

The Psychological Effects of Metabolic Crisis on Patients who Suffer from Fatty-Acid Oxidation Disorders and Their Families



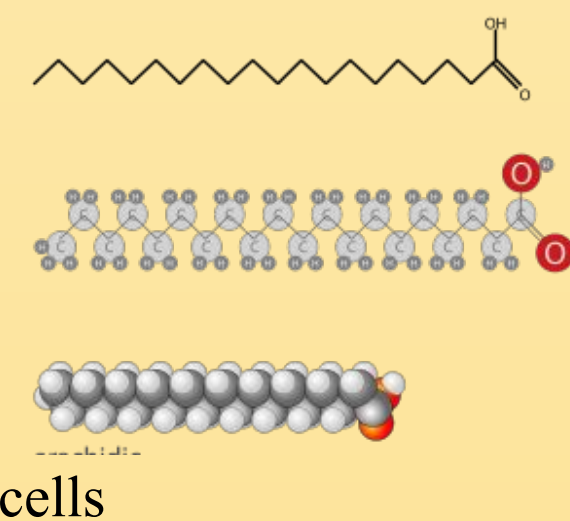
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Abstract

Fatty acid oxidation disorders (FOD) are classified as genetic mitochondrial disorders that can cause morbidity and mortality in infants. Surveys analyzed the everyday life of individuals with FODs in the online community. Results found a significant correlation between the medical, physical, and psychological effects in individuals and families who suffer from fatty-acid oxidation disorders.

