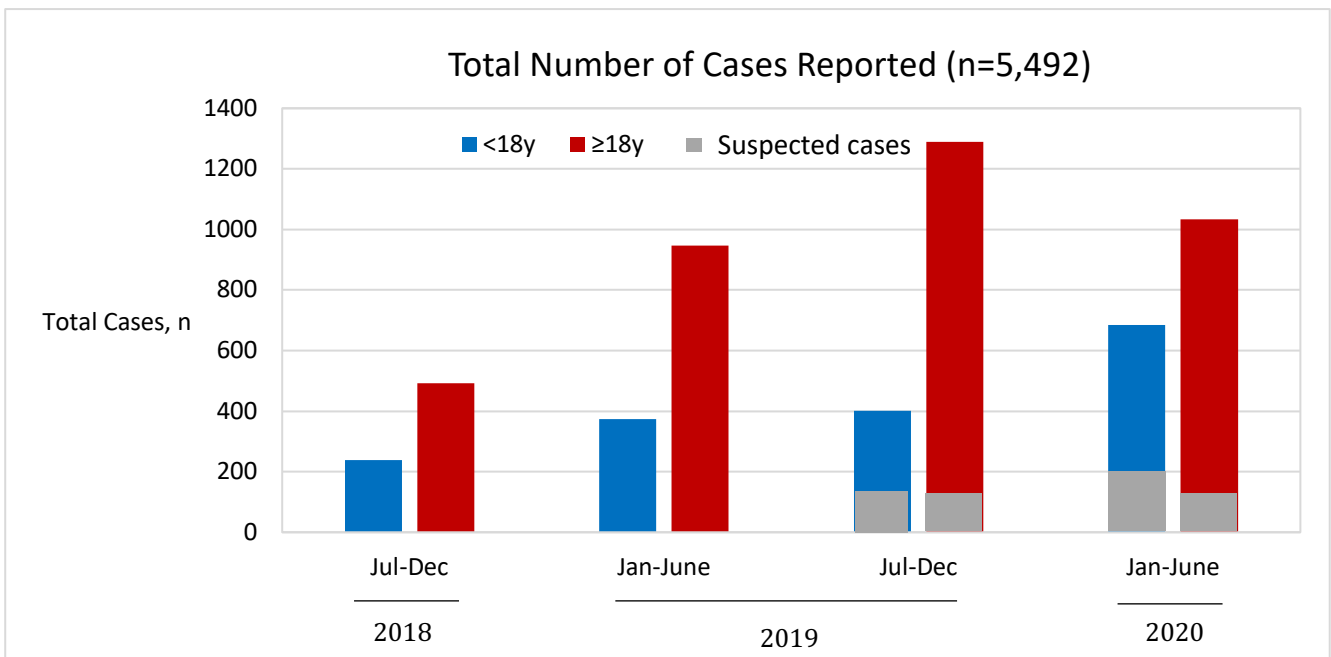


Fig. 2 – The temporal variation in the cases reported between July 2018 and June 2020. A total of 3,748 adult cases and 1,744 paediatric cases were reported. From the last quarter of 2019, the platform also collected information on the proportion of cases that were considered to be 'suspected' and the proportion that were 