

**European Registries for Rare Endocrine Conditions (EuRRECa):
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 platform which acts as a registry that 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 2.5-year period from July 2018 to December 2020 inclusive.

Methods

The e-REC platform issues invitations to clinicians who have participated in e-REC from July 2018 to December 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

By December 2020, a total of 57 centres from 22 countries had reported on the e-REC. Of these 57 centres, 24 are in Endo-ERN only, 22 are in Endo-ERN and ERN-BOND, 2 are in ERN-BOND only and 9 are in no ERNs.








Country	Centres	Country 2	Centres 3
 Austria	3	 Latvia	1
 Belgium	5	 Lithuania	1
 Croatia	1	 Luxembourg	1
 Czechia	2	 Netherlands	6
 Estonia	1	 Poland	1
 France	5	 Portugal	1
 Germany	8	 Romania	1
 Greece	1	 Slovenia	1
 Hungary	1	 Spain	2
 Israel	1	 Sweden	2
 Italy	7	 UK	5

Table 1 – Number of centres in each country that are active (i.e. have submitted 1 or more returns between July 2018 and December 2020).

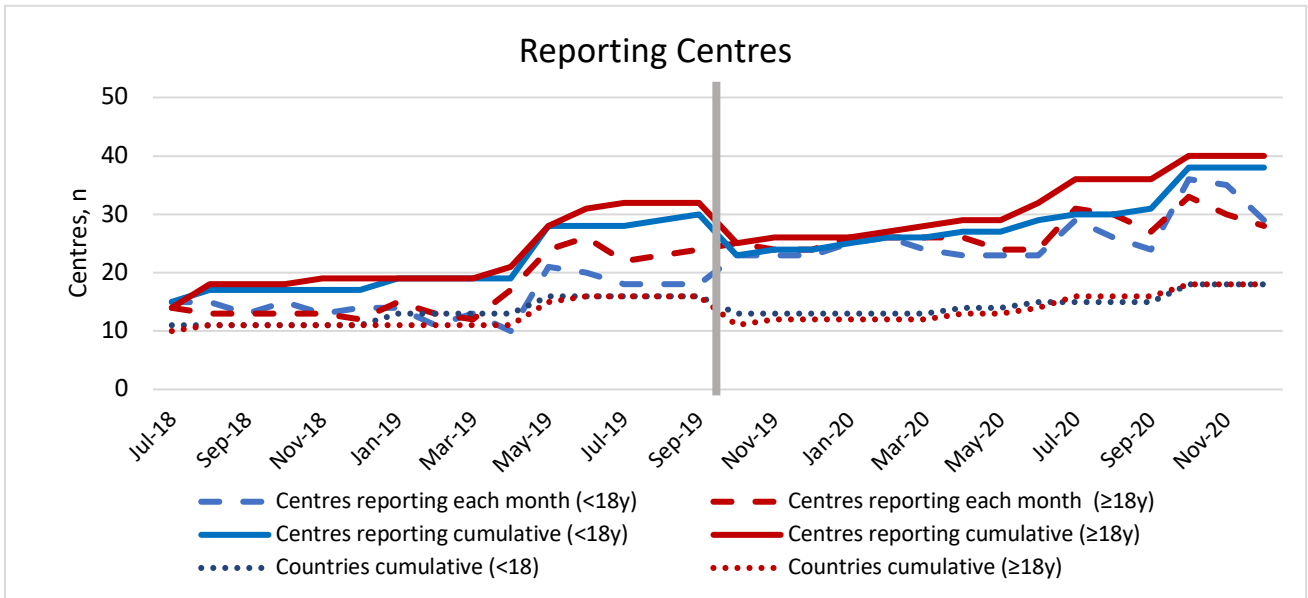


Fig.1 – Change in the number of centres that have been actively reporting between July 2018 and December 2020. Currently, 41 paediatric centres and 46 adult centres have actively participated in e-REC. The vertical line indicates the shift to the current bespoke reporting platform from the original REDcap platform.

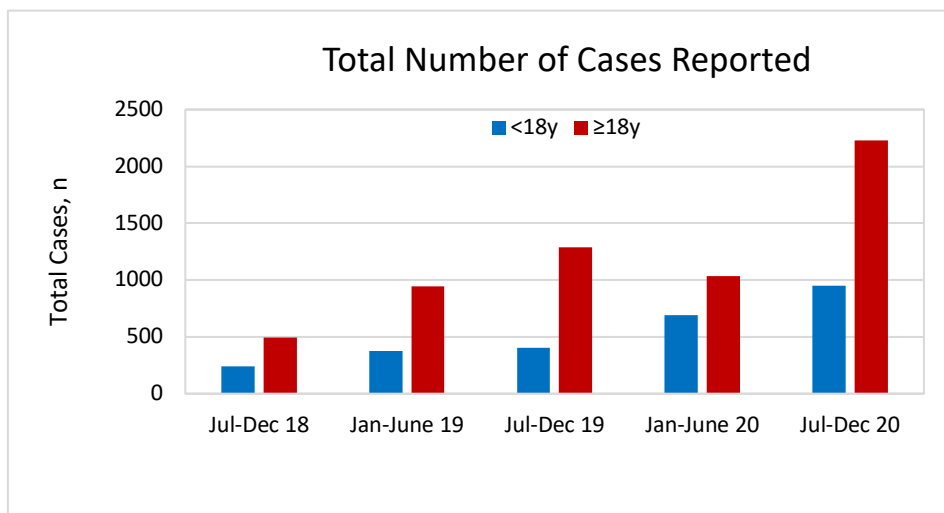


Fig. 2a – The change in cases reported between July 2018 and December 2020. A total of 5,987 new cases in adults and 2,652 new cases in children have now been reported.

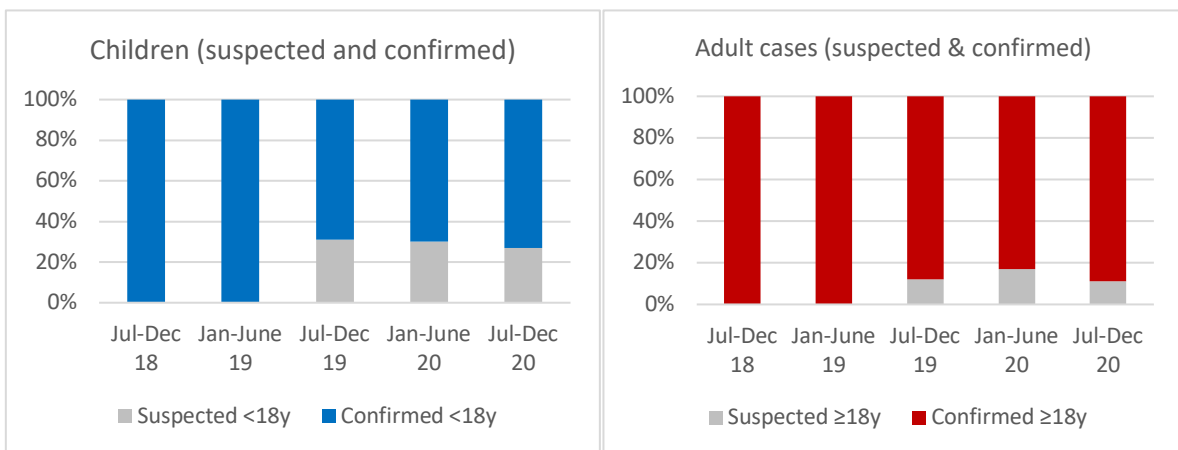


Fig 2b. Since the last quarter of 2019, the platform collects information on the proportion of cases that are considered to be 'suspected' and the proportion that are considered to be 'confirmed'. Suspected cases (in grey) are shown as a percentage of total cases for children (blue) and adults (red).

