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. The e-REC platform also assisted 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 4.5-year period from July 2018 to December 2022 inclusive.

Methods

The e-REC platform issued invitations to clinicians who had registered to participate in e-REC from July 2018 to December 2022 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 2022, a total of 66 centres from 22 countries had reported on the e-REC. Of these 66 centres, 27 are in Endo-ERN only, 18 are in Endo-ERN and ERN-BOND, 4 are in ERN-BOND only and 17 are in no ERNs.

Country	Centers	Country	Centers
Austria	5	Israel	1
Belgium	7	Italy	12
Croatia	1	Latvia	1
Czechia	1	Lithuania	1
Egypt	1	Luxembourg	1
Estonia	1	Netherlands	7
Finland	1	Portugal	1
France	2	Romania	1
Germany	9	Spain	4
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 December 2022).

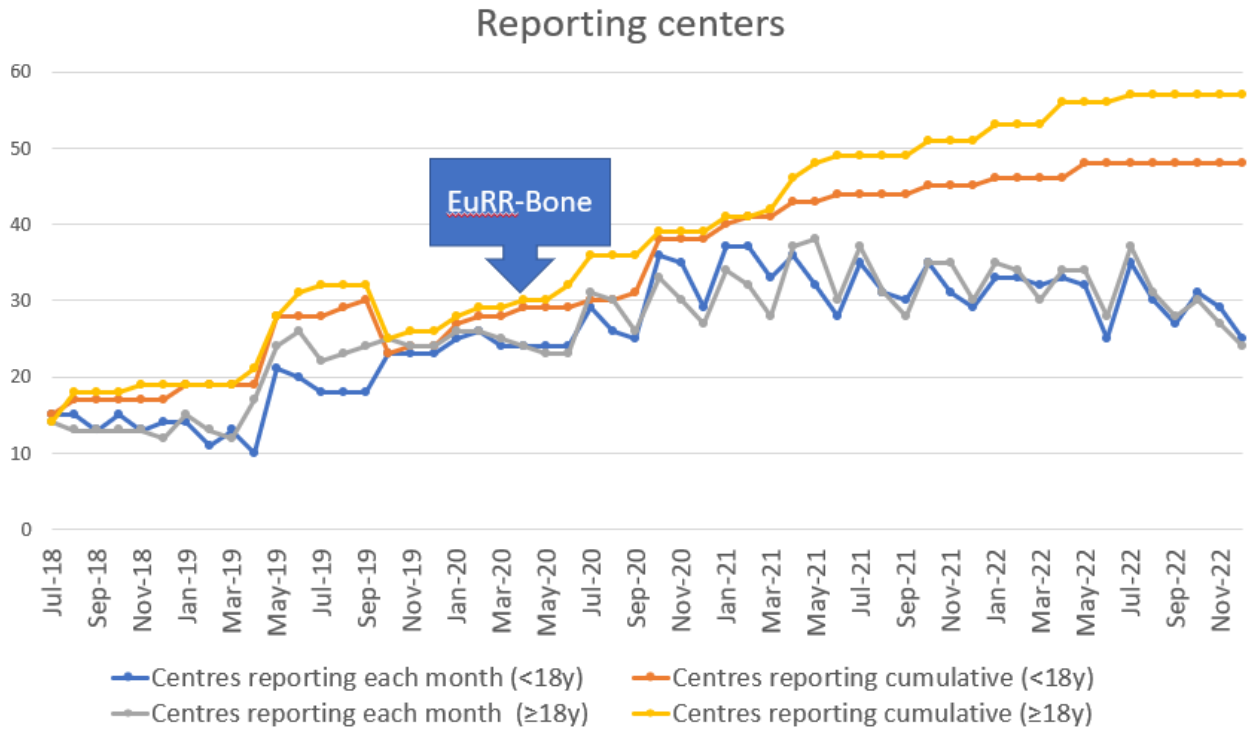


Fig.1 – Change in the number of centres that have been actively reporting between July 2018 and December 2022. Currently, 48 paediatric centres and 57 adult centres have actively participated in e-REC. The arrow indicates the launch of the EuRR-Bone project (April 2020).

