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Original Research Article

Genetic Counselling Empowers Parents of Children with Intellectual Disabilities: A Fragile X Syndrome Perspective

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Abstract

Background: Intellectual disabilities (ID) profoundly affect individuals and their families, leading to financial strain, emotional distress, and limited access to healthcare and education. Fragile X Syndrome (FXS), the most common inherited cause of ID, remains largely underdiagnosed in Indonesia due to limited awareness and resources.
Objective: This study aimed to assess the impact of genetic counselling on parental empowerment, defined as improvement in decision-making ability, emotional regulation, cognitive understanding, and future-oriented hope, as measured by the Genetic Counselling Outcome Scale - 24 (GCOS-24).
Methods: This pre-experimental study employed a before and after counselling. A total of 238 parents of children with ID from four special schools in Jakarta participated. Empowerment was measured using GCOS-24. Purposive sampling was applied. Paired t-tests were conducted to evaluate changes in empowerment scores before and after counselling, while one-way ANOVA was used to assess whether these changes differed based on parental educational level.
Results: The mean GCOS-24 score increased significantly from 106.79 (SD = 16.36) before counselling to 125.11 (SD = 15.42) after counselling ($p < 0.001$). Only 27.3% of parents were aware of genetic disorders, reflecting their limited baseline knowledge. Parents with primary school education showed the greatest improvement in empowerment scores compared to those with higher education levels (F = 4.035, p = 0.008).
Conclusions: Genetic counselling significantly enhanced parent empowerment, as evidenced by increased GCOS-24 scores. These findings emphasize the importance of expanding genetic counselling services and educational initiatives in Indonesia to improve awareness and support for families managing ID.

Keywords: *Fragile X Syndrome; genetic counselling; intellectual disability; parental empowerment; empowerment*
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INTRODUCTION

Intellectual disability (ID) profoundly affects individuals and their families, posing challenges such as financial strain, emotional distress, and limited access to educational and healthcare services. Families must often navigate caregiving responsibilities while balancing work, leading to job insecurity and added pressures on siblings. ID arises from genetic and non-genetic causes, necessitating a multidisciplinary approach for effective management.[1] Among genetic causes, Fragile X Syndrome (FXS) is the most frequently inherited form of ID. FXS results from mutations in the *Fragile X Messenger Ribonucleoprotein* 1 (*FMR1*) gene, essential for neurodevelopment. The prevalence of FXS in Indonesia ranges from 0.9% to 1.9% among individuals with ID, with higher rates observed in those with Autism Spectrum Disorder (ASD).[2,3]

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Notably, a study in a rural Javanese village revealed a high prevalence of 45%, suggesting genetic clustering and the influence of localized genetic factors.[2,4] Genetic counselling plays a crucial role in supporting families of children with ID by providing comprehensive information about the condition, its causes, prognosis, and management options. It also addresses recurrence risks and facilitates parental empowerment of their child's diagnosis.[5,6] The process often includes before and after counselling sessions to address both medical and emotional needs. Studies highlight the importance of continued FXS screening and cascade testing to identify carriers and affected individuals, aiding family planning and clinical decisions.[2,3] However, awareness of FXS in Indonesia remains limited, and access to genetic counselling is hindered by logistical and infrastructural challenges, particularly in rural areas.[7] Early diagnosis and interventions significantly improve outcomes for individuals with ID. Programs such as behavioral therapy, speech therapy, and occupational therapy are most effective when initiated early.[8] Molecular testing, including CGG repeat expansion analysis in the FMR1 gene, remains critical for confirming FXS diagnoses and informing family decisions.[9] Despite advancements in diagnostic technologies like TP-PCR and next-generation sequencing, widespread adoption is limited by funding and technical barriers.[10,11] Parental empowerment of a genetic diagnosis significantly impacts decisions about care, education, and family adaptation. Studies show that parents often struggle with feelings of denial or guilt, affecting their emotional well-being and caregiving approaches.[12,13] Genetic counselling addresses these challenges by providing targeted interventions to improve understanding, correct misconceptions, and offer emotional support. Tools like the Genetic Counselling Outcome Scale (GCOS-24) measure the impact of counselling on emotional regulation, decisional control, and hope, which together represent key dimensions of parental empowerment. Empowerment is defined as the increased ability of parent to make informed decisions, regulate emotions, understand their child's condition, and maintain hope for the future.[14,15] This study investigates the influence of genetic counselling on parental empowerment in families managing FXS in Indonesia. By assessing changes in parental knowledge, emotional adaptation, and decision-making, this research seeks to inform healthcare practitioners and policymakers in developing effective interventions to support affected families.