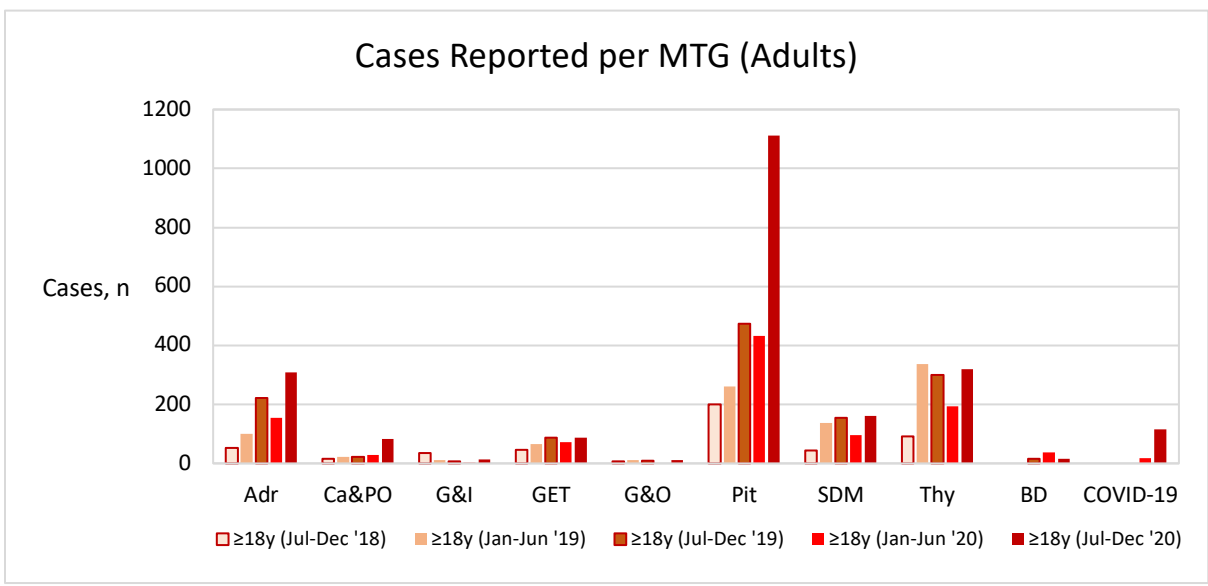
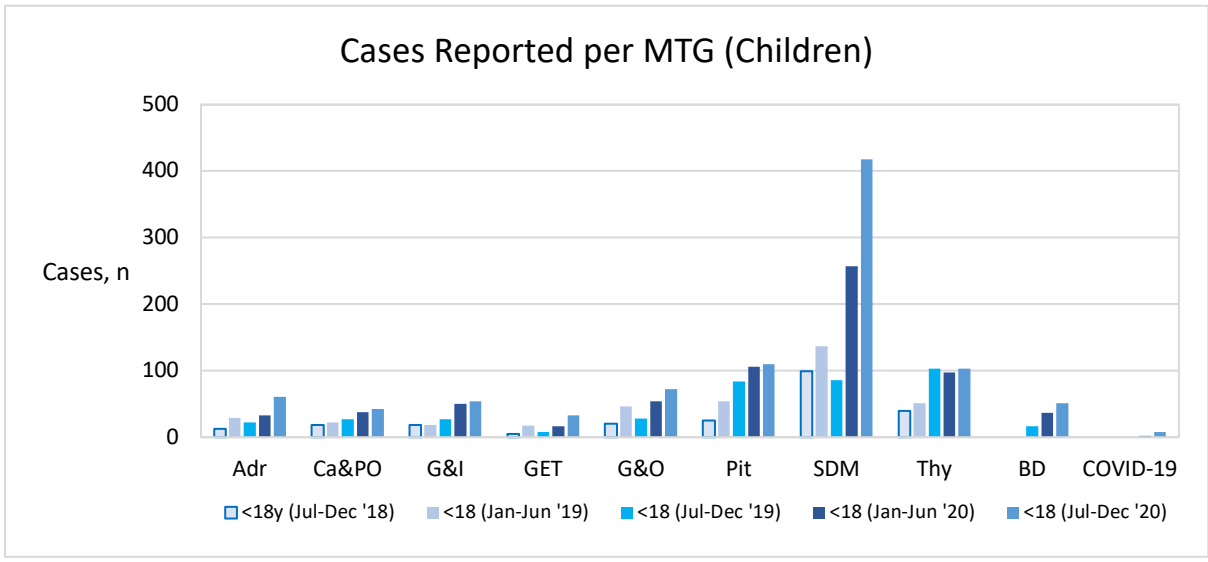


Fig.3 – Cases reported per Main Thematic Group (MTG) between July 2018 and December 2020. Conditions within the ‘Sex Development’ and ‘Pituitary’ condition groups were most commonly reported amongst children and adults, respectively.