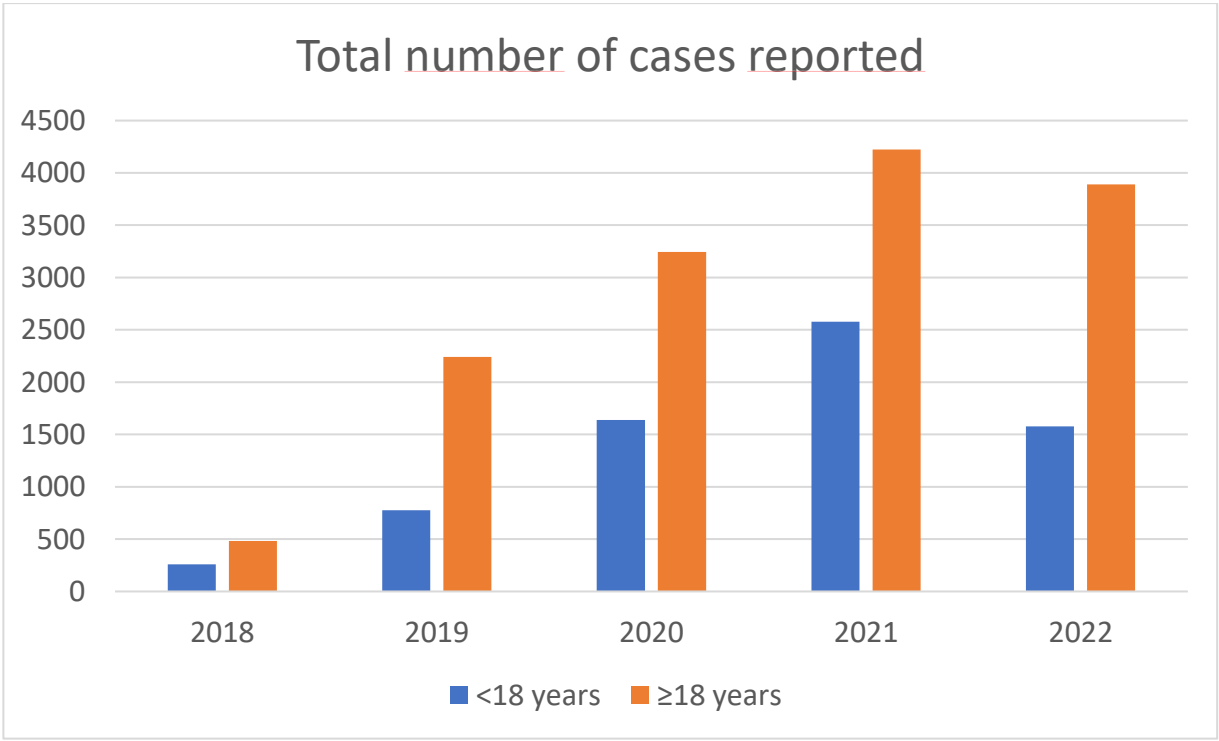
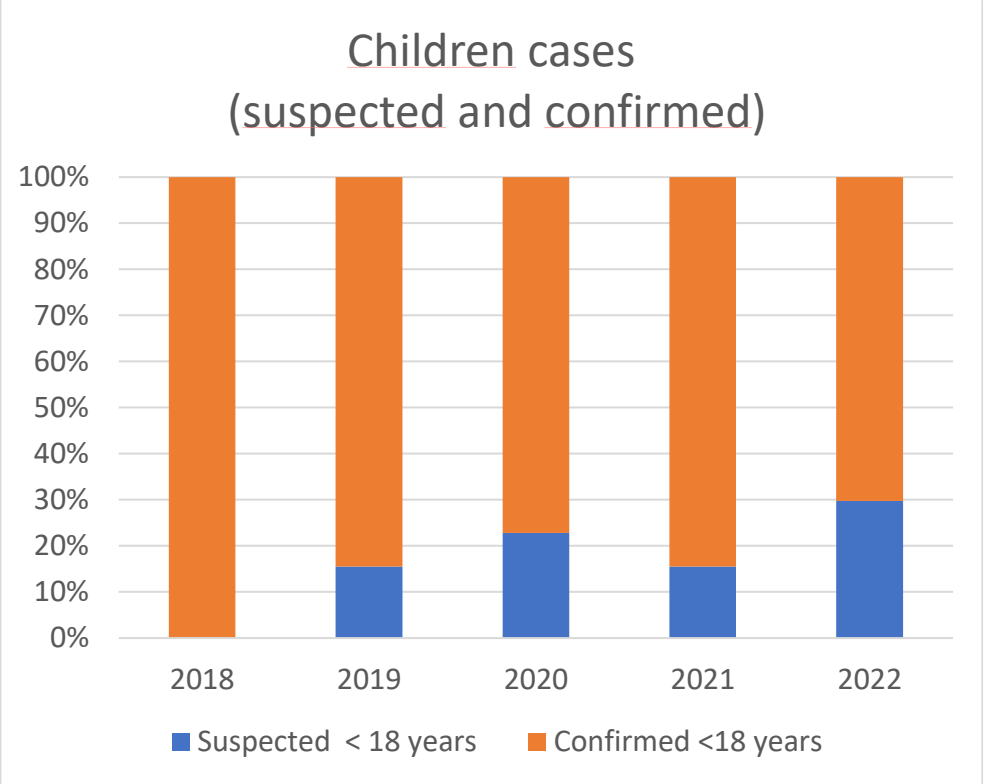


Fig. 2 – The change in cases reported between July 2018 and December 2022. A total of 14,100 new cases in adults and 5997 new cases in children have now been reported.



