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EuRRECa Registry: Genetic Obesity Survey Results

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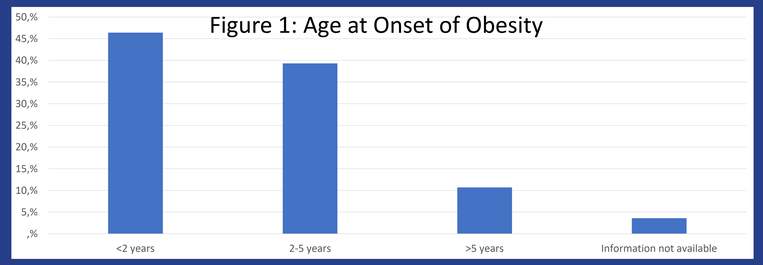
BACKGROUND

The diagnosis and treatment of patients with rare diseases is often difficult as most clinicians do not encounter them. Therefore, centralization and collaboration between centres of expertise is necessary. European Reference Networks (ERN's) such as RareEndoERN provide a platform for this, with one of its main thematic groups having a specific focus on rare growth and obesity disorders. Genetic obesity encompasses a heterogeneous group of conditions, classically divided into non-syndromic and syndromic obesity. These disorders are extremely rare and can be challenging to diagnose. More knowledge on the clinical presentation and the core features is needed. Using the EuRRECa's e-reporting platform (e-REC) which was established in July 2018, we aimed to assess routine clinical data in order to understand the diagnostic processes and clinical outcome of cases with Genetic Obesity.

METHODS

A survey was sent out to centres that registered patients with genetic obesity in the EuRRECa Registry. The questionnaire used a secure on-line tool (Webropol). All information provided was kept in compliance with the UK Data Protection Act (2018) and General Data Protection Regulation (GDPR 2016/679). No personally identifiable data were collected. Data on reporting centre, diagnosis, patient characteristics, comorbidities and treatment were collected.

Figure 1: Age at Onset of Obesity



RESULTS

Seven European centers registered 37 cases (35 children/2 Adults) of which 32 surveys were completed. Of these 37 cases, there were 3 Prader-Willi syndrome (PWS) and 1 pseudohypoparathyroidism (PHP) cases registered as part of the rare genetic obesity survey. 57 PWS and 44 PHP cases were also reported separately in the growth & obesity and calcium/phosphate thematic groups respectively.

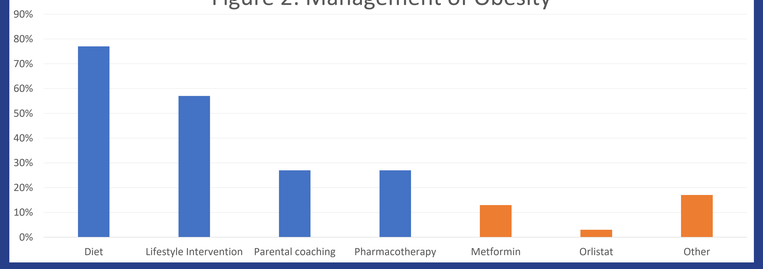
For our analysis, PWS and PHP cases were excluded. Diagnosis was confirmed in 78% (25/32) of which >50% were found to have an abnormality in a obesity gene (table 1). Median Age of onset was 2 years (14/29) (figure 1) with a median age at last review of 12yrs and a median BMI of +3.21 SDS

Reported clinical features included hyperphagia 67%, developmental delay 37%, endocrine dysfunction 33%; short stature 20% and tall stature 10%. Reported comorbidities were: Acanthosis nigricans 26%; dyslipidaemia 13%; hypertension 13%; diabetes mellitus 10%; sleep apnoea 10%; non-alcoholic liver disease 3%. Reported treatment included diet 77%; Lifestyle Intervention 55%; Parental coaching 26%; pharmacotherapy 26%; Metformin 3%; Orlistat 3% (figure 2). Other medications which were mentioned included setmelanotide; none of the patients had bariatric surgery.

Obesity Gene	N 13 (%)	BMI SDS at presentation Median (range)	BMI SDS at last review Median (range)
Leptin receptor deficiency	4(30.8)	4.7(4.4-6.6) N=3	4.4 (3.8-5.3) N=4
Melanocortin 4 receptor deficiency	5(38.5)	4.3 (2.8-4.4) N=4	4.4 (3.7-4.5) N=4
Prohormone convertase 1 deficiency	1(7.7)	3.6 N=1	3.6 N=1
Pro-opiomelanocortin deficiency	3(23.1)	2.37 N=1	3.9(3.3-4.0) N=3

Country	No of cases reported in e-REC (<18years)
Netherlands	18
UK	9
Belgium	3
Italy	2
Estonia	2
Czech Republic	1

Figure 2: Management of Obesity



CONCLUSION

Using surveys through registries such as EuRRECa can enable clinicians to collaborate and collect data on genotype/phenotype and clinical outcomes in rare conditions, aiding in the development of management strategies, including clinical trials. Genetic causes of obesity remain rare or often not identified and this form of collaboration between centres across Europe will ultimately improve patient care

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