MATERIALS AND METHODS

Study Design

This pre-experimental study, part of our project on screening FXS, evaluated parental empowerment of children with intellectual disabilities through before counselling and after counselling sessions. This study involved parents of children with intellectual disabilities enrolled in four state special schools (SLBN) across Jakarta: SLBN 3 Central Jakarta, SLBN 10 West Jakarta, SLBN 11 South Jakarta, and SLBN 9 North Jakarta. Inclusion criteria included:

a) parents of boys diagnosed with intellectual disabilities and enrolled in one of the participating SLBN schools, as the study was conducted alongside a FXS screening project that primarily targeted male children.
b) willingness to share their experiences through a questionnaire, and
c) ability to communicate in Bahasa Indonesia.

Exclusion criteria included:

a) parents of children diagnosed with Down syndrome,
b) parents of children with multiple congenital anomalies, and
c) parents who declined to participate.

Sample Size and Sampling Technique

A total of 256 parents were invited for initial screening, as part of our project on screening FXS. The sampling technique used was purposive sampling, ensuring participants were selected based on their relevance to the study while accommodating time and resource constraints. Schools were chosen based on their willingness to participate, and the number of respondents was determined by those available and meeting the study criteria.

Measures

Data collection utilized the Genetic Counselling Outcome Scale-24 (GCOS-24), a validated patient-reported outcomes measure designed specifically for clinical genetics services. The GCOS-24 evaluates empowerment across five key areas: decisional control, cognitive control, behavioral control, emotional regulation, and hope for the future.[16] Each item is rated on a 7-point Likert scale, which captures a range of responses: 1 (Strongly Disagree), 2 (Disagree), 3 (Somewhat Disagree), 4 (Neutral), 5 (Somewhat Agree), 6 (Agree), and 7 (Strongly Agree). Total scores range from 24 to 168, with higher scores indicating greater levels of empowerment.

In addition to GCOS-24, a brief self-report questionnaire was administered prior to counselling to assess participants' baseline knowledge of genetics. This included three simple yes/no questions developed by the research team, addressing: (1) awareness of genetic disorders, (2) understanding that genetic disorders can be inherited, and (3) prior experience with or access to genetic testing. This tool was intended for descriptive purposes and to provide context for interpreting empowerment scores.

The GCOS-24 has shown strong psychometric reliability, exhibiting excellent internal consistency (Cronbach's alpha = 0.87) along with high test-retest reliability (r = 0.86), and sensitivity to change over time.[17] In this study, the questionnaire was translated into Bahasa Indonesia through a forward translation process conducted by a certified sworn translator. To ensure semantic and conceptual equivalence with the original version, a backward translation was then performed by a different certified sworn translator, following international guidelines for cross-cultural adaptation.

Assessments were conducted at two time points: before and after the genetic counselling sessions. Genetic counselling was delivered by trained professionals and

covered key aspects including the etiology, implications, and management of intellectual disabilities, with a focus on Fragile X Syndrome. An increase in GCOS-24 scores after-counselling was interpreted as an improvement in parental empowerment, demonstrating the effectiveness of the counselling sessions.

Ethical Considerations

This study adhered to ethical research standards, ensuring informed consent was obtained from all participants. Ethical approval was granted by the institutional ethics review board at Universitas YARSI (Approval Number: 309/KEP-UY/EA.10/IX/2024). Participant confidentiality was maintained throughout the study, and participation was entirely voluntary.

Data Analysis

Quantitative data analysis was conducted using paired t-tests to compare GCOS-24 scores before and after genetic counselling, providing insights into changes in parental empowerment. An increase in GCOS-24 scores of 10.3 points or more was interpreted as clinically meaningful. This benchmark was defined as the Minimum Clinically Important Difference (MCID) for the scale, indicating as a significant positive change in empowerment as perceived by the participants.[18] It reflects an improvement in key dimensions such as decision-making, emotional regulation, and overall confidence in managing their child's condition following genetic counselling. One-Way ANOVA was employed to evaluate the relationship between parental educational levels and empowerment scores. Post-hoc analysis using Tukey HSD and LSD methods was conducted to identify significant differences between groups. Statistical significance was set at $p < 0.05$.

RESULTS

Parent Demographics

A total of 256 parents were initially invited for the screening process, with 18 excluded for not meeting the inclusion criteria. Among these, 4 parents had children with Down syndrome, and 14 declined to participate. A total of 238 parents met the inclusion criteria and successfully completed the study. The demographic characteristics of the parents are summarized in Table 1. This includes age distribution, gender, educational level, and employment status. The majority of participants were aged 41–50 years (50,4%) and predominantly female (88,7%). Educational levels varied, with 61,3% having completed high school, while 84% were unemployed.