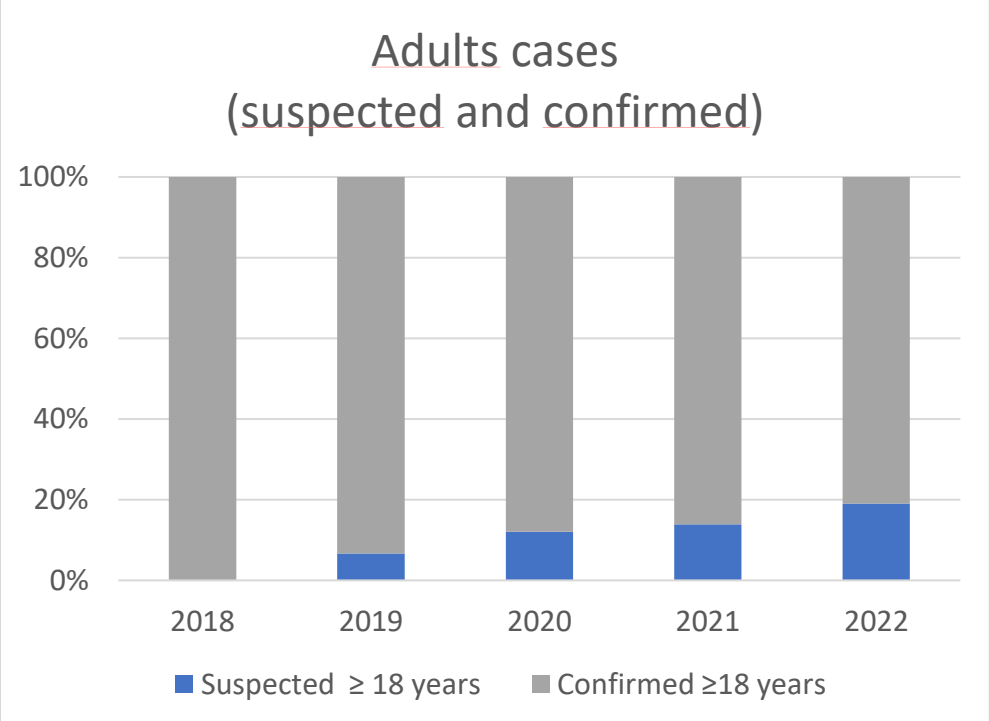
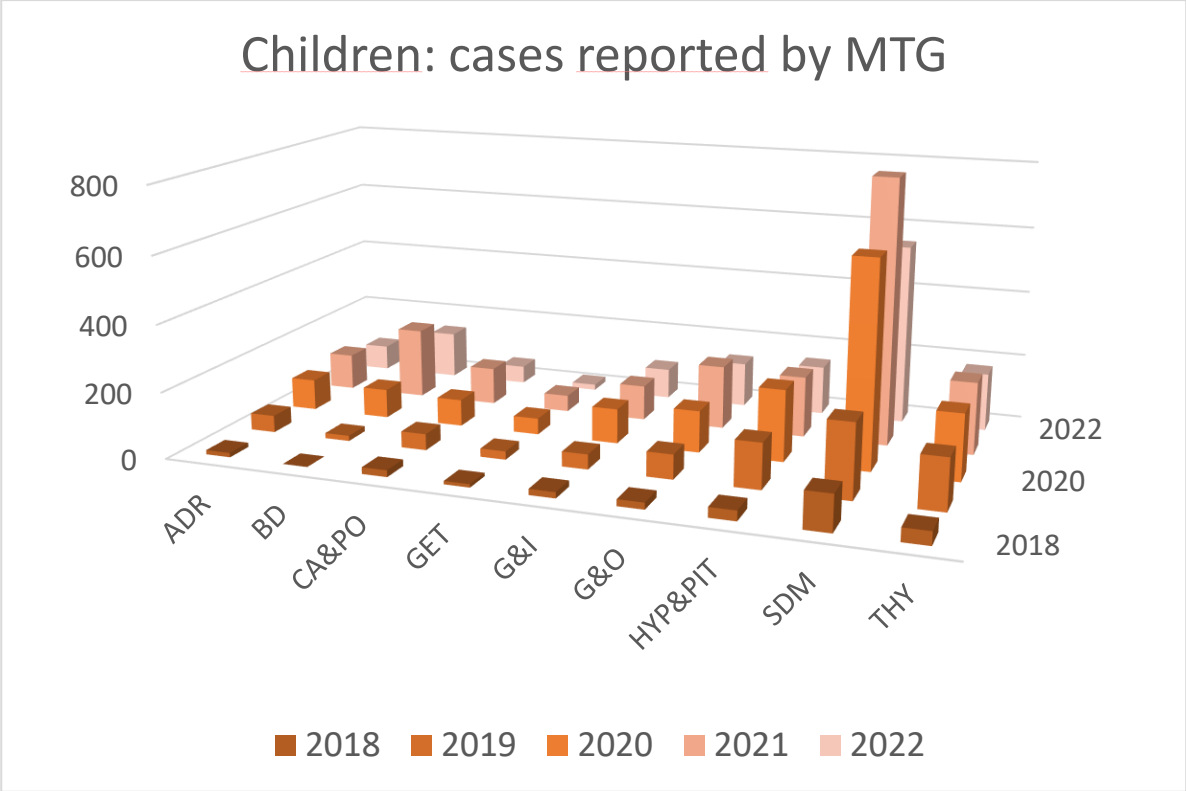


Fig 3. Since the last quarter of 2019, the platform collects information on the proportion of cases that are considered to be 'suspected' and the proportion that are considered to be 'confirmed'. Suspected cases (in blue) are shown as a percentage of total cases for children (orange) and adults (grey). The platform also allows updating of previously reported suspected cases to either confirmed or excluded.