'confirmed'. The former are identified on the panel as the grey cases.

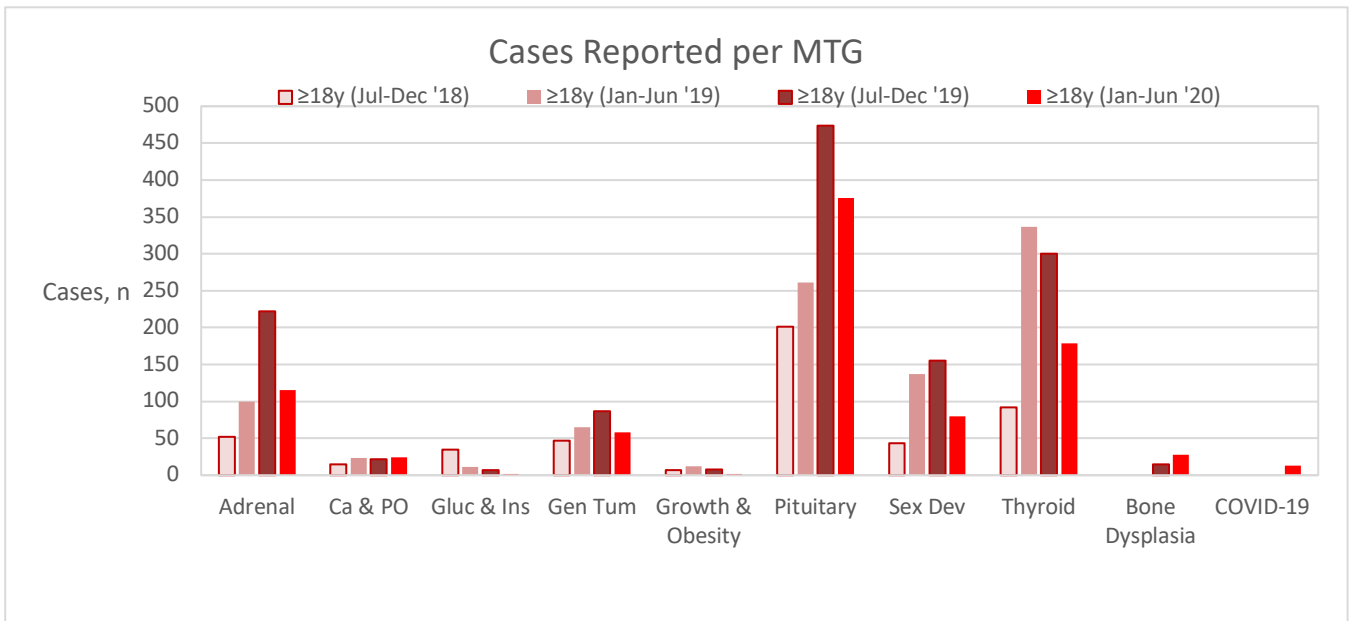
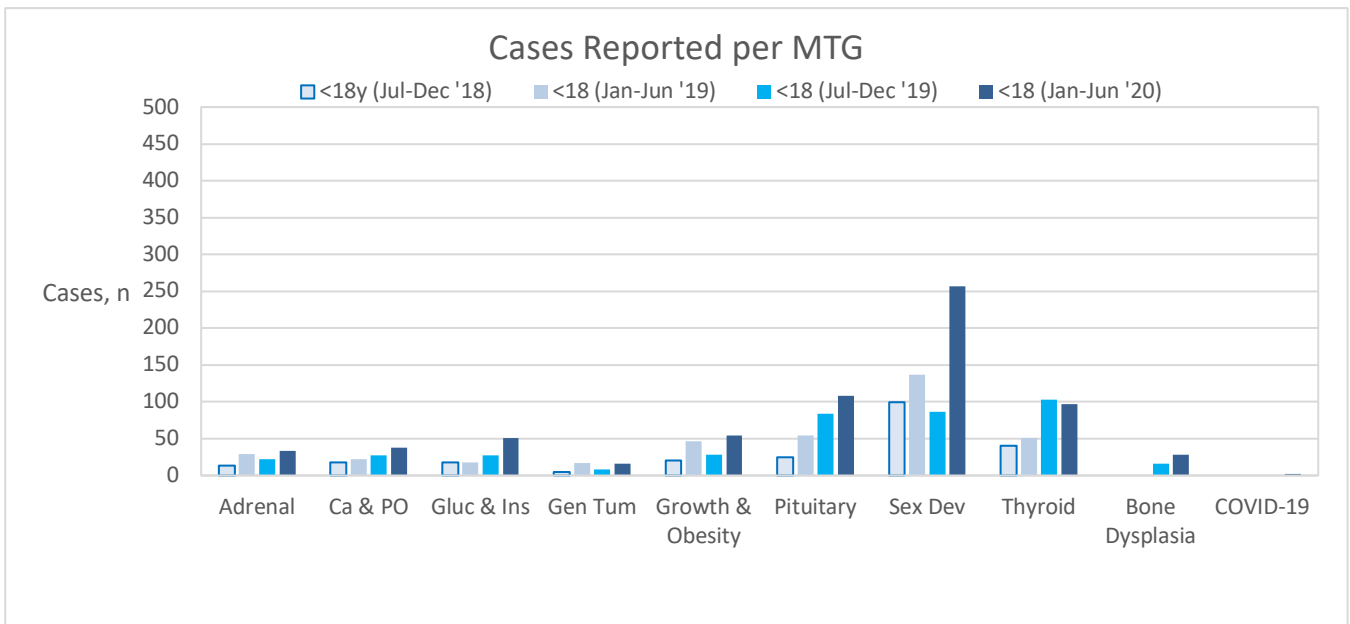


Fig.3 – Cases reported per Main Thematic Group (MTG) between July 2018 and June 2020. Conditions within the ‘Sex Development’ and ‘Pituitary’ condition group were most commonly reported amongst children and