

**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 3-year period from July 2018 to June 2021 inclusive.

Methods

The e-REC platform issued invitations to clinicians who had registered to participate in e-REC from July 2018 to June 2021 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 June 2021, a total of 60 centres from 22 countries had reported on the e-REC. Of these 60 centres, 23 are in Endo-ERN only, 16 are in Endo-ERN and ERN-BOND, 3 are in ERN-BOND only and 18 are in no ERNs.


Country	Centres	Country ²	Centres ³
 Austria	5	 Israel	1
 Belgium	7	 Italy	9
 Croatia	1	 Latvia	1
 Czechia	1	 Lithuania	1
 Egypt	1	 Luxembourg	1
 Estonia	1	 Netherlands	6
 Finland	1	 Portugal	1
 France	2	 Romania	1
 Germany	8	 Spain	3
 Greece	2	 Sweden	2
 Hungary	1	 UK	4

Table 1 – Number of centres in each country that are active (i.e. have submitted 1 or more returns between July 2018 and June 2021).

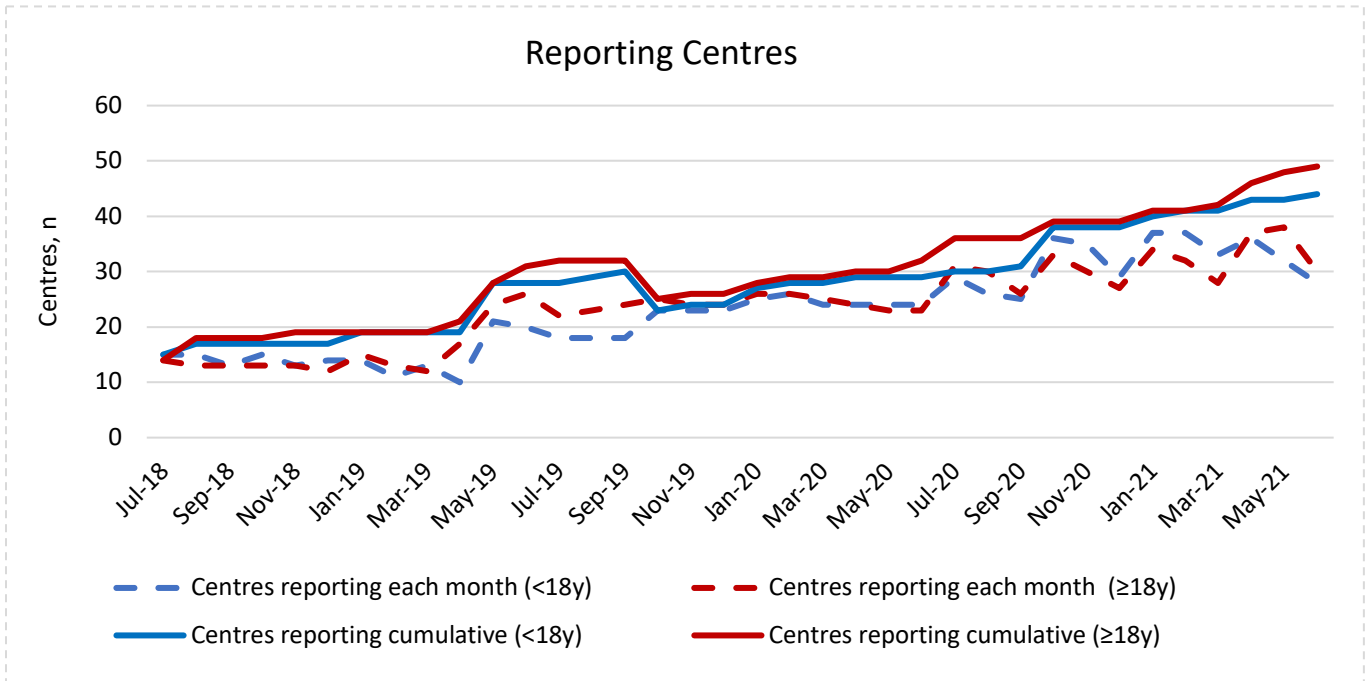


Fig.1 – Change in the number of centres that have been actively reporting between July 2018 and June 2021. Currently, 44 paediatric centres and 49 adult centres have actively participated in e-REC.

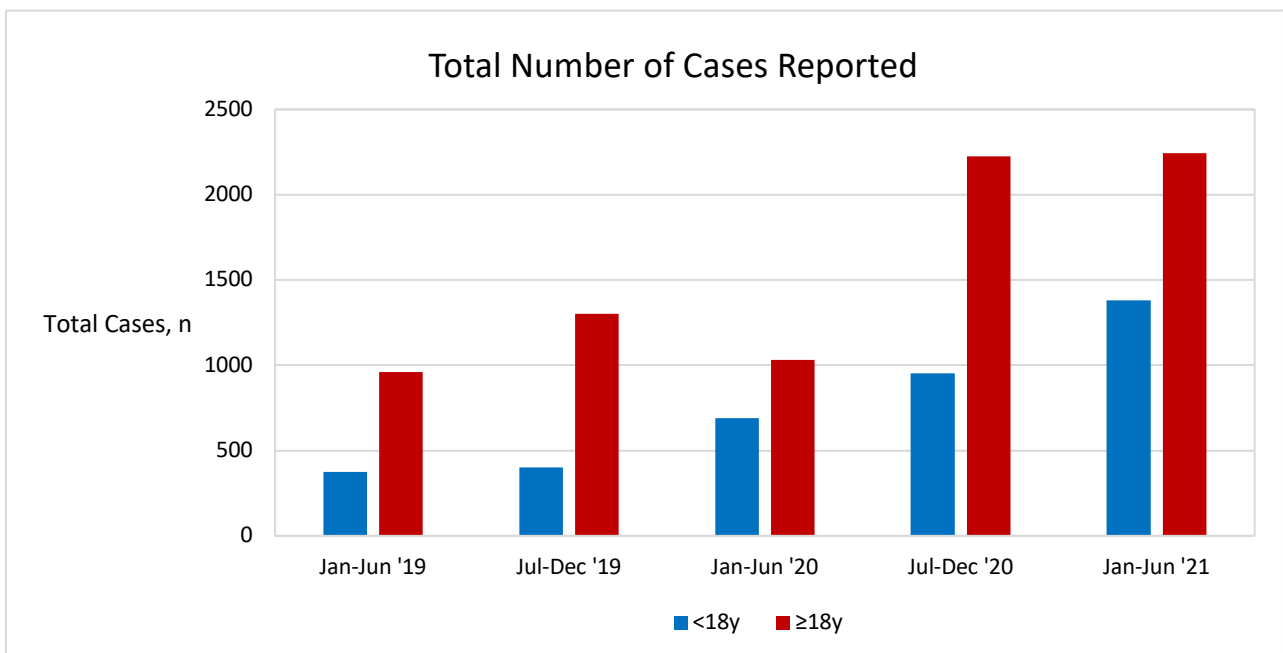


Fig. 2a – The change in cases reported between July 2018 and June 2021. A total of 8,247 new cases in adults and 4,056 new cases in children have now been reported.

