

Assessing patient empowerment in caregivers of patients with autism spectrum disorder

by

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Background: Autism spectrum disorder (ASD) is a neurodevelopmental disorder commonly referred to many genetics' clinics. When considering genetic counselors' role in the diagnostic pathway of this specific patient population, it is important to consider the point at which a genetic counselor is involved and the outcomes of genetic counseling. The Genetic Counseling Outcome Scale (GCOS-24) is a survey designed to measure empowerment in patients who receive genetic counseling services. The aim of this study is to (1) measure a baseline empowerment level of the caregivers of patients with ASD seen at UPMC Children's Hospital of Pittsburgh Division of Genetic and Genomic Medicine, (2) identify factors that may impact the selected empowerment domains including caregiver strain and appointment timing, and (3) assess the satisfaction of caregivers regarding the timing of their genetic counseling appointment in relation to the patient's diagnosis of ASD

Methods: A survey designed to collect information regarding satisfaction with appointment time and caregiver strain, and all questions from the empowerment domains of Hope, Emotional Regulation, and Behavioral Control were distributed to 46 caregivers of patients with an ASD diagnosis who were scheduled to see a genetic counselor and physician or nurse practitioner in the Division of Genetic and Genomic Medicine at UPMC Children's Hospital of Pittsburgh.

Results: 15 individuals responded to the survey with 13 completing the GCOS-24 portion of the questions. Empowerment scores ranged from 51 to 91 with an average of 66.62 (SD 11.72).

Caregiver strain ranged from 0 to 7 with an average of 2.86 and when compared to empowerment scores, there was a negligible correlation between the two ($r = -0.0900$). When analyzing satisfaction with appointment timing, there was no statistical difference in empowerment scores between individuals who were "happy/okay" and "neutral" regarding the amount of time they waited for an appointment ($p = 0.5210$). Zero individuals stated they were unhappy with their appointment timing.

Conclusion: Neither caregiver strain nor satisfaction with appointment timing showed significant correlation with empowerment scores. It is possible that the range in empowerment scores is due to factors outside of what was measured in this study.

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1.0 Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that mainly affects an individual's ability to have successful social communications and causes restricted interests and repetitive behaviors. The average age of diagnosis is around four and a half years, however, the most recent updates to the *Diagnostic and Statistical Manual of Mental Disorders-5th edition* included changes to the diagnostic criteria for ASD that aimed to allow for earlier diagnosis (Hodges, Fealko, & Soares, 2020). While autism can be a symptom of many conditions, the etiology is not entirely understood. In some cases, autism can occur as a characteristic of a larger genetic condition. When families search for a genetic cause of their child's autism, there unfortunately is not a standard care pathway that provides access to genetic services. According to the American College of Medical Genetics and Genomics (ACMG), every individual with ASD should be referred for a clinical genetics evaluation and any testing ordered should be based on said clinical evaluation. Genetic counseling is recommended regardless of the results of genetic testing, and results should inform clinical management (Schaefer & Mendelsohn, 2013). Studies suggest, however, that regardless of existing recommendations, there are inconsistencies in clinical practice. These inconsistencies can lead to delays or other barriers to individuals receiving an autism diagnosis, appropriate support services, and medical services including genetic testing and counseling (Amiet, Couchon, Carr, Carayol, & Cohen, 2014; Chen, Xu, Huang, & Dhar, 2013; Hurt et al., 2019; Savatt & Myers, 2021; Yusuf et al., 2021).

The aim of this study is to (1) measure a baseline empowerment level of the caregivers of patients with ASD seen at UPMC Children's Hospital of Pittsburgh Division of Genetic and Genomic Medicine, (2) identify factors that may impact the selected empowerment domains

including caregiver strain and appointment timing, and (3) assess the satisfaction of caregivers with the timing of their genetic counseling appointment in relation to the patient's diagnosis of ASD. The survey created for this study includes questions from the Genetic Counseling Outcome Scale (GCOS-24) (M McAllister, Wood, Dunn, Shiloh, & Todd, 2011). The GCOS-24 measures the patient-reported outcome measure of empowerment by asking questions that belong to five different sub-dimensions: cognitive control, decisional control, behavioral control, emotional regulation, and hope (M McAllister et al., 2011). Measuring empowerment is particularly important in this patient population due to the high levels of uncertainty and stress in receiving genetic testing results, and often lengthy diagnostic odyssey (Yusuf et al., 2021). This study includes all questions from the latter three sub-dimensions plus additional questions to collect information on caregiver strain and satisfaction with appointments times. The survey was distributed to caregivers of individuals with clinically diagnosed ASD prior to their genetic counseling visit. While the GCOS-24 has been validated for use in any genetic counseling patient population, the survey has been adapted for patients of the ASD community and other related neurodevelopmental conditions (mGCOS-24). This survey was constructed to evaluate the use of genetic services and the efficacy of receiving genetic testing results in this specific population (Yusuf et al., 2021). Upon review of the literature, we are not aware of any research examining caregiver empowerment in the ASD population. The baseline empowerment level in this specific population, along with information regarding the factors that may impact empowerment is important because it will be helpful in illuminating how genetic counselors can best serve this population. The information the survey can provide regarding satisfaction with the time between receiving an autism diagnosis and attending a genetics appointment can help to create a more standard pathway at UPMC Children's Hospital of Pittsburgh.

1.1 Specific Aims

Specific Aim 1: Develop and distribute a survey to caregivers of children up to age 17 with an autism diagnosis, using three sub-dimensions of the Genetic Counseling Outcome Scale, before their genetic counseling session to measure the empowerment domains of behavioral control, emotional regulation, and hope.

Specific Aim 2: Analyze data to identify factors that impact the measured empowerment domains before receiving genetic counseling, including caregiver strain and appointment timing.

Specific Aim 3: Assess the satisfaction of parents with the timing of their genetic counseling appointment in relation to their child's autism diagnosis.

2.0 Literature Review

2.1 Autism Spectrum Disorder

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in reciprocal social interactions and communications, and repetitive and restricted patterns of play. Characteristics of autism can be noticed by 18 months, however, the average age of diagnosis in the United States is about four and a half years according to the CDC. Their estimates indicated that the prevalence of ASD is about 1:44 (Maenner et al., 2021) . Although it is typically reported that males are four times more likely to be affected, it is worth noting that women are largely underdiagnosed. A meta-analysis completed by Loomes et. al in 2017 revealed that the ratio is likely closer to 3:1. Loomes et. al reported an apparent gender bias in patients receiving a diagnosis. When female patients met ASD criteria, they were less likely to receive a diagnosis (Loomes, Hull, & Mandy, 2017). Previously, autism spectrum disorder was under a category of diagnoses titled pervasive developmental disorders (PDD). This included autistic disorder, Asperger syndrome, disintegrative disorder, and pervasive developmental disorder not otherwise specified (PDD-NOS). In the *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition* (DSM-5), published in 2013, these diagnoses were combined under a diagnosis of autism spectrum disorder (Hyman, Levy, Myers, COUNCIL ON CHILDREN WITH DISABILITIES, & PEDIATRICS, 2020).

Obtaining a clinical diagnosis of ASD is considered essential in ensuring appropriate medical and therapeutic services. A diagnosis requires the use of a screening tool as well as a comprehensive developmental evaluation. While there is no standard screening tool, the most

commonly used tool is the Modified Checklist for Autism in Toddlers (M-CHAT) (Batshaw, 2019; Hyman, Levy, Myers, COUNCIL ON CHILDREN WITH DISABILITIES, & PEDIATRICS, 2020). Gathering information for a diagnosis often requires input from multiple individuals on the child's care team, however, the final diagnosis is often made by a developmental pediatrician, a child neurologist, child psychologist, or a child psychiatrist (Batshaw, 2019). The CDC reports that concerns about development were noticed by age three in 85% of children with ASD; despite this, 39% of children with an ASD diagnosis did not receive a developmental evaluation until after age four. The American Academy of Pediatrics (AAP) states that all patients with ASD should be offered a genetic evaluation including a chromosome microarray, Fragile X testing, and other cytogenetic or molecular testing when indicated (Hyman, Levy, Myers, COUNCIL ON CHILDREN WITH DISABILITIES, PEDIATRICS, et al., 2020). AAP also recommends that families with a child with autism have a conversation with their provider to discuss current knowledge of the genetics of ASD, recurrence risks, and the risks and benefits of genetic testing in the context of ASD. These recommendations are matched by the American College of Medical Genetics and Genomics (ACMG) and are included in the guidelines for the diagnostic process (Schaefer & Mendelsohn, 2013). Even with these recommendations and guidelines in place, it is reported that 17.4%-41.2% of children with ASD have undergone genetic testing in the past ten years (Zhao et al., 2021).

2.2 Empowerment

In 2010 Marion McAllister, Graham Dunn, and Chris Todd described a new construct to be used for measuring patient outcomes in clinical genetic services. They completed a systematic

review of validated outcome measures that were being utilized in clinical genetics and determined that there were 67 different outcome measures being used, in some cases only one time in one study. Patient outcomes were typically assessed by measuring information retention by the patient, and their knowledge on what was discussed by the provider, psychological constructs like anxiety and depression, the effectiveness of informed decision making by the patient, and strictly patient satisfaction. With this review, McAllister et al. discovered that there was no validated construct available that could be used to measure currently recognized benefits as well as additional potential benefits of clinical genetic services. The team developed their own construct: empowerment. They defined empowerment as “as set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future.” Their original model included four dimensions “the beliefs that one (1) can make important life decisions in an informed way (Decision Making), (2) has sufficient information about the conditions, including risks to oneself and ones relatives, and any treatment, preventions and support available (Knowledge and Understanding), (3) can make effective use of the health and social care systems for the benefit of the whole family (Instrumentality) and (4) can look to the future with hope for a fulfilling family life, for oneself, one’s family and/or one’s future decedents (Future orientation).” To test the validity, relevance, and importance of empowerment in clinical genetic services, they completed a qualitative study in which they used both focus groups and individual interviews with patients and only focus groups with geneticists. Participants were tasked with explaining their own views of the benefits and harms of clinical genetic services, as well as reading and critiquing information describing the empowerment construct and it’s four dimensions. With the responses to this study, they were able to refine the empowerment construct by renaming the dimensions, and add a fifth dimension. The team’s final decision was to rename the dimensions to reflect Perceived Personal

Control (PPC) terminology. Cognitive, decisional, and behavioral control, were included, Hope replaced the fourth dimension of Future Orientation, and an additional dimension of Emotional Regulation was added (Marion McAllister, Dunn, & Todd, 2011).

2.3 The Genetic Counseling Outcome Scale

The Genetic Counseling Outcome Scale (GCOS-24) was originally developed by M McAllister, AM Wood, G Dunn, S Shiloh and C Todd in 2011. The initial article introducing the GCOS-24 is entitled “The Genetic Counseling Outcome Scale: a new patient-reported outcome measure for clinical genetic services.” In this article, McAllister et al. explain that prior to the development of the GCOS-24, there was no standard way to measure outcomes of clinical genetic services. In other specialties, patient reported outcomes (PROMs) are used to measure Health Related Quality of Life (HRQoL). PROMs are questionnaires that are designed to capture HRQoL and are read and completed by the patient themselves. However, McAllister et al. posit that genetic services are often unlikely to alter health status, so it would not be as beneficial to measure this in the genetics patient population. Instead, the authors propose measuring empowerment. They define empowerment as decisional control, cognitive control, behavioral control, emotional regulation, and hope.

McAllister et al. created an 84-item questionnaire based on their own previous qualitative data, the PPC items, and the emotional representations sub-scale of the revised illness perceptions. This questionnaire was distributed to individuals of different support groups. Exploratory factor analysis and parallel analysis were used to select the twenty-four questions that would effectively measure the five domains of empowerment to create the GCOS-24. After determining validity and

sensitivity results showed they had developed a PROM specific to the genetics patient population, the GCOS-24, that could be used to measure patient-reported empowerment and how it may change over time (M McAllister et al., 2011).

While the GCOS-24 was designed to measure the change in empowerment gained from genetic counseling (M McAllister et al., 2011) it can also be used to measure baseline empowerment. A study completed by Palmer et al through the Undiagnosed Disease Network (UDN) used the GCOS-24 to measure baseline empowerment in 102 individuals (Palmer et al., 2018). The sample was composed of 35 affected adults and 67 healthy parents (either a mother or father) of affected children seeking care through the UDN. Participants took the GCOS-24 prior to their first in-person evaluation at the UDN. The survey was administered at two following timepoints; however, this study was only analyzing the first timepoint as a baseline. The results showed an average score of 112.66 on the GCOS-24 with a significantly lower score in adults than parents. Palmer et al. also found that their participant scores matched the scores of previous studies of families and patients (Palmer et al., 2018)

2.3.1 The modified Genetic Counseling Outcome Scale for autism spectrum disorders and related conditions

In 2020 Afqah Yusuf et al. set out to validate a modified version of the GCOS-24 for patients with autism spectrum disorders and related neurodevelopmental disorders receiving routine genetic testing (Yusuf et al., 2021). In their study, Yusuf et al. provides the recommendation for a care pathway as of 2013. It states that the care pathway should begin with a referral placed by the primary care provider for a chromosome microarray along with a pre- and post-test discussion with a genetic counselor regardless of results. The care pathway should

continue with a referral to a clinical geneticist? depending on results. The geneticists should then be incorporated in continuation of care along with the primary care provider after a diagnosis is made. Their study examines the discrepancies in the referral process and diagnostic care pathway for patients with autism spectrum disorder across different locations and facilities.

In their study design, Yusef et al. adapted the survey into a version that could be provided to parents of patients with ASD and related disorders to assess empowerment in the context of pursuing genetic testing in the current care pathway recommendation. A developmental pediatrician and a psychologist were employed to re-word GCOS-24 questions that required revision to create the mGCOS-24 (modified genetic counseling outcome scale). Some revisions included replacing the generic “the condition” in questions with “the neurodevelopmental condition”. 113 individuals participated in the study. An analysis of their responses showed that the mGCOS-24 was reliable and valid within this population for “assessing the anticipated impact of genetics results.” (Yusuf et al., 2021)

2.4 Genetic Counseling Considerations for Patients with Autism

2.4.1 Perceptions of ASD etiology and recurrence risk

There are a number of counseling considerations for a genetic counselor seeing a patient or family diagnosed with autism. Among them, is the patient or family’s perception of what causes autism. It’s widely known that there are many claims as to why a person may develop autism, some of which not supported by scientific evidence. In 2009 Selkirk et al. published a study that provides information on parental perceptions of ASD etiology (Selkirk, McCarthy Veach, Lian,

Schimmenti, & LeRoy, 2009). The study was designed to collect information on the perception of ASD etiology and recurrence risks and the effect this had on family planning. A survey was sent to individuals with one or more children with autism through the Autism Society of Minnesota list-serve. Two hundred and twenty-five individuals completed the 27 multi-part question survey collecting information on the following: their child's diagnosis, the parents' perceptions of the cause of the ASD and recurrence risk, family planning choices, whether or not there was a family history of ASD, their history of working with genetics professionals, and parent demographics. The survey provided a list of 12 commonly reported causes of ASD as well as an "other" option for which a family could type in their own response. Participants were permitted to select multiple causes. The top five selected causes were Genetic Influences (72.7%), Other (41.8%), Don't know (30.1%), Vaccinations (27.3%), and Improper brain development (24.1%). Selkirk et al.'s study also determined that the numerical perception of recurrence risks was inaccurate for the majority of respondents (Selkirk et al., 2009).

2.4.2 Complexities of negative results and variants of uncertain significance on genetic testing

Many families of a child with ASD see genetic testing a potential way to end a "diagnostic odyssey" (Yusuf et al., 2021). When testing yields results that do not provide an answer, it can contribute to the stress that many families feel (Giarelli & Reiff, 2015). Giarelli et al. conducted a study composed of 48 mothers of children with autism who received chromosome microarray analysis (CMA). They interviewed the mothers to determine similarities in thought or appreciation of results from the genetic testing. Interviews were coded to create principal themes, conceptual categories, and subordinate themes. Among mothers who had received negative results and

variants of uncertain significance (VUS), the principal theme was that something they were expecting or hoping to learn from results or the testing process was “missing”. The “missing information” ranged from genetic/genomic information about ASD, what exactly is being tested with a CMA, to a clear explanation of the use of results, implications of results for the future, and questioning the worth of the science (Giarelli & Reiff, 2015). The identified subordinate theme was “disappreciation.” Many of the mothers made statements that implied after received a negative or VUS results, they had less appreciation for the science as well as its value to them (Giarelli & Reiff, 2015). This study notes however that there were differing levels of pre- and post test counseling provided to the interviewed mothers as well as differing levels of the involvement of a genetic counselor in each case. The study itself was completed for nurses who may interact with families of children receiving genetic testing and encourages them to speak with a genetic counselor before discussing the CMA and its results with a family, or including a genetic counselor in the discussion with the family.

A patient or parent may feel differently about an uncertain result depending on their understanding of the VUS and how the information was presented to them (Clift et al., 2020; Giarelli & Reiff, 2015; Jez, Martin, South, Vanzo, & Rothwell, 2015; Skinner et al., 2018). Common negative feelings toward VUSs across multiple specialties include feelings of shock, frustration, and hopelessness. While common positive feelings include optimism, relief, and hope for the future (Clift et al., 2020).

Another study completed by Jez et al. in 2015 examines the parental perspectives of a VUS on a CMA for parents of children with developmental delay/intellectual disability/autism spectrum disorder (DD/ID/ASD) (Jez et al., 2015). The main goal of the study was to assess parental understanding of the VUS, their perceived value of the result, and perceptions of child

vulnerability and parental stress. There were 30 participants in total, each with a child who had CMA completed at the recommendation of their pediatrician or another specialist. All ordering providers were given a binder of documentation and educational material explaining basic genetic concepts, the VUS detected, a summary of known literature, any associated features, and recommendations for follow-up, all written at a middle school level. This binder was referred to as report documentation. 26 of the 30 respondents had received the report documentation after their results disclosure and 24 of 30 respondents reported that they felt they received “adequate information” about the results. The results of the study showed that individuals who received adequate information had a better understanding of the meaning of a VUS and indicated they were not overwhelmed with the information provided. 20 out of 30 respondents stated they felt they received adequate support to cope with results, and 14 of those individuals reported that support was provided by a genetic counselor. In contrast to the Giarelli et al. study, the parents in this study reported that the VUS provided some explanation and “parental relief for their child’s diagnosis” (Jez et al., 2015).

2.4.3 The Diagnostic Odyssey

The idea of a diagnostic odyssey is not unique to families of children with autism; however, it is a common experience among this specific population. A study completed in 2017 by Lappé et al. was designed to examine factors that may play a role in how parents experience the diagnostic odyssey of obtaining a clinical ASD diagnosis (Lappé et al., 2018). Lappé et al. completed interviews of 44 parents of 25 children with a confirmed diagnosis of autism (in some cases parents were interviewed separately) and were able to identify three themes that they believe describe three phases of the diagnostic odyssey in receiving an ASD diagnosis. They determined the diagnostic

odyssey begins with the initial change in the parents' perception and expectations of their child's development, is followed by the parents beginning to encounter barriers and increased wait time for services related to receiving the diagnosis, and continues with adjustment to the systems used to care for and provide support for families once the ASD diagnosis is received. The interviews revealed that throughout the start of the diagnostic odyssey many families felt conflicted by what they saw in their own child and what they were being told by family, friends, and pediatricians. Lappé et al. state that their study provides ways in which the diagnostic odyssey are influenced by parents' own previous experiences as well as previous experiences with other children, family members, friends, and different healthcare providers (Lappé et al., 2018).

2.5 Caregiver Strain

Caregiver strain, parental burden, or burden of care, all describe the stress a primary caregiver feels when caring for a child or family member with complex emotional and behavioral needs (Ana Maria Brannan, Heflinger, & Bickman, 1997; Durán-Pacheco et al., 2022; Lindly, Shui, Stotts, & Kuhlthau, 2021; Patel, Arya, Agarwal, Gupta, & Agarwal, 2022). While it's not the only way to measure caregiver strain, the Caregiver Strain Questionnaire (CGSQ), was developed by Brannan et al. to specifically measure the term they adopted as caregiver strain (Ana Maria Brannan et al., 1997). They defined caregiver strain as “the demands, responsibilities, difficulties, and negative psychic consequences of caring for relatives with special needs. In particular...parents and other caregivers (e.g. foster parents, relatives) who have primary responsibility for the needs of children with mental, emotional, and behavioral problems” (Ana Maria Brannan et al., 1997). Since the development of the 21 question CGSQ, it has been validated

and determined reliable for measuring strain specifically for caregivers of children with autism (Khanna et al., 2012) and has been shortened, and validated, twice to a CGSQ-Short Form 11 (Brennan, Babinski, & Waschbusch), and CGSQ-Short Form 7 (Ana María Brannan, Athay, & de Andrade, 2012) in order to decrease the time it takes to administer the survey to caregivers, and allow for more ease in continuation data collection.

The original CGSQ asks parents to rate how much of a problem each item is on a scale ranging from “not at all” to “very much”. It includes prompts such as “interruption of personal time,” “missing work or neglecting others,” and “disruption of family routines” (Ana Maria Brannan et al., 1997). One way in which this could potentially be impacted is by the number of visits required for individuals with emotional and behavioral issues compared to those without. Cummings et al. completed a review of the amount of health services utilized by children with ASD when compared to those without (Cummings et al., 2016). The goal of this study was to compare the use of preventative health services, primary, specialty, and acute care services between the ASD and non-ASD population, after adjusting for comorbid physical and/or mental health conditions. Preventative care included vaccinations, flu shots, any vaccine other than flu shots, and well-child visits. Primary care included well-child visits. Specialty visits referred to speech, occupational, and/or social skills therapy, physical therapy, psychotherapy, and neurology. Outpatient appointments were also counted and included. Acute care included any visit to an emergency department (ED) or an inpatient stay. Data was taken from an ASD Registry that included five healthcare systems, all participating in National Institute of Mental Health’s Mental Research Network. The sample included 8,325 youth with ASD and 83,195 comparison youth, matched based on age, gender, months of enrollment in the health system, having prescription drug coverage, and the healthcare system of enrollment. Analysis of results showed that individuals

with ASD were two times as likely to be diagnosed with a genetic disorder, and four to five times as likely to have a diagnosis of mental health disorder including ADHD, ODD/Conduct disorder, anxiety, depression, etc. (Cummings et al., 2016). Results showed that younger children, ages 3-9, had almost two more visits to their pediatrician during a year than those without ASD. While older youth, ages 10-17, were less likely to receive any well-child visits. Younger youth also had 4.8 more outpatient visits than those without ASD, while older youth had 2.2 more outpatient visits.

One study completed in 2021 assessed the change in caregiver strain over time and the factors that may contribute to improved strain (Lindly et al., 2021). Lindly et al. used data previously collected through the Autism Speaks Autism Treatment Network (ATN) Registry Call-Back Assessment (RCBA) study. As part of this larger study, the ATN RCBA administered the full 21 CGSQ to parents of children with a confirmed ASD diagnosis. Lindly et al. used 368 children who had CGSQ data at two different time points one to two years apart, T1 and T2, in order to appropriately measure a change in caregiver strain. Contrary to their hypothesis, the strain felt by caregivers was not statistically different between T1 and T2. Their analysis showed that 47.9% of parents showed an increase in strain, 45.7% of parents showed a decrease, and 6.4% of parents remained at the same level (Lindly et al., 2021).

Another study completed by Patel et al. used the Burden Assessment Schedule (BAS) to assess burden felt by primary caregivers and the Hindi version of the Abbreviated World Health Organization Quality of Life (WHOQOL-BREF) to assess quality of life (QoL) in caregivers of children with autism in India (Patel et al., 2022). The BAS includes 40 items across nine domains and measures the burden on caregivers of a chronically ill family member (Thara, Padmavati, Kumar, & Srinivasan, 1998). Patel et al. excluded one domain from the study as it only pertains to

caring for a mentally ill spouse. The study included 40 caregivers of children with ASD. Overall results showed the adjusted burden on the BAS to be significantly high in the participants, suggesting a high degree of burden in the mothers, with higher burden on families who fell into low-income groups. QoL was also significantly impacted in the “physical health” and “psychological” domains (Patel et al., 2022).

2.6 Diagnostic care pathway for autism spectrum disorders

The American College of Medical Genetics and Genomics (ACMG) updated previously published guidelines for the diagnostic care pathway for individuals with ASD in 2013. The updated guidelines state that the approach should be stepwise and should be customized based on the individual presenting. According to ACMG, the pathway must start with an accurate diagnosis of ASD including an audiogram, and ensuring that the individual have established care with a primary-care provider. Every individual with ASD should be referred for a clinical genetics evaluation and any testing ordered should be based on said clinical evaluation. Genetic counseling is recommended regardless of results of testing, and treatment/follow-up should be based on the results (Schaefer & Mendelsohn, 2013).

Studies suggest that despite having recommendations in place, there are inconsistencies in clinical practice that lead to a delay in receiving a diagnosis and appropriate services (Amiet et al., 2014; Chen et al., 2013; Hurt et al., 2019; Savatt & Myers, 2021).

2.6.1 Satisfaction with wait times for appointments

Appointment wait times, described as the time a patient has to wait before they can be seen by a genetics provider, varies based on the subspecialty (Maiese, Keehn, Lyon, Flannery, & Watson, 2019). Jenkins et al. reported results from a survey that outlines average wait times for a non-emergent patient (Jenkins et al., 2021). This wait time was across all specialties and reported to be greater than three months for 39% of respondents. Maiese et al. reports that in 2015 children's hospitals had the longest wait times with 39.4% waiting more than three months for a non-emergent appointment. Some genetic counselors report wait times upwards of eight months, and feel that the wait time is a possible deterrent to patients (Boothe, Greenberg, Delaney, & Cohen, 2021).

A review of recommendations from parents to pediatric appointment scheduling, states that many parents feel frustrated with wait times and the management of wait lists (Kallos et al., 2021), however, there is little data regarding measured patient satisfaction, especially in the context of a clinical genetics setting. One study by Kalb et al. found that increased wait times, defined as the difference between referral date and prospective or scheduled appointment date, increased the likelihood of families canceling initial appointments in an Outpatient Pediatric Autism Clinic. Patient satisfaction with these increased wait times was not measured in the study (Kalb et al., 2012).

3.0 Manuscript

3.1 Background

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in reciprocal social interactions and communications, and repetitive and restricted patterns of play. The CDC estimates that the prevalence of ASD is about 1:44, with the average age of diagnosis being about four and a half years in the United States (Maenner et al., 2021). Although it is typically reported that males are four times more likely to be affected, it is worth noting that women are largely underdiagnosed and some studies suggest the ratio is closer to 3:1 (Loomes et al., 2017). Previously, autism spectrum disorder was under a category of diagnoses titled pervasive developmental disorders (PDD). This included autistic disorder, Asperger syndrome, disintegrative disorder, and pervasive developmental disorder not otherwise specified (PDD-NOS). In the *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition* (DSM-5), published in 2013, these diagnoses were combined to all fall under a diagnosis of autism spectrum disorder (Hyman, Levy, Myers, COUNCIL ON CHILDREN WITH DISABILITIES, & PEDIATRICS, 2020).

The American Academy of Pediatrics (AAP) recommends that all patients with ASD be offered a genetic evaluation and that families of a child with autism have a conversation with their provider to discuss current knowledge of the genetics of ASD, recurrence risks, and the risks and benefits of genetic testing in the context of ASD. These recommendations are matched by the American College of Medical Genetics and Genomics (ACMG) and are included in the guidelines for the diagnostic process (Schaefer & Mendelsohn, 2013). Studies suggest that despite the

recommendations in place, there are inconsistencies in clinical practice, which leads to a delay in receiving a diagnosis and appropriate services (Amiet et al., 2014; Chen et al., 2013; Hurt et al., 2019; Savatt & Myers, 2021). Due to the delay in receiving an autism diagnosis, as well as any potential genetic diagnosis, many patients experience a diagnostic odyssey (Lappé et al., 2018).

When determining a genetic counselor's role in the diagnostic odyssey or diagnostic care pathway for this specific patient population, it's important to analyze where in the pathway the genetic counselor falls, as well as the outcome of the counseling session. In other specialties, patient outcomes are typically assessed by measuring information retention by the patient, their knowledge regarding what was discussed by the provider, psychological constructs like anxiety and depression, the effectiveness of informed decision making by the patient, and strictly patient satisfaction. In 2010 Marion McAllister, Graham Dunn, and Chris Todd described a new construct to be used for measuring patient outcomes in clinical genetic services. This new construct was titled "empowerment" and is defined as "as set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future" (Marion McAllister et al., 2011). McAllister et al. separated the empowerment construct into five domains: decisional control, cognitive control, behavioral control, emotional regulation, and hope (Marion McAllister et al., 2011). Through an additional study, M. McAllister, A.M. Wood, G. Dunn, S. Shiloh and C. Todd developed the Genetic Counseling Outcome Scale (GCOS-24). The GCOS-24 is a survey consisting of 24 questions spanning the five domains and is typically used before and after a genetic counseling session to measure the change in empowerment.

The aim of this study is to (1) measure a baseline empowerment level of the caregivers of patients with ASD seen at UPMC Children's Hospital of Pittsburgh Division of Genetic and Genomic Medicine, (2) identify factors that may impact the selected empowerment domains

including caregiver strain and appointment timing, and (3) assesses the satisfaction of caregivers with the timing of their genetic counseling appointment in relation to the patient's diagnosis of ASD. This study includes all questions from the behavioral control, emotional regulation, and hope sub-dimensions of the mGCOS-24 plus additional questions to collect information on caregiver strain and satisfactions with appointments times. Caregiver strain, parental burden, or burden of care, all describe the stress a primary caregiver feels when caring for a child or family member with complex emotional and behavioral needs (Ana Maria Brannan et al., 1997; Durán-Pacheco et al., 2022; Lindly et al., 2021; Patel et al., 2022). In this study caregiver strain will refer to the number of additional specialties a patient sees. Appointment time refers to the time between the patient receiving a clinical diagnosis of ASD and when their genetic counseling session took place. The survey was distributed to caregivers of individuals with clinically diagnosed autism prior to their genetic counseling visit. While the GCOS-24 has been validated for use in any genetic counseling patient population, the survey has been adapted for patients of the ASD community and other related neurodevelopmental conditions (mGCOS-24). This survey was constructed to evaluate the use of genetic services and the efficacy of receiving genetic testing results in this specific population (Yusuf et al., 2021). Upon review of the literature, we are not aware of any research examining caregiver empowerment specifically in the ASD population. The baseline empowerment level in this specific population, along with information regarding the factors that may impact empowerment is important because it will be helpful in illuminating how genetic counselors can best serve this population. Assessing caregiver satisfaction with the time between receiving an autism diagnosis and attending a genetics appointment can help to create a more standard and patient-centered pathway at UPMC Children's Hospital of Pittsburgh.

3.2 Materials and Methods

3.2.1 Study design and participants

This study used a quantitative approach to survey caregivers about their empowerment as well as the factors that could potentially be contributing to this empowerment. Participants of the study were caregivers of children diagnosed with ASD being seen in the Medical Genetics Clinic at UPMC Children's Hospital of Pittsburgh. These individuals were sent an email including a brief description of the study along with a link and QR code to the survey. Caregivers were required to be 18 years of age or older. The ASD patients were required to be 17 years of age or less with an ASD diagnosis documented in their electronic medical record system. Outside of these requirements there were no specific exclusion criteria. Prior to implementation, this study was approved by the Institutional Review Board of the University of Pittsburgh Graduate School of Public Health. Informed consent was obtained by all participants and all information obtained was anonymous and remained deidentified for the study.

3.2.2 Survey development and distribution

The survey developed for this study included questions addressing basic demographic information such as ethnicity and gender, along with questions designed to collect information regarding appointment time and caregiver strain. All questions were either multiple choice or fill-in-the-blank. Additionally, the GCOS-24 questions in the subdimensions of behavioral control, emotional regulation, and hope were also included (M McAllister et al., 2011). These questions were answered using the 7 Point Likert Scale, as intended by the GCOS-24. In order to ensure

readability and accessibility to the largest audience, the survey was developed and written to meet the recommendations of the American Medical Association and National Institutes of Health that patient educational materials to be written at a sixth-eighth grade reading level (Rooney et al., 2021). When analyzed using an online readability tool, the survey achieved a Flesch-Kincaid Grade Level of 6.89 and a Flesch Reading Ease score of 60.71 (Readable.com (<http://readable.com>) by Added Bytes, 2022). The Flesch-Kincaid Grade level falls within the recommended sixth-eighth grade reading level which, according to the readability tool, makes it accessible to 85% of the general public. A Flesch Reading Ease score of 60.71 reaches the suggested threshold of 60+, making it easily understood by individuals between ages 13-15 (Readable.com (<http://readable.com>) by Added Bytes). In total, the survey had 20 separate questions; however, due to the use of skip-logic, there was a possibility for less questions. Qualtrics XM was used for the survey design and distribution.

Caregivers were identified for recruitment by reviewing the weekly schedule for the Medical Genetics Clinic between March 15th, 2022 and April 29th, 2022. A brief review of the diagnosis codes and problems list in the medical record system was completed for qualifying patients scheduled to see a geneticist or nurse practitioner along with a genetic counselor. Patients selected had a documented diagnosis of Autism, Autism Spectrum Disorder, or a related diagnosis including either of the two. The email address listed in the patient's electronic medical record was used to send the recruitment email, which included a link and QR code for the survey. Potential participants were contacted one day to two weeks from their appointment date and were sent one reminder email the Sunday before their appointment.

3.2.3 Data analysis

Responses were anonymously recorded and saved in the Qualtrics XM system. Statistical analysis of the responses was completed through Microsoft Excel as well as Stat/SE 16.1. Empowerment scores were calculated based on values associated with the 7 Point Likert Scale where “strongly disagree” was given a value of one and “strongly agree” was given a value of seven. Questions were reverse coded when necessary. The values were totaled to create an Empowerment score for each participant. The highest possible score was 105 and the lowest was 15. Higher scores indicate a higher level of empowerment. Descriptive statistics was used for categorical and quantitative data.

3.3 Results

3.3.1 Sample demographics

A total of 46 surveys were distributed. After two responses were discarded from data due to lack of sufficient completion, there was a total response rate of 32% (15/46). Of those responses, 13 (13/15) completed the GCOS-24 portion of the survey. Table 1 shows the full breakdown of the respondent and ASD patient demographics. The majority of respondents identified themselves as mothers (40%) and the majority of patients with ASD were males (60%). Around 72% of the ASD patients described themselves as White or a combination of White and another race. The age of the ASD patient was calculated from month and year of birth for the patient and the month and year the survey was taken i.e., March or April of 2022. For birth months that were during the

month the survey was taken, the full month was included into the calculation, and it was assumed the birthday had passed. Seven (47%) patients were between the age of zero and 71 months, five (33%) were between the ages of 72 and 131 months, two (13%) were between 132 and 191 months, and one (7%) was between 192 and 251 months old. The average age of the patients at the time of survey was 88.3 months, or seven years and three months, with the oldest patient being 211 months (17 years and seven months), and the youngest being 33 months (two years and nine months). The respondents provided the month and year that the patient was diagnosed with ASD. The age at diagnosis was determined using the month and year of birth, again including the full month if it was the month of the birthday. The majority (80%) of the patients were diagnosed before 71 months (age five), with the remaining patients being diagnosed between 72 and 131 months (between six and 10 years old). The average age of diagnosis was 40.5 months, or three years and four months. This is about 12 months earlier than the average age listed in the literature.

Table 1: Respondant and ASD Patient Demographics (N = 15) ¹

Caregiver	n	%
Mother	6	40%
Father	0	0%
Grandparent	3	20%
Legal Guardian	0	0%
Prefer to Self-Describe	0	0%
Blank	6	40%
Total	15	
Gender	n	%
Male	9	60%
Female	6	40%
Non-Binary	0	0%
Transgender Male	0	0%
Transgender Female	0	0%
Prefer to Self-Describe	0	0%
Blank	0	0%
Total	15	-
Ethnicity	n	%

¹ some totals are more than 15 as respondents could select more than one answer

American Indian	0	0%
Asian	1	6%
Black Or African American	3	17%
Hispanic Or Latino	1	6%
Native Hawaiian	0	0%
White	13	72%
Prefer to Self-Describe	0	0%
Blank	0	0%
Total	18 ¹	-
Patient Age in Months	n	%
0-71	7	47%
72-131	5	33%
132-191	2	13%
192-251	1	7%
Blank	0	0%
Total	15	100%
Mean	88.3	
Patient Age at Dx in Months	n	%
0-71	12	80%
72-131	3	20%
132-191	0	0%
192-251	0	0%
Blank	0	0%
Total	15	100%
Mean	40.5	

3.3.2 Empowerment Scores

Empowerment scores were calculated for each participant by totaling the values assigned to each GCOS-24 response. Thirteen of the 15 respondents completed this portion of the survey. Table 2 shows that scores ranged from 51 to 91 with a mean of 66.62. A previous study completed by the Undiagnosed Disease Network (UDN) used the GCOS-24 to observe baseline empowerment scores in their patients before they underwent an evaluation (Palmer et al., 2018). Their study included both adults affected with an undiagnosed disease, as well as parents of affected children. The mean empowerment score for the 67 (67/102) parents involved in this study was 116.5 (SD 18.1). In comparison to this study, Palmer et al.'s study used all five empowerment domains adding an additional nine questions. This increases the total possible empowerment scores a minimum of 24 and a maximum of 168.

Table 2: Summary Statistics of Participant Empowerment Scores.

	n	Mean (SD)	Min	Max
Empowerment Score	13	66.62 (± 11.72)	51	91

Both current age and the age at ASD diagnosis had a weak negative correlation with empowerment scores. Figure 1 shows that as the age of the patient at the time of the survey (current age) increases, the empowerment score of their caregiver decreases ($r = -0.3443$). This same trend is reflected in the age that the patient received their ASD diagnosis ($r = -0.3641$).

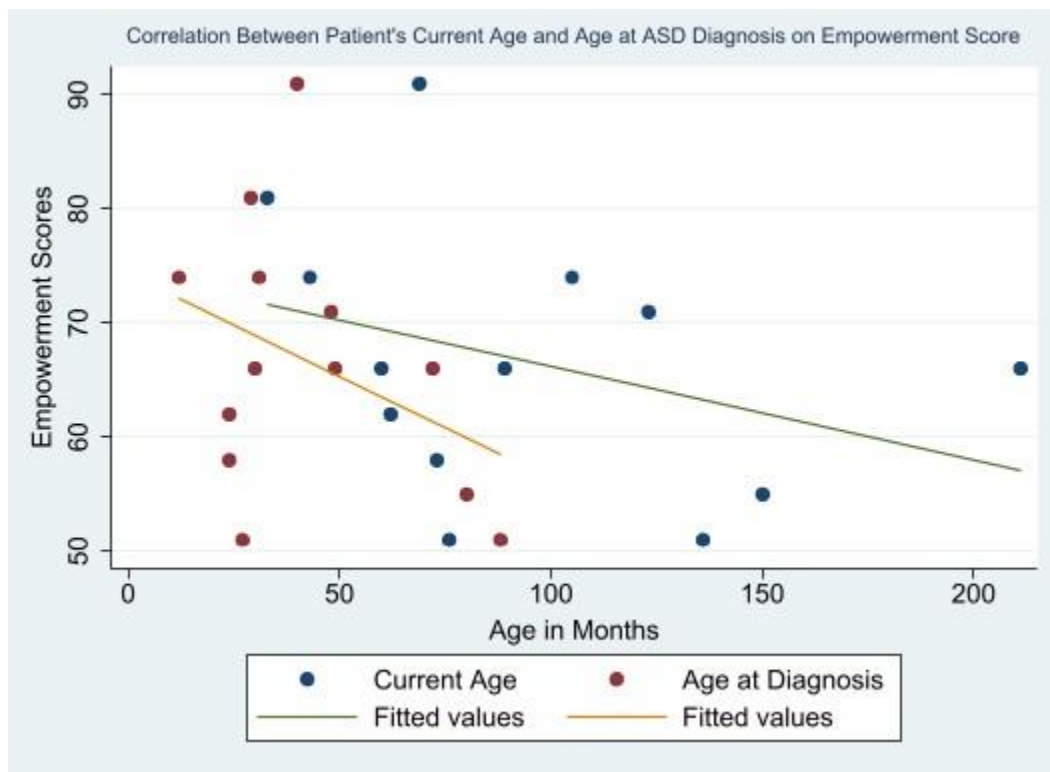


Figure 1: Correlation Between Patient's Current Age and Age at ASD Diagnosis with Empowerment Scores

Interestingly, the mean empowerment score between individuals who had met with a genetic counselor before (6/15) and those who had not (7/15), were similar. The mean scores were 66.33 (± 15.97) and 66.85 (± 7.88), respectively. However, the range was wider in individuals who

had met with a counselor. Table 3 shows that participants who met with a genetic counselor in the past had empowerment scores that ranged from 51 to 91, while those who have not had scores that ranged from 51 to 74. Figure 2 shows the relationship between meeting with a genetic counselor and empowerment scores. This figure shows the median score is much lower in the group that had previously met with a genetic counselor. The distribution of the empowerment scores is also more spread out in this group as well.

Table 3: Summary of Empowerment Scores Based on Meeting with a Genetic Counselor.

Met With a Genetic Counselor	n	Mean (SD)	Min	Max
No	7	66.86 (± 7.88)	51	74
Yes	6	66.33 (± 15.97)	51	91



Figure 2: Relationship Between Empowerment Scores and Meeting with a Genetic Counselor

In order to evaluate similarities in responses between the participants, the 7 Point Likert Scale was reduced to three main categories. Strongly agree, slightly agree, and agree, were combined to one unifying “agree” category. Disagree was condensed similarly, and neither agree nor disagree was left as “neutral”. Table 4 shows the number of participants who selected each category for each GCOS-24 item of the survey. Questions where 50% or more of the participants selected similar responses are highlighted. The most similarity was seen in the Hope domain; participants had higher than 50% similarity on all four questions within this domain. The least similarity was seen in the Emotional Regulation domain. Out of the three questions, zero of them had 50% or higher similar responses. Participants had 50% or higher similar responses to six out of the eight questions in the Behavioral Control domain.

Table 4: Percentage Similarity in GCOS-24 Survey Responses

Question	Agree % (n)	Neutral % (n)	Disagree % (n)
Behavioral Control²			
I can change how this condition affects my family in the future (2 ³)	61.5 (8)*	30.8 (4)	7.7 (1)
I don’t know where to go to get the medical help I/my family need (s) (5) ⁴	30.4 (4)	23.1 (3)	46.2 (6)
I can control how this condition affects my family (7)	61.5 (8)*	7.7 (1)	30.8 (4)
I am able to cope with having this condition in my family (9)	84.6 (11)*	7.7 (1)	7.7 (1)
I know how to get the non-medical help I/my family need(s) (e.g., educational, financial, social support) (15)	38.5 (5)	7.7 (1)	53.8 (7)*
I can explain what the condition means to people in my family who may need to know (16)	69.2 (9)*	23.1 (3)	7.7 (1)
I don’t know what I can do to change how this condition affects me/my children (17) ⁴	53.8 (7)*	15.4 (2)	30.8 (4)

² the empowerment domain,

³ item number within the GCOS-24

⁴ reverse coded questions

* 50% or more of the participants had similar responses

I am powerless to do anything about this condition in my family (22) ⁴	23.1 (3)	30.8 (4)	46.2 (6)
Emotional Regulation²			
when I think about the condition in my family, I get upset (4) ⁴	38.5 (5)	30.8 (4)	30.8 (4)
having this condition in my family makes me feel anxious (11) ⁴	46.2 (6)	23.1 (3)	30.8 (4)
I feel guilty because I (might have) passed this condition on to my children (21) ⁴	38.5 (5)	23.1 (3)	38.5 (5)
Hope²			
I can see that good things have come from having this condition in my family (6)	53.8 (7)*	23.1 (3)	23.1 (3)
I feel positive about the future (8)	76.9 (10)*	7.7 (1)	15.4 (2)
I am hopeful that my children can look forward to a rewarding family life (19)	92.3 (12)*	7.7 (1)	0.0 (0)
I am able to make plans for the future (20)	61.5 (8)*	30.8 (4)	7.7 (1)

3.3.3 Caregiver Strain and Patient Satisfaction

In this study, caregiver strain was defined by the number of other specialties involved in the ASD patient's care. The survey provided a list of major specialties at UPMC Children's Hospital of Pittsburgh, and the respondents were asked to select all specialties they had seen, or will see, more than one time. The study refers to the number of specialties as a caregiver strain. The average caregiver strain was 2.86, however, the caregiver strain ranged from 0 to 7. One respondent was excluded from the average score and range due to the stage at which the survey was uncompleted. Figure 3 shows that with a Pearson's Correlation Coefficient of $r = -0.0900$, the correlation between caregiver strain and empowerment is negligible (Schober, Boer, & Schwarte, 2018).