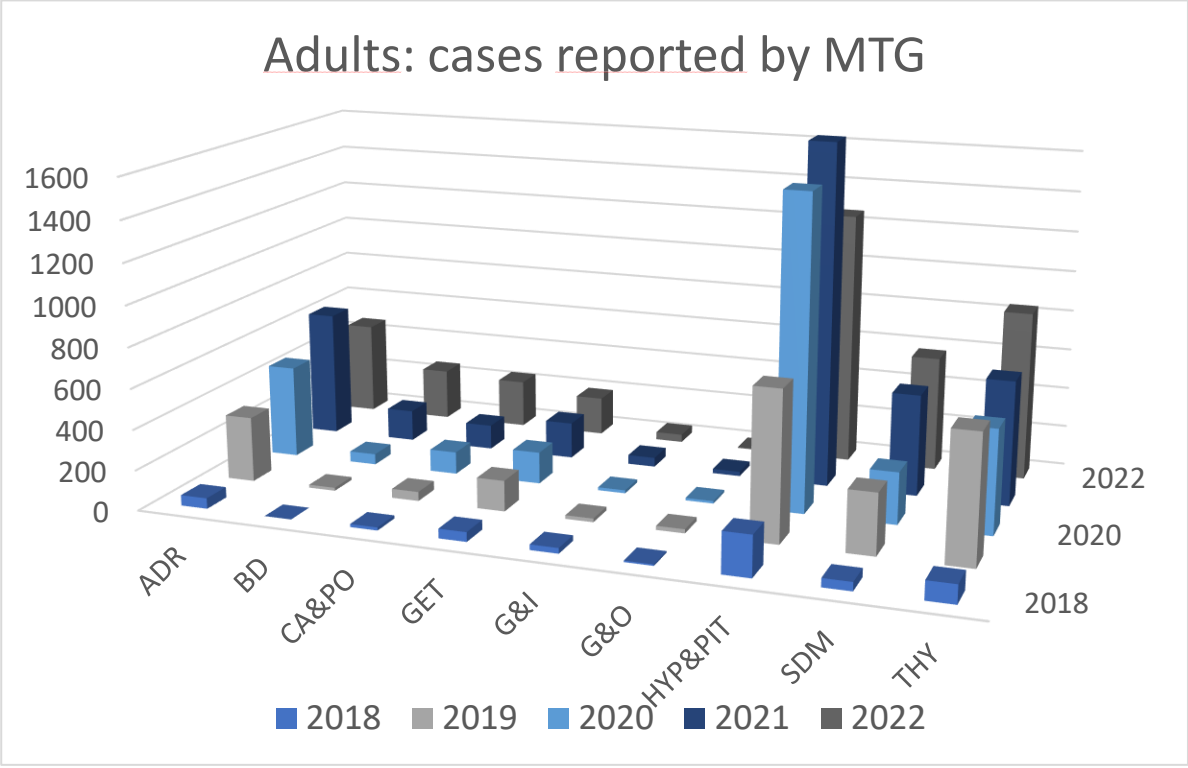


Fig.4 – Cases reported per Main Thematic Group (MTG) between July 2018 and December 2022. Conditions within the 'Sex Development' and 'Pituitary' condition groups were most commonly reported amongst children and adults, respectively. However, in general, the increase in reported conditions is across the majority of MTGs.

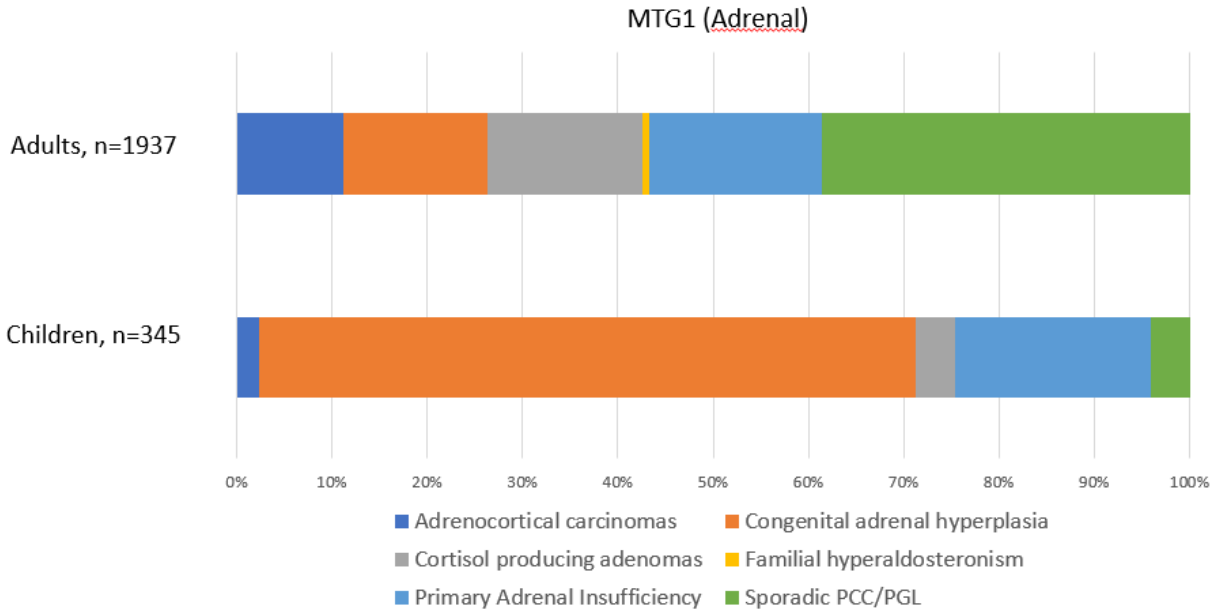


Fig.5 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG1 (adrenal).