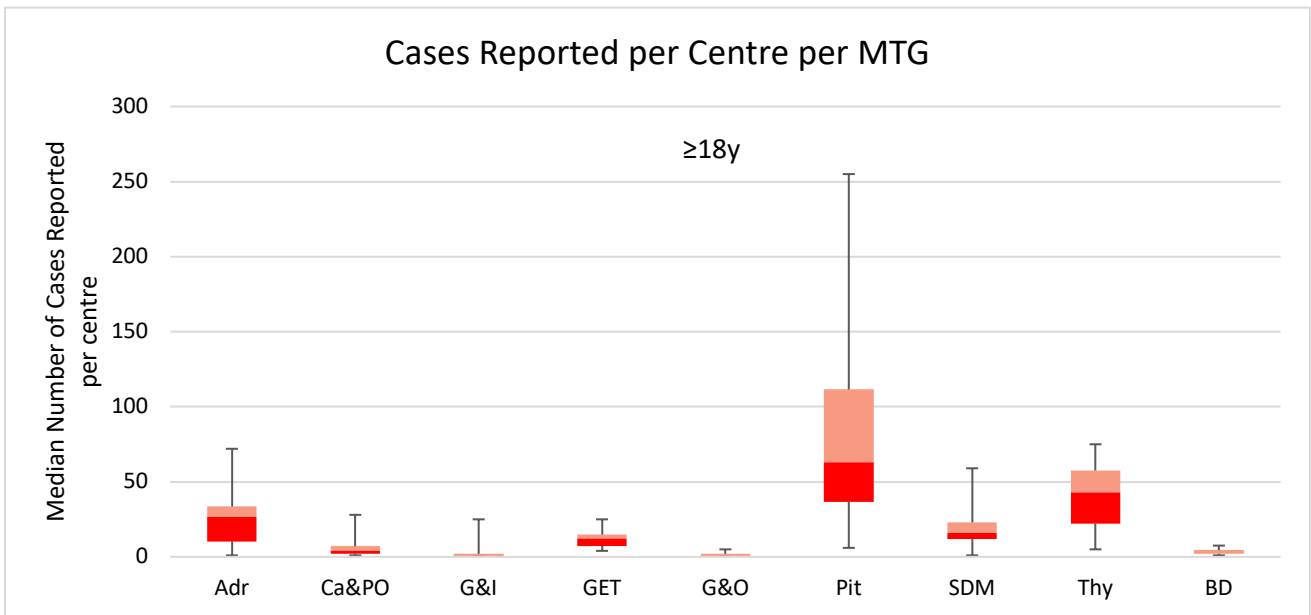
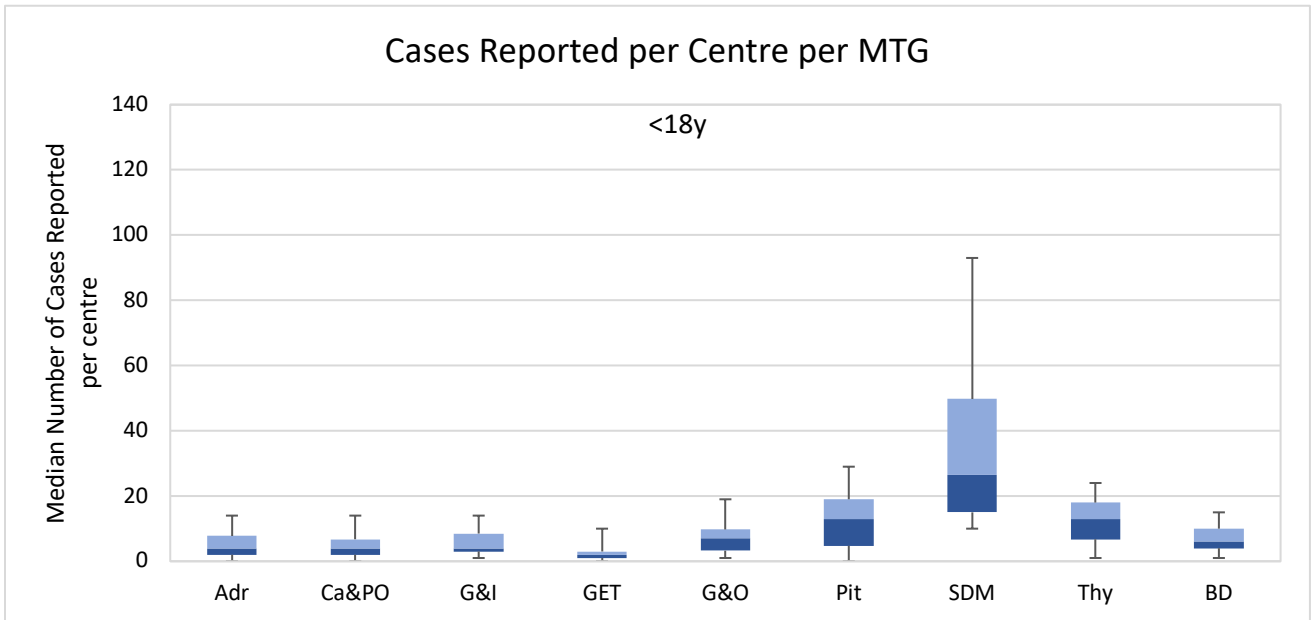
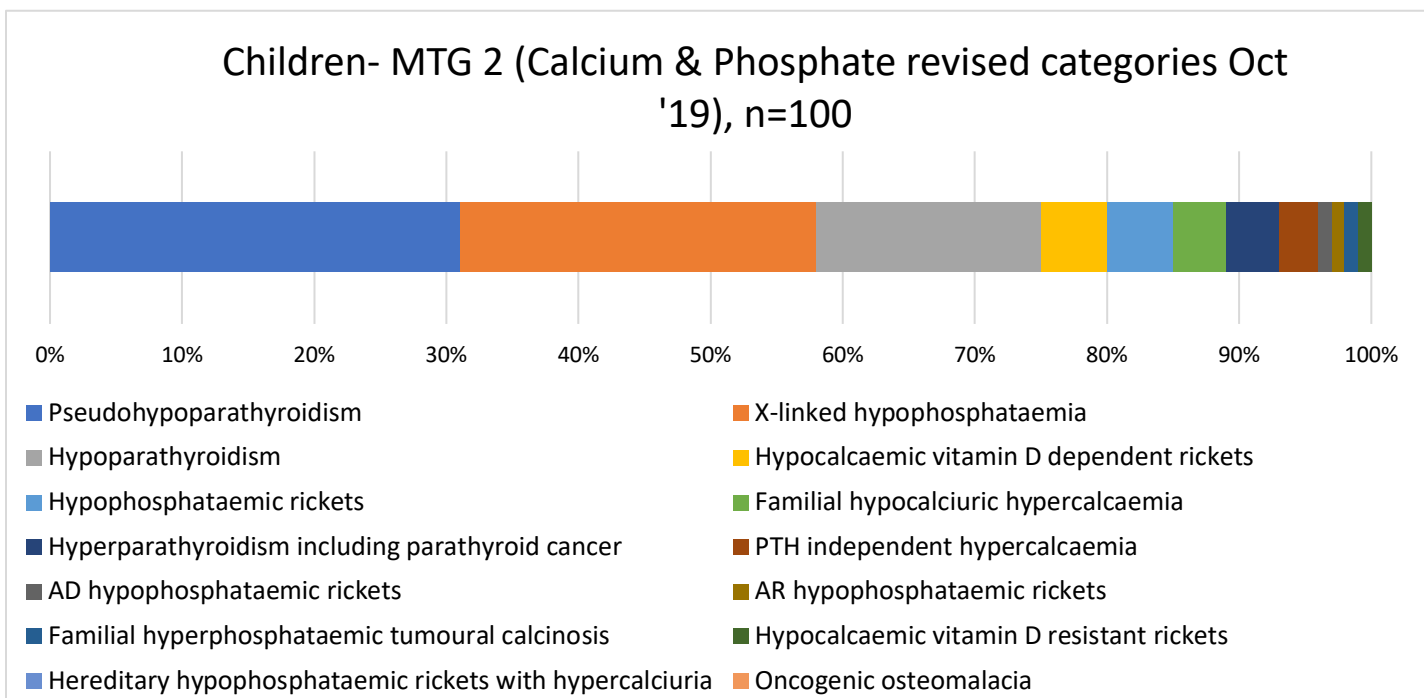
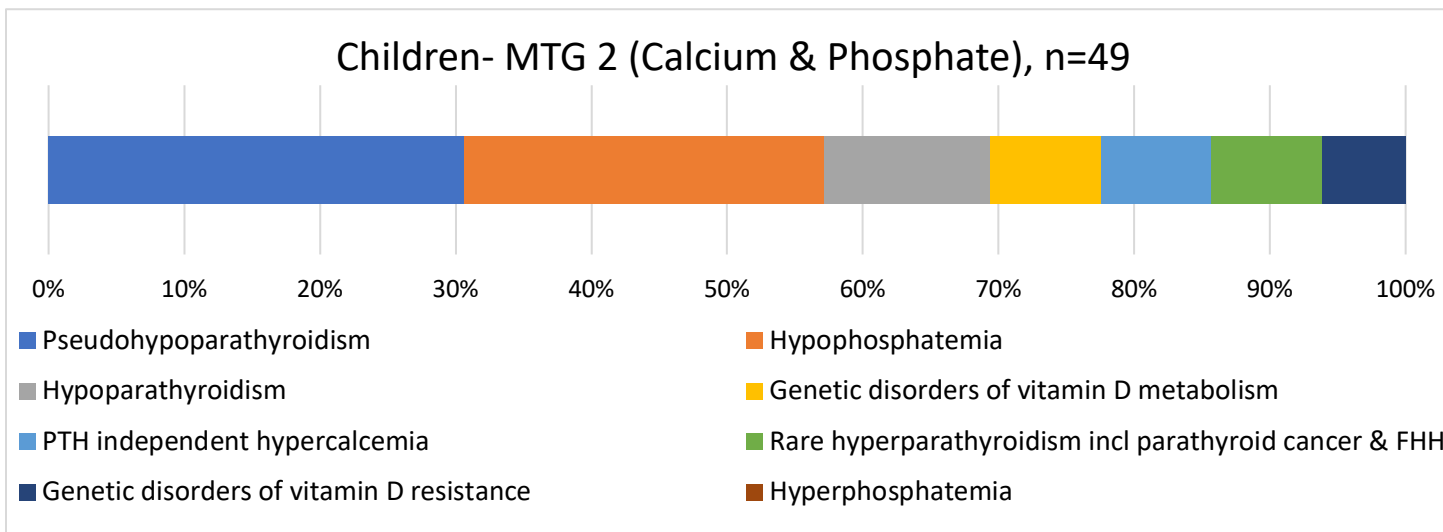
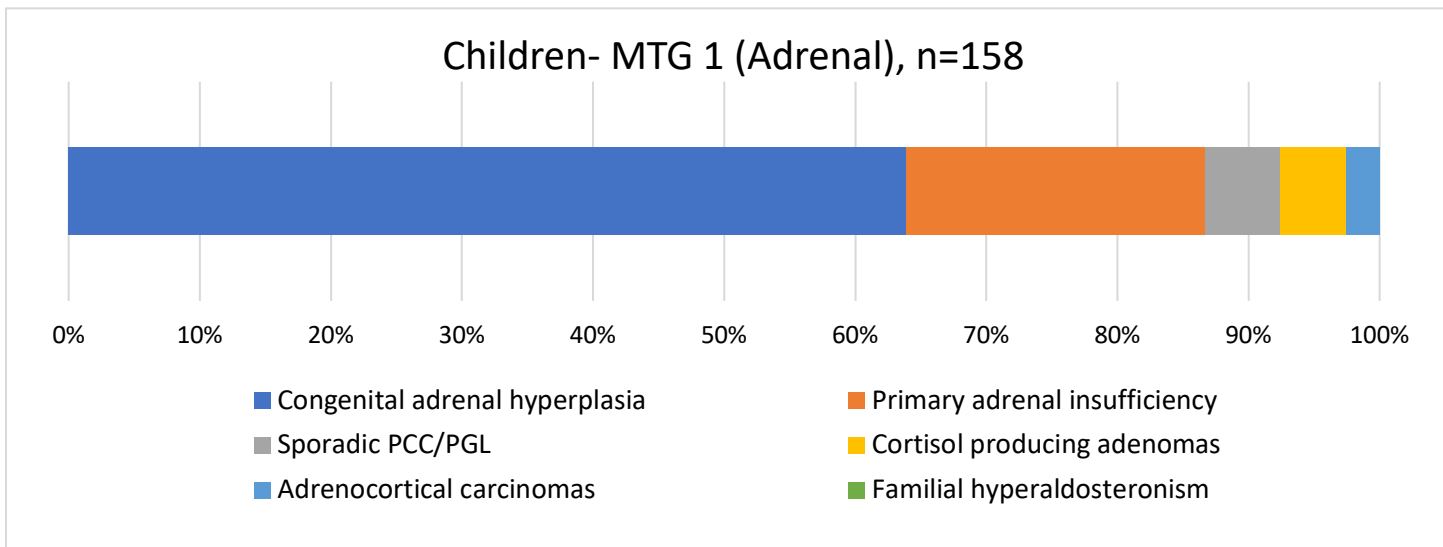
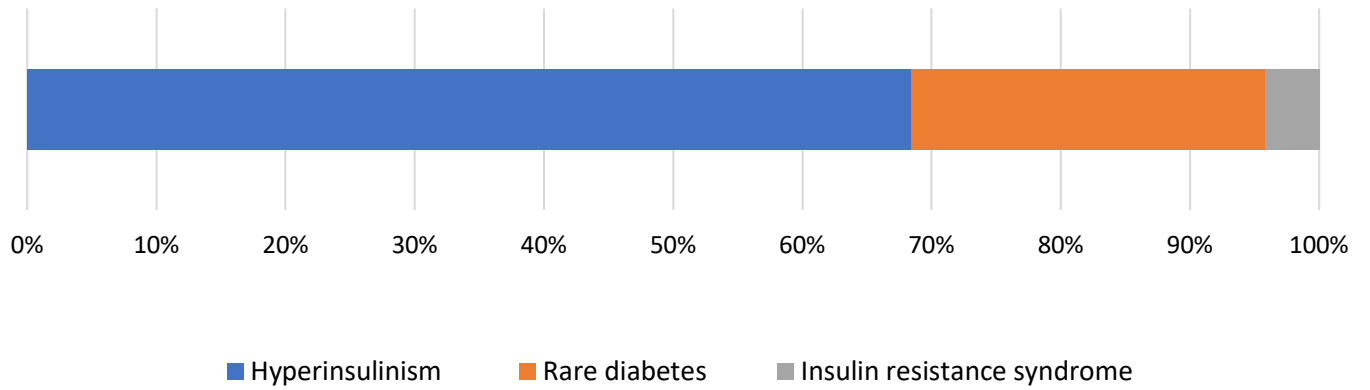


Fig.4 – Cases reported per centre per MTG between July 2018 and December 2020.

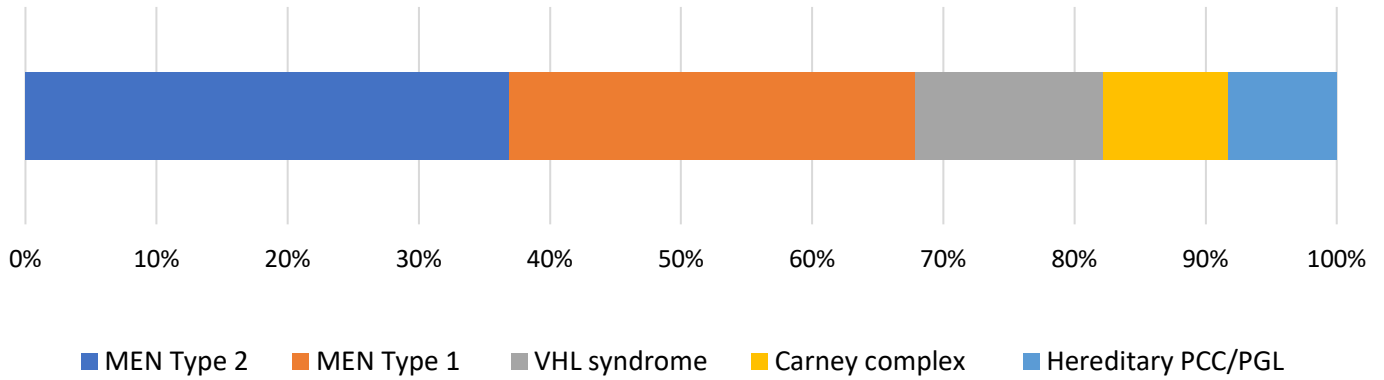
Fig.5a – Commonly reported conditions in children between July 2018 and December 2020; cases, n.



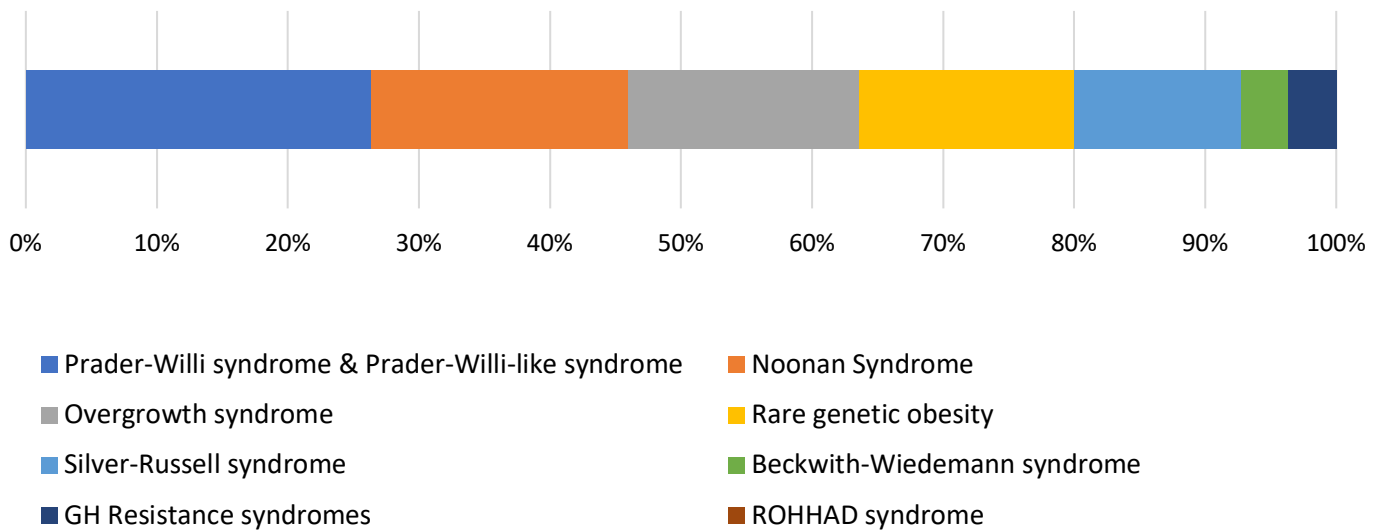
Children- MTG 3 (Glucose & Insulin), n=168



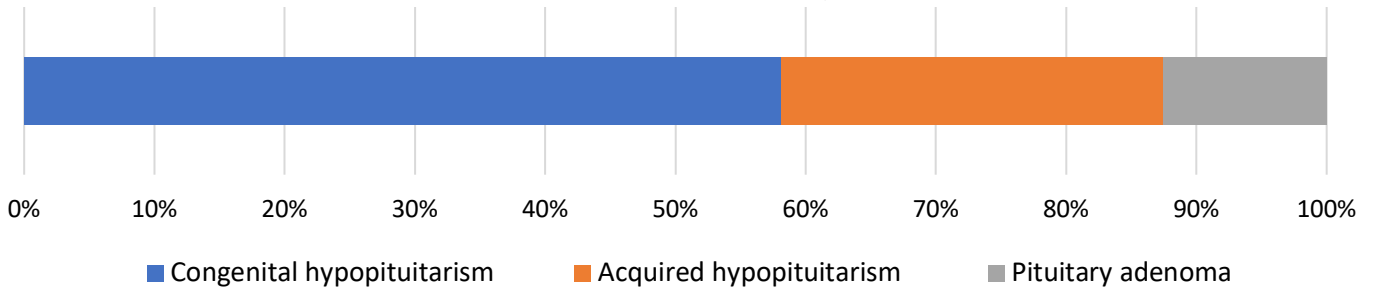
Children- MTG 4 (Genetic Endocrine Tumours), n=84



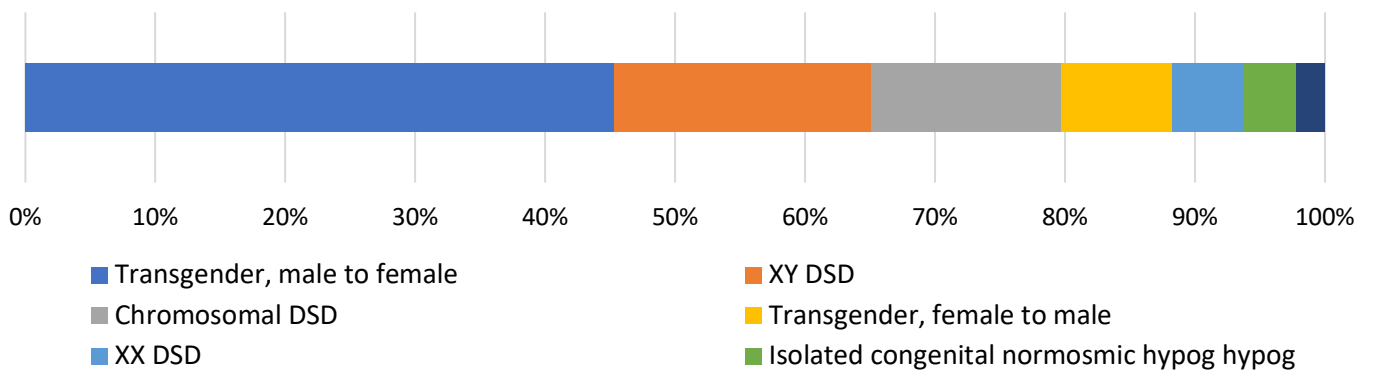
Children- MTG 5 (Growth & Obesity), n=220



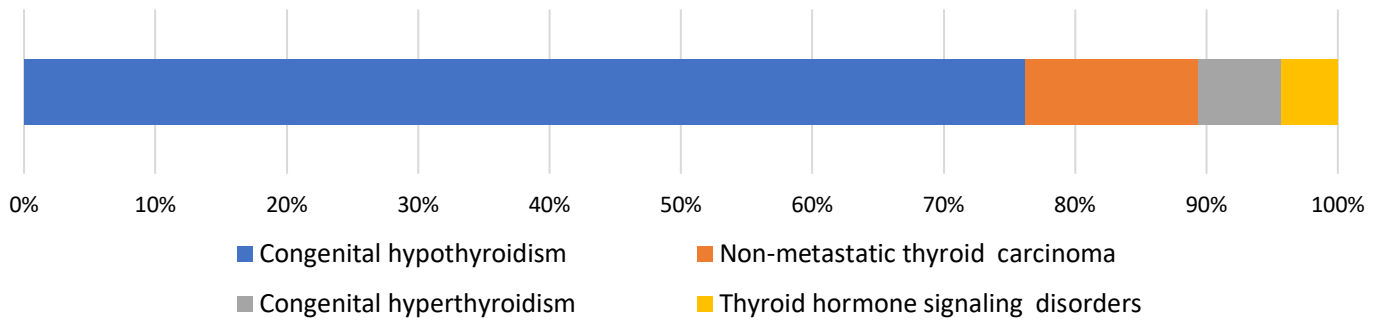
Children- MTG 6 (Pituitary), n=384



Children- MTG 7 (Sex Development), n=1002



Children- MTG 8 (Thyroid), n=394



Children- Bone Dysplasia (n=104)

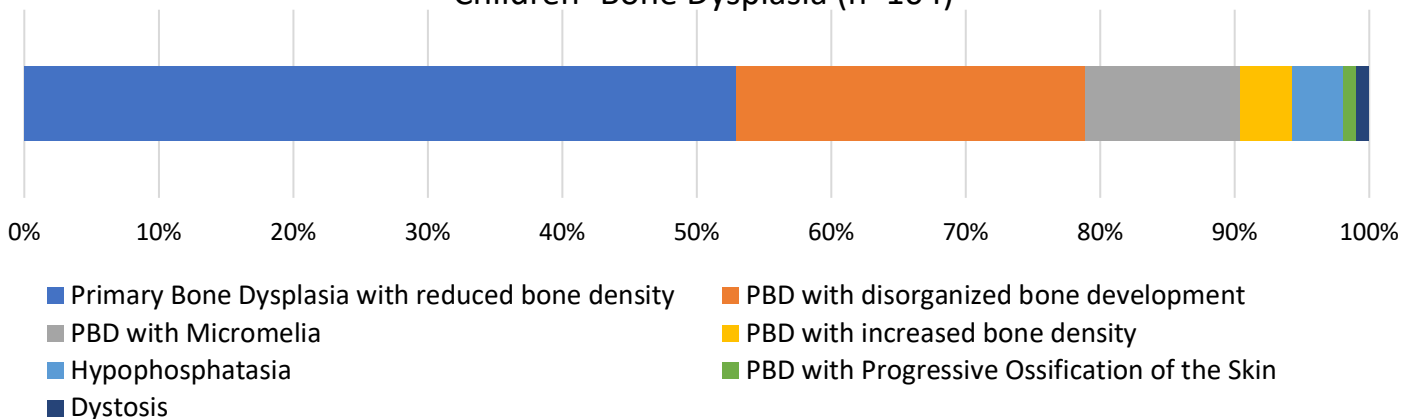
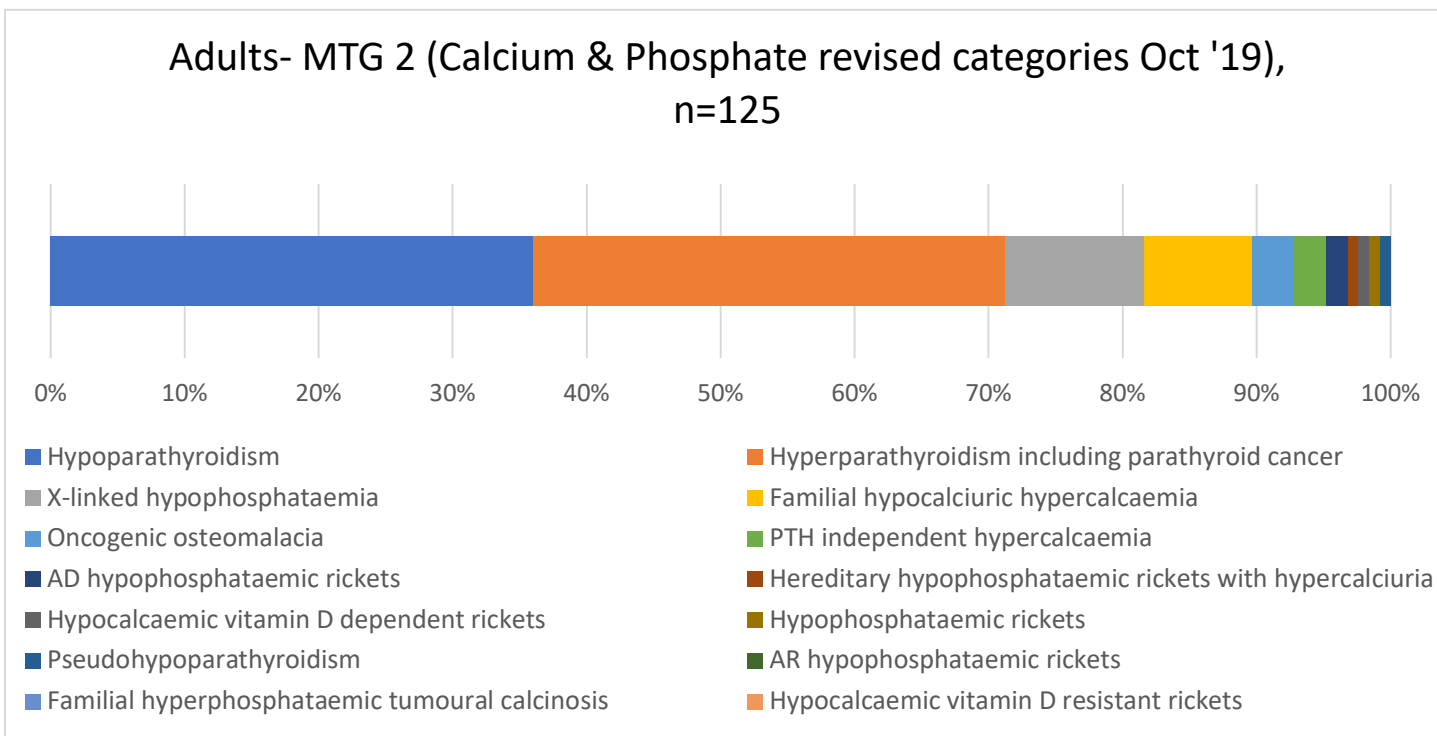
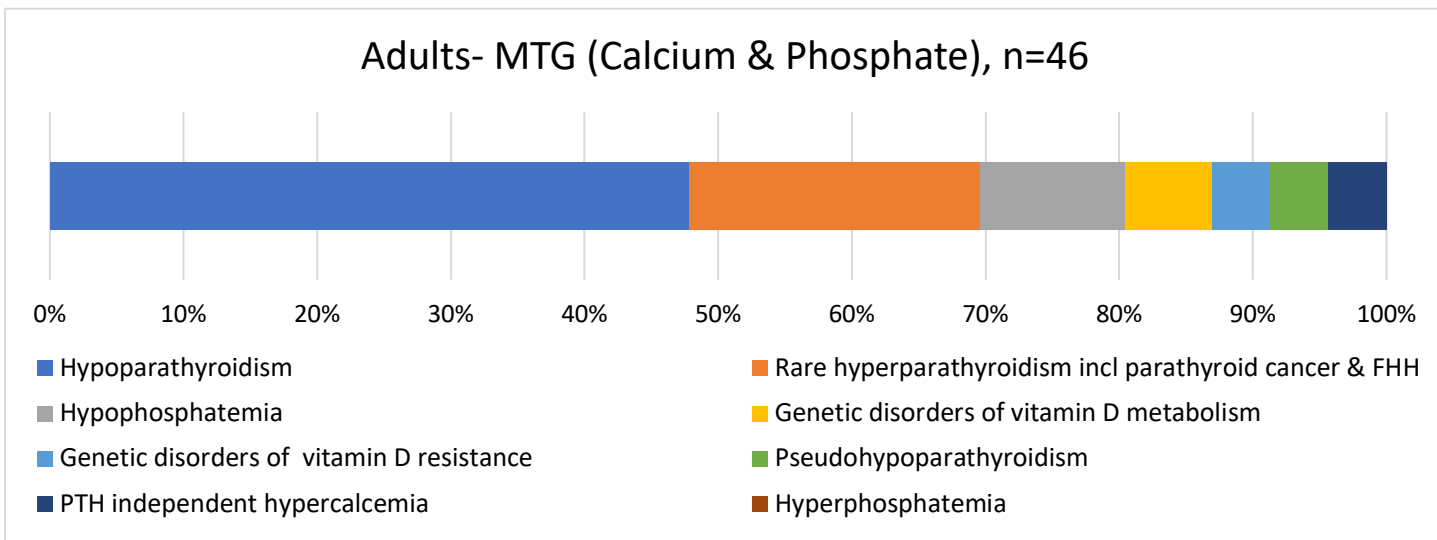
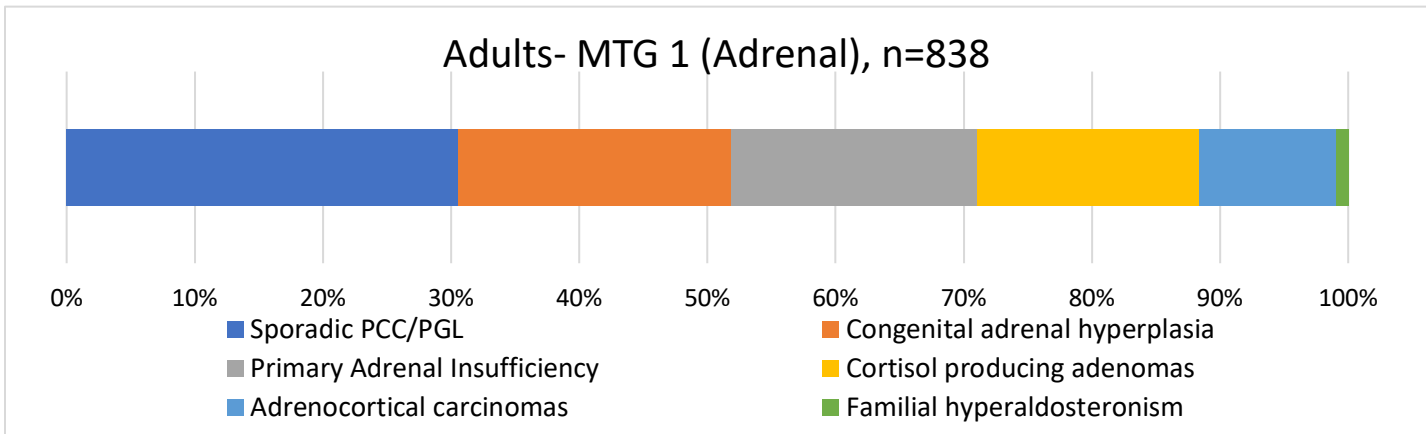
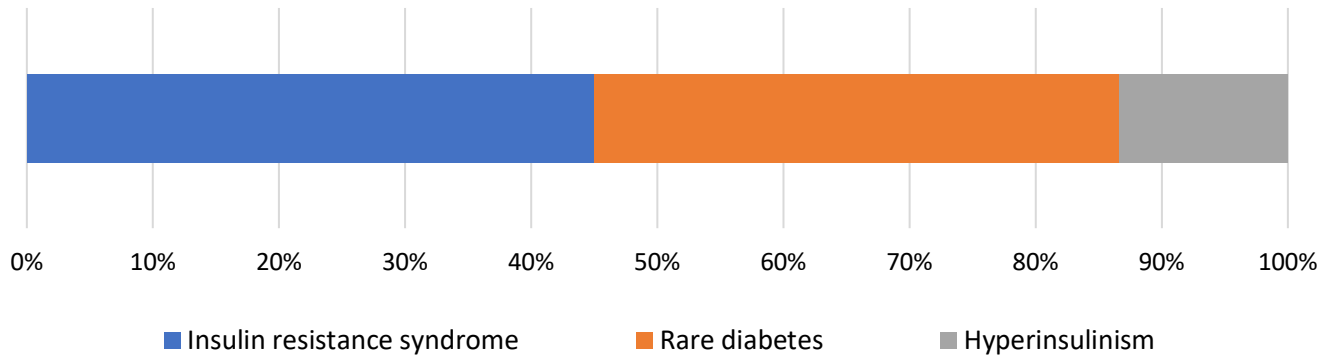