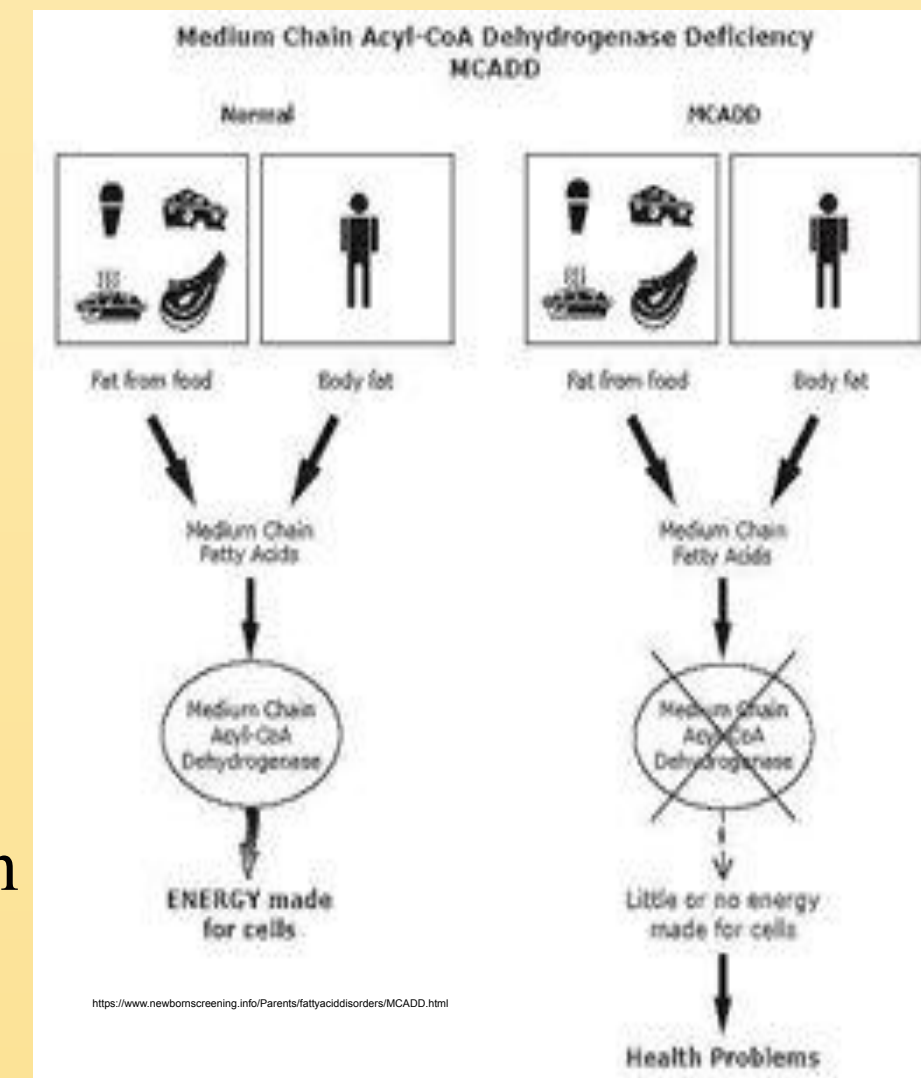
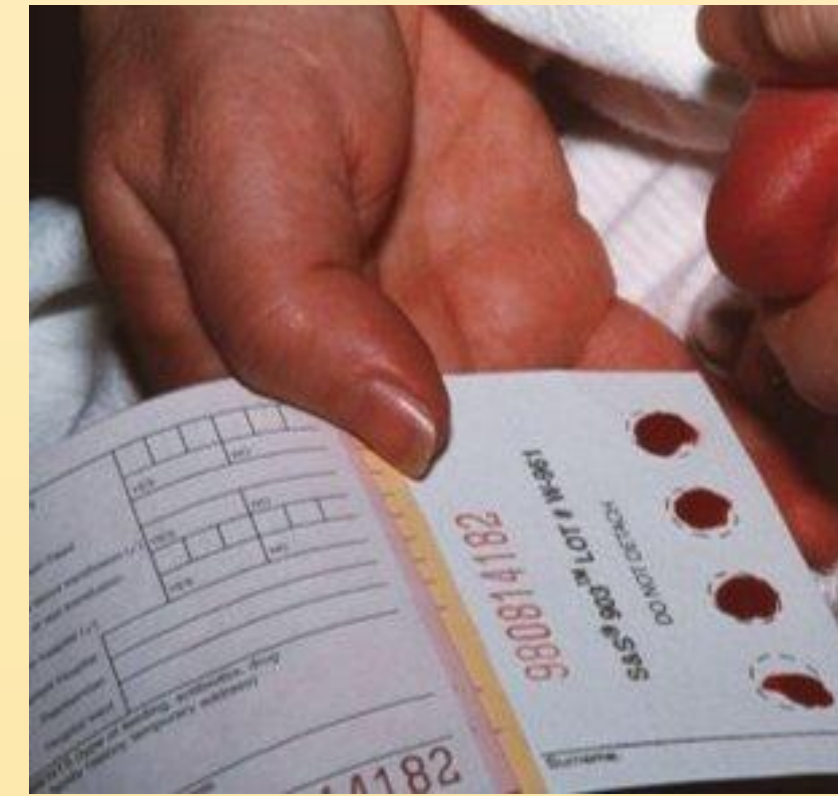
Introduction

- Metabolism: the series of chemical changes within the body that result in the creation of energy
 - 3 Steps of Metabolism
 - Ingestion
 - Digestion
 - Production
- Creation of ATP allows:
 - Cellular Signal Transduction
 - Other Metabolic Processes
 - Aids in major cellular functions
- Fatty-Acids
 - Most Basic Form of Fat
 - Aids in Brain Function & Growth
 - Metabolized by mitochondria of liver cells
- Metabolism Produces:
 - Ketones
 - ATP
 - CO₂
- Newborn Screening System (NBS)
 - Series of diagnostic tests to analyze inherited genetic disorders of infants (12)
 - MCADD determined by elevated C8 levels in the bloodstream
- Fatty-Acid Oxidation Disorders
 - Inherited Genetic Disorders