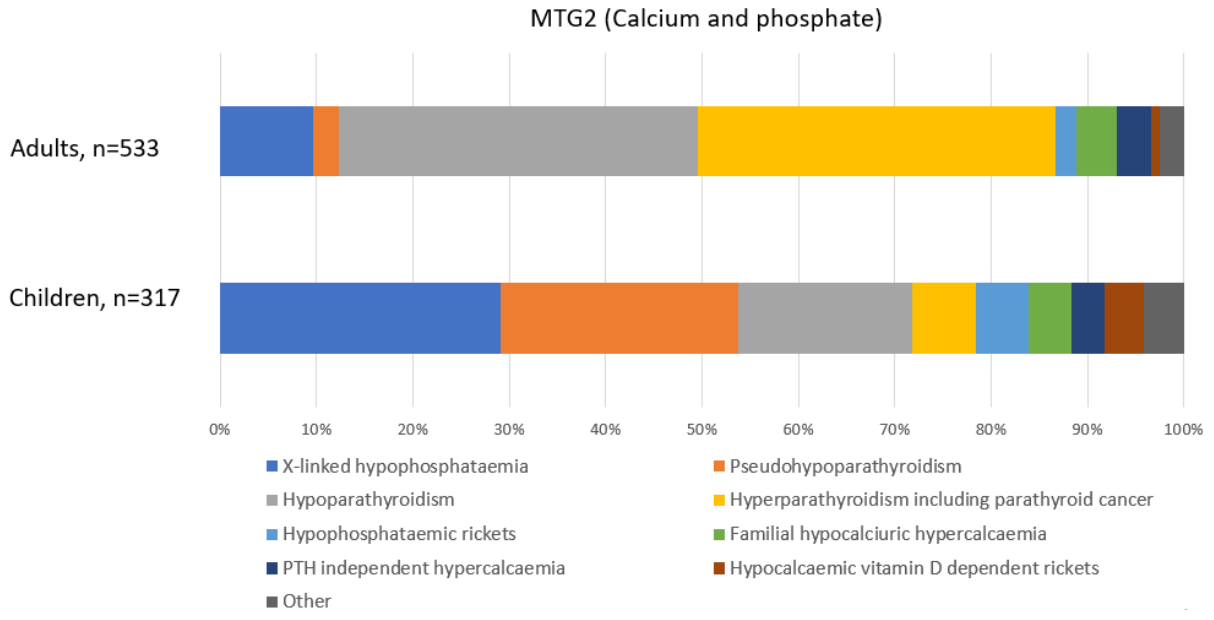


Fig.6 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG2 (Calcium and phosphate).

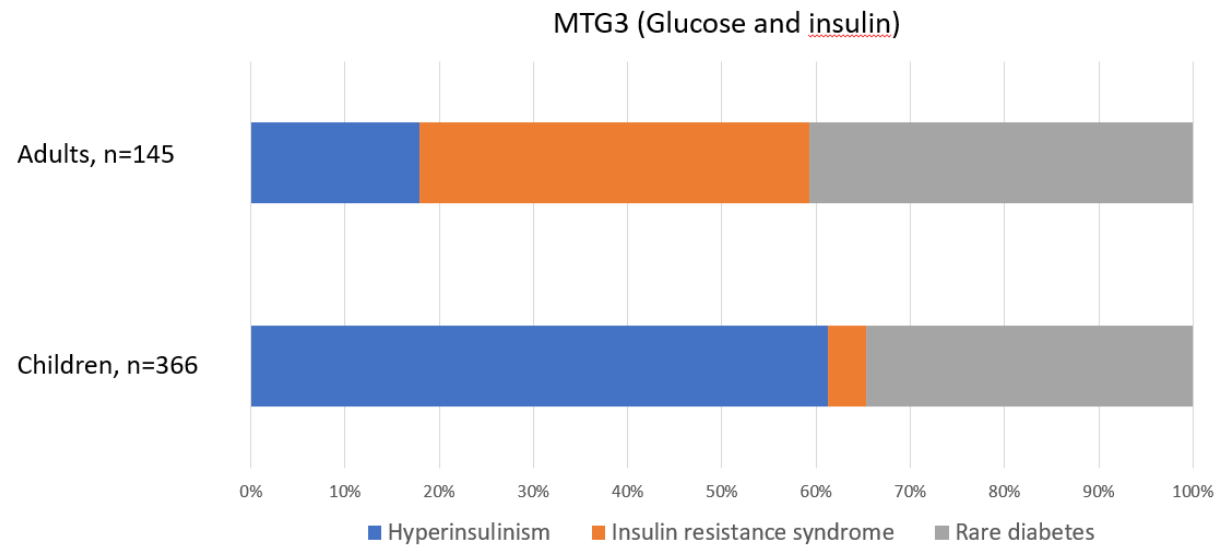


Fig.7 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG3 (glucose and insulin).

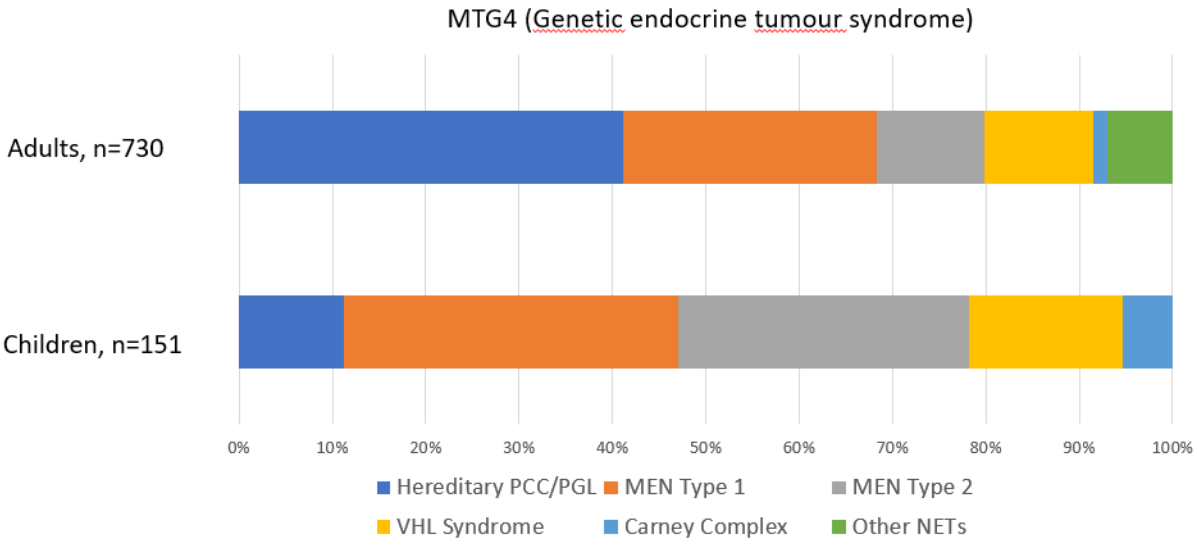


Fig.8 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG4 (Genetic endocrine tumour syndrome).

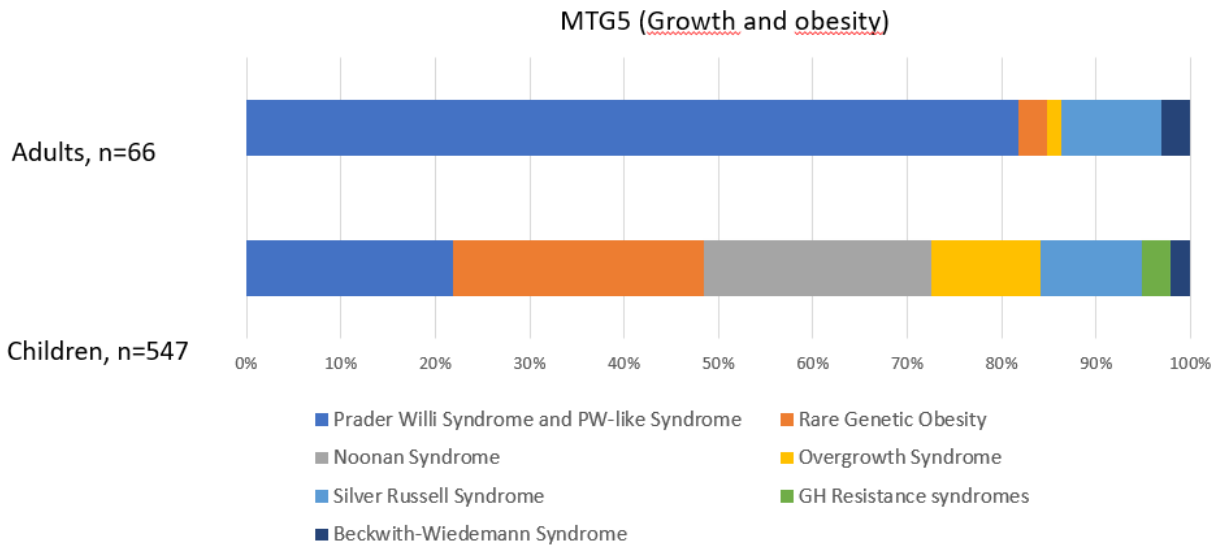


Fig.9 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG5 (Growth and obesity).