Respondents were also asked to select whether they felt "happy/okay," "unhappy," or "neutral" about the time they waited for an appointment. This time refers to the amount of time the patient waited for an appointment after calling to schedule their appointment. Respondents

provided additional information on whether they accepted the first available appointment time, and if they did not, they could provide a reason. Out of the 14 participants that answered this question, responses were split evenly between feeling “happy/okay” and “neutral” and zero participants reported they were “unhappy.” Thirteen of the 14 participants who responded to this question completed the survey to provide an empowerment score. Comparing individuals who responded with “happy/okay” and “neutral” showed no statistical difference in empowerment scores ($p = 0.5210$).

An additional question required participants to rate their satisfaction with the length of time between when the patient was diagnosed with ASD and when they were being seen in genetics, using the same “happy,” “unhappy,” or “neutral” scale. Although this time ranged from one year to 11 years, 79% (11/14) respondents reported that they felt neutral. Zero participants reported feeling unhappy about their appointment wait time or their time from diagnosis to genetics appointment.

Table 5: Empowerment Scores and External Factors that may Impact them

Participant ID	Empowerment Score	Caregiver Strain	Satisfaction W/ Appointment Wait Time	Satisfaction W/ Time from Dx to Seeing Genetics
CHP1	51	0	happy/okay	neutral
CHP2	.*	3	neutral	neutral
CHP3	66	1	happy/okay	neutral
CHP4	58	3	happy/okay	neutral
CHP5	66	2	neutral	neutral
CHP6	74	1	neutral	happy
CHP7	74	4	happy/okay	neutral
CHP8	81	4	happy/okay	happy
CHP9	62	3	neutral	happy
CHP10	91	1	neutral	neutral
CHP11	51	2	happy/okay	neutral
CHP12	71	3	happy/okay	neutral
CHP13	55	7	neutral	neutral
CHP14	66	6	neutral	neutral

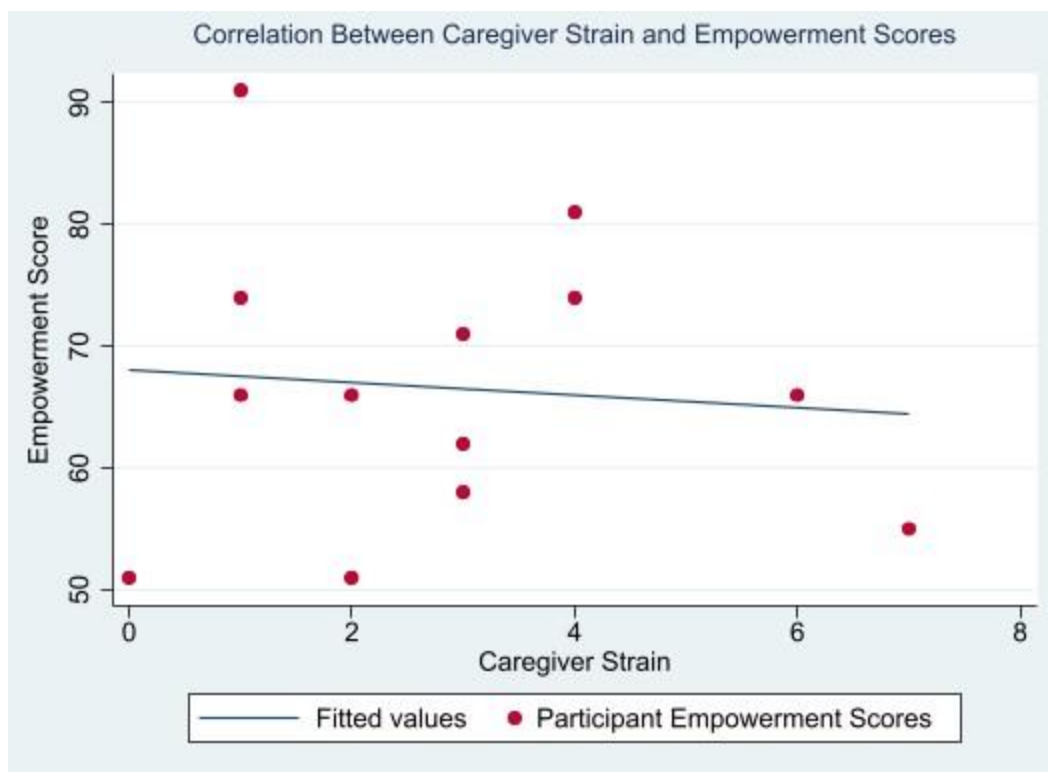


Figure 3: Correlation Between Caregiver Strain and Empowerment Scores

3.3.4 Additional Gathered Data

Respondents were also asked to report whether or not they had already completed genetic testing, if so, what type, and the results of testing if available/known. Of the 15 respondents, 53% (8/15) had genetic testing and 40% (6/15) had not. One respondent reported they were unsure if testing had been completed. Options for genetic testing were provided and participants could select as many as the patient had completed. These options included karyotype, chromosome microarray (CMA), Fragile X testing, whole exome sequencing (WES), panel testing, and the option to write in additional testing, and/or select “I’m not sure.” The most commonly reported testing in the above mentioned eight participants, was Fragile X testing with five participants. This was followed by four individuals selecting CMA, three selecting WES, and four selecting “I’m not sure.” If

results were available, the participants could select one or more of the following choices as a result type: normal/negative, a microdeletion or microduplication syndrome, variant of uncertain significance or inconclusive result, Fragile X, Angelman syndrome, Rett syndrome, Williams syndrome, Prader-Willi syndrome, Cowden syndrome (PTEN related disorder), tuberous sclerosis (TSC), I'm not sure, or Other with the option to write in a different result. For participants that reported testing had been completed, and results were available (6/8), two of them reported normal/negative results, two reported a variant of uncertain significance or inconclusive result, and two selected other and typed in their own results.

Table 6: Empowerment Scores with Genetic Testing and Results

Participant ID	Empowerment Score	Genetic Testing	SNP	WES	Panel	Karyotype	Fragile X	Not sure	Results Known	Results
CHP1	51	No								
CHP2	-	No								
CHP3	66	No								
CHP4	58	Yes	X				X		Yes	Normal/negative
CHP5	66	No								
CHP6	74	Yes						X	No	
CHP7	74	Yes					X	X	Yes	Variant of uncertain significance or inconclusive result
CHP8	81	Unsure								
CHP9	62	Yes	X	X			X		Yes	Other
CHP10	91	No								
CHP11	51	Yes	X	X			X		Yes	Variant of uncertain significance or inconclusive result
CHP12	71	Yes						X	Yes	Normal/Negative, Other
CHP13	55	No								
CHP14	66	Yes						X	No	
CHP15	-	Yes	X	X			X		Yes	-

3.4 Discussions

3.4.1 Participants

The majority of participants identified themselves as mothers (40%) with grandparents being the only other selected type of caregiver (20%). Out of the total 15 responses, six individuals left the caregiver type blank so it's difficult to draw any conclusions from this portion of the demographics. However, comparing these results to other studies focused on autism, the GCOS-24, and genetic testing in children, there is an obvious trend of mothers and female caregivers making up the majority of the responses (Chen et al., 2013; Giarelli & Reiff, 2015; Palmer et al., 2018; Selkirk et al., 2009; Yusuf et al., 2021). Nine of the respondents marked that the ASD patient were male (60%), and the remaining six (40%) were female. This gives a male to female ratio of 3:2. The most commonly reported ratio in the literature is 4:1, although some individuals suggest a ratio close to 3:1 due to the fact that females are largely undiagnosed (Loomes et al., 2017). It is interesting to see that in this population the ratio is closer to 1:1 than may have been expected. If this ratio could be replicated in a larger study, it would be important to take patient gender into consideration, and see if this could play a role in the empowerment score of the caregiver.

3.4.2 Empowerment Scores

An empowerment score is typically derived from all five domains included in the GCOS-24 (M McAllister et al., 2011). In this study, cognitive control and decisional control questions were excluded because it was felt these questions applied to genetic conditions that are generally seen as more severe or life limiting, or focused on the idea of pursuing genetic testing. Given the

timing of the administration of this survey, many families may not have yet considered genetic testing before their appointment. Removing 10 questions from the GCOS-24 lead to a lower overall empowerment score, however it allowed the ability to only collect data on the specific dimensions of patient empowerment that this study was assessing. Table 4 highlights that many of GCOS-24 questions had higher than 50% similarity in responses. The question with the highest similarity was “I am hopeful that my children can look forward to a rewarding family life” with 12 out of the 13 respondents agreeing with this statement and only one responding with feeling neutral. This result is similar to what was observed in the parental group of a similar UDN study (Palmer et al., 2018) and matches parents’ feelings regarding the importance of conveying hope in a counseling session when dealing with an ASD diagnosis (Selkirk et al., 2009). Caregivers were most evenly split on the question “when I think about the condition in my family, I get upset” with five participants agreeing with this statement, four stating they felt neutral, and four disagreeing. Neither the GCOS-24 nor this survey collect specific information on what component of the condition is causing the respondent to be upset, however, previous studies show that having a child with autism can cause complicated feelings of guilt, frustration and confusion on the potential cause (Lappé et al., 2018; Selkirk et al., 2009). Comparing responses to the question “I know how to get the non-medical help I/my family need(s) (e.g., educational, financial, social support” is also important. The majority of respondents disagreed with this statement. Struggling to find services is commonly reported in this patient population (Lappé et al., 2018; Maenner et al., 2021). However, it is worth noting that six of the seven individuals who disagreed with this statement had seen a genetic counselor at least one time or more prior to their upcoming appointment.

3.4.3 Factors contributing to Empowerment

The purpose of this study was to examine the impact of two specific factors on patient empowerment: caregiver strain and appointment timing. Caregiver strain was defined in this study as the number of other specialties the ASD patient follows with, or plans on following with for more than one visit. The hypothesis prior to the study was that caregiver strain would correlate either with significantly higher empowerment scores due to families having a larger support system, or significantly lower empowerment scores due the stress of managing multiple doctors' appointments. The data showed that the correlation between caregiver strain and empowerment scores was negligible. The strain, or "burden" as it can often be referred to, in caring for an individual with autism, has been shown to have an impact on the caregivers psychological health as well as overall wellbeing (Patel et al., 2022) so this negligible correlation was surprising. It is worth noting, however, that there are more in-depth ways to measure caregiver strain. In the future it may be beneficial to combine a validated survey to measure caregiver strain or caregiver quality of life with the GCOS-24 to determine whether there is any correlation with empowerment scores.

The survey asked participants to rate their satisfaction with the amount of time they waited for their appointment after calling to schedule (i.e., how far in advance they were scheduled), as well as their satisfaction with the time from the patient receiving the ASD diagnosis to the time of their scheduled genetics appointment. In both sections, zero participants stated that they felt unhappy with their appointment timing. These were unexpected results because in previous studies parents have reported feeling frustrated with wait times and the management of wait lists (Kallos et al., 2021). In addition, some genetic counselors feel as though longer wait times can be a deterrent for patients to seek genetic counseling (Boothe et al., 2021). In a future study, it could be

helpful to have patients provide the amount of time they waited for an appointment to have a better understanding of the rates of satisfaction.

In this study, six out of the 13 individuals who had empowerment scores had previously met with a genetic counselor. The average empowerment scores between those who had previously met with a genetic counselor and those who had not showed almost no difference. Figure 2 shows that individuals who had not previously met with a genetic counselor had a moderately positive correlation while those who had previously met with a genetic counselor had a negligible correlation. While this is interesting to see, no true conclusion can be made without additional research. It is possible that prior to their first appointment their empowerment score was lower than what is depicted in this survey. In the future, with a larger sample, it would be helpful to compare true baseline empowerment scores with one another.

3.4.4 Study Limitations

The design chosen for this study allowed for participants to leave multiple responses blank. With a small sample size, these blank responses made some comparisons difficult. In a future study it would be helpful to better utilize the Qualtrics features and require an answer on certain questions. For example, the purpose of the study was to look at caregiver empowerment, so having the type of caregiver would have been helpful in determining trends, but with 40% of the respondents having left this portion blank, comparisons between caregiver type weren't possible.

In order to capture the largest sample size, only an ASD diagnosis was required. This means it is possible that some patients had a diagnosis of ASD but were referred for a genetics evaluation for an unrelated reason. It is not known how this affects empowerment scores, but future studies

should include referral reasons in the survey, or potentially exclusion criteria that focuses the study on ASD as the main referral reason.

Another limitation to the study is the method chosen to measure caregiver strain. The definition of caregiver strain was created for this study, however, there are other validated and more in-depth ways to measure caregiver strain or caregiver burden. Incorporating these measurement tools with the GCOS-24 could be helpful in gathering more robust data. This study chose to focus on specific factors that could have an effect on empowerment scores, however, there are many additional factors that weren't collected that could have been playing a role. For example, this study did not collect information on socioeconomic status, insurance coverage, the distance a patient lived from the hospital, a potential diagnostic odyssey, etc., all of which are known to affect caregivers of individuals with autism and caregivers in general (Giarelli & Reiff, 2015; Lappé et al., 2018; Patel et al., 2022; Zhao et al., 2021).

3.5 Conclusion

This study was designed to assess patient empowerment in the caregivers of patients with autism spectrum disorder using three out of the five domains of empowerment in the Genetic Counseling Outcome Scale. Additionally, the study was analyzing the possible impact of caregiver strain and satisfaction with appointment timing on patient empowerment. There were 15 participants in the study, with 13 out the 15 completing the GCOS-24 portion of the survey. Empowerment scores ranged from 51 to 91 with an average of 66.62 (SD 11.72). The average age of the ASD patient at the time the survey was taken was 88.3 months (seven years and three months) and the average age at ASD diagnosis was 40.5 months (three years and four months).

Both current age and age at ASD diagnosis had a weak negative correlation with empowerment scores ($r = -0.3443$, $r = -0.3641$ respectively). Having met with a genetic counselor previously also showed little impact on average empowerment scores. Participants who met with a genetic counselor had an average empowerment score of 66.33 (SD 15.97) and a negligible correlation. Participants who have not met with a genetic counselor had an average empowerment score of 66.86 (SD 7.88) and a moderately positive correlation with empowerment score. No true conclusion can be drawn from these scores because there is no baseline data available for the individuals who had previously met with a genetic counselor. It is impossible to tell if their scores are lower, but increased from a baseline empowerment score. After condensing responses to the GCOS-24 portion of the survey into three main categories of agree, disagree, and neutral, higher than 50% similarity was seen on multiple questions. Participants had the most similarity in the domain of Hope and the least similarity in the domain of Emotional Regulation. The majority of participants (12/13) agreed with the statement “I am hopeful that my children can look forward to a rewarding family life” and participants were the most dissimilar in their responses to the statement “I know how to get the non-medical help I/my family need(s) (e.g., educational, financial social support)”. Five participants agreed, one participant was neutral, and seven participants disagreed with this statement.

Caregiver strain ranged from 0 to 7 with an average of 2.86 and when compared to empowerment scores, there was a negligible correlation between the two ($r = -0.0900$). It is worth noting that caregiver strain can be measured through more validated and in-depth measures and additional research would need to be completed to determine any true correlation between caregiver strain and empowerment score.

When analyzing satisfaction with appointment timing, there was no statistical difference in empowerment scores between individuals who were "happy/okay" and "neutral" regarding the amount of time they waited for an appointment ($p = 0.5210$). Zero participants stated they were "unhappy" with the length they waited for their appointment, and the amount of time between receiving their ASD diagnosis and being seen for an evaluation in the Division of Genetic and Genomic Medicine.

4.0 Research Significance to Genetic Counseling and Public Health

The construct of empowerment was made specifically for clinical genetics services. The development of the GCOS-24 made it possible to measure empowerment in individuals who are receiving genetic counseling (M McAllister et al., 2011). Being able to track this value in patient cohorts has the potential to shed light on areas where patients require more support. Patients with autism spectrum disorder are known to experience a diagnostic odyssey when it comes to receiving a clinical diagnosis, as well as finding a potential underlying genetic cause (Lappé et al., 2018). Lower scores in certain domains could help the genetic counselor better evaluate where specific patients need assistance.

The prevalence of ASD is increasing (Maenner et al., 2021) and as patients become more aware of genetic testing and genetic counseling, the referrals to genetics services will continue to increase. Previous studies have reported that many parents have questions and misunderstandings about genetic testing for ASD and the process (Giarelli & Reiff, 2015; Zhao et al., 2021), but also wished their genetic counselor was more up-to-date on available tests and research, possible etiologies, and basic features of ASD (Selkirk et al., 2009). Genetic counselors need to be prepared to address the patient/family's concerns and tailor content. Having these individualized discussions can mean the difference between a poor experience and a well-served patient.

The information from this study also has important implications for public health. With ASD being a common referral reason or symptom of individuals referred for a genetics evaluation, genetic counselors have the opportunity to play a larger role as a public health official. A main part of genetic counseling is ensuring families are aware of support services and resources. The results from this study indicated that the majority of participants did not know how to gain access

to these resources. Genetic counselors have the ability to connect families with local resources and services that will benefit the patient and the entire community. As more patients utilize these resources it will increase awareness of ASD and the availability of resources in general.

According to the CDC, the average age of diagnosis for ASD is about four and a half years old, although 42% of individuals with ASD received a developmental evaluation by age three (Centers for Disease Control and Prevention, August 27, 2019). The average age of diagnosis in this study was three years and four months and the majority of individuals were diagnosed before age five. This means the individuals in this study received a developmental evaluation earlier than more than half of the general population with ASD. This is relevant to both the realm of genetic counseling and public health because it shows that genetic counselors are often meeting patients earlier on in their diagnosis. By establishing this relationship earlier, it allows the counselor to assist the family in their understanding of the possible etiologies of the patient's ASD and hopefully set realistic expectations for any known prognosis.

5.0 Public Health Essay: Analysis of Comorbidities in the Autism Population

5.1 Background

The National Institute of Child Health and Human Development, under the NIH, created the Autism Center of Excellence (ACE) Program in order to support and fund large scale studies on autism spectrum disorder. The groups leading the study in turn provide the data to the National Database for Autism Research (National Institute of Child Health and Human Development). The main goal of this program is to collect features of ASD and find causes and treatments for the disorder.

The aim of this analysis is to use a data set provided by the National Institute of Mental Health (NIMH) to determine trends of features and comorbidities in the provided ASD population. The findings from this analysis will be helpful in future studies examining the change in patient empowerment in families of a child with autism after receiving genetic counseling, and any correlation between specific features and comorbidities. The analysis informed the survey in the larger study to ensure a comprehensive list of specialties are included in the survey questions. The goal of the study is to determine the number of participants that meet the definition of children with medical complexities (CMC). There is no current definition of CMC. This study will use the definition described by Cohen et al. (Cohen et al., 2011). Having the analysis of the data provided by the NDA will allow for comparison between this study and the general population.

5.1.1 Research Questions

The research questions for this study were as follows:

1. Which, if any, comorbidities/features are the most common?
2. Is there a body system(s) that has a higher rate of involvement?
3. Out of the provided conditions, which one was most commonly selected?

5.2 Children with Special Healthcare Needs

Children with Special Healthcare Needs is defined as “those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally.” Each year the Health Resources and Services Administration’s (HRSA) Maternal and Child Health Bureau (MCHB) distributes the National Survey of Children’s Health (NSCH). The survey includes the Children with Special Healthcare Needs (CSHCN) Screening and is designed to collect information on the general health and healthcare needs of children between the ages of newborn to 17 (Cohen et al., 2011; Maternal and Child Health Bureau, 2020). The survey allows for assessment of all children who meet CSHCN criteria across the areas of prescription medication, use of services, specialized therapies, and functional difficulties due to an ongoing condition (Data Resource Center, 2012).

The most recent data available from the NSCH states that 18.5% of children in the United States had a special healthcare need varying in severity. The survey reported more than 20 different conditions with autism in the top 10 prevalence percentages at 13% (Maternal and Child Health

Bureau, 2020). The data also showed that 42.7% of CSHCN lived in a medical home, had higher rates of emergency room visits, were more likely to need healthcare coordination on a weekly basis, and were more likely to miss greater than seven days of school due to illness or injury.

5.2.1 Child with Medical Complexity

A child or children with medical complexity (CMC) is a subset of the category of CSHCN. CMC are considered the more severe or medically fragile group of CSHCN (Cohen et al., 2011). While there is no set definition for CMC, the most widely recognized is provided by Cohen et al. in an article published in 2011 that set out to create a framework in which to identify and define CMC. They propose a framework that includes four domains: needs, chronic conditions, functional limitations, and healthcare use. Needs, refers to the many healthcare service needs required by CMC. Chronic conditions are included because CMC have one or more diagnosed or undiagnosed chronic clinical conditions. In CMC, functional limitations are limitations to body structure, function, or performance that are considered to be severe. Healthcare use refers to the high utilization of healthcare resources throughout their life time (Cohen et al., 2011).

Cohen et al. discuss that, due to advancements in healthcare, individuals who are CSHCN and CMC are increasing in number because they are more likely to live into older ages. A better understand of CMC will lead to more informed avenues of care.

5.3 NIH Autism Center of Excellence Program

The Autism Center of Excellence (ACE) Program was created in 2017 by the National Institutes of Health (NIH). The goal of the ACE Program was to create a way for participating facilities to collaborate and contribute to an autism spectrum disorder dataset. The dataset is housed in National Institute of Mental Health (NIMH) Data Archive (NDA) and includes the National Database for Autism Research (NDAR). Participating ACE centers, located throughout the United States, conduct multidisciplinary studies in order to increase knowledge on potential causes and treatments of ASD (National Institute of Child Health and Human Development).

5.4 Methods

The data set was obtained from the National Institute of Mental Health Data Archive (NDA). Permission to access the data set was requested and provided through the University of Pittsburgh and the NDA. This provided access to 23 different studies involving patients with autism. Each of the studies used the ACE subject medical history form to collect information on their participants. In total there are 4,700 respondents being used in this analysis. The ACE subject medical history form was designed by the ACE Program to establish a standard method of information collection for the NDA (National Institute of Mental Health Data Archive). The medical history form was either completed through a medical record review during the original study, or patient-reported during a physical evaluation, depending on the study. The form includes questions for five different domains: diagnostic history, prenatal/early postnatal history, developmental history, current medication, review of systems, and mental health. The inclusion

criteria vary between each study, but one main criterion is that the individual has a diagnosis of autism spectrum disorder. Data was analyzed using Microsoft excel functions and epitools.com was used for all z-tests.