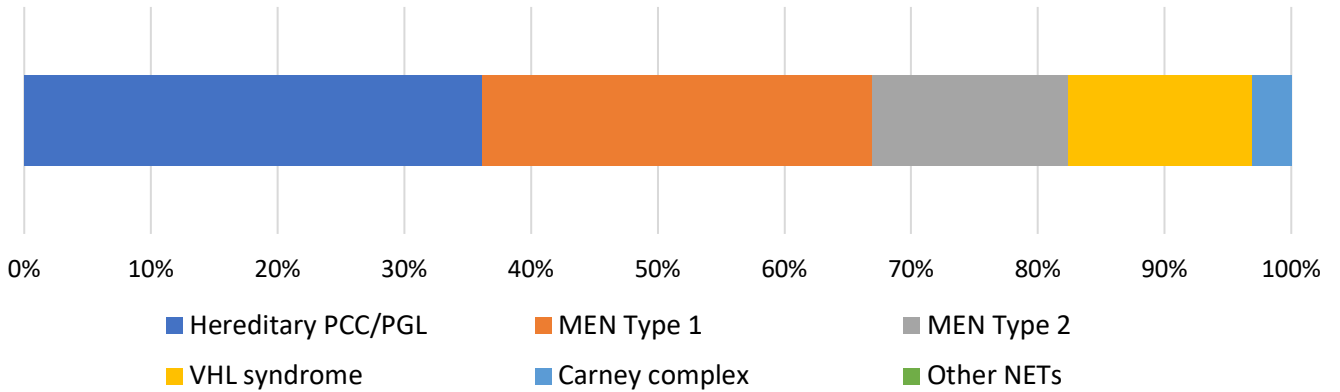
Fig.5b – Commonly reported conditions in adults between July 2018 and December 2020; cases, n.



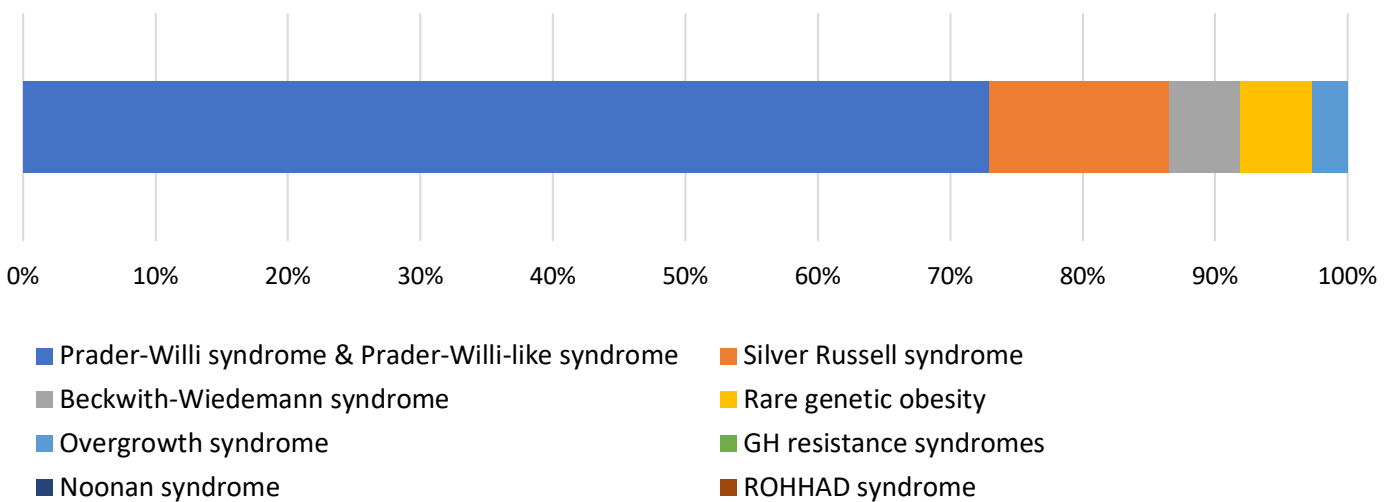
Adults- MTG 3 (Glucose & Insulin), n=60



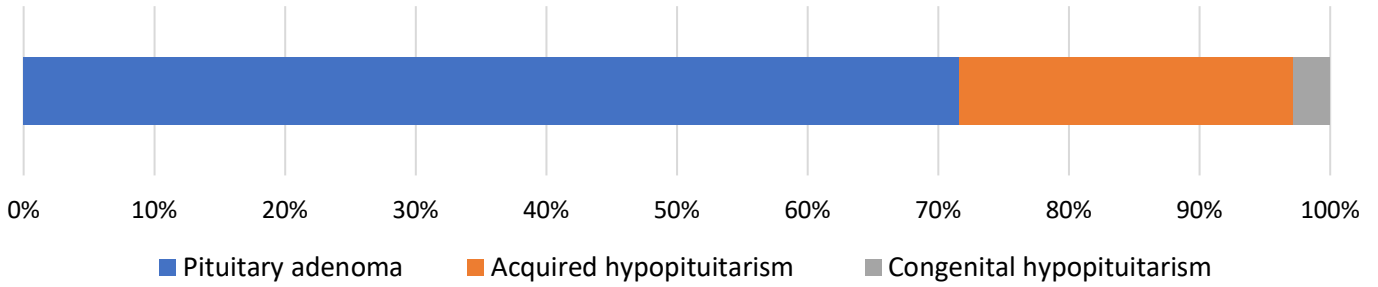
Adults- MTG 4 (Genetic Endocrine Tumours), n=357



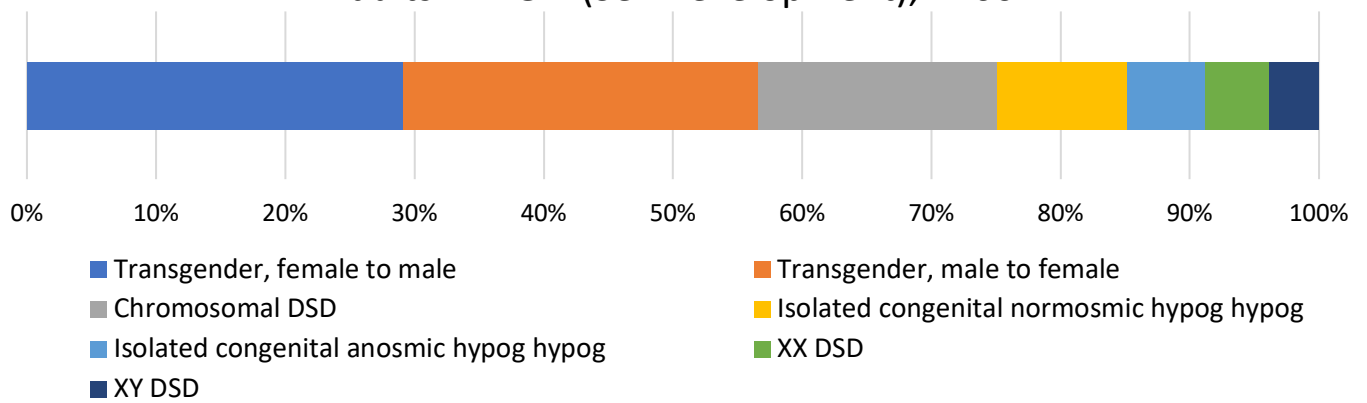
Adults- MTG 5 (Growth & Obesity), n=37



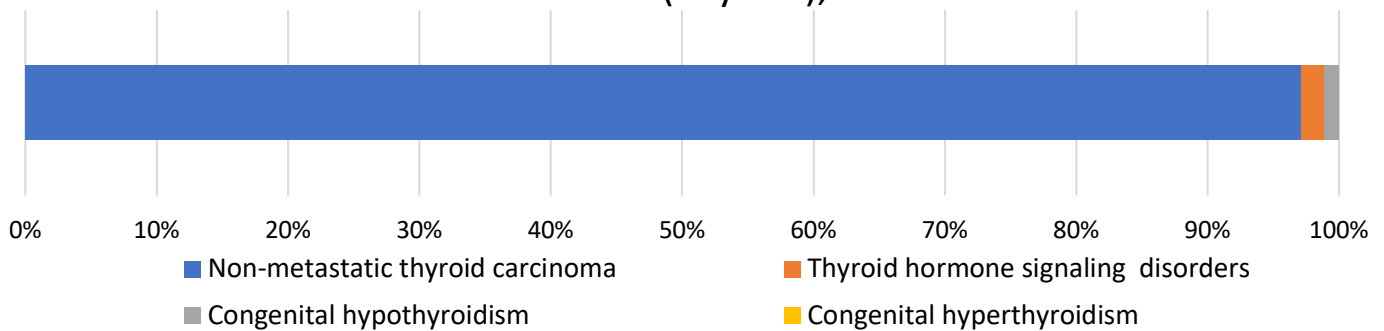
Adults- MTG 6 (Pituitary, n=2480)



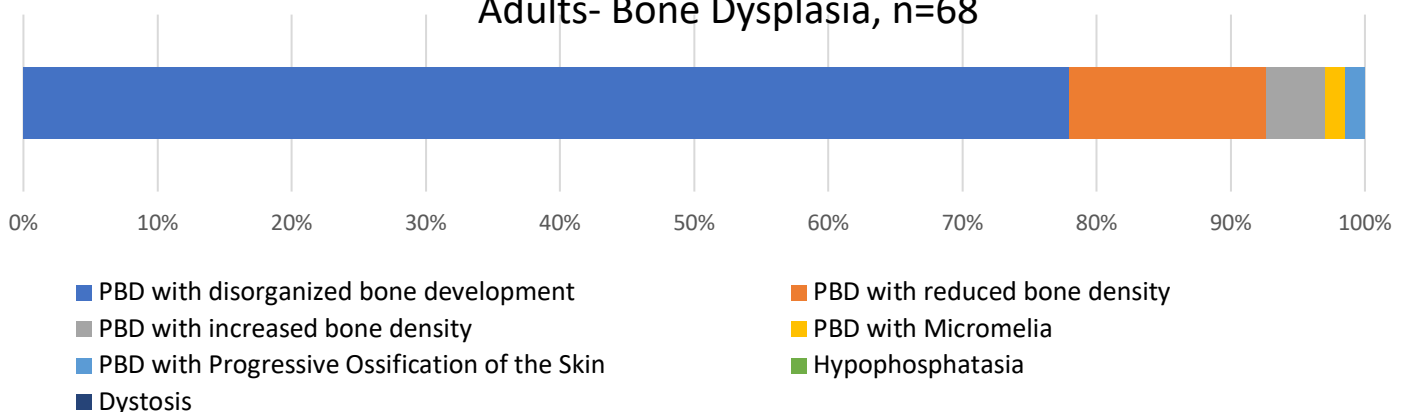
Adults- MTG 7 (Sex Development), n=601



Adults- MTG 8 (Thyroid), n=1236



Adults- Bone Dysplasia, n=68



Interpretation of Findings

- The e-REC platform is a simple registry platform that can be used to capture information on new encounters with patients with rare endocrine and 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.
- During 2020, the number of new cases that were being reported at participating centres did not show a decline.
- There is wide variability in the number of cases encountered for different conditions amongst different centres.
- The proportion of cases that have a suspected diagnosis are higher in children compared to adults and may reflect the groups of conditions that are reported more often in these two groups.

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 now available to investigators for further analysis and research.

Jillian Bryce, Salma Ali, Ana Priego, Natasha Appelman-Dijkstra, Faisal Ahmed
EuRRECa Work Package 6
March 2021