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Editorial Note

South Asian Journal of Management Research (SAJMR), is a scholarly journal that publishes

scientific research on the theory and practice of management. All management, computer

science, environmental science related issues relating to strategy, entrepreneurship, innovation,

technology, and organizations are covered by the journal, along with all business-related

functional areas like accounting, finance, information systems, marketing, and operations. The

research presented in these articles contributes to our understanding of critical issues and offers

valuable insights for policymakers, practitioners, and researchers. Authors are invited to publish

novel, original, empirical, and high quality research work pertaining to the recent developments

& practices in all areas and disciplined.

Cross-functional, multidisciplinary research that reflects the diversity of the management science

professions is also encouraged, the articles are generally based on the core disciplines of

computer science, economics, environmental science, mathematics, psychology, sociology, and

statistics. The journal's focus includes managerial issues in a variety of organizational contexts,

including for profit and nonprofit businesses, organizations from the public and private sectors,

and formal and informal networks of people. Theoretical, experimental (in the field or the lab),

and empirical contributions are all welcome. The journal will continue to disseminate knowledge

and publish high-quality research so that we may all benefit from it.

Dr. Pooja M. Patil

Editor

South Asian Journal of Management Research (SAJMR)

March, 2024

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College of Urban Development and Engineering,

Ethiopian Civil Service University, Addis Ababa, Ethiopia

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Behavioral Health Implications of Autoinflammatory Disease Assessment

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Abstract

Yao's Syndrome is a rare genetic disorder characterized by changes to the NOD2 gene. Due to the rarity of the condition, we do not have much research on the topic. This decreases the knowledge about the disease for both healthcare providers, and patients with the condition. Because there is not a lot of research/knowledge, medical students do not learn about this which is challenging for patients because they are going to professionals who lack knowledge which leads to patients feeling isolated. This is a qualitative cohort study using a survey using the Likert scaling method. Key findings of the survey showed that the average individual with this condition struggles with daily pain, anxiety, and feelings of isolation and lack of support due to this illness. It is our hope that with this knowledge moving forward, we are able to advocate for patients to gain the support that they need, and that we can provide training on this topic for medical professionals. In doing so, we plan to use this to help better prepare medical professionals and equip them with the knowledge of rare disorders to better serve their patients in a well-rounded manner, treating patients as more than just a statistic.

Keywords: Yao's Syndrome, Autoinflammatory Disorder, Chronic Illness, Genetics

Introduction

Yao's Syndrome is a chronic and recurrent autoinflammatory disease characterized by episodic fevers and multiorgan inflammation. It manifests with severe pain in joints, abdomen, chest, and other body regions. Its global prevalence is estimated at 1 in 100000 to 1 in 100000 individuals. The inheritance pattern appears complex, with no identified singular genetic cause, although the NOD2 gene is implicated, and familial cases have been reported. This syndrome affects both genders and spans various age groups, through it exhibits a predilection for females. Treatment primarily focuses on alleviating pain and inflammation, commonly involving NSAIDS and Corticosteroids; However, personalized therapeutic approaches are recommended to address individual patient needs (Yao Syndrome: Medlineplus Genetics (N.D). 17 patients with Yao's syndrome or a related NOD2 mutation disorder were surveyed on a variety of questions involving their disease and how it impacts their daily life. Results of the survey demonstrated the urgent need for increase understanding and awareness of autoinflammatory conditions, as patients frequently endure years of pain and symptoms without proper diagnosis or treatment.

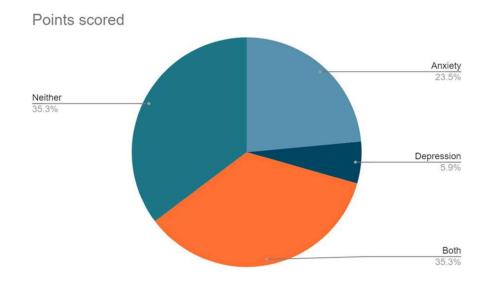
Literature Review

Given the rarity of Yao's disease and related NOD2 mutation disorders, many patients experience extended diagnostic delays, sometimes spanning decades. The survey revealed a wide spectrum in the duration of time between symptom onset and diagnosis, ranging from a mere two months to as long as 56 years. However, more than half of the surveyed patients remained undiagnosed for a minimum of ten years. This underscores the importance for heightened awareness and education regarding autoinflammatory diseases, ensuring that prospective patients do not endure significant diagnostic delays and subsequently receive timely treatment for their symptoms. Patients were also surveyed regarding the extent to which Yao's or a related NOD2 mutation syndrome. Affects their daily mobility, using a 1-5 scale where 1 signifies no impact and 5 signifies severe impact. The average rating for the severity of impact was 3.8/5. None of the survey respondents related the impact as less than 3/5 in severity, while over half them rated as 4 or 5/5 in severity. This emphasizes the urgent need for greater disease awareness, given that despite its rarity, those affected endure significant pain and disruptions in their lives. Furthermore, patients were surveyed to gauge the impact of their autoinflammatory disease on their quality of life, using the same 1-5 scale. The average score was 4.2/5, with none of the participats rating their disease impact on quality of life as less than 3/5 in severity. Additionally, over one-third of participants rated the impact as 5/5. Lastly, patients were surveyed regarding the influence of their disease on their mental health, employing the same 1-5 scale where 5 indicate a severe impact. The average score was 3.8/5, and only one out of the 17 participants rated the

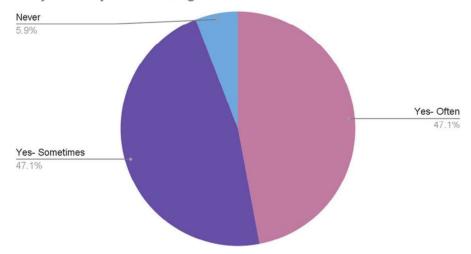
diseases on their mental health, as less than 3/5 in severity. This highlights that autoinflammatory diseases such as Yao's syndrome not only impact patients physically but also affect their mental wellbeing subsequently; participants were queried in the survey about comorbid diagnoses of anxiety and/or depression. Approximately one-third reported both anxiety and depression, while another third reported neither diagnosis, and the remaining third indicated either, anxiety or depression. Intriguingly, these finding did not align with responses from the previous question concerning the impact of autoinflammatory disease on mental health. While 16 out of 17 participants reported that their autoinflammatory disease affected their mental health to a severity of at least 3/5 on the rating scale, only 11 out of the 17 participants reported concurrent mental health diagnoses of anxiety and/or depression. While the cause of this discrepancy remains unclear, it indicates that mental health in underrecognised, resulting in under diagnosis and under treatment. Given the impact of autoinflammatory diseases on mental well-being enhancing disease awareness and understanding should also encompass improved integration of mental health treatment. Participants were then asked about how their autoinflammatory disease impacts their medication adherence and usage on 1-5 scale with 1 being no impact and 5 being severe impact. Five participants (29.4%) answered 1 for the question while six (35.3%) answered 3. The rest of the responses were scattered with 2 having the next most responses at 3 (17.6%). The responses are surprising in how five participants answered 1, so the question could have been worded poorly where the participants weren't fully sure what was being asked. Due to the question being asked in non-layman's terms, the question then arises if the question was not completely understood due to the use of the term "Medication Adherence." The researchers came to the conclusion that based on the way the question was worded; this had a large impact on the reliability of the question. In the future, the research team will go to greater lengths to screen questions, and word them in a way that is of universal understanding to those not in the healthcare field, As we all are, in order to increase comprehension and accessibility when answering the assessment. This question in particular yielded surprising results, as half of respondents answered that they feel they are always taking too little, or too much medication in order to treat their disease. One outlier responded never to this question. The next question asked participants to rate on the same 1-5 scale how daily pain impacts their quality of life an astonishing seven (41.2%) participants responded with a 5, and the next highest response was six (35.5%) people responding with a 4. This is not surprising considering the severe and painful side effects of Yao's syndrome. These results contradict the previous question. It was surprising how many people didn't believe their disease affected their medication adherence and usage where there could be larger implications of how people may not be receiving the pain medication they need. Of course, again, it's important to note that the previous question could have been worded poorly, which could have explained the contradictory results. Pain was assessed again in the next question. Participants were asked to rank their daily pain with an autoinflamatory disease on a scale from 1-10 with 1 being minimal pain and 10 being extreme pain. Six people (35.5%) responded with a 7 and five people (29.4%) responded with and 8. These results further clarify the previous question because the participants were able to quantify the daily pain they experience and how it affects their quality of life. These people are in significant pain for most of their lives, and that affects how they are able to live their lives. People were then asked about if they ever worry about taking too much or too little medication to treat symptoms instead of the disease. The results were split fairly evenly with eight people (47.1%) saying that they often worry and eight people saying they sometimes worry. It is not known which side participants were leaning towards (either too little or too much), so this could be something that could be assessed in future research. What is important to not however is that a majority of participants answered that they do worry in some manner about their medication usage taking too much medication can be worrisome because the side effects of being on too many medication can negatively harm a person's health or the person may become addicted depending on the medication they take. On other hand, taking too little medication can be harmful as well because the person would not be treating their pain symptoms. They would be in constant pain daily, and that could have negative consequences on their social and professional life. The next question connects with the previous question concerning the implications of taking too much or too little medication. People were asked about if they ever worry about job security due to their inflammatory disease causing them to miss work they were asked to respond on a scale from 1-5 with 1 being no impact and 5 being severe impact. Over half of the respondents (58.8% or 10 people) answered with a 5. These results show that many people who have this disease are negatively affected by it in terms of job security. They worry about losing their job due to factors such as immense pain or sudden flares to add onto this point, their worries about taking too much or too little medication could also factor into their worries about retaining their job. If they take too much medication, they may not be able to work properly due to side effects of such as feeling unfocused. Then, if they take too little medication, they wouldn't be able to work because of immense pain they're in. There are many factors that can affect how someone worries about various aspects of their life, and these results show that job security is one of those aspects. Many also fear that during flares, they will deal with brain fog, poor performance at work, and poor attendance due to the negative impact of their health on their occupation. Even when patients are able to attend work, oftentimes they have a fear of underperforming due to the physical limitations of the disease they suffer from. As this type of disease is newly classified, and rare, with not a lot of research on the subject, often those who suffer are misunderstood and suffer in silence. In further analyzing the following question, "Do you ever worry about job security due to your autoinflammatory disease causing you to miss work, on a scale of 1-5, with 1 being no impact and 5 being severe impact?" 10 respondents shared that they would rate their employment as minimally impacted, further shedding light on the disparities that many with disabilities face on a daily basis when it comes to the topic of financial stability and job security. The next question at hand aimed to discuss the feelings of hopelessness that many with autoinflammatory diseases, specifically NOD2 specific mutations, such as Yao's syndrome face. Patients were asked to rate their sense of hopelessness experienced in correlation with this type of systemic diseases. On a scale of 1 to 5, with 5 being the most severe impact, 15 respondents gave a response of 3-5 in severity. 29% of respondents shared a score of 5, heavily implicating Yao's syndrome as a disease that is not well understood, and that the support needed for patients suffering is often not there, leaving in its absence a feeling of severe hopelessness. In asking about implications of the NOD2 related gene mutation autoinflammatory syndromes, and the impact of the pain on mental health, the assessment revealed that as pain worsens, so does the mental health of 15/17 patients. The intense amount of pain felt daily by patients with this type of rare disease impacts not only physical health, but has a positive correlative relationship with their mental health status as well. As physical pain worsens, so does mental health. Likewise, on days when pain is not as severe, mental health is not as drastically impacted in a negative manner. The final question on the assessment poses a response on the support felt by those experiencing this type if condition. Out of 17 terms of others being able to understand what they are going through. 5 other respondents shared that they feel adequately supported by their family and friends. 3 respondents shared that they gain adequate support from online support groups. 2 respondents gain the support needed from their health care providers (Rheumatologists). Rheumatologists are leading specialist type that primarily support and treat those with type of disease; through the patients see many other specialists as well for symptom management, as this is a disease that impacts the entire body. One respondent shared that they gain the needed support from a therapist. One outlier did not answer the question.

Methodology:

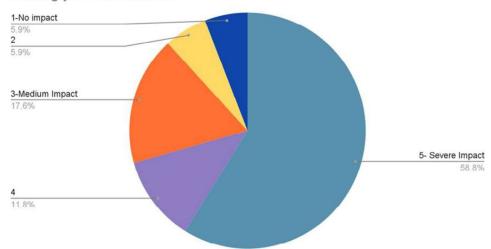
The BHIADA was a qualitative study in which patients with Yaos's Syndrome (NOD2 autoinflammatory disease) actively chose to participate in the survey shared with them. The participation in this study was not compensation based. The survey was based on a Likert Scale with scoring from 1-5 in all areas, except for the area of pain score, which went to 10 with one being the least impact, and 5 being the greatest of impacts. Figures



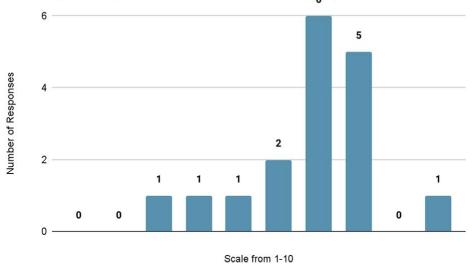
Do you worry about taking too much medication of too little?



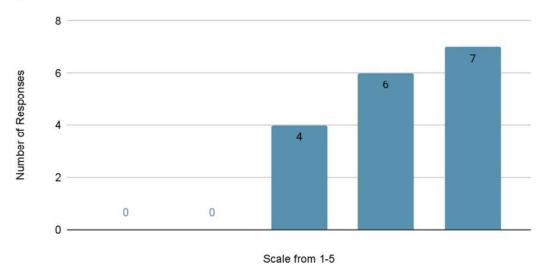
Do you ever worry about job security due to your autoinflammatory disease causing you to miss work?



Ranking of Daily Pain with an Autoinflammatory Disease



Severity of Impact of Autoinflammatory Disease on Quality of Life



Conclusion:

In conclusion, Yao's syndrome and other autoinflammatory syndrome and diseases are rare, and rarely researched due to the small number of individuals in the population impacted by these diseases. As the research has stated, this type of syndrome not only impacts the physical health of those impacted on a daily basis, but their mental and behavioral health as well. Many are left feeling not only in pain, but also undersupported and hopeless. Due to the rare nature of this disease, many go half of their lives in pain before being diagnosed, as there are very few specialists in the world equipped with the knowledge and training to be aware of this type of genetic abnormality, let alone to diagnose it. In shedding light on the behavioral health impact of autoinflammatory diseases life Yao's syndrome, it is our hope to spread awareness and broaden the research done in order to serve this underserved and rate community of patients.

REFERENCES

Our team would like to thank all the people fighting this rare disease who gave of themselves, in both time and input, in order to further our research and give back to those struggling with Yao's Syndrome.

References

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