The MC4R pathway, which is a component of the central melanocortin pathway, plays a vital role in regulating appetite and energy balance, and mutations in this pathway can result in rare genetic disorders of obesity.

References:

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Acknowledgments:

- This registry captures data entered by the patient; caregiver; and treating, care providers, and the healthcare system
- The Registry Coordinating Center is responsible for obtaining written consent and are informed that they have the right to
- The TEMPO registry enrolls patients with rare genetic disorders of obesity characterized by dysfunction of the melanocortin-4 receptor signaling pathway, a component of the central melanocortin pathway, and

Study Design and Methodology

The TEMPO registry is a voluntary, prospective, open-ended registry of individuals with rare genetic disorders of obesity in which the MC4R pathway is implicated in early-onset severe obesity and insatiable/excessive hunger

This registry captures data entered by the patient, caregiver, and

Electronic surveys are HIPAA (Health Insurance Portability and Accountability Act of 1996) compliant

Eligible participants are referred to the TEMPO study in either of the following ways:
- Referral by healthcare provider to coordinating centers
- Inclusion after positive identification through an industry-sponsored genetic screening study

Key Inclusion Criteria

- Patients aged ≥2 years with the following criteria are eligible for inclusion:
  - Body mass index (BMI) >14 times that of the age- and sex-adjusted 95th percentile value (in patients aged 2 to 17 years)
  - BMI >40 kg/m² (in patients aged ≥18 years)
- Patients are eligible if homozygous, compound heterozygous, heterozygous, or composite heterozygous mutations upstream or downstream of the MC4R are detected (Figure 1)

Key Exclusion Criteria

- Patients with syndromic disorders of obesity such as Bardet-Biedl syndrome
- Individuals identified with mutations consistent with Prader-Willi syndrome, Bardet-Biedl syndrome, or Alström syndrome are referred to existing registries

Enrollment

- Following enrollment, baseline electronic surveys are completed
- Patients and caregivers complete the baseline healthcare provider survey tool, reporting patient baseline demographics and disease characteristics
- Patients and caregivers complete their respective baseline survey tools, answering questions on the burden of disease

Baseline surveys collect the following information:

- Demographics
- Medical history
- Pedigree of obesity history
- Genetic testing
- Symptoms and complaints present
- Resource utilization
- Development and education
- Social and emotional impact

Annual Follow-ups

- Enrolled patients, caregivers, and healthcare providers are contacted annually by the Registry Coordinating Center to complete an online follow-up survey, which includes a smaller subset of questions from the baseline survey
- The period of follow-up is open ended