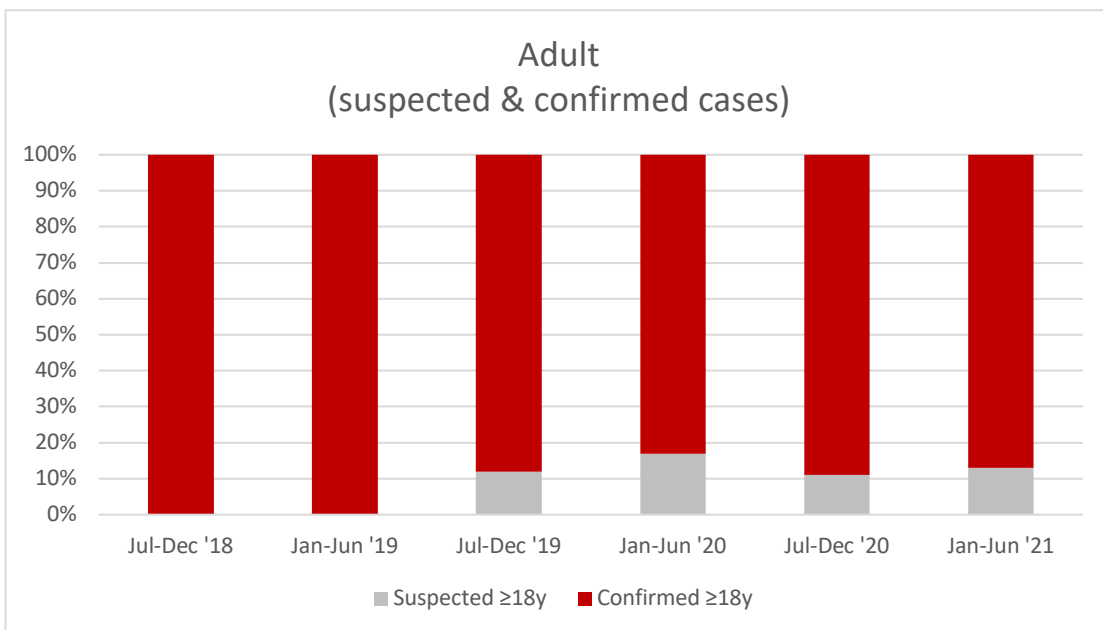
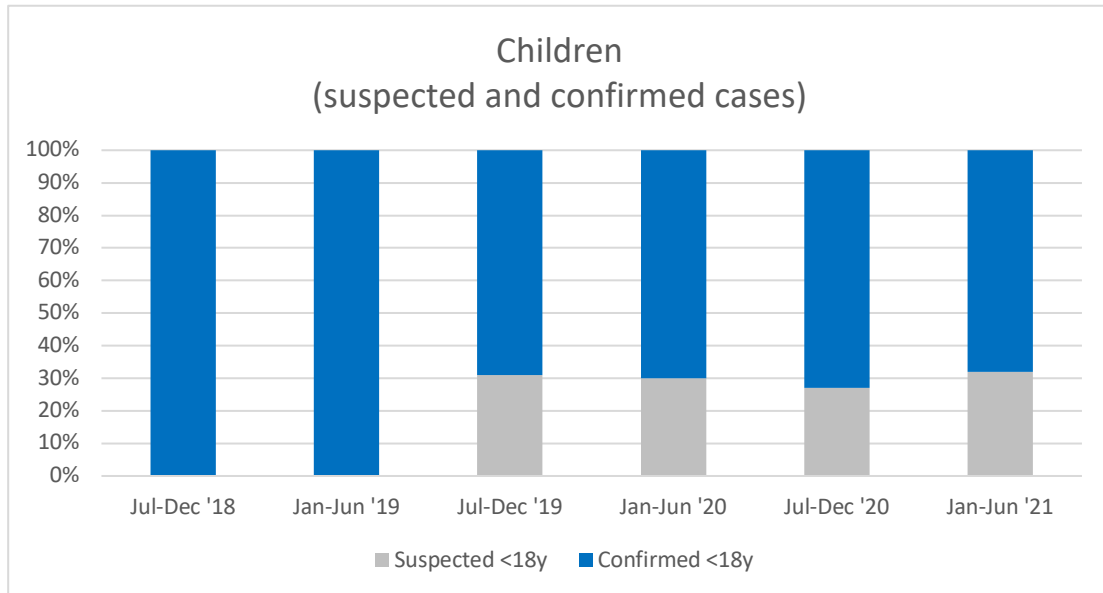


Fig 2b. Since the last quarter of 2019, the platform collects information on the proportion of cases that are considered to be 'suspected' and the proportion that are considered to be 'confirmed'. Suspected cases (in grey) are shown as a percentage of total cases for children (blue) and adults (red).

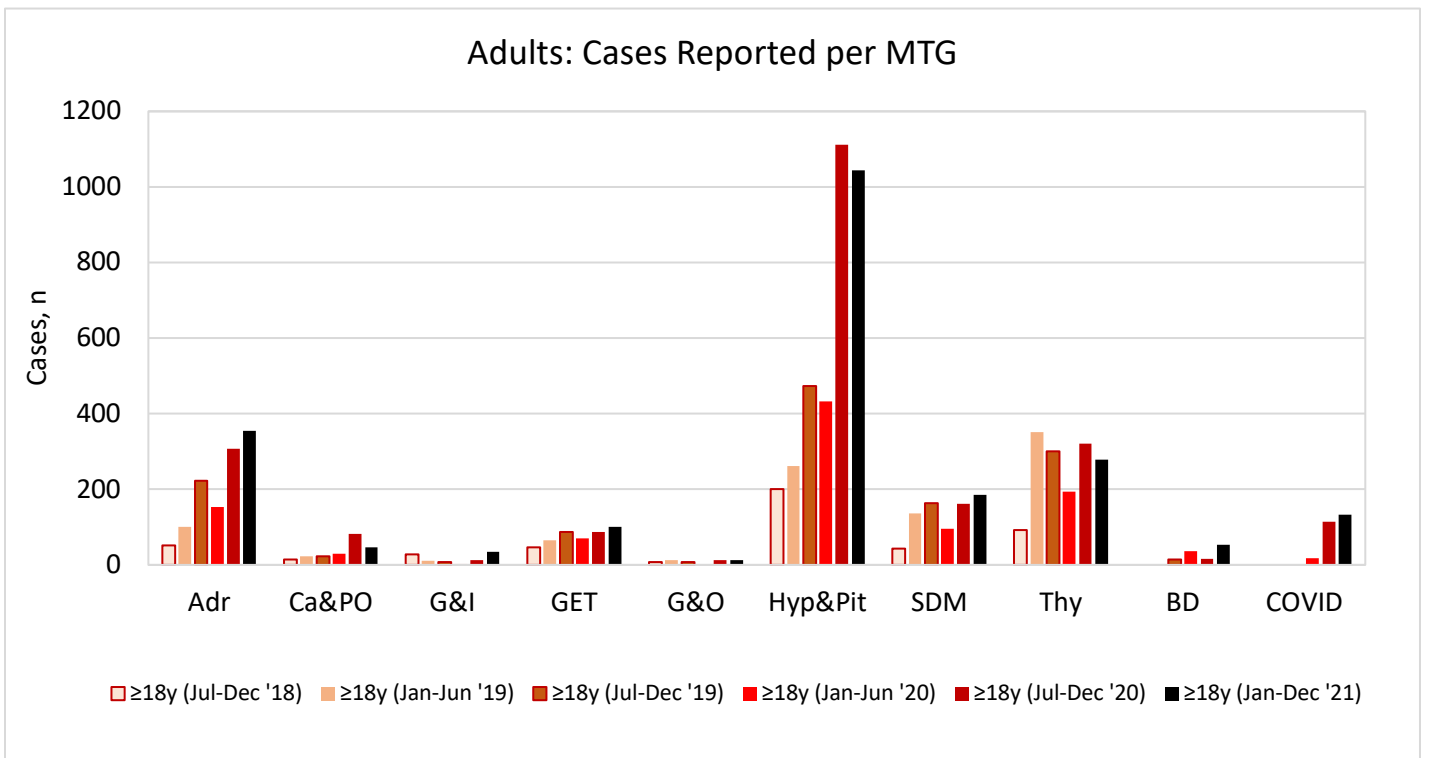
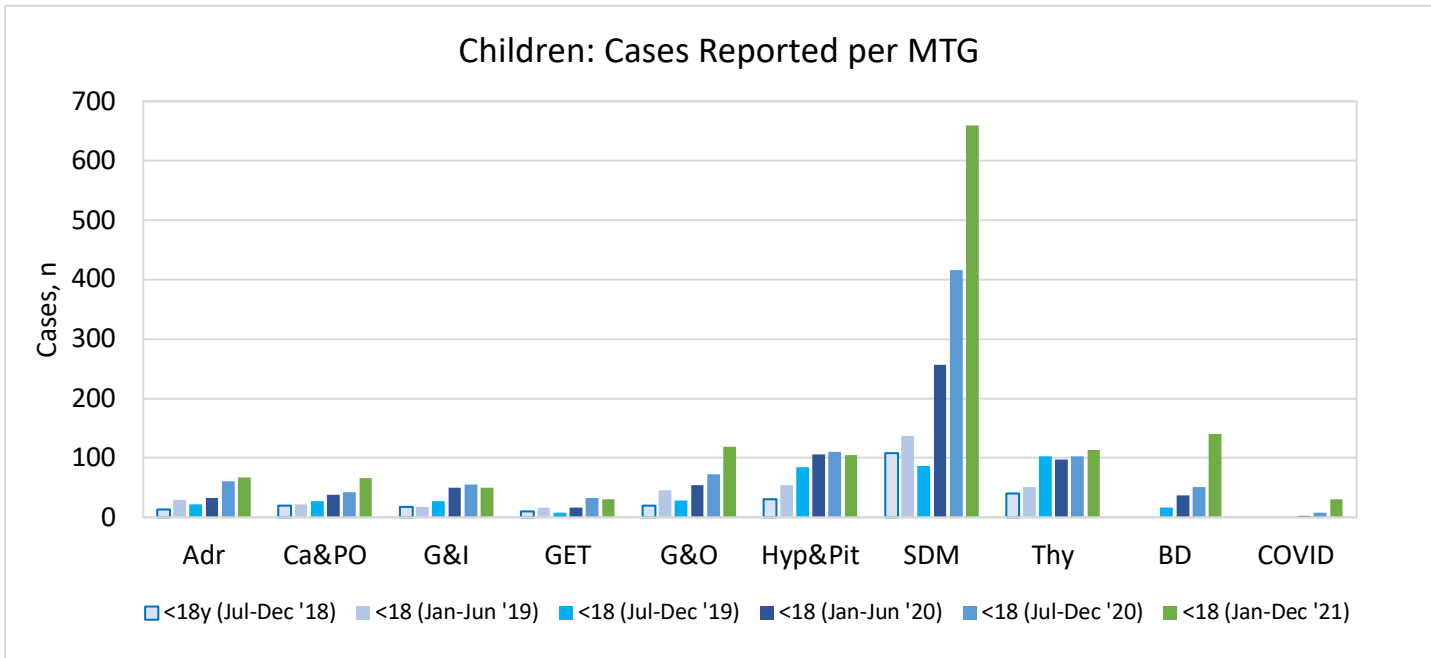


Fig.3 – Cases reported per Main Thematic Group (MTG) between July 2018 and June 2021. Conditions within the ‘Sex Development’ and ‘Pituitary’ condition groups were most commonly reported amongst children and adults, respectively. However, in general, the increase in reported conditions is across all MTGs.

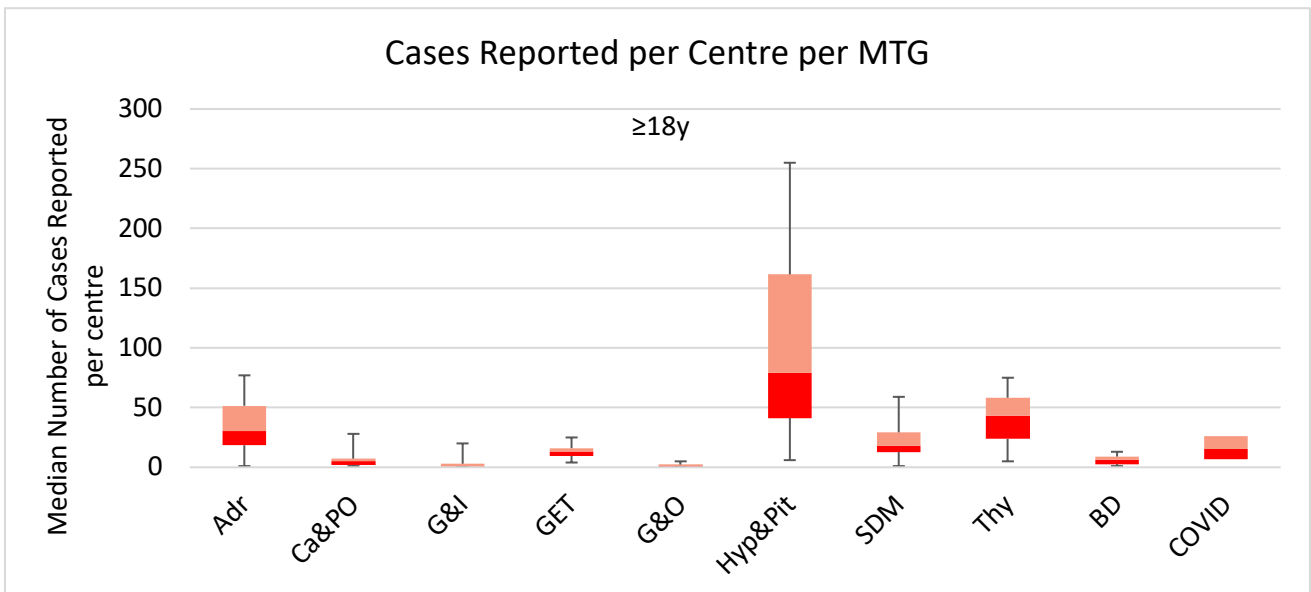
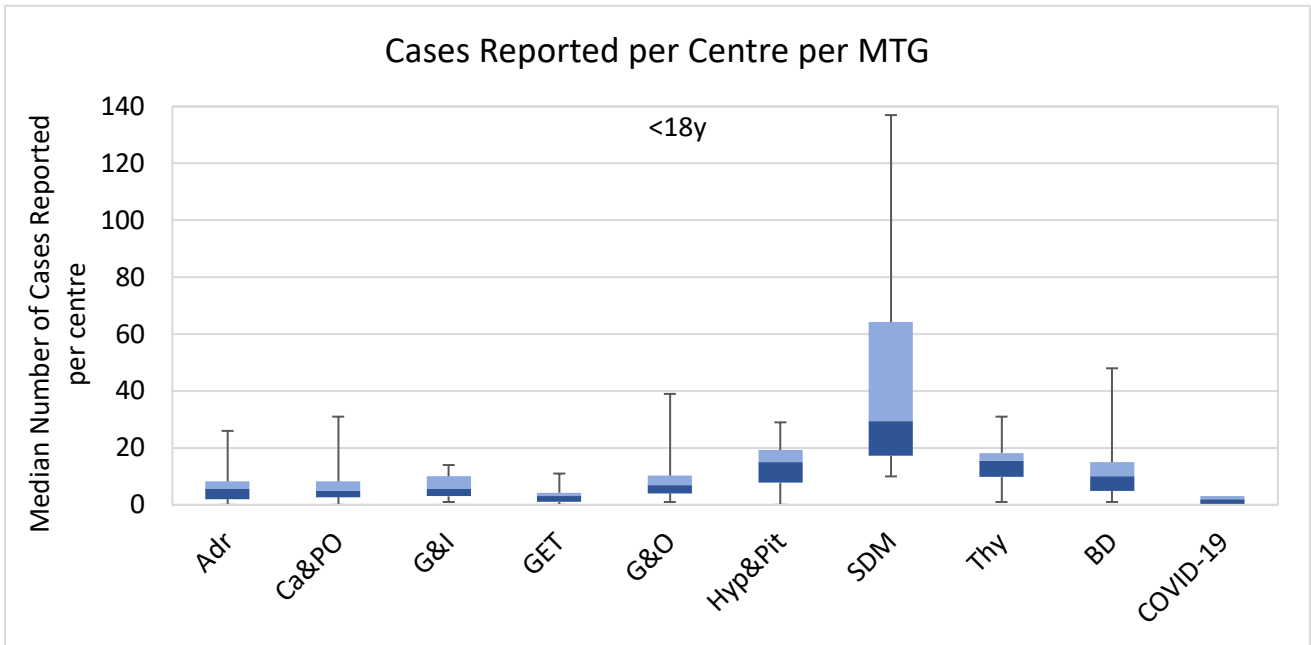
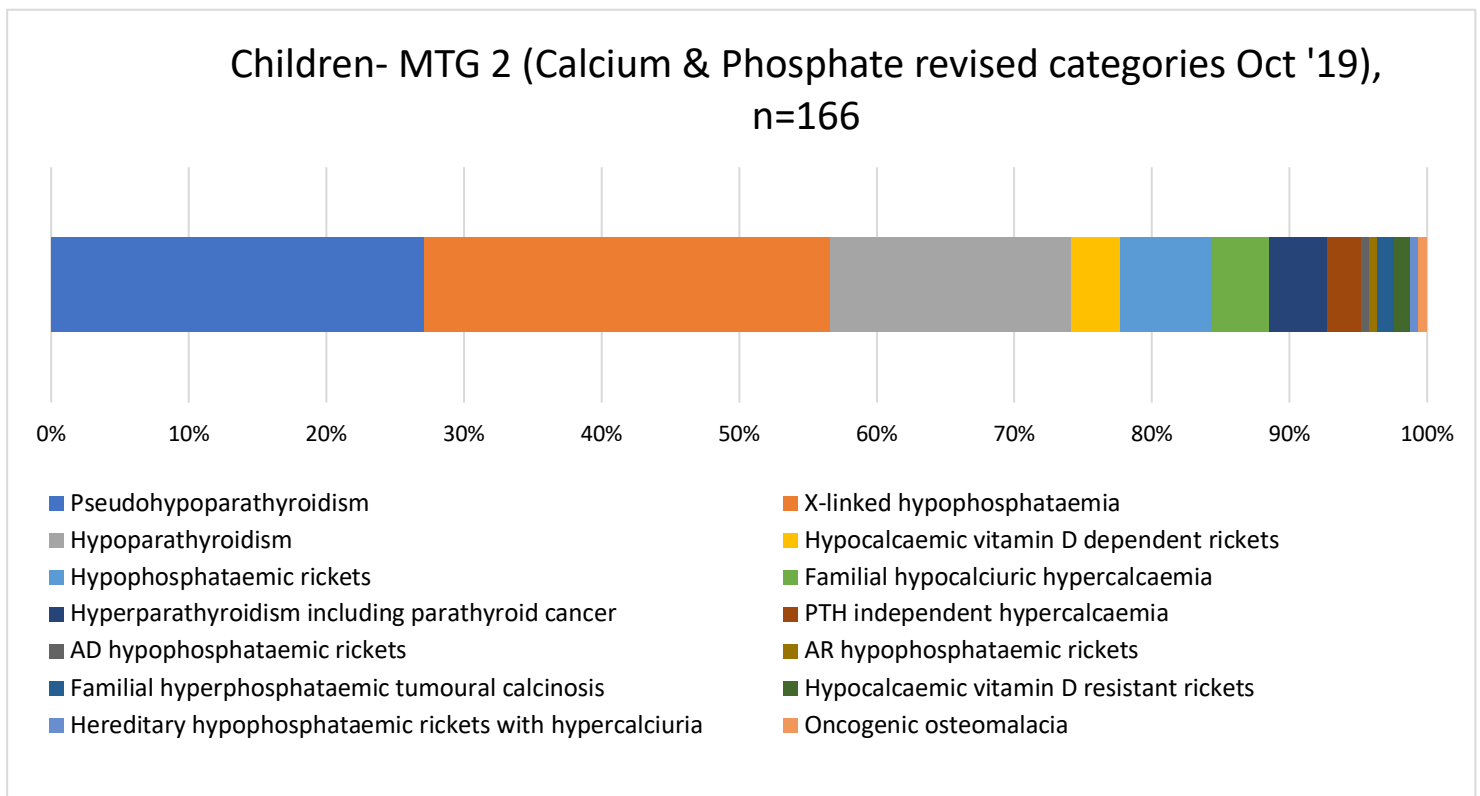
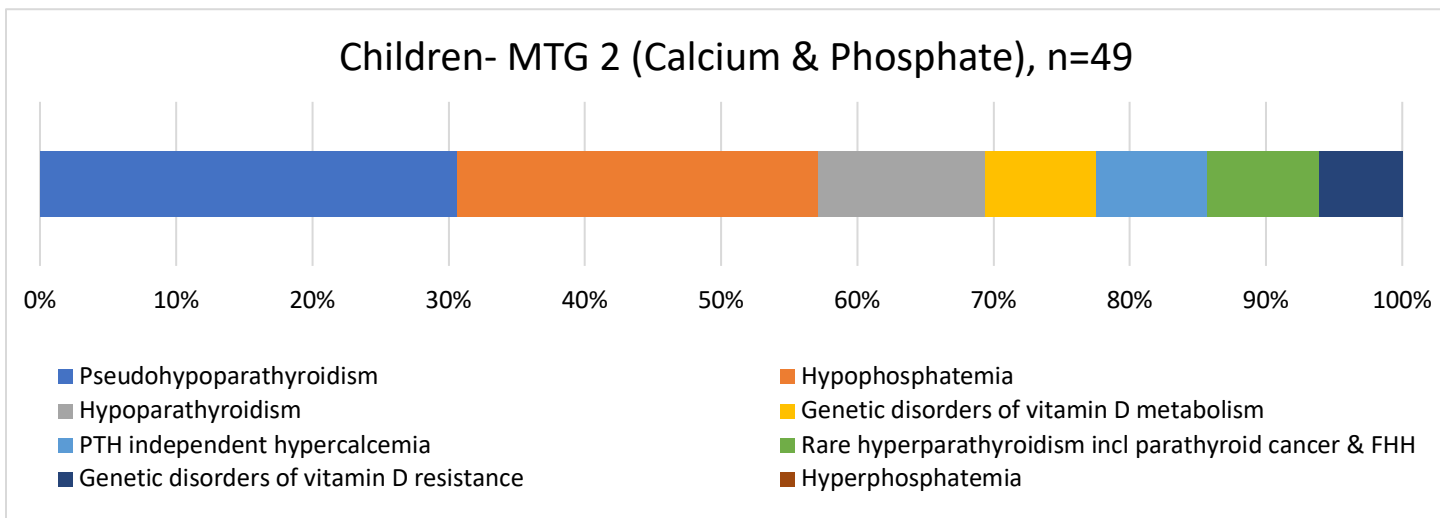
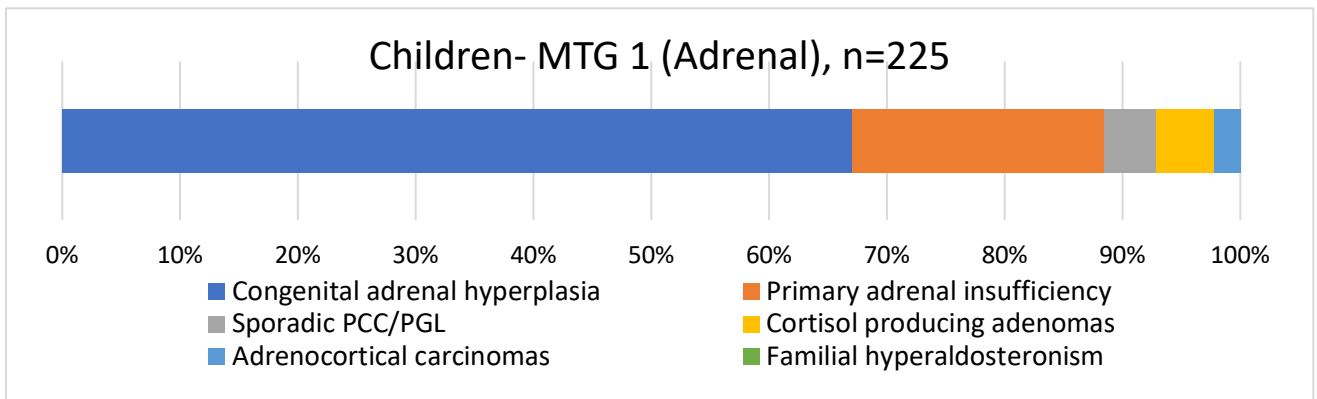
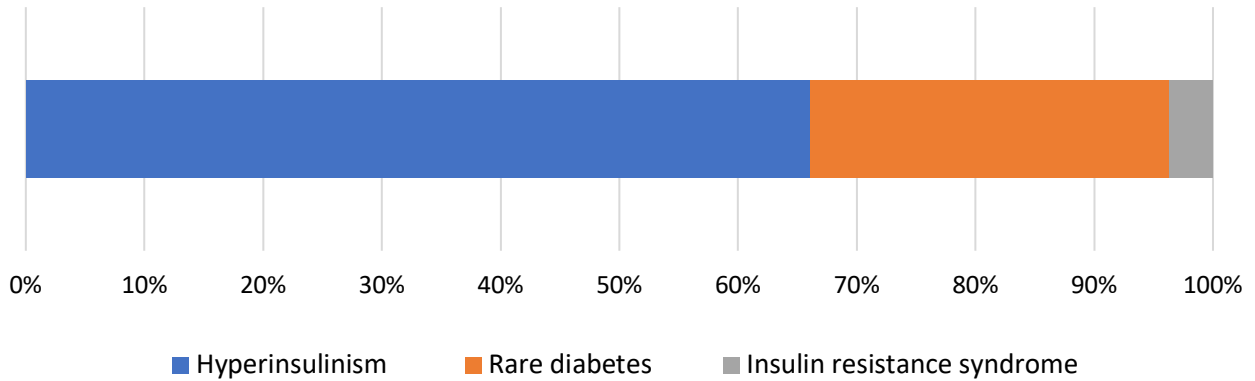


Fig.4 – Cases reported per centre per MTG between July 2018 and June 2021.

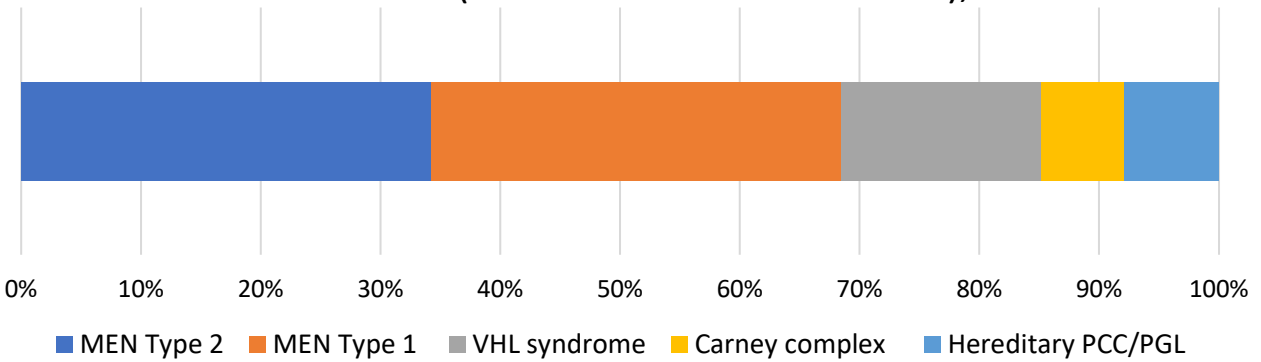
Fig.5a – Commonly reported conditions in children between July 2018 and June 2021; cases, n.



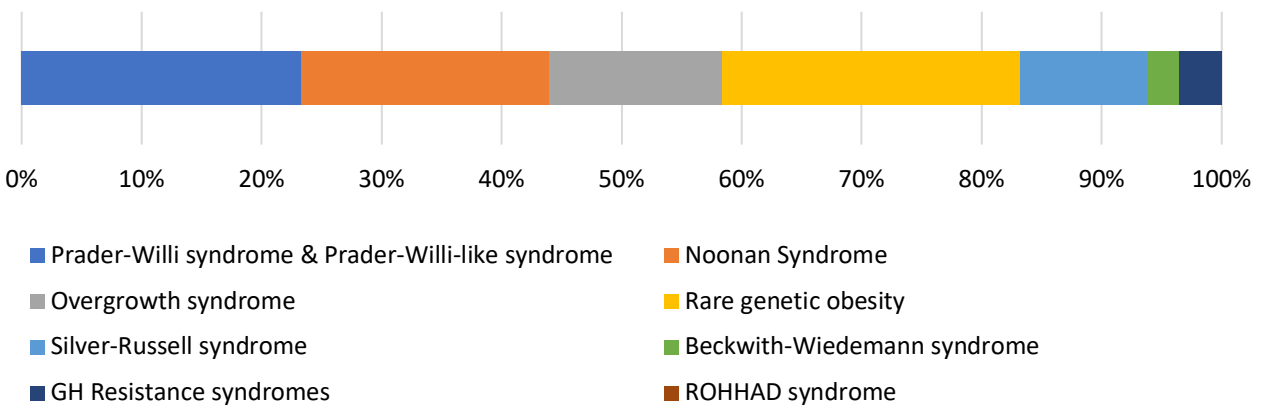
Children- MTG 3 (Glucose & Insulin), n=218

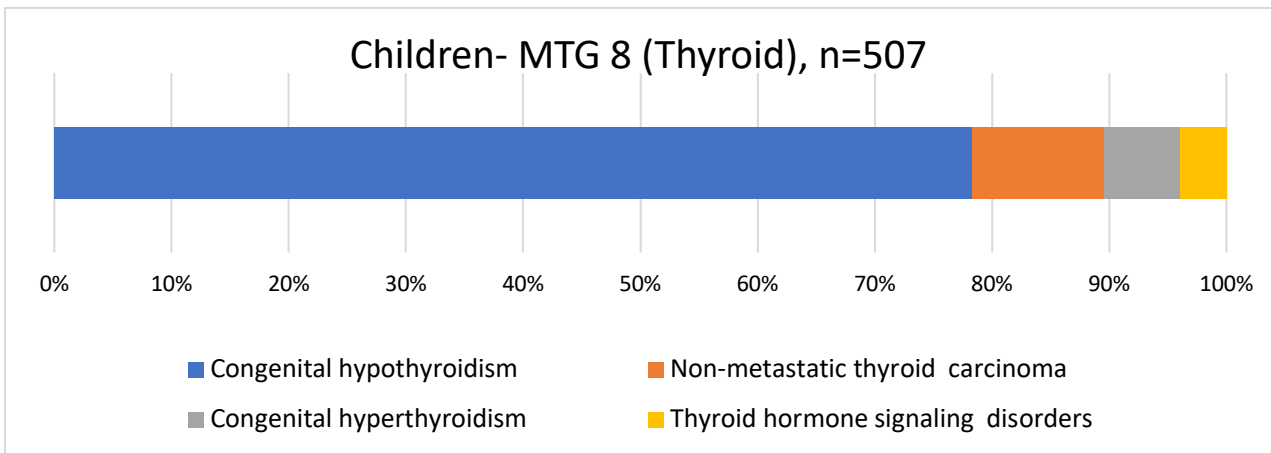
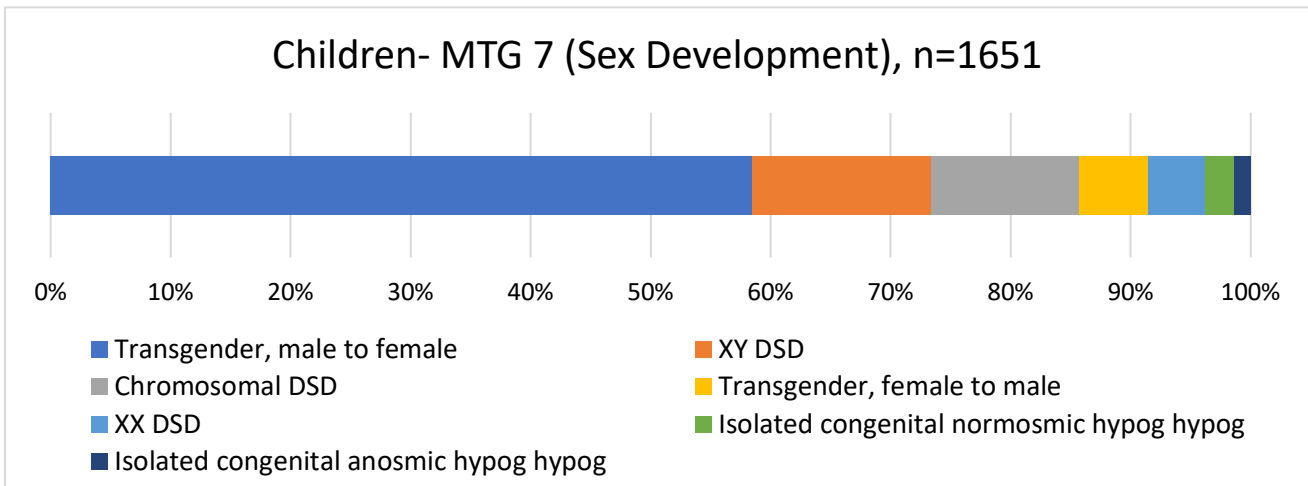
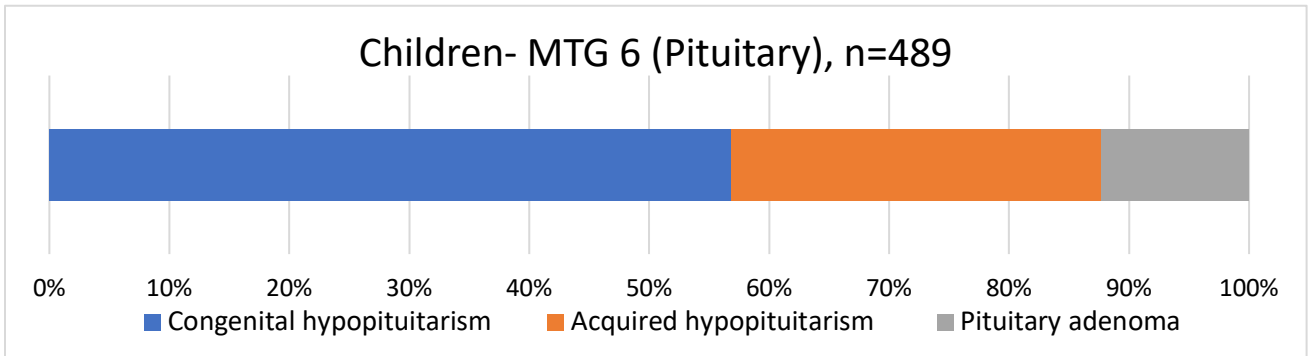


Children- MTG 4 (Genetic Endocrine Tumours), n=114

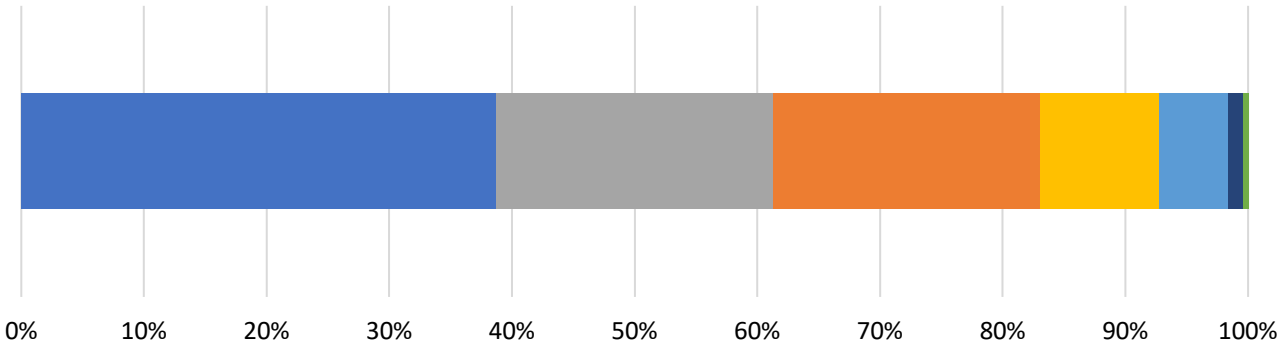


Children- MTG 5 (Growth & Obesity), n=339



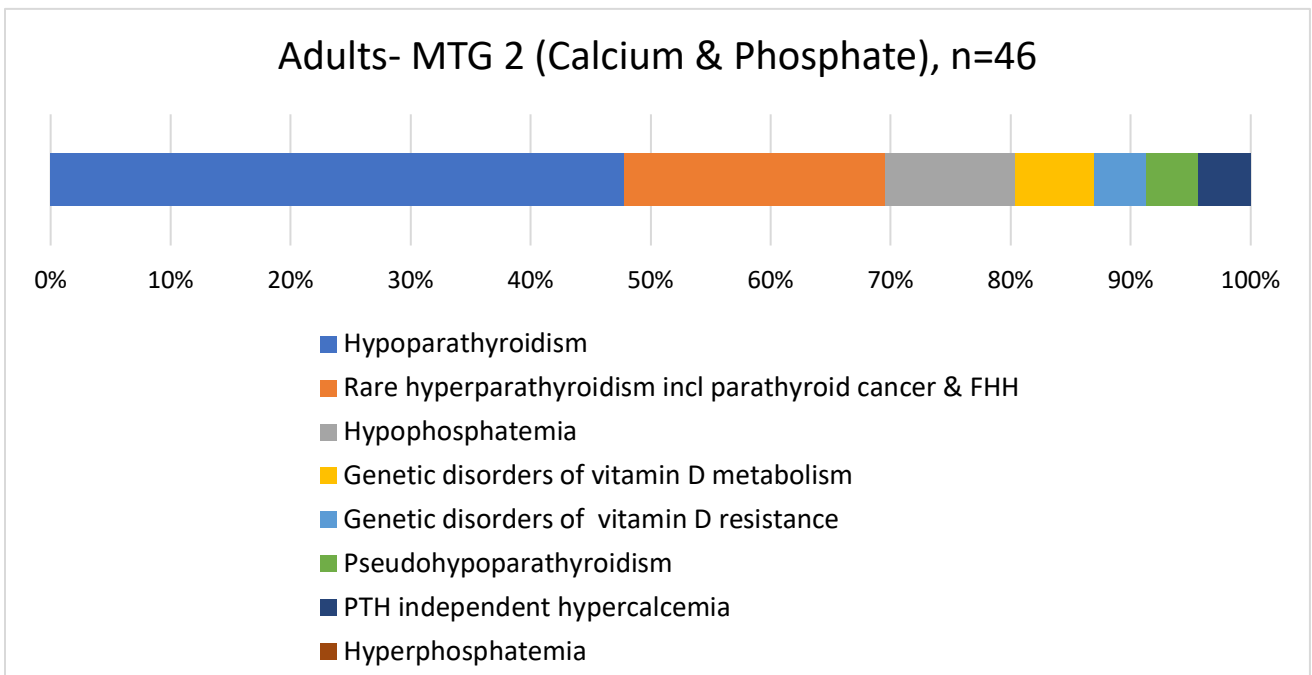
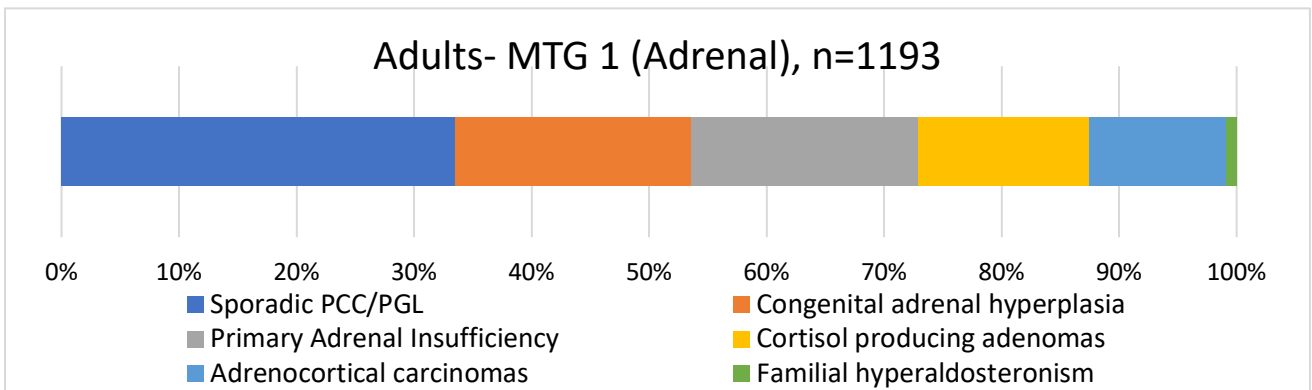


Children- Bone Dysplasia, n=244

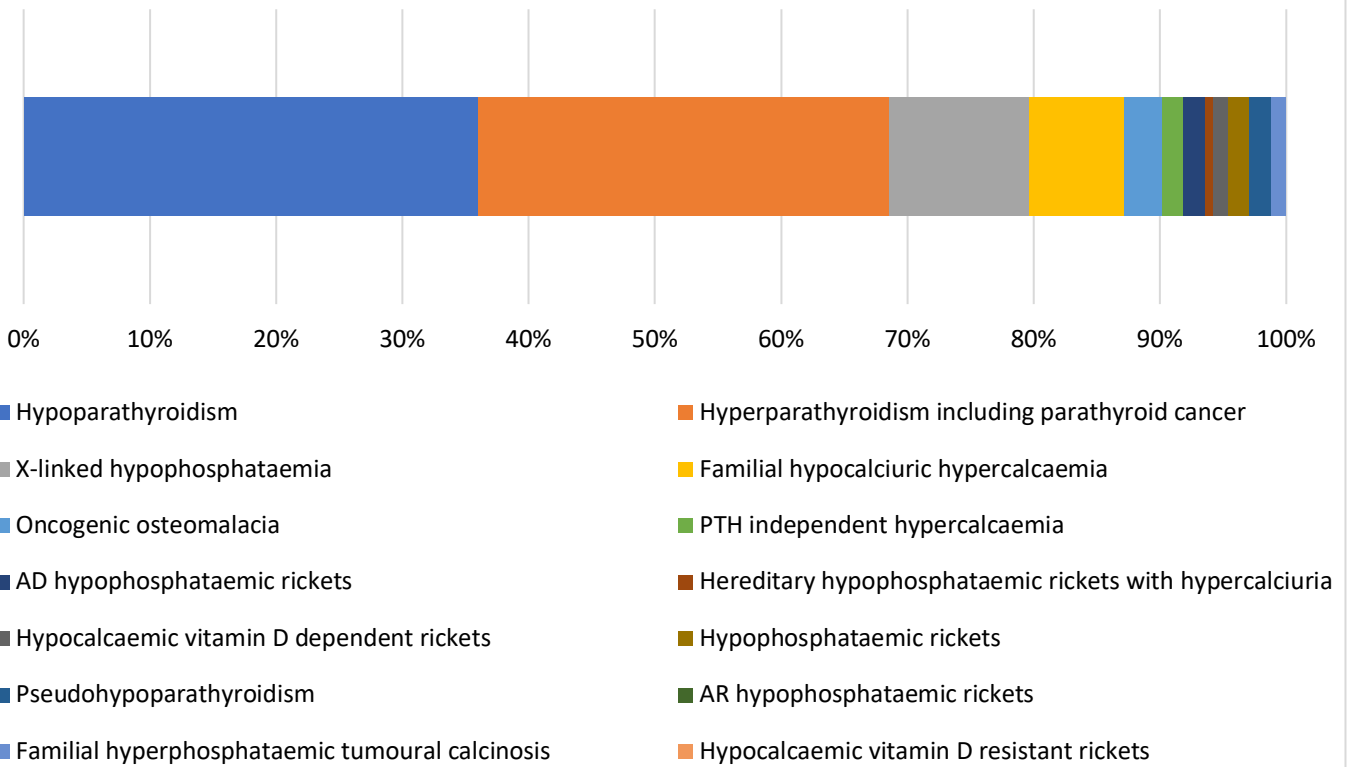


- Primary Bone Dysplasia with reduced bone density
- PBD with Micromelia
- PBD with disorganized bone development
- PBD with increased bone density
- Hypophosphatasia
- Dystosis
- PBD with Progressive Ossification of the Skin

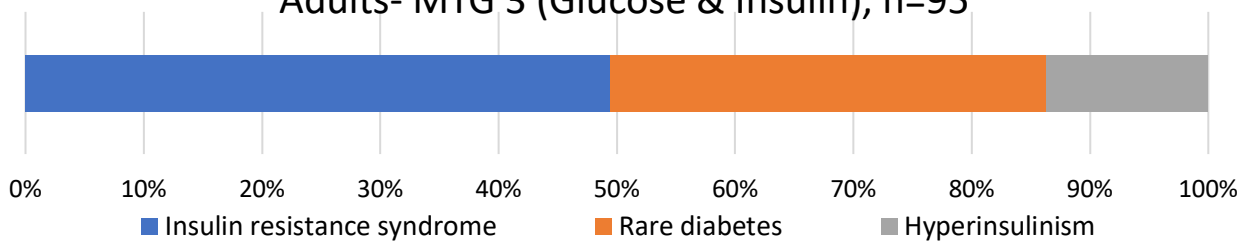
Fig.5b – Commonly reported conditions in adults between July 2018 and June 2021; cases, n.



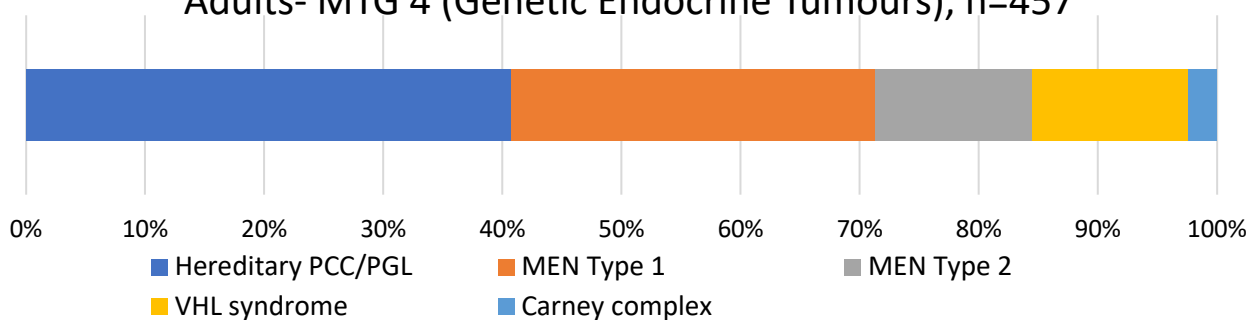
Adults- MTG 2 (Calcium & Phosphate revised categories Oct '19), n=172



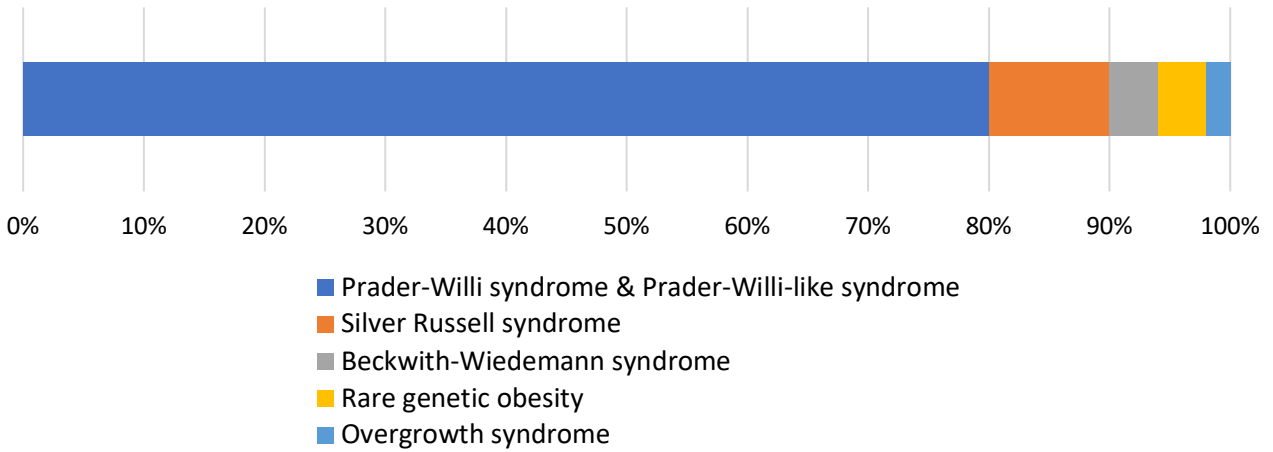
Adults- MTG 3 (Glucose & Insulin), n=95



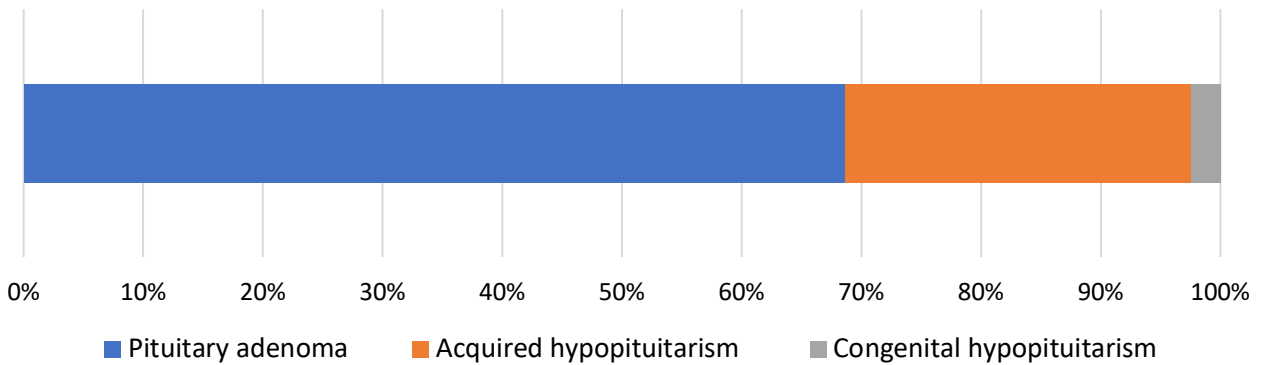
Adults- MTG 4 (Genetic Endocrine Tumours), n=457



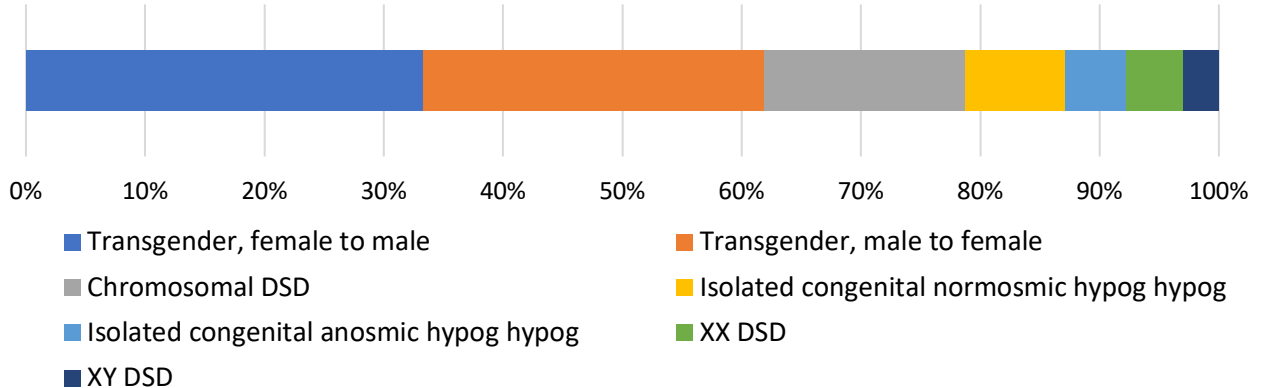
Adults- MTG 5 (Growth & Obesity), n=50

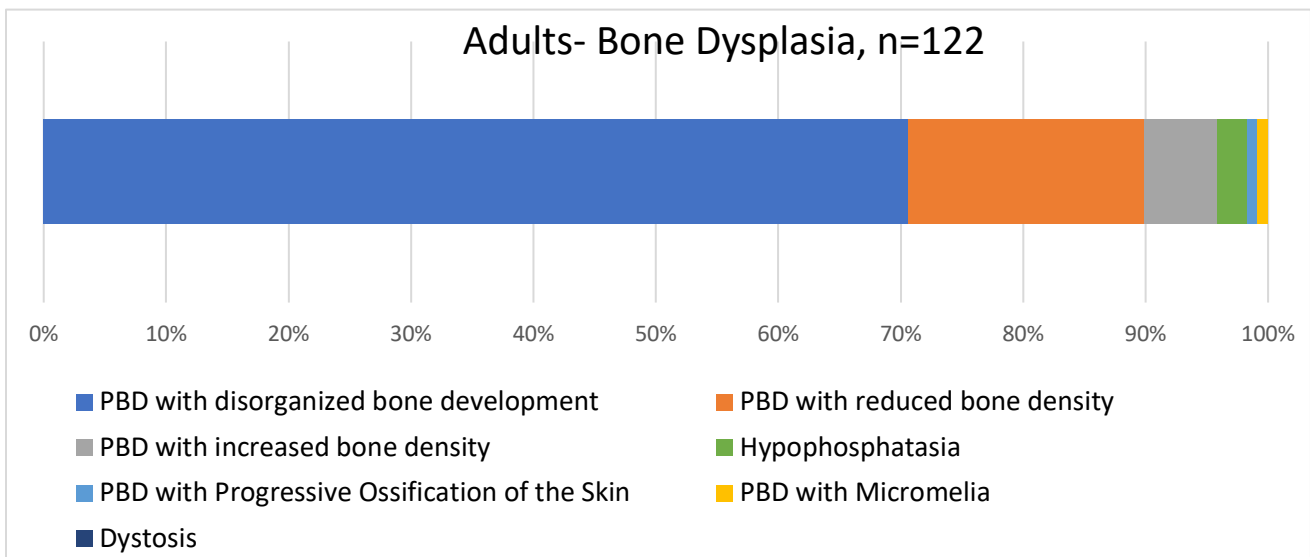
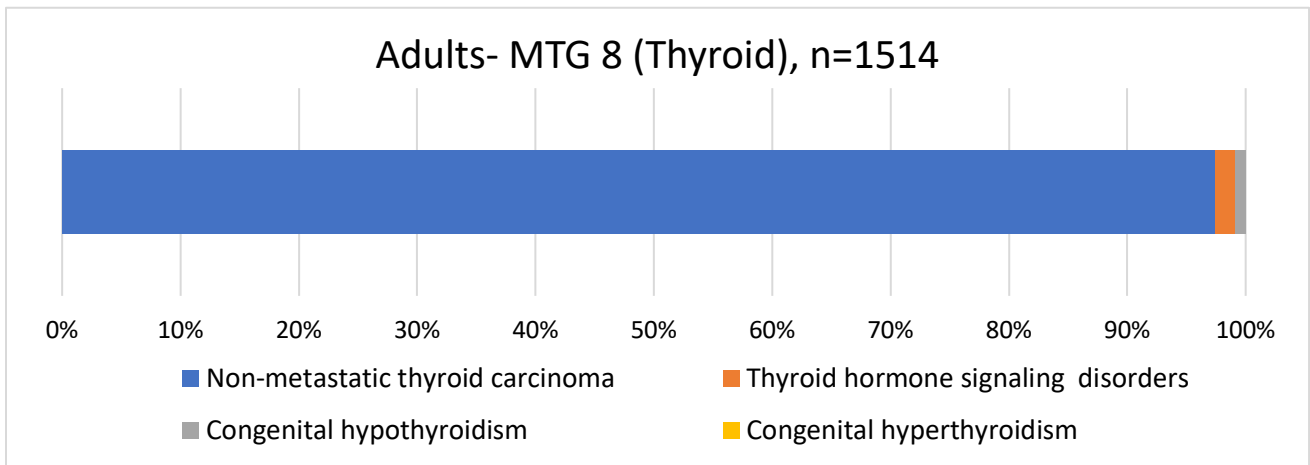


Adults- MTG 6 (Pituitary, n=3524)



Adults- MTG 7 (Sex Development), n=786





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 2021, 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.

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 EuRRECa Work Package 6
 October 2021