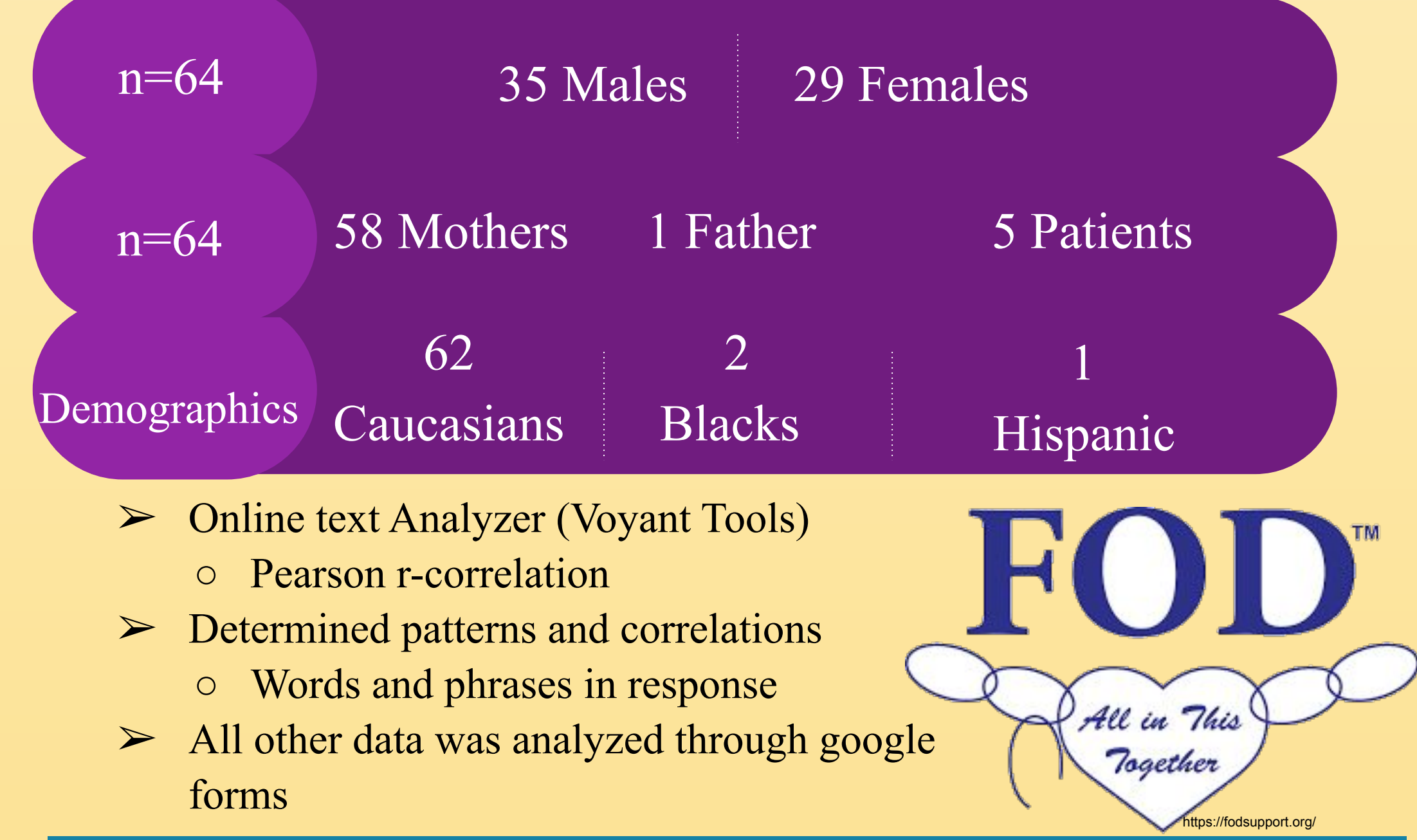
Literature Review

- Parents experienced anxieties during genetic screening due to pretest expectations
 - Marteau et al. 1998
- Mothers and Fathers presented to be equally stressed
 - Waisbren et al. 2003
- Caregiver may experience psychological distress, psychiatric illness, physical illness,
 - Nurs et al 2009
- Increased awareness of FOD, particularly in emergency treatments
 - Piercy et al. 2017
- Despite earlier diagnosis, sudden deaths were not avoided
 - Janeiro et al. 2019



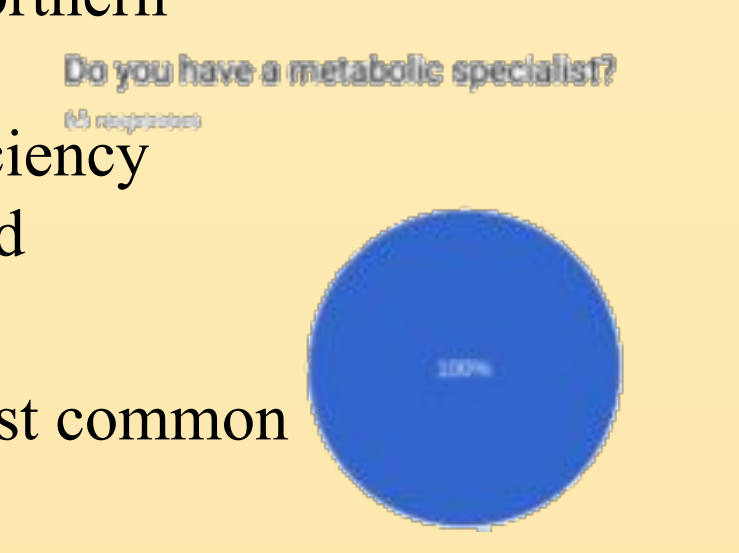
Methodology

- Participants were selected through Facebook support group
- Contacted group administrator for authorization
- Gave brief overview of experimental goals
- Participants emailed me directly if interested or with any follow-up questions
 - Participant must have an FOD or be related and be direct care of someone with an FOD
 - Given until the September 2018 to complete survey



Discussion

- Data supported all hypotheses
 - 95% of participants were caucasian
 - According to the National Organization for Rare Diseases (NORD), MCADD is most prevalent in people of northern Europe⁽⁴⁾
 - 68% of participants had MCAD deficiency
 - According to the New England Consortium of Metabolic Programs, MCADD is the most common metabolic disorders⁽¹⁰⁾
- All Participants had a metabolic specialist
 - The rarity of these genetic disorders cause an increased monitoring by a specialist



Conclusion

- Data supported all hypothesis
- This research directly influences the level of care that individuals with fatty-acid oxidation disorders receive during a time of metabolic crisis
- Through future research in addition to this research, more accurate genetic screens and medical treatment can be improved

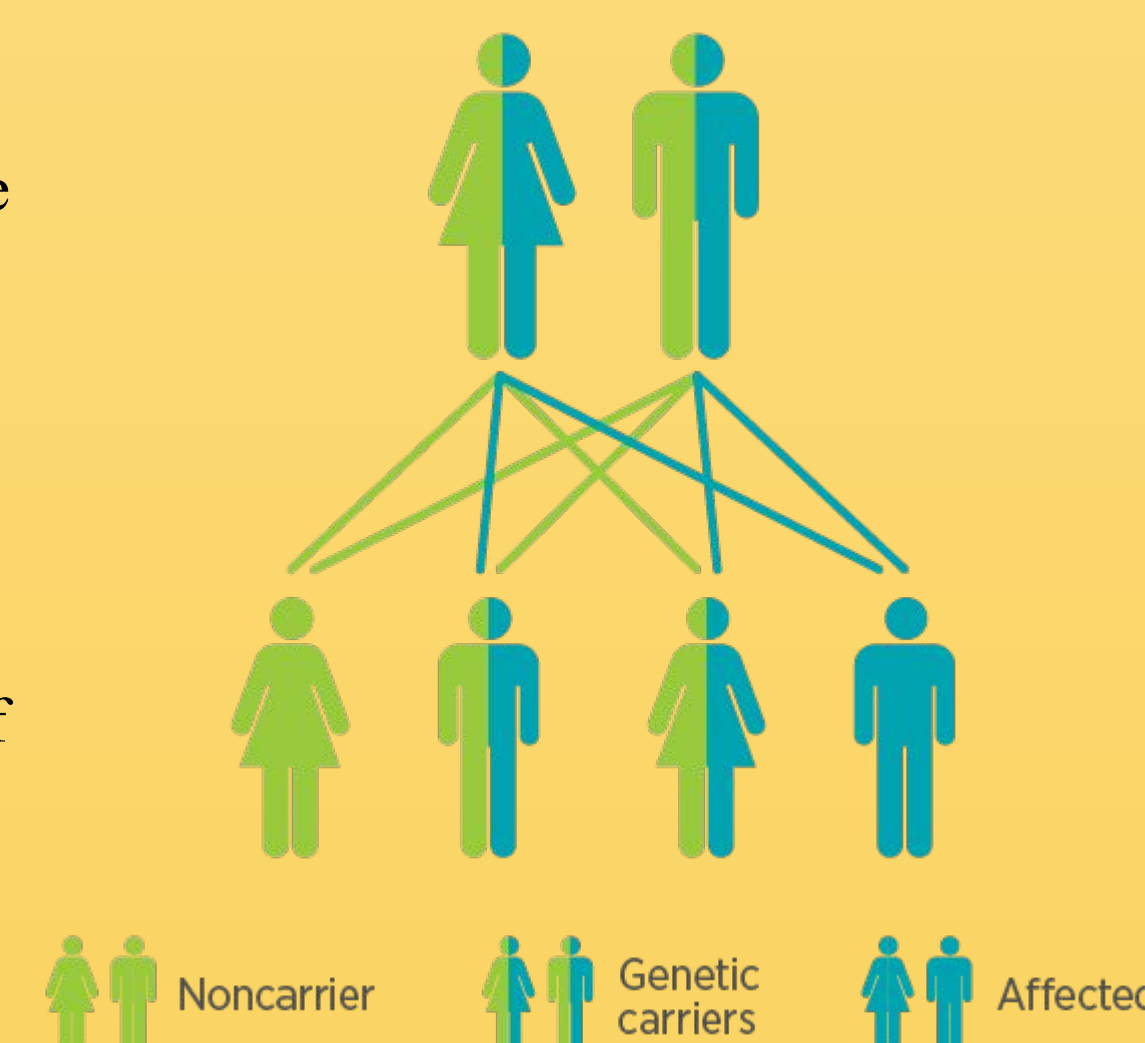


Gap in the Research



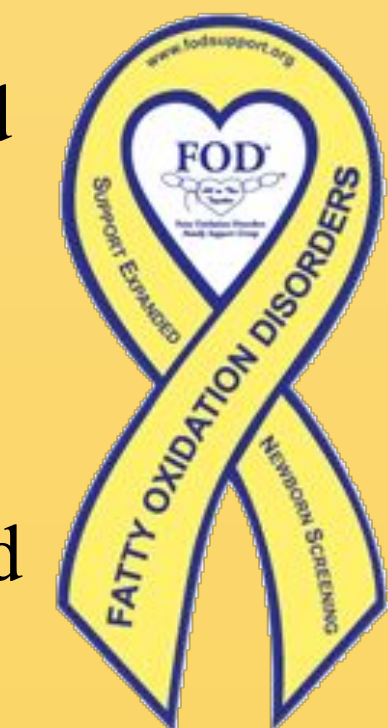
Literature Review

- Non-Carriers reported a general sense of well-being and relief in regards to future childbearing
 - Williams et al. 1997
- Carriers reported feelings of hopelessness concerning the health of their offspring
 - Williams et al. 1997