adults, respectively. For adults, the increase in reported cases over time was evident in several MTGs.

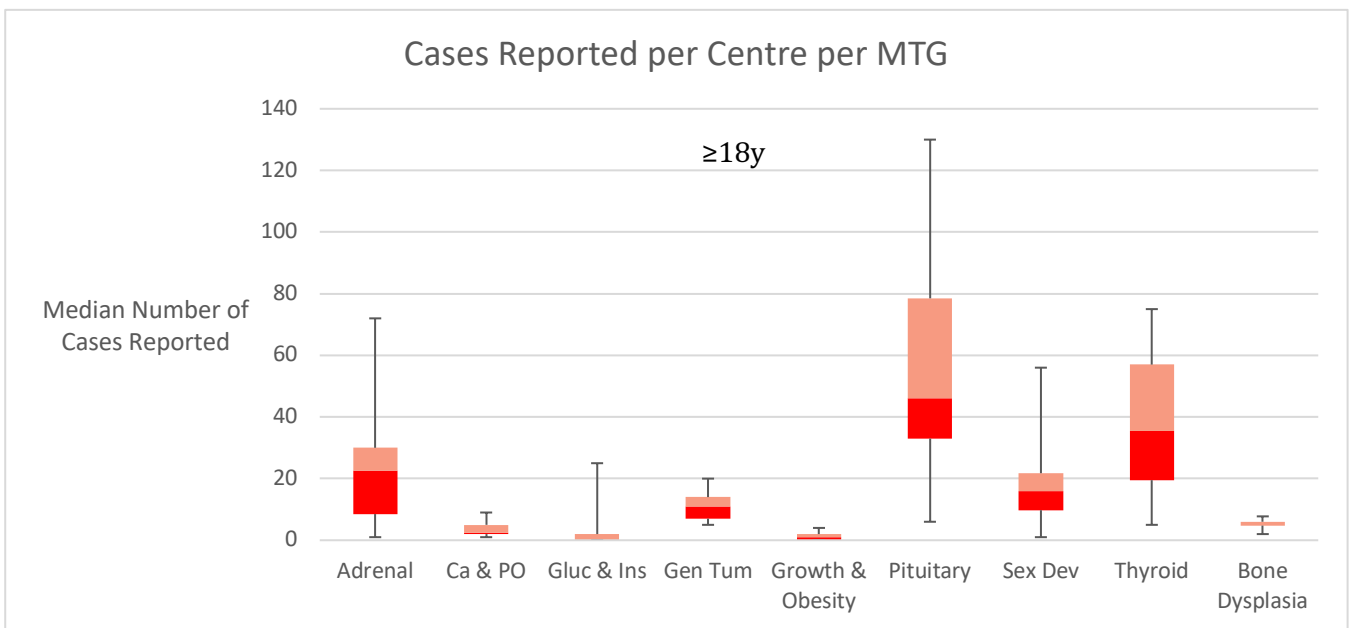
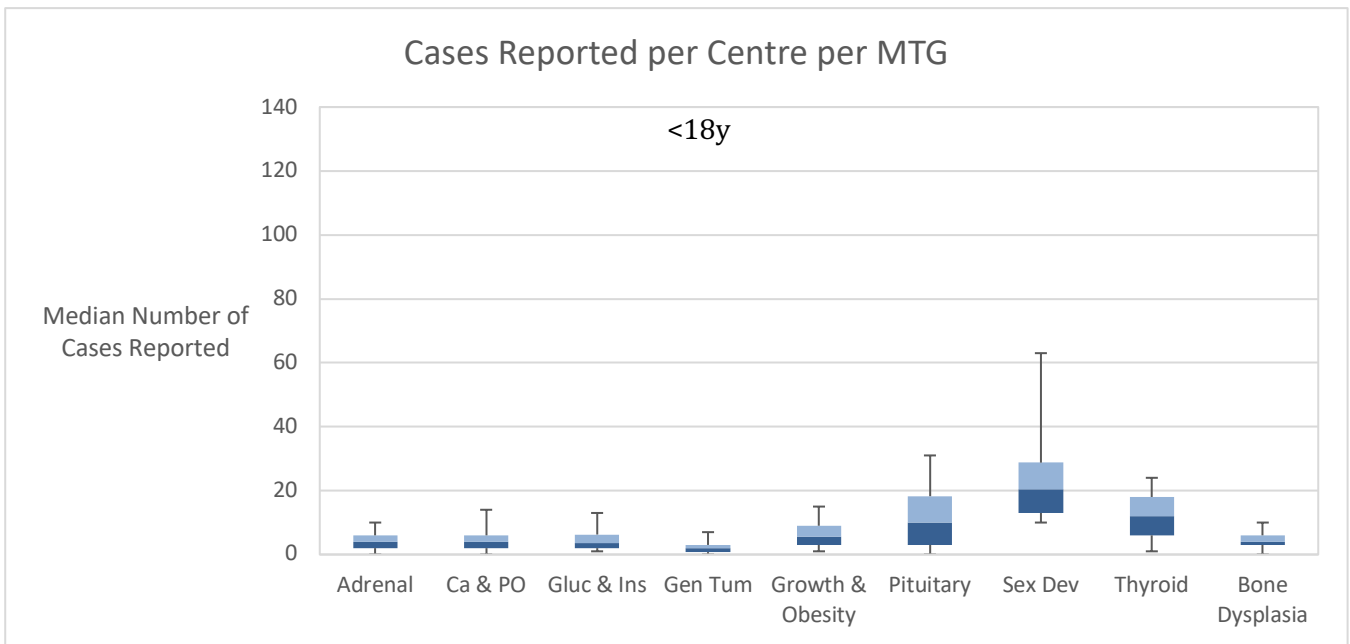
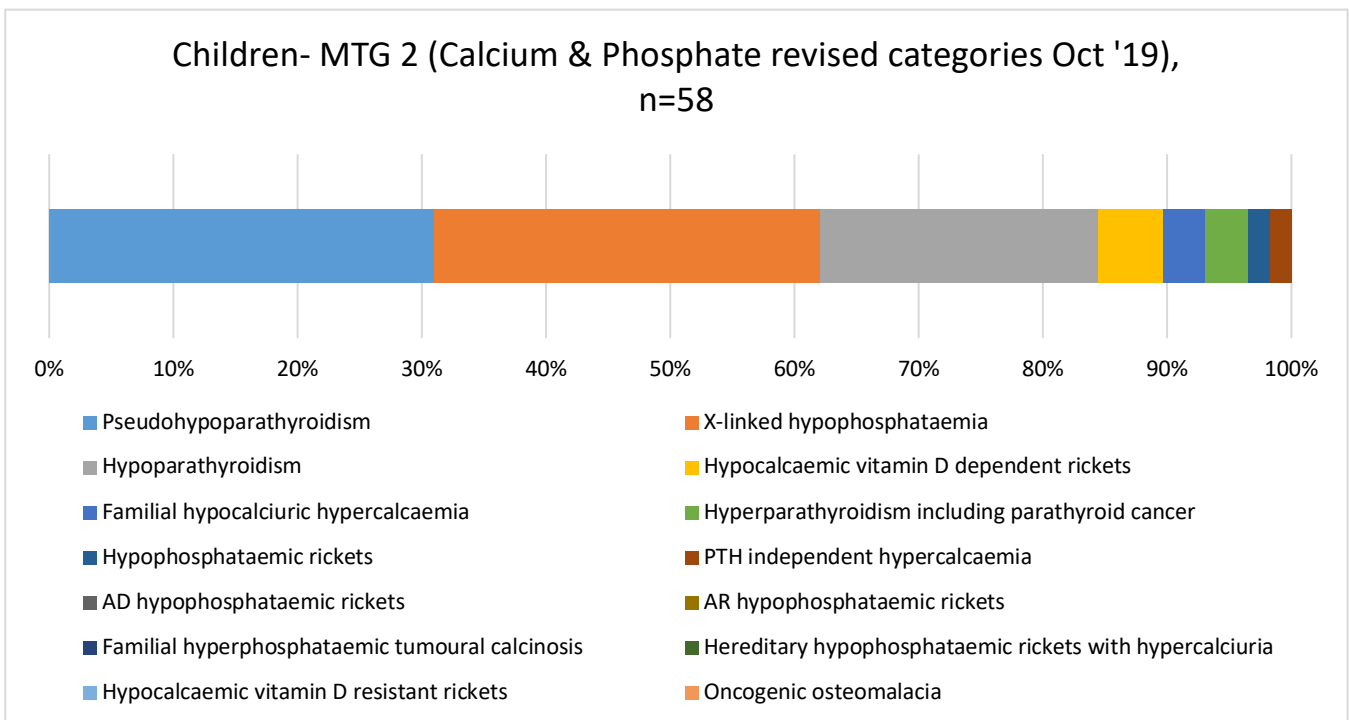
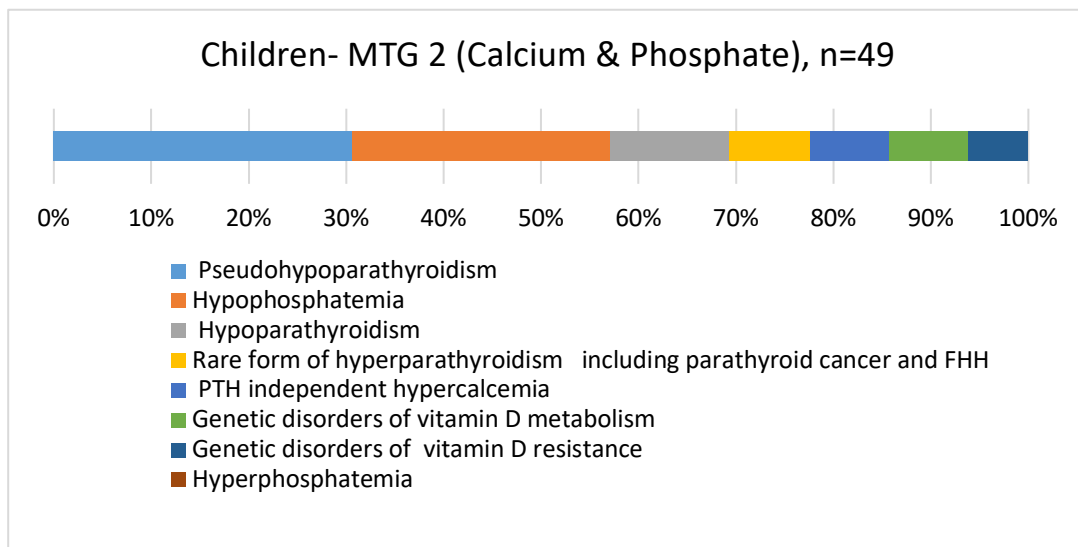
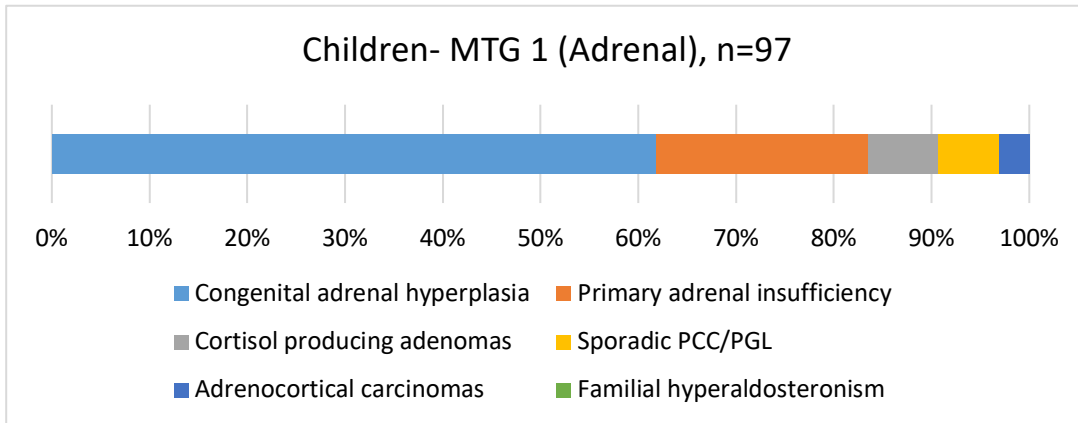
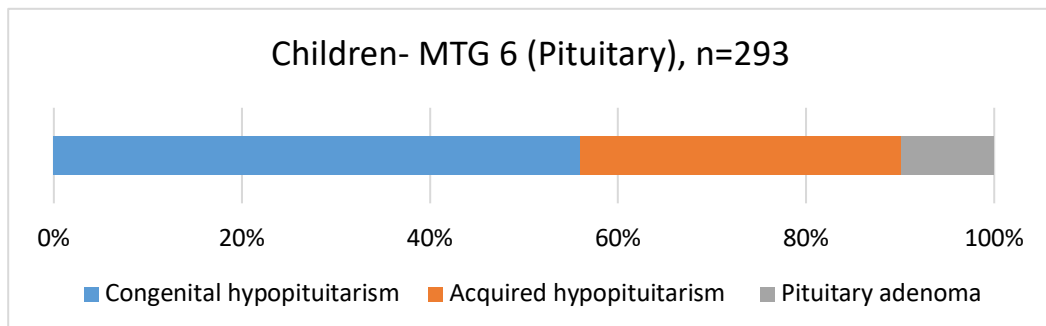
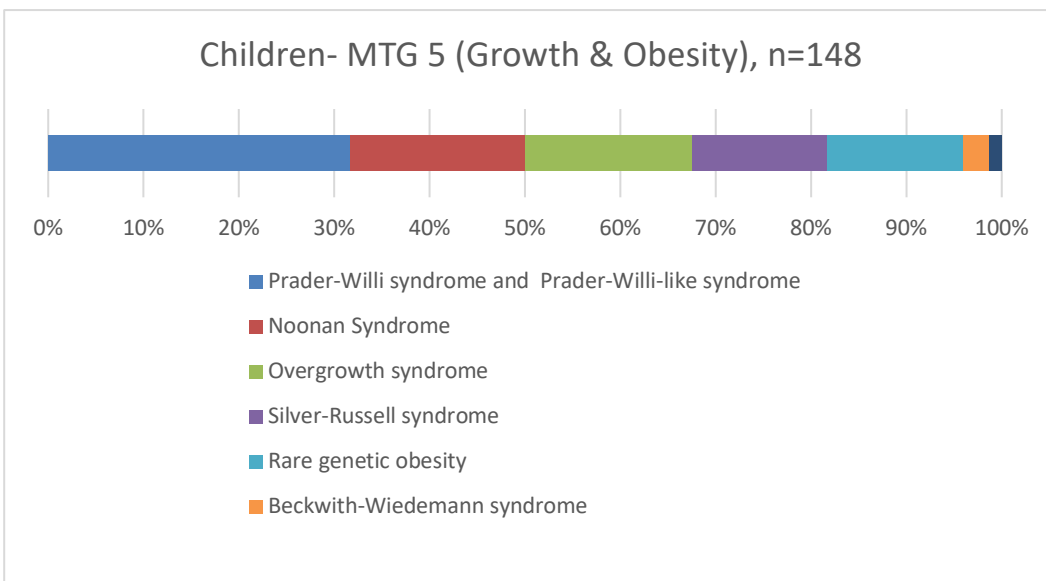
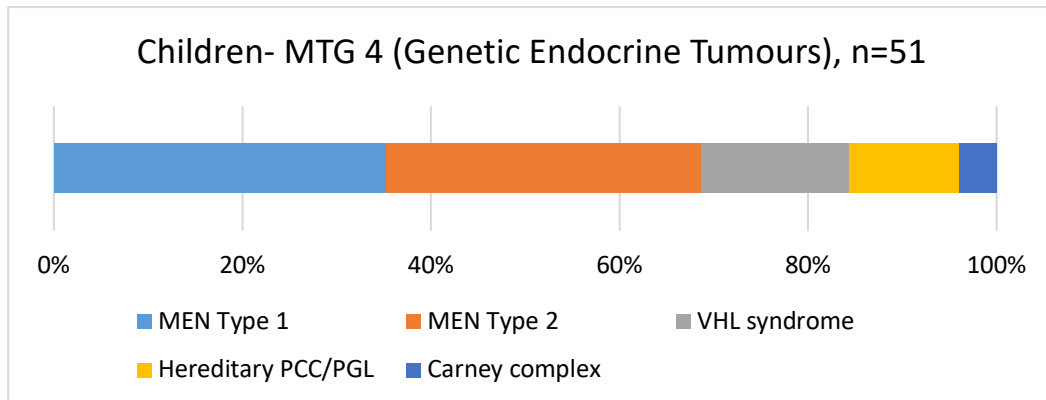
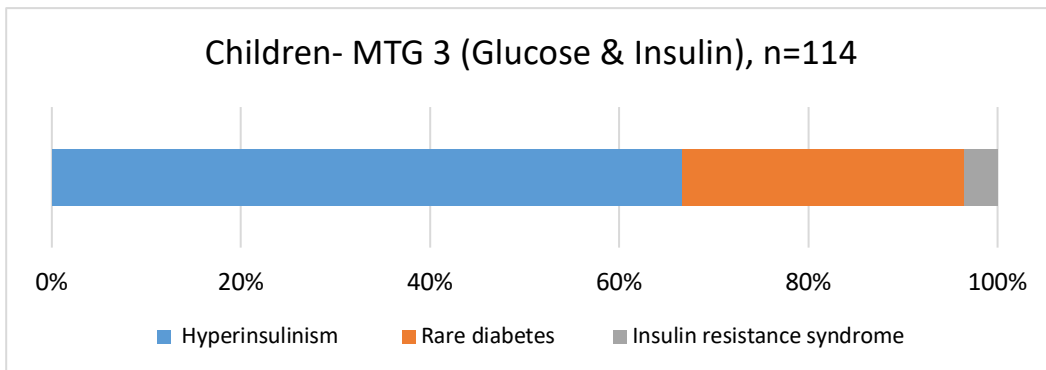


Fig.4 – Cases reported per MTG between July 2018 and June 2020. There is wide variation in the number of cases reported per centre, particularly amongst adults.

Fig.5a – Commonly reported conditions in children between July 2018 and June 2020; cases, n.