5.5 Results

5.5.1 Sample demographics and review of systems

The data set is comprised of 3241 (69%) males and 1459 (31%) females as seen in Figure 4. The ages of patients involved in the study range from less than one month to 97 years old, with the majority of individuals being 18 years old or less. Responses to the review of systems portion of the data were counted to determine the number of respondents who selected “Yes”, “No”, “NK” (not known), or left blank. “Yes” indicates the individual does have the feature or comorbidity in question, “No” indicates the individual does not, and “NK” is used when the person completing the form (i.e., the caregiver or a member of the study) is unsure of whether or not the individuals has the feature or comorbidity in question. The number of responses left blank for each feature was subtracted from the total number of respondents to create the net response. The percentage of respondents that selected “Yes” for each feature was determined using the net responses. Features that had higher than a five percent response rate of “Yes” were included in the analysis. Figure 5 shows that 12 of the 51 features meet this criterion. The feature with the highest percentage of “Yes” responses was for Birthmarks (e.g., café-au-lait spots, white spots) with 23%, followed by eczema with 21.67%, and sleep disrupted patterns with 20.03%.

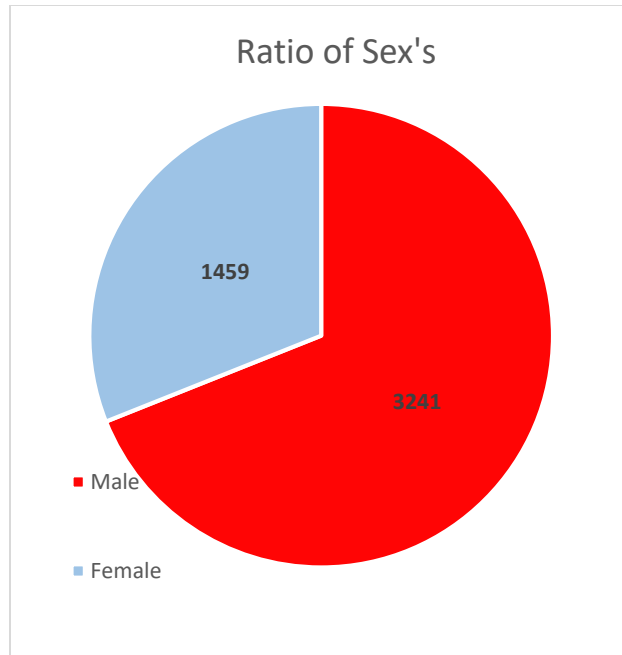


Figure 4: Ratio of Sexes

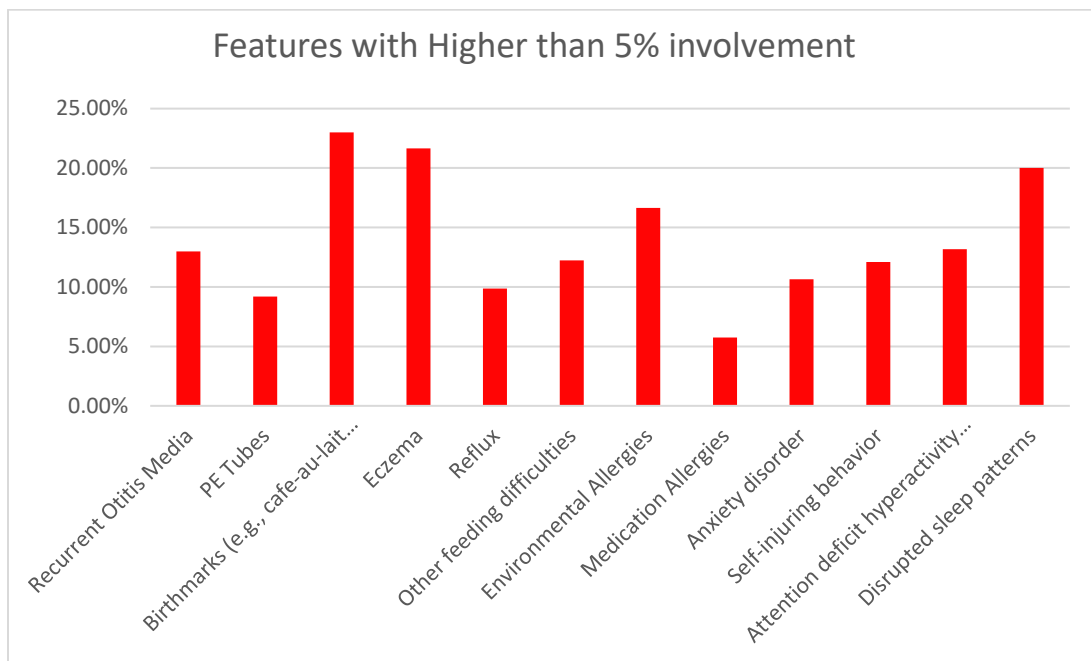


Figure 5: Features with Higher than 5% Involvement

5.5.2 Sample and general public comparisons

The percentage of “Yes” responses for each feature above 5% was compared to the general population prevalence and a z-test was used to determine statistical significance. Table 6 shows the general population prevalence, the z-value, and the p-value for each feature. The z-test could not be completed on three of the features due to an inability to locate literature listing an accepted prevalence in the general population. Based on the z-test, the percentage for nine of the 12 features were determined to be statistically significant in this population.

Table 7: Comparison of Features Between ACE Sample and General Population

Feature	General Population	Data Set	Sample Size	95% CI	z-Value	p-Value
RAOM	11.80%	12.98%	4699	0.1202 - 0.1394	2.50	0.0122
PE Tubes	8.90%	9.20%	4306	0.9119 - 0.9281	6.30	<0.0001
Birthmarks	unable to determine					
Eczema	17.10%	21.67%	3803	0.2036 - 0.2298	7.50	<0.0001
GERD	20-60% (used 40 as average)	9.87%	4700	0.9838 - 0.9902	82.10	0
Feeding Difficulties	20-50% (used 35 as average)	12.24%	4274	0.1126 - 0.1322	31.20	0
Environmental Allergies	41.70%	16.64%	4700	0.1558 - 0.177	34.80	0
Medication Allergy	10%	5.74%	4700	0.0508 - 0.064	9.70	0
Anxiety Disorder	19.10%	10.63%	3404	0.0959 - 0.1167	12.60	0
Self-Injury	unable to determine					
ADHD	9.40%	13.17%	3879	0.1211 - 0.1423	8.00	<0.0001
Disrupted Sleep Patters	unable to determine					

Figure 6 shows the “Yes” response rate for each body system included in the survey. The body system with the highest percentage of “Yes” responses out of the net responses was skin with 14.76%, followed by ears with 9.70%, and gastrointestinal (GI) with 8.13%. The ACE subject

medical history form included a choice of five genetic disorders that respondents could select as a diagnosis for the individual. 202 out of the total 4,700 respondents selected one of the genetic disorders. Figure 7 shows the distribution of each genetic disorder. The disorder with the highest number of respondents is tuberous sclerosis with a total of 161 individuals. Down syndrome had the second greatest number of respondents with 31. Rett syndrome was selected as a diagnosis by five respondents, Fragile X syndrome was selected four times, and Neurofibromatosis type 1 was selected once.

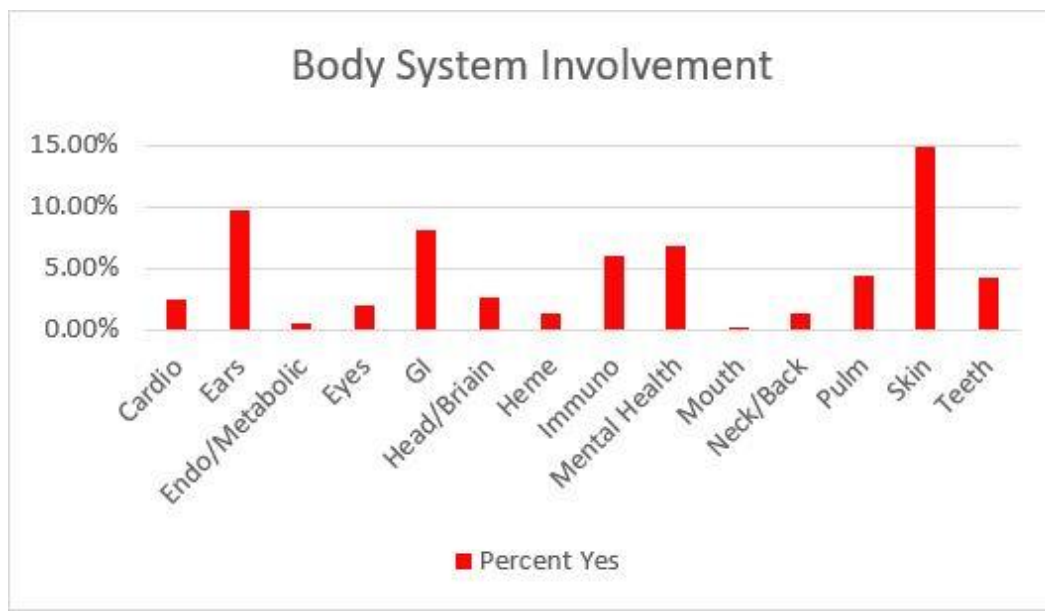


Figure 6: Body System Involvement

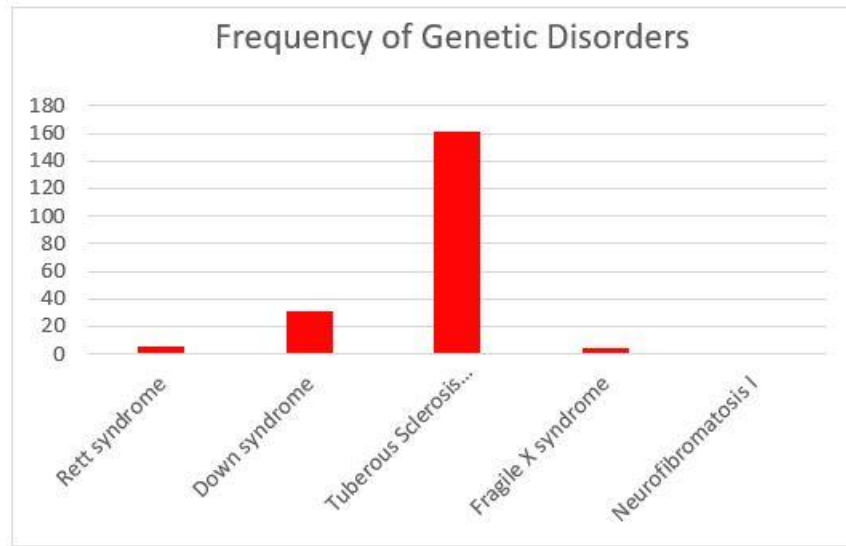


Figure 7: Frequency of Genetic Disorders

5.6 Conclusions and Implications

The data provided had a larger proportion of males, which is to be expected. In the general population, women are four times less likely to receive a diagnosis of autism, even when diagnostic criteria is met (Loomes et al., 2017). Due to this, the majority of data available characterizes the phenotype of males with autism. In order for the data to be the most widely applicable, it is important to take this disproportion of the sexes into consideration when proposing causes and treatments for autism.

The analysis of this dataset highlights the features or comorbidities that were most common (had a higher than 5%) in this population. This information can help gear treatments or services toward the areas that are most often impacted in this patient population. This information is also helpful to consider when giving a diagnosis of autism to a patient. The phenotype for autism is known to be widely variable, so expanding on this phenotype could provide more information for

families whose child does not fit a typical? diagnosis of autism. The p-value provided by the z-test performed on each of these features indicated that they were statistically significant values. The results from this study are congruent with what has been previously reported. Among the features and comorbidities with higher than 5% involvement in this study, Hodges et al. list sleep disorders, gastrointestinal disorders, anxiety, and ADHD as conditions to that can commonly occur with ASD (Hodges et al., 2020).

One limitation to this study is in the interpretation of the review of systems questions. For example, the feature with the highest “Yes” response rate was Birthmarks (e.g. café-au-lait spots, white spots). Some respondents may have selected “Yes” assuming this applied to freckles or normal variations in skin pigmentation, while others may have only thought to select “Yes” if their child had unusual skin pigmentation such as multiple café-au-lait macules. The difference in interpretation could impact the “Yes” response rate for each feature, and in turn affect the body systems that showed the highest percentage of “Yes” responses.

Differences in the way the data was collected is another limitation to the study. The data set is derived from a compilation of 23 studies. In some of these studies, the survey was completed by a medical record review. In others, it was completed by verbal report of the patient’s caregiver. Different collection methods across studies could not only impact question interpretation, but also general understanding of medical terminology.

Another limitation to the interpretation of these results lies in the responses to which genetic disorder was selected most often by the respondents. It is not clear why these five conditions were chosen, however, it is worth noting that one of the 23 studies contributing data was to solely examine autism in a tuberous sclerosis population. Of the 4,700 respondents, 202

responded to the genetic disorder portion of the ACE subject medical history form, 161 of them selected tuberous sclerosis as the patient's genetic diagnosis.

Appendix A IRB Approval



EXEMPT DETERMINATION

Date:	March 10, 2022
IRB:	STUDY21110166
PI:	Julia Knapo
Title:	Assessing patient empowerment in caregivers of patients with autism spectrum disorder
Funding:	None

The Institutional Review Board reviewed and determined the above referenced study meets the regulatory requirements for exempt research under 45 CFR 46.104.

Determination Documentation




Determination Date:	3/10/2022
Exempt Category:	(2)(i) Tests, surveys, interviews, or observation (non-identifiable)
Determinations:	
Approved Documents:	<ul style="list-style-type: none">• Survey QuestionsV2_Version_0.07.docx, Category: Data Collection;• Recruitment letter_Version_0.04.docx, Category: Recruitment Materials;

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, [Amy Fuhrman](#).

Please take a moment to complete our [Satisfaction Survey](#) as we appreciate your feedback.


Appendix B IRB Request: Public Health Essay


Access to NIMH Data Archive

 3  

Any help would be greatly appreciated!

Julia Knapo (she/her/hers)
Genetic Counselor Assistant
UPMC Children's Hospital of Pittsburgh
MSGC, MPH Student
University of Pittsburgh,
Graduate School of Public Health
Class of 2022

 See 2 more messages





**Barone, Jean Marie**
Mon 10/18/2021 4:04 PM

To: Knapo, Julia Marilyn

If you will be getting de-identified data from the archive, then you wouldn't need a submission to the IRB unless required by NIMH. You can upload this message in place of the IRB approval letter.

Jeannie

...

    ...

Appendix C Survey Recruitment Letter/Email

Hello,

My name is Julia Knapo and I am a second-year genetic counseling student at the University of Pittsburgh Graduate School of Public Health. I am reaching out to you about an opportunity to participate in a research study for my graduate thesis. The goal of this study is to gather information from the caregivers of an individual with autism to assess the empowerment of parents and caregivers of patients with autism. Participating in the study would involve taking a brief (5-10 mins) survey before your upcoming appointment in genetics. The responses to the survey will be anonymous and there will be no data saved that would be able to track your responses back to you. There are no foreseeable risks associated the study and there will not be any direct benefits to you either. Whether or not you choose to participate in the study will not impact your care and there will not be any financial compensation.

Participation is voluntary and you may withdrawal at any time by contacting me. The link below will take you to the survey. The survey can be taken on any computer or mobile device that has access to the internet. Scanning the QR code will take you to the survey as well.

https://pitt.co1.qualtrics.com/jfe/form/SV_7WFczPSjy2foGai



Please do not hesitate to contact me with any questions or concerns. I can be reached at jmk318@pitt.edu

Thank you for your time and consideration of this opportunity,

Julia Knapo

Appendix D Survey

By clicking “Continue” you consent to participation in this graduate thesis research study. Your participation is voluntary. There will be no change to the patient's care or treatment, regardless of your choice to participate. Participation will involve taking a survey before your upcoming genetic counseling session. The survey should take 5-10 mins to complete. You are able to quit the study at any time by not finishing the survey. The only potential risk associated with participation is the time burden of taking the survey. Results of the study will not be shared with you directly. If the results from the study are published, all information will remain de-identified. Your results will not be able to be traced back to you. There is no financial payment for participating. There are no invasive or non-invasive procedures, or collections of a sample needed for this study.

☐ Continue

▼  Display this question

If By clicking “Continue” you consent to participation in this graduate thesis research study. Your... Continue Is Selected

▼  Skip to

End of Survey If No Is Selected

Are you, the caregiver, 18 years of age or older?

☐ Yes

☐ No

How are you related to the patient?

☐ Mother

☐ Father

☐ Grandparent

☐ Legal Guardian

☐ Prefer to self-describe

What is the patient's gender?

☐ Male

☐ Female

☐ Non-binary

☐ Transgender Male

☐ Transgender Female

☐ Prefer to self describe

Skip to

End of Survey if No Is Selected

At the time you are taking this survey, is the patient younger than 18 years old (Age 0-17)

☐ Yes

☐ No

What is the patient's birth month and year?
(Please enter the month as a two digit number. For example January would be 01, November would be 11. Please enter the year as a four digit number, for example 2022)

★ Month

★ Year

What is the patient's ethnicity? (Select all that apply):

☐ American Indian

☐ Asian

☐ Black or African American

☐ Hispanic or Latino

☐ Native Hawaiian or other Pacific Islander

☐ White

☐ Prefer to self-describe

When was the patient diagnosed with autism?
(Please enter the month as a two digit number. For example January would be 01, November would be 11. Please enter the year as a four digit number, for example 2022)

★ Month

★ Year

Skip to

Have you ever met with a genetic coun... if No Is Selected

Skip to

Have you ever met with a genetic coun... if Unsure Is Selected

Has the patient ever had genetic testing?

☐ Yes

☐ No

☐ Unsure

▼  Display this question

If Has the patient ever had genetic testing? Yes Is Selected

What kind of genetic testing has the patient had (select all that apply)?

- ☐ Chromosome Microarray (may have been called SNP Array)
- ☐ Whole Exome Sequencing
- ☐ Panel Testing
- ☐ Karyotype
- ☐ Fragile X
- ☐ I'm not sure
- ☐ Other

Do you have results of the genetic testing?

- ☐ Yes
- ☐ No

▼  Display this question

If Do you have results of the genetic testing? Yes Is Selected

What were the results of their genetic testing? (Select all that apply)

- ☐ Normal/Negative
- ☐ Fragile X
- ☐ Rett Syndrome
- ☐ Angelman Syndrome
- ☐ Williams Syndrome
- ☐ Prader-Willi Syndrome
- ☐ Cowden Syndrome (PTEN related disorder)
- ☐ Tuberous sclerosis (TSC)
- ☐ A Microdeletion or Microduplication Syndrome
- ☐ Variant of uncertain significance or inconclusive result
- ☐ I'm not sure
- ☐ Other

 Skip destination

 Skip destination

Have you ever met with a genetic counselor to discuss the patient's health concerns?

****A genetic counselor is a healthcare professional that usually collects information about family history and medical history to help someone understand their chance of having a genetic condition. They can help explain genetic concepts, genetic testing, and the results of testing****

- ☐ Yes
- ☐ No
- ☐ Unsure

▼  Display this question

If Have you ever met with a genetic counselor to discuss the patient's health concerns? **A genetic... Yes Is Selected

How many times have you met with a genetic counselor to discuss the patients health concerns?

- ☐ one (1) time
- ☐ two (2) times
- ☐ three (3) times
- ☐ four (4) or more times

When is the patients upcoming genetics appointment scheduled?

(Please enter the month as a two digit number. For example January would be 01, November would be 11. Please enter the year as a four digit number, for example 2022)

★ Month

★ Year

When it came to scheduling the patient's appointment, did you take the first available appointment?

☐ **Yes**

☐ Yes

☐ **No (select reasoning)**

☐ I chose to wait longer

☐ That date did not work for me

☐ Other, please specify:

Regarding your upcoming appointment, which of the following applies to you?

- ☐ I'm happy/okay with the amount of time we are waiting
- ☐ I'm unhappy about the amount of time we are waiting
- ☐ I feel neutral about the amount of time we are waiting

Regarding the time between the patient's diagnosis and your upcoming appointment in genetics, which of the following applies to you?

- ☐ I'm happy with the amount of time between the patient's diagnosis and our appointment
- ☐ I'm unhappy with the amount of time between the patient's diagnosis and our appointment
- ☐ I feel neutral about the amount of time between the patient's diagnosis and our appointment

Please select all of the specialties/departments that the patient has seen or will see more than one time.

- ☐ Cardiology – deals with heart conditions
- ☐ Cleft-Craniofacial – deals with issues with the shape or development of the skull, cleft palate, etc.
- ☐ Endocrinology – deals with hormone issues, issues with growth, metabolism, and puberty
- ☐ ENT or Otolaryngology – deals with ear, nose, and throat conditions
- ☐ Gastroenterology – deals with problems in digesting food such as constipation, diarrhea, nausea
- ☐ Hematology/Oncology – deals with blood or bleeding disorders and cancers
- ☐ Neurology or the Child Development Unit (CDU) – deals with conditions of the head or brain like headaches, migraines, seizures, and developmental delay
- ☐ Rheumatology – deals with autoimmune or autoinflammatory conditions, or problems such as joint pain, joint swelling, and recurrent fevers
- ☐ Pulmonology – deals with lung or breathing issues

Please answer the next few questions about yourself, and not your child/the patient.

Using the scale, please choose the option that best fits how much you agree with the statement on the left. If the statement does not apply to you, please choose "Neither Agree or Disagree"

	Strongly Disagree	Disagree	Slightly Disagree	Neither Agree nor Disagree	Slightly Agree	Agree	Strongly Agree
I can change how this condition affects my family in the future	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I don't know where to go to get the medical help I/my family need(s)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I can control how this condition affects my family	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I am able to cope with having this condition in my family	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I know how to get the non-medical help I/my family need(s) (e.g., educational, financial, social support)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

I can explain what the condition means to people in my family who may need to know	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I don't know what I can do to change how this condition affects me/my children	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I am powerless to do anything about this condition in my family	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
When I think about the condition in my family, I get upset	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Having this condition in my family makes me feel anxious	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I feel guilty because I (might have) passed this condition on to my children	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I can see that good things have come from having this condition in my family	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I feel positive about the future	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I am hopeful that my children can look forward to a rewarding family life	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I am able to make plans for the future	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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