Goal of Research

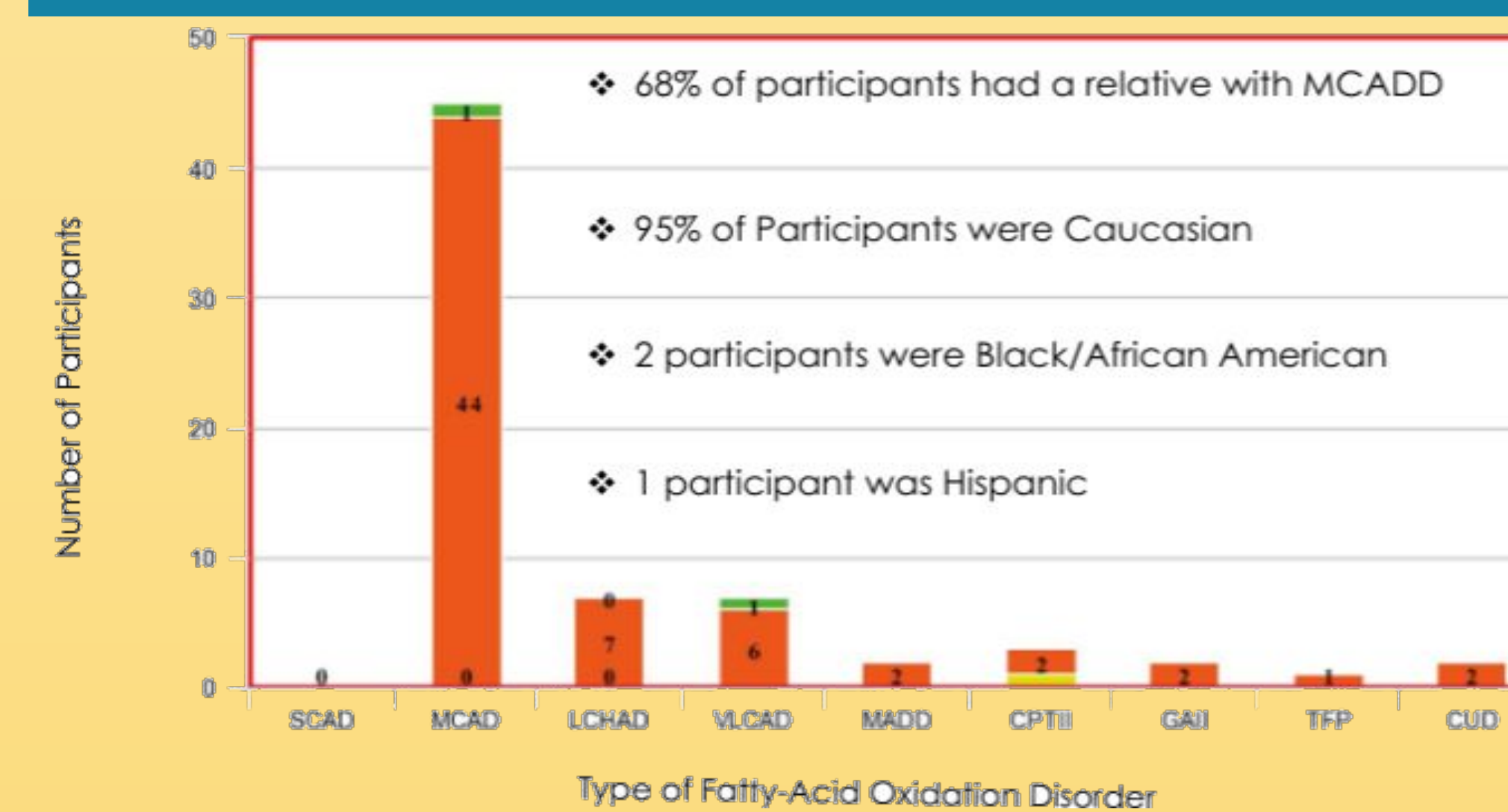
To acquire information about parents, families and effected individuals with fatty-acid oxidation disorders involving treatment history and medical accommodations



Hypotheses

- Most patients will be caucasian
- Most patients will have MCADD
- Most will have a metabolic specialist
- There will be similarities between patients in regards to emergency medical situations

Results



- Participants were questioned about symptoms during times of illness
 - 73.8 % Lethargy
 - 35.4% Weakness in Muscles
 - 30.8% Brain Fog
 - 26.4% Confusion
- Age of diagnosis was also taken into consideration
 - 87% diagnosed during infancy through the NBS
 - 5% diagnosed during childhood
 - 5% diagnosed during adolescents
 - 3% diagnosed during adulthood

Term 1	Term 2	Correlation (r)	Significance (p)
Hospitalization	Infection	0.93700426	0.00006367539
Hospitalization	Read	0.93700426	0.00006367539
Hospitalization	Treatment	0.93700426	0.00006367539
Hospitalization	Visits	0.93700426	0.00006367539
Ill	Illnesses	0.93700426	0.00006367539

Future Research

- Limitations
 - Survey Sample Size
 - Voluntary Survey Bias
- Future Research
 - Administer secondary survey regarding key life events
 - Reconnect with survey participants about residing state or country

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- My Science Research Peers



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