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AI in Healthcare

**FOXG1-GPT: A ChatGPT-Powered Assistant for Supporting Parents and Caregivers of FOXG1 Syndrome Patients**

Video Recording: https://utexas.hosted.panopto.com/Panopto/Pages/Viewer.aspx?id=947bfc21-4d15-48ca-990c-b2440131ef1e

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FOXG1 GPT assistant aims to help parents and caregivers of FOXG1 syndrome patients

ABSTRACT

FOXG1 syndrome is a rare genetic condition associated with neurodevelopmental challenges, requiring caregivers and parents to navigate complex medical, genetic, and management information. We introduce FOXG1-GPT, a ChatGPT-based conversational assistant designed to provide accessible, accurate, and empathetic support. FOXG1-GPT is tailored to address questions about FOXG1 syndrome, including its genetic underpinnings, effective management strategies, and medication-related concerns. This paper describes the design, functionality, and potential applications of FOXG1-GPT, emphasizing its role in empowering caregivers and bridging gaps in patient care. FOXG1 syndrome is a very complex disease. Patients usually suffer from Seizures, Sleep disturbances and issues, Cortical vision impairment, low muscle tone, acid reflux issues, failure to thrive, constipation. Due to this complex nature of disease, managing disease is very difficult. Parents are often left helpless due to lack of knowledge about this disease in healthcare industry among doctors and due to lack of medicines that work. FOXG1 research foundation is a non-profit organization working to find a cure for this disease. This Chatbot aims to use all publicly available information about the disease be it research papers that are published, using transcripts from the videos available on YouTube about this disease, as well as any public information provided by research foundation working on this disease. With this information and power of LLM’s, we are trying to help parents and caregivers of the foxg1 syndrome.

KEYWORDS

FOXG1 syndrome, Seizures, ChatGPT, genetic information, disease management, caregiver support, rare diseases, support

1. Introduction

FOXG1 syndrome is a rare neurodevelopmental disorder caused by mutations in the FOXG1 gene. It presents a spectrum of symptoms, including intellectual disabilities, movement disorders, and seizures. Parents and caregivers of FOXG1 patients often face challenges in accessing reliable information and managing day-to-day care.

Chatbots powered by large language models (LLMs) like ChatGPT have demonstrated potential as tools for disseminating information and providing personalized support. This paper introduces FOXG1-GPT, a specialized chatbot designed to support caregivers and parents of FOXG1 patients by offering disease-specific information, genetic insights, and management advice. The objective of FOXG1-GPT is to enhance the caregiving experience by providing timely, accurate, and empathetic assistance.

2. Background and Related Work

Existing resources for caregivers of rare diseases, such as online forums, support groups, and medical literature, can be fragmented and difficult to navigate. Additionally, parents and caregivers may not have the skill and time to read scientific publications and research papers to deduce the information they need to care for the patient. Recent advancements in artificial intelligence have enabled the creation of domain-specific chatbots capable of addressing these challenges.

Several chatbots have been deployed in healthcare settings to provide patient education, symptom management, and mental health support. However, few, if any, focus on rare genetic conditions like FOXG1 syndrome. FOXG1-GPT aims to fill this gap by leveraging the capabilities of ChatGPT to deliver highly specialized support.

3. System Design

A screenshot of a chat

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**3.1 Architecture** FOXG1-GPT is a GPT assistant app created using ChatGPT 4o model and some specific data sources are fed into this app, including publicly available:

* Medical literature on FOXG1 syndrome
* Input from geneticists, neurologists and doctors
* Curated FAQs from parent support groups

**3.2 Features**

* **Disease Information**: Provides details on FOXG1 syndrome, its symptoms, and progression.
* **Genetic Support**: Answers questions about genetic mutations, inheritance patterns, and testing. Helps parents understand their specific mutation and if there is any correlation between disease type and mutations.
* **Medication Guidance**: Highlights medications commonly prescribed for symptom management, along with potential side effects and contraindications. Also finding medications which work for similar symptoms in other diseases that are worth trying in this disease.
* **Caregiving Strategies**: Offers practical advice on daily care routines, therapy options, and resources for emotional support. Additionally, various government assistance programs available for caring for this disease. Therapy interventions that show promises in helping patients.
* **Empathetic Interaction**: Employs techniques to ensure responses are compassionate and sensitive.

**4. Methodology**

**4.1 Data Collection**

Reference data for FOXG1-GPT includes publicly available:

* Peer-reviewed research articles on FOXG1 syndrome
* Patient case studies
* Expert interviews
* Community-driven insights from caregiver forums

**4.2 Evaluation Metrics**

The chatbot’s performance is evaluated using:

* Comparison with validated medical sources.
* Measured through user satisfaction surveys.
* Assessed via caregiver feedback on ease of use and relevance.

**5. Results and Discussion**

Preliminary testing of FOXG1-GPT demonstrates:

* Really good alignment with verified medical sources.
* Caregivers report increased confidence in managing care.
* Reduces time spent searching for information.

Challenges include maintaining up-to-date medical knowledge and ensuring that complex genetic concepts are communicated effectively to a non-specialist audience. Also, there are many private patient support groups exist which contain mass amount of information which is not accessible to this GPT app. If that information is available regarding de-identified parent conversations regarding medicines, seizures, therapy interventions, habits of patients, visual environment adaptations to help with CVI, chatbot will be even more powerful in helping parents and caregivers.

**6. Conclusion and Future Work**

FOXG1-GPT represents a novel application of AI in supporting caregivers of patients with rare diseases. By integrating medical expertise with empathetic communication, the chatbot addresses critical gaps in caregiving resources. Future work will focus on:

* Expanding the knowledge base to include video transcripts for publicly available videos regarding disease, disease management,
* Incorporating feedback loops for continuous improvement.
* De-identified information regarding patient symptoms and medicines

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REFERENCES

[1] The postnatal injection of AAV9-FOXG1 rescues corpus callosum agenesis and other brain deficits in the mouse model of FOXG1 syndrome Shin Jeon1,2,5 ∙ Jaein Park1,5 ∙ Shibi Likhite3,5 ∙ Ji Hwan Moon1,4 ∙ Dongjun Shin1 ∙ Liwen Li1 ∙ Kathrin C. Meyer3 kathrin.meyer@nationwidechildrens.org ∙ Jae W. Lee1 jlee269@buffalo.edu ∙ Soo-Kyung Lee

[2] The clinical and sleep manifestations in children with FOXG1 syndrome Lee-Chin Wong 1 2, Cheng-Hsien Huang 3 4, Wan-Yun Chou 5, Chia-Jui Hsu 6, Wen-Che Tsai 7, Wang-Tso Lee 1 8

[3] Expanding genotype–phenotype correlations in FOXG1 syndrome: results from a patient registry Elise Brimble1\* , Kathryn G. Reyes2 , Kopika Kuhathaas3 , Orrin Devinsky4 , Maura R. Z. Ruzhnikov5 , Xilma R. Ortiz‑Gonzalez6 , Ingrid Schefer7 , Nadia Bahi‑Buisson8 and Heather Olson9 on behalf of the FOXG1 Research Foundation

[4] https://foxg1research.org/foxg1-key-papers

Conference Name:ACM Woodstock conference

Appendix

Few sample interaction with FOXG1-GPT

A screenshot of a medical report

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A screenshot of a cell phone

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A screenshot of a medical information

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A screenshot of a medical document

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