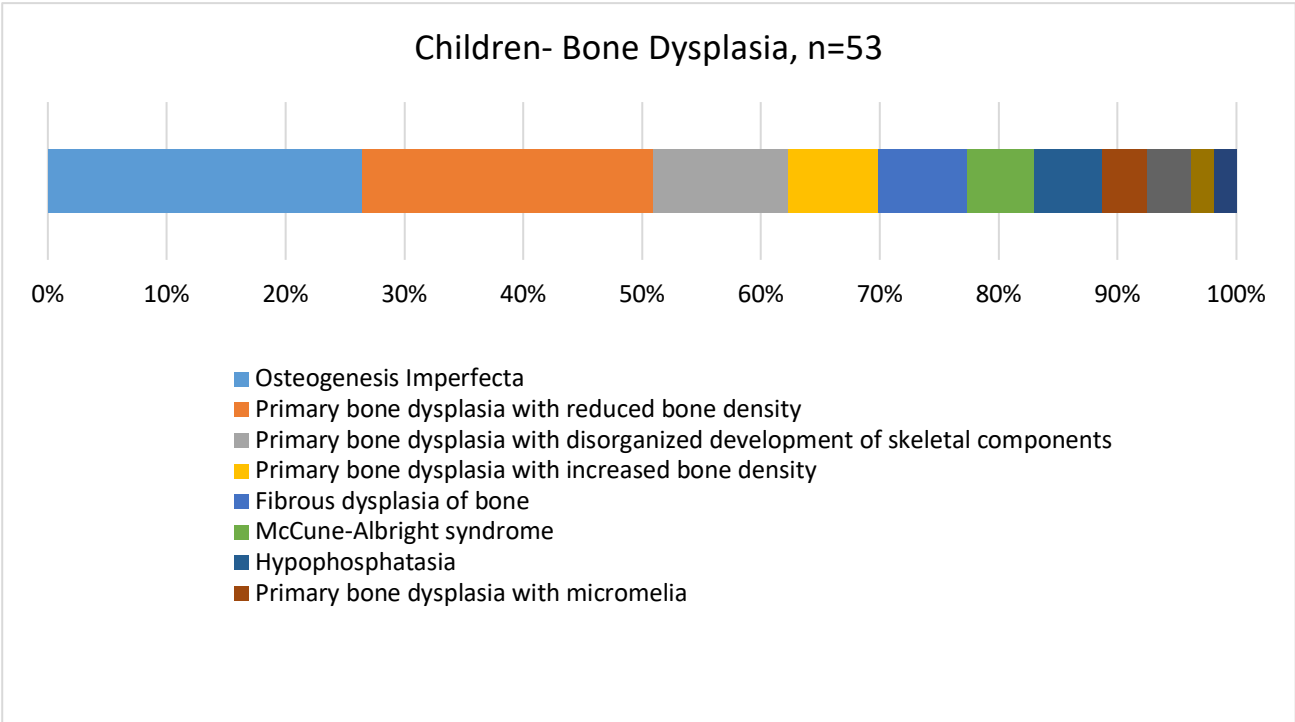
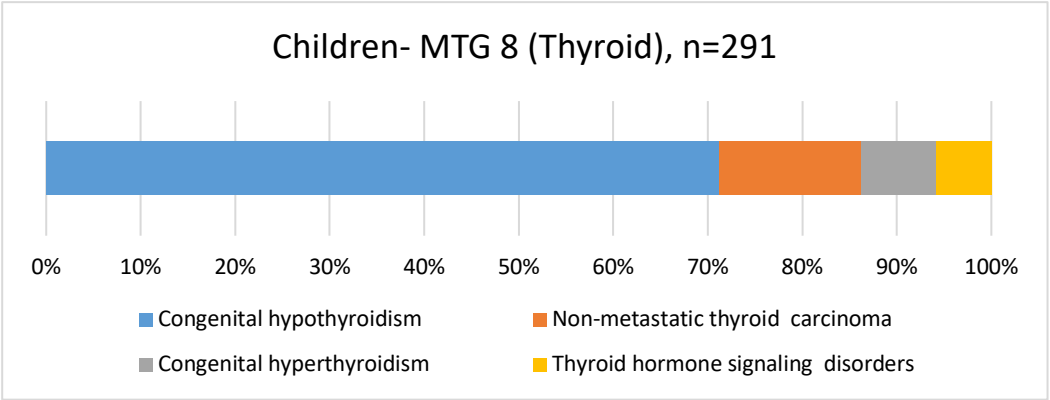
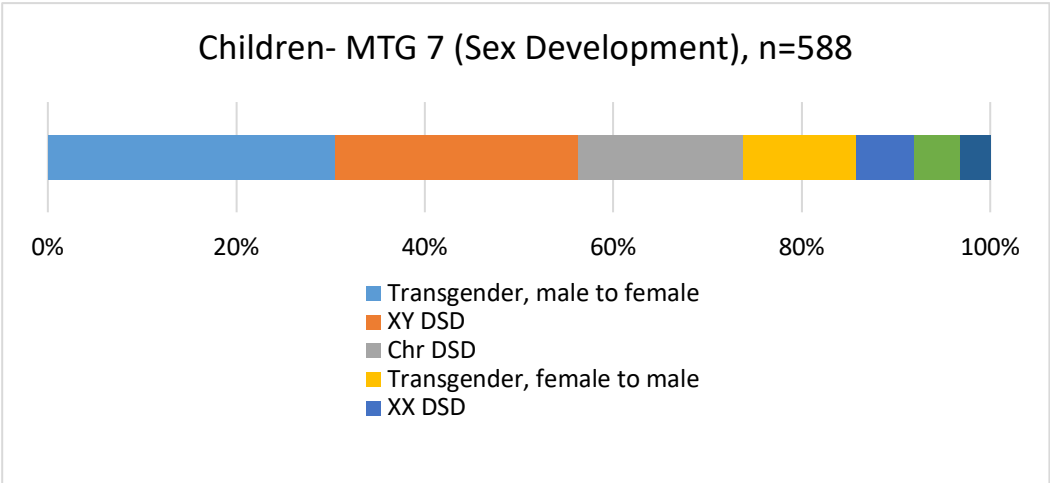
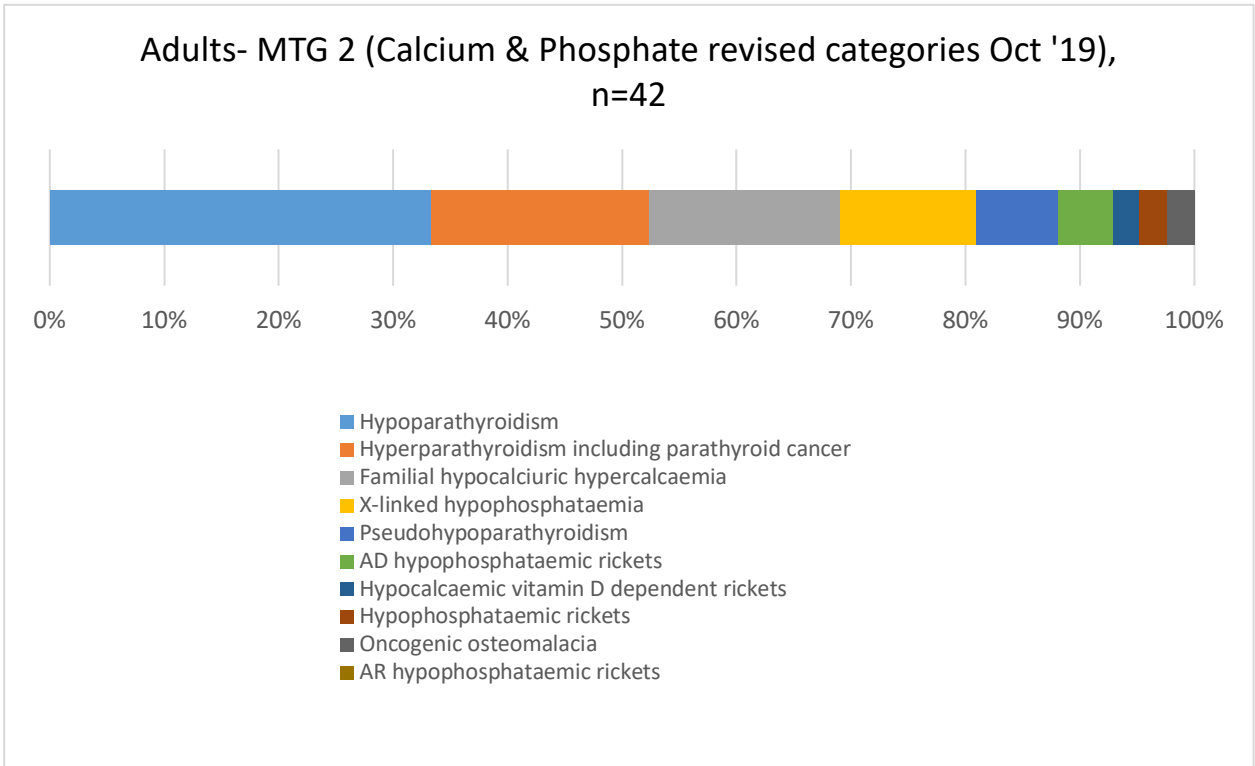
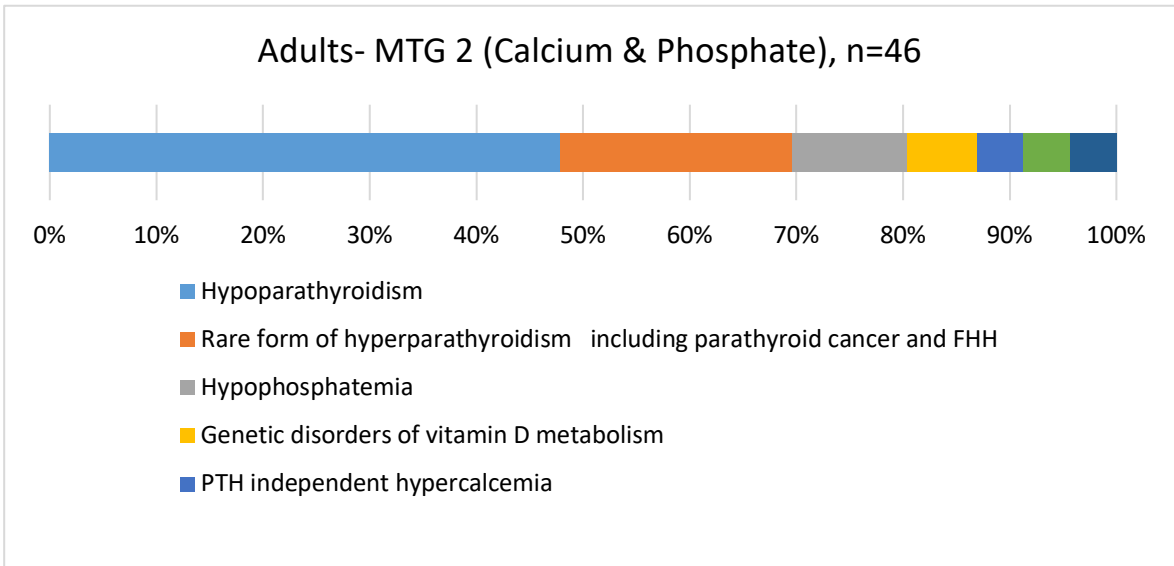
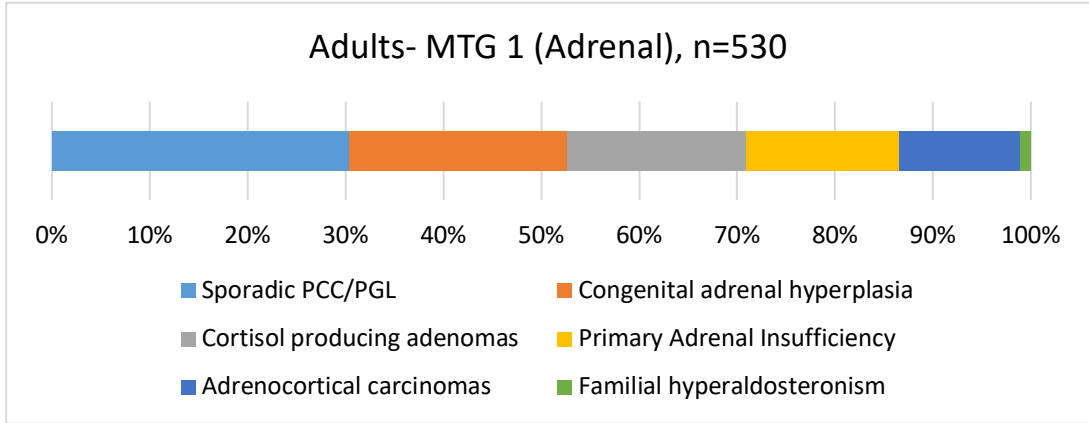
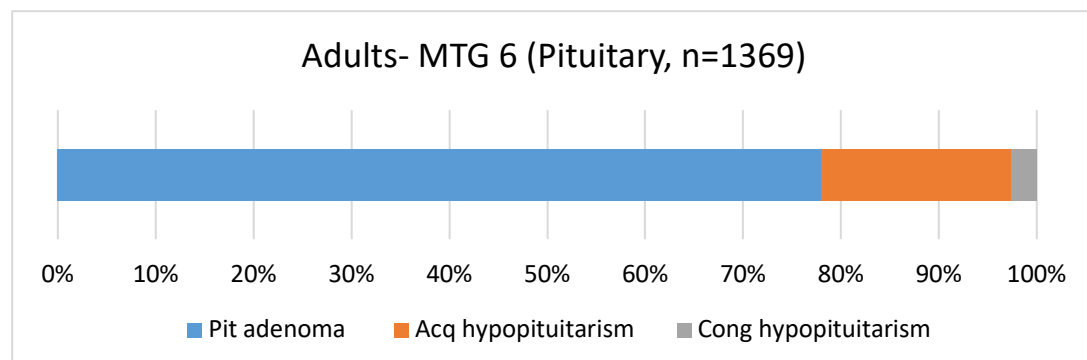
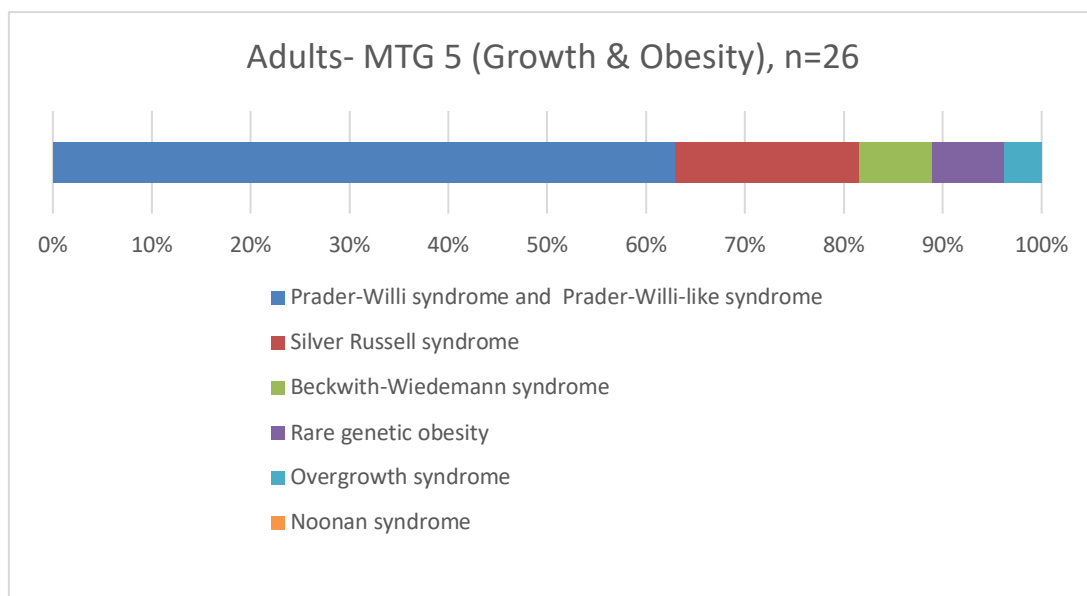
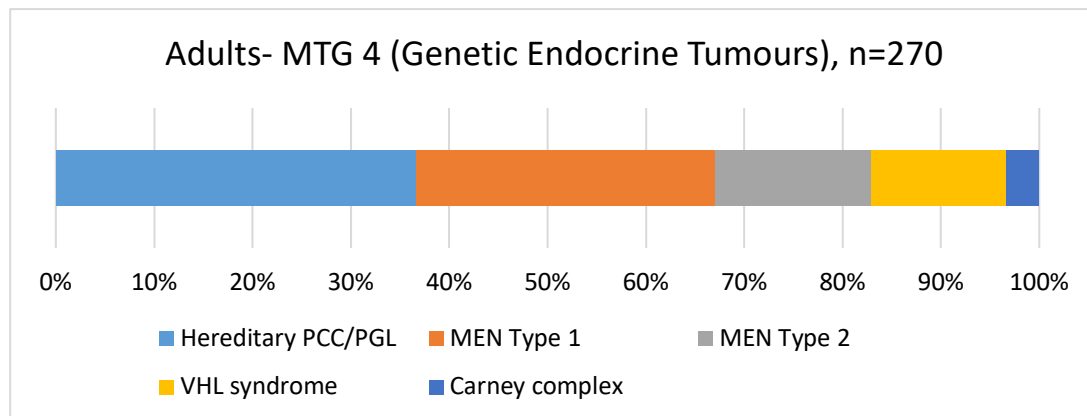
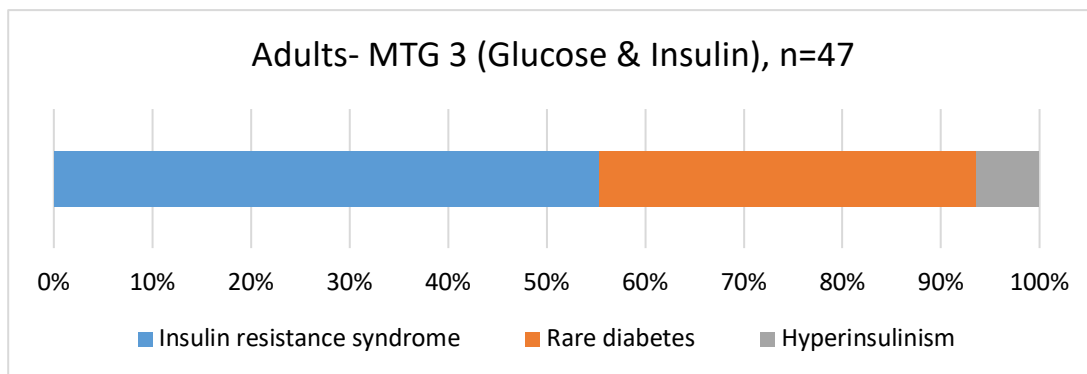
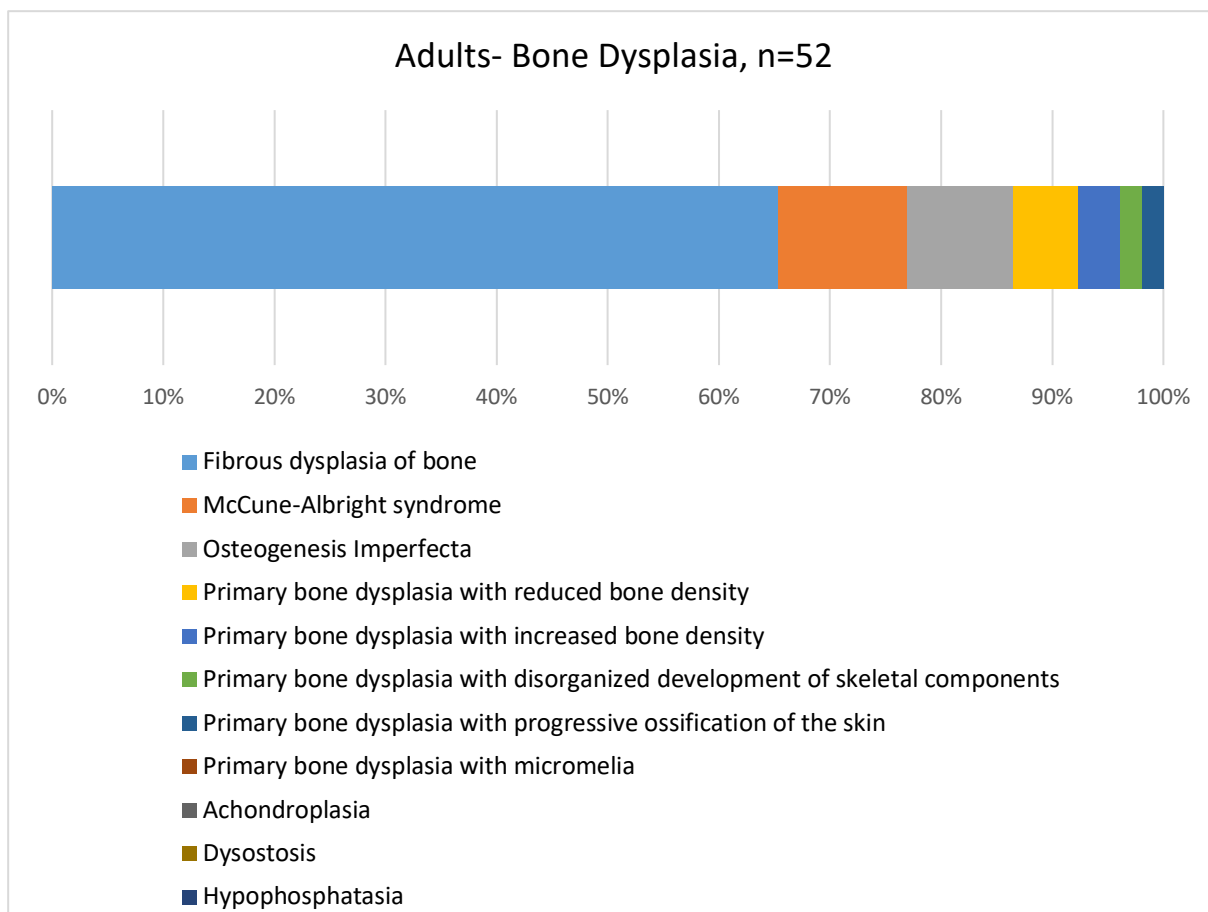
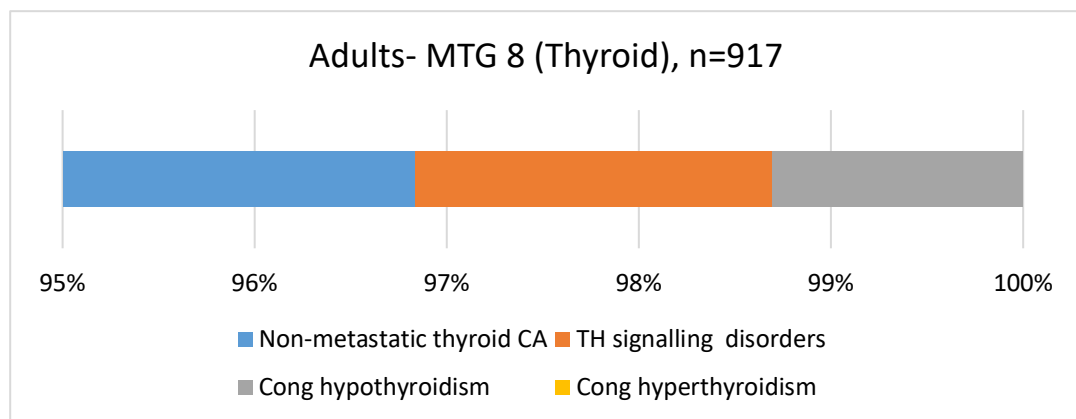
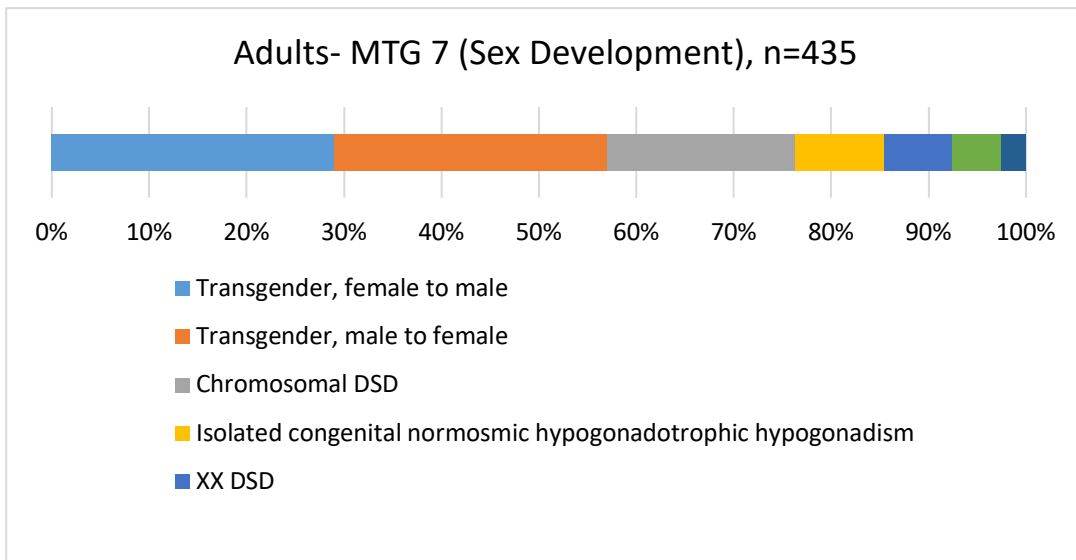


Fig.5b – Commonly reported conditions in adults between July 2018 and June 2020; cases, n.







Interpretation of Findings

- The e-REC platform is a simple platform that can be used to capture information on new encounters with patients with rare endocrine and bone conditions, including more recently, COVID-19 infections in people with rare endocrine or bone conditions.
- Although the number of centres that have registered to use the platform continues to increase, the number of centres that are actively reporting cases remains steady.
- Over the last six months, there has been a marked increase in number of cases being reported in children with rare conditions.
- There is wide variability in the number of cases encountered for different conditions amongst different centres.
- Amongst children, conditions within the 'Sex Development' condition group were most commonly reported; amongst adults, conditions within the 'Pituitary' and 'Thyroid' condition groups were most commonly reported.

Recommendations

- The use of the platform should continue to be disseminated widely.
- Members of ERNs should be able to utilize the platform for continuous reporting.
- Data are available to the EuRRECa Expert Working Groups for further analysis and investigate variations in diagnoses and presentations.
- Subject to the data sharing agreement developed by the EuRRECa Data Access Committee, more detailed data are available to interested parties external to EuRRECa.

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EuRRECa Work Package 6
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