e-Reporting Of Rare Endocrine Conditions (e-REC)

Instructions

- The electronic survey that you will receive every month will enquire whether over the previous month you have encountered a **new** case of any of the Endo-ERN conditions.
- The survey contains one 'card' for each main thematic group. There are 2 surveys, one for the paediatric age group (<18yrs) and one for adults (≥18). You can complete as many cards as you want but, as a minimum, please complete those that you have signed up for and select 'Not Applicable' for those that you prefer not to complete.
- The whole survey will not take more than two minutes if you have the information to hand.
- The list of conditions are hyperlinked to the Orphacode online definitions (see below).
- It is advisable that, at each centre, there are either several clinicians who are nominated to report on specific conditions or there is one team member who is nominated to receive the survey(s) and reports on behalf of all the clinicians at the centre.
- On reporting a case, no identifiable or partially identifiable information will be transferred to the Office for Rare Conditions in Glasgow.
- The Office will provide the centre with unique IDs for the reported cases and these should be stored locally at the centre to link with the case, if required in the future.
- A separate exercise will be performed later to map existing cases.
- This survey is approved by the Information Governance Office of NHS Greater Glasgow & Clyde & the National Research Ethics Service of Scotland.
- For further information and to sign up, visit **eurreca.net/e-rec/** or e-mail **info@eurreca.net**

Stage 1
All participants will receive an email asking them to complete the survey around Day 15 in the month after the reporting month.

Stage 2
The survey contains reporting cards from MTG1 Adrenal to MTG8 Thyroid. If you have not signed up to report on any group of conditions (MTG), select 'Not Applicable' and submit and the next card will open. If you want to save and return later you must note the return code for the specific MTG card.

Stage 3
Participants will return the survey notifying of any cases seen in the reporting month or "nothing to report". A reminder e-mail will be issued 15 days and 30 days after the first email after which no more reminders will be sent.

Stage 4
Participants will be asked to keep a record of patients reported locally for quality control.

Stage 5
The EuRRECa team at the Office for Rare Conditions in Glasgow will issue unique IDs for each notified patient which should be stored in the local records.

Stage 6
All the data gathered will be shared with Endo-ERN for its official reports to the EU.

Please notify the Office for Rare Conditions of any changes to the conditions your centre reports on or any change in reporting staff.

Endo-ERN main thematic groups, conditions and their orphacodes

Main Thematic Group	Condition	Orphacode
MTG1 ADRENAL	Sporadic PCC/PGL	ORPHA276624
MTG1 ADRENAL	Cortisol producing adenomas	ORPHA443287
MTG1 ADRENAL	Adrenocortical carcinomas	ORPHA1501
MTG1 ADRENAL	Primary adrenal insufficiency	ORPHA101959
MTG1 ADRENAL	Congenital adrenal hyperplasia	ORPHA418
MTG1 ADRENAL	Familial hyperaldosteronism	ORPHA235936
MTG2 CALCIUM & PO	Rare form of hyperparathyroidism including parathyroid cancer and FHH	ORPHA181408
MTG2 CALCIUM & PO	PTH independent hypercalcemia	ORPHA300547
MTG2 CALCIUM & PO	Hypoparathyroidism	ORPHA181405
MTG2 CALCIUM & PO	Pseudohypoparathyroidism	ORPHA79445
MTG2 CALCIUM & PO	Hypophosphatemia	ORPHA89937
MTG2 CALCIUM & PO	Hyperphosphatemia	ORPHA306661
MTG2 CALCIUM & PO	Genetic disorders of vitamin D metabolism	ORPHA289157
MTG2 CALCIUM & PO	Genetic disorders of vitamin D resistance	ORPHA93160
MTG3 GLUC & INS	Rare diabetes	ORPHA101952
MTG3 GLUC & INS	Hyperinsulinism	ORPHA276525
MTG3 GLUC & INS	Insulin resistance syndrome	ORPHA181368
MTG4 GENETIC ENDO TUMOURS	MEN Type 1	ORPHA652
MTG4 GENETIC ENDO TUMOURS	MEN Type 2	ORPHA653
MTG4 GENETIC ENDO TUMOURS	Carney complex	ORPHA1359
MTG4 GENETIC ENDO TUMOURS	Hereditary PCC/PGL	ORPHA29072
MTG4 GENETIC ENDO TUMOURS	VHL syndrome	ORPHA892
MTG5 GROWTH	Silver Russell syndrome	ORPHA813
MTG5 GROWTH	Beckwith-Wiedemann Syndrome	ORPHA116
MTG5 GROWTH	Prader Willi syndrome and Prader Willi-like syndrome	ORPHA739
MTG5 GROWTH	Noonan Syndrome	ORPHA648
MTG5 GROWTH	GH resistance syndromes	ORPHA633
MTG5 GROWTH	Overgrowth syndrome	ORPHA93460
MTG5 GROWTH	Rare genetic obesity	ORPHA77828
MTG6 PITUITARY	Pituitary adenoma	ORPHA99408
MTG6 PITUITARY	Congenital hypopituitarism	ORPHA95494
MTG6 PITUITARY	Acquired hypopituitarism	ORPHA95502
MTG7 SEX DEV	Chromosomal DSD	ORPHA325546
MTG7 SEX DEV	XY DSD	ORPHA98085
MTG7 SEX DEV	XX DSD	ORPHA2982
MTG7 SEX DEV	Isolated congenital anosmic hypogonadotrophic hypogonadism	ORPHA478
MTG7 SEX DEV	Isolated congenital normosmic hypogonadotrophic Hypogonadism	ORPHA432
MTG7 SEX DEV	Transgender, male to female	ICD-10 F64
MTG7 SEX DEV	Transgender, female to male	ICD-10 F64
MTG8 THYROID	Thyroid hormone signaling disorders	ORPHA183631
MTG8 THYROID	Congenital hypothyroidism	ORPHA442
MTG8 THYROID	Congenital hyperthyroidism	ORPHA424
MTG8 THYROID	Non-metastatic thyroid carcinoma	ORPHA100088