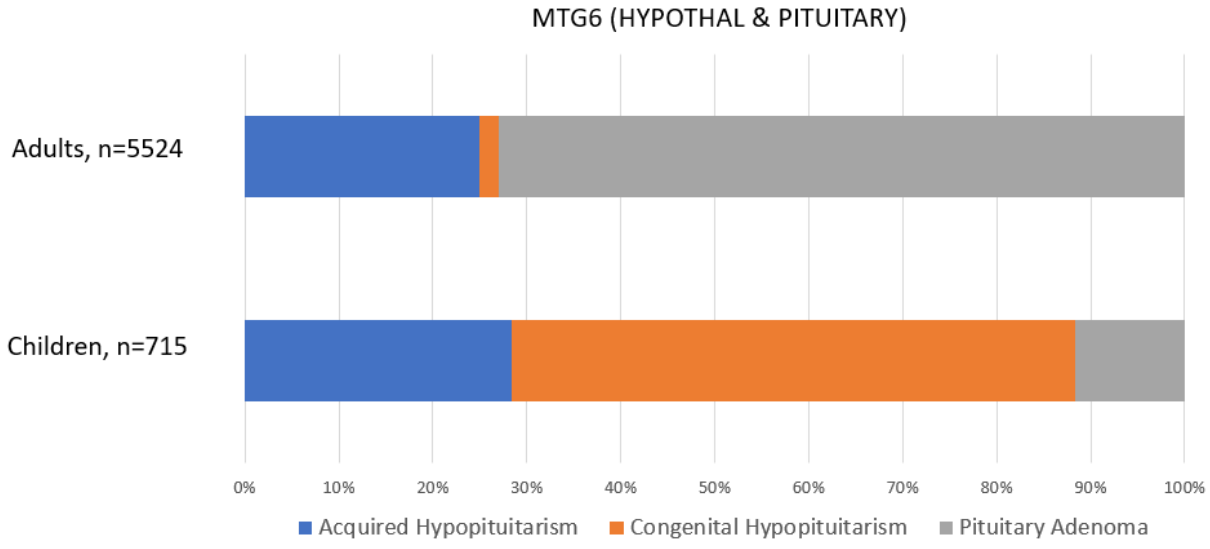


Fig.10 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG6 (Hypothalamus and pituitary).

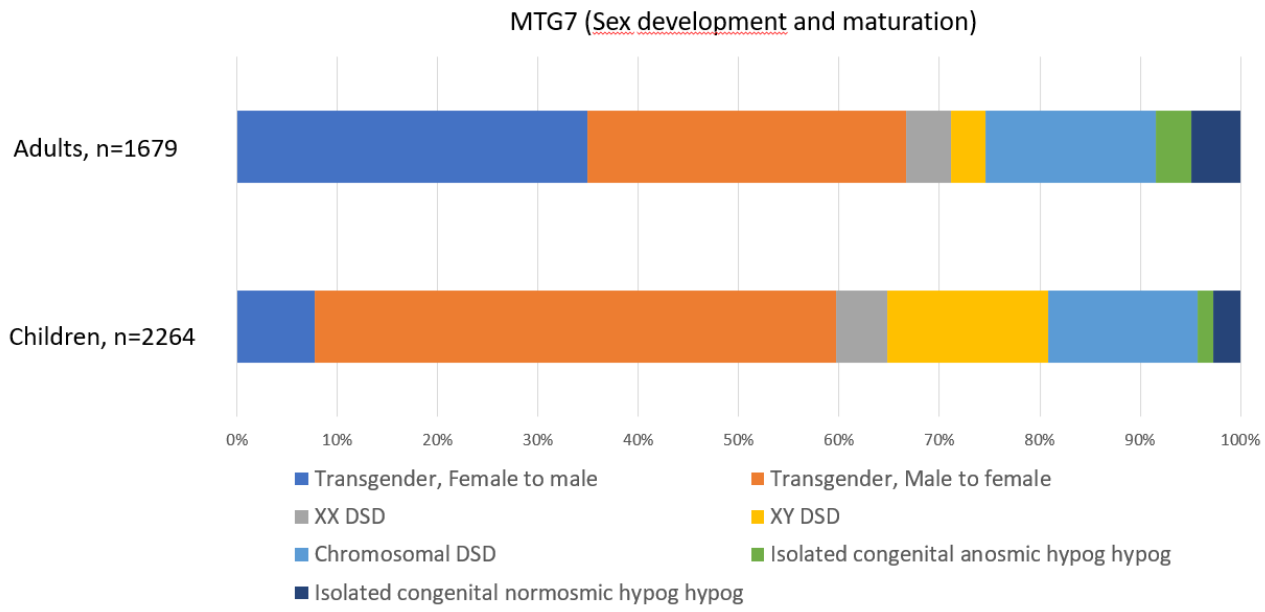


Fig.11 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG7 (Sex development and maturation).

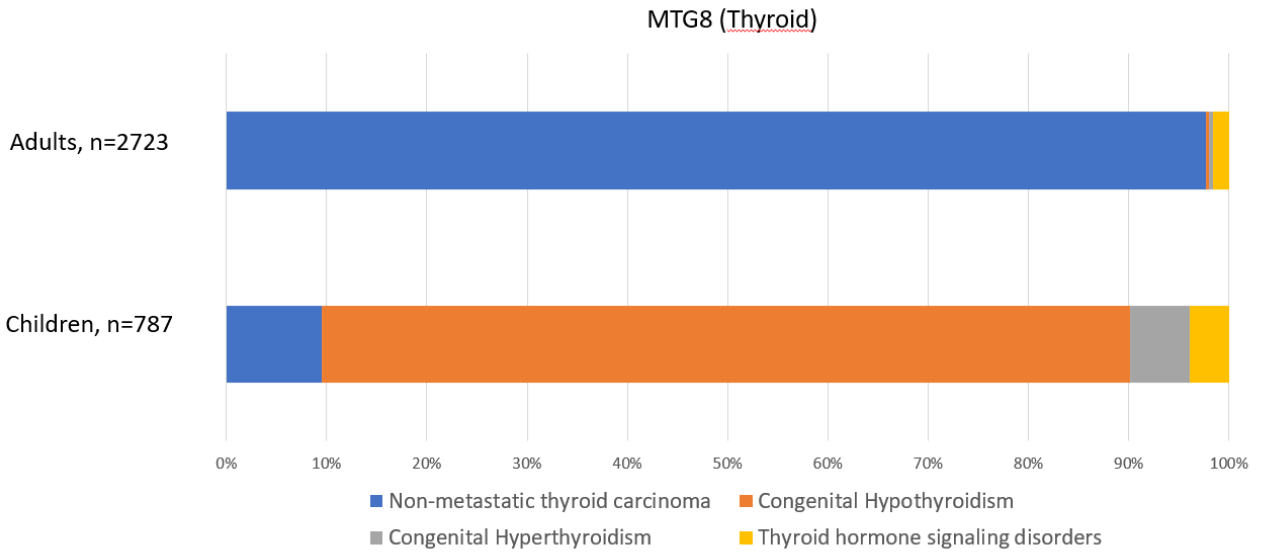


Fig.12 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG8 (Thyroid).

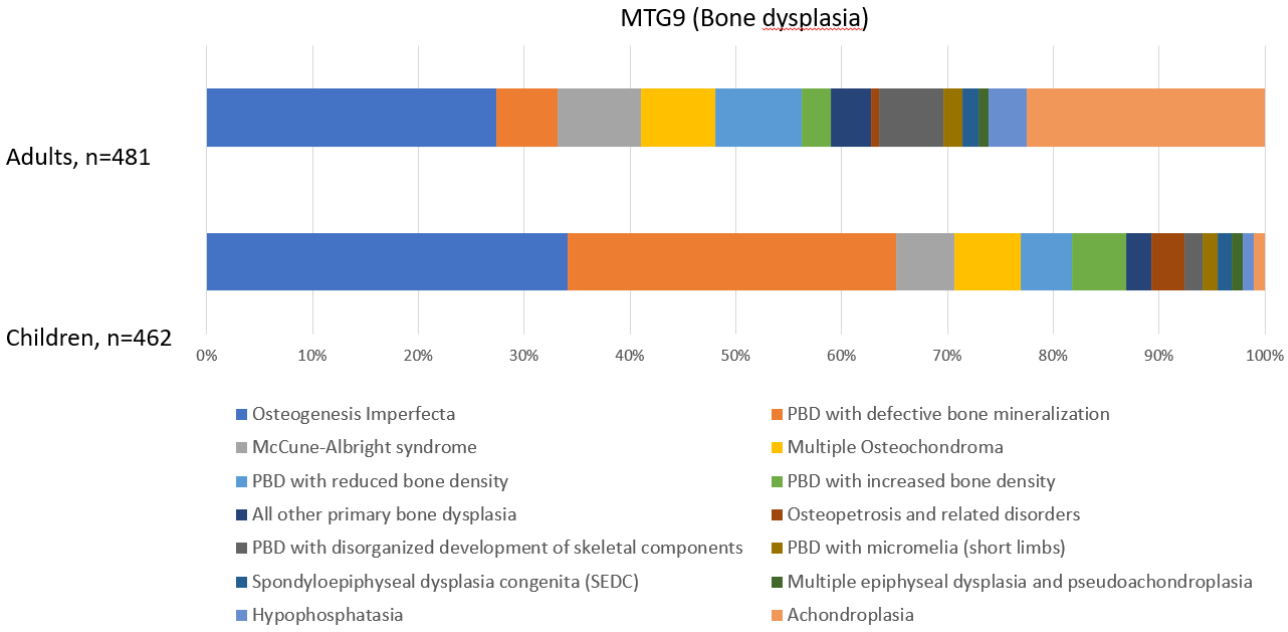


Fig.12 – Commonly reported conditions in children and adults between July 2018 and December 2022 within MTG9 (Bone dysplasia).

	<18 years					≥18 years				
Condition group	N of centers, reporting in this MTG	N of cases in this MTG	Median (cases)	Min	Max	N of centers, reporting in this MTG	N of cases in this MTG	Median (cases)	Min	Max
Adrenal	29	345	6	0	51	30	1937	34,5	0	457
Ca & Ph	35	317	4	0	64	32	533	6.5	0	85
Gluc & ins	26	366	5	0	101	22	145	0.5	0	35
Gen endo tumours	23	151	4	0	25	29	730	14.5	0	129
Growth & obesity	30	547	6	0	121	13	66	0.5	0	33
Hypothal & Pituitary	33	715	7.5	0	277	32	5524	54	0	1112
SDM	32	2264	11	0	1258	17	1679	26	0	382
Thyroid	30	787	8	0	192	26	2723	38.5	0	402
Bone dysplasia	28	462	12	0	53	22	481	6	0	202

Table 2 – Cases reported per centre per MTG between July 2018 and December 2022.

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.
- 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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