Baseline Knowledge of Genetics

At baseline, 27,3% of participants reported awareness of genetic disorders, but only 29% of these individuals understood that such disorders could be inherited. None of participants children had never undergone genetic testing, reflecting a low level of awareness about genetic testing and its importance. Financial burden was identified as a key barrier, as genetic testing is only available in major capital cities on Java Island and is not covered by national insurance. These findings highlight significant gaps in knowledge about genetics and the urgent need for targeted educational initiatives and improved accessibility to genetic testing services.

GCOS-24 Before Counselling and After Counselling Scores

The effectiveness of genetic counselling was evaluated by comparing GCOS-24 scores before and after the intervention. These results, summarized in Table 2, demonstrate a significant improvement in parental empowerment following genetic counselling. The mean GCOS-24 score before genetic counselling was 106.79 (SD = 16.36), with a range of 43–143. After counselling, the mean score increased to 125.11 (SD = 15.42), ranging from 93–165. The increase in mean scores ($\Delta = 18.32$) was statistically significant ($p < 0.001$), indicating a substantial enhancement in parental empowerment as a direct result of the counselling intervention.

Table 2. Mean GCOS-24 Before Counselling and After Counselling

Variable	Mean	SD
GCOS-24 Before Counselling	106.79	16.36
GCOS-24 After Counselling	125.11	14.42
Change in GCOS-24 Score	18.32	18.48

The GCOS-24 results showed significant improvements in parental empowerment following genetic counselling. Agreement with the statement "I understand the impact of the condition on my child" increased from 55% before counselling to 72.3% after counselling, while clarity about the purpose of attending

Table 1. Demographic characteristics of parents.

Variable	Categories	Total (n =256)	Percentage
Age	< 20 years	5	2.1%
	21-30 years	4	1.7%
	31-40 years	53	22.3%
	41-50 years	120	50.4%
	>50 years	56	23.5%
Gender	Female	211	88.7%
	Male	27	11.3%
Educational Level	Elementary or Lower	31	13%
	Middle School	36	15.1%
	High School	146	61.3%
	Diploma or University	25	10.5%
Employment Status	Unemployed	200	84%
	Part-time	17	7.1%
	Full-time	30	12.6%
Total		**256**	**100%**

Effectiveness of Genetic Counseling Session

Table 3. Summary of Paired T-Test and One-Way ANOVA Results

Analysis Type	Variable/Group	Mean Score	Std. Dev	Test Statistic	df	p-value
Paired T-Test	Before Counselling	106.79	1,9%	t = -15.295	237	< 0.001
	After Counselling	125.11	15.42			
One - Way ANOVA	Elementary or Lower	27.74		F = 4.035		0.008
	Middle School	21.25				
	High School	15.99				
	Diploma or University	2516.04				
Post - Hoc Results	Primary vs High School					0.007
	Primary vs Diploma/Undergraduate					0.017

genetic counselling improved from 50% to 68.5%. Confidence in decision-making also increased, with agreement rising from 50.8% to 60.1%. Furthermore, 68.1% of parents after counselling reported knowing how to access non-medical support, compared to 51.3% before counselling. Optimism about the future improved from 57.6% to 64.7%, reflecting the positive impact of genetic counselling. A full table that explains the increase in GCOS-24 can be provided upon request.

Table 3 presents the results of the statistical analyses conducted to evaluate the effectiveness of genetic counselling in improving parental empowerment. The paired t-test results demonstrate a significant increase in GCOS-24 scores after-counselling ($p < 0.001$), underscoring the intervention's effectiveness in fostering parental understanding and emotional adaptation. The mean score improved from 106.79 (SD = 16.36) before counselling to 125.11 (SD = 15.42) after counselling.

The one-way ANOVA analysis further highlights significant differences in score improvements across education levels ($F = 4.035$, $p = 0.008$). Parents with primary or lower education achieved the highest mean score improvement (27.74), significantly outperforming those with higher education levels. Post-hoc comparisons revealed that the differences between primary education and high school ($p = 0.007$) and primary education and diploma/undergraduate levels ($p = 0.017$) were statistically significant. These findings suggest that parental educational background plays a critical role in the magnitude of counselling effectiveness.

DISCUSSION

This study highlights the critical role of genetic counselling in enhancing parental empowerment in managing children with intellectual disabilities, particularly Fragile X Syndrome (FXS). The significant increase in GCOS-24 scores after counselling underscores the effectiveness of genetic counselling in addressing the multifaceted needs of parents, including emotional adaptation, cognitive understanding, and decision-making capabilities.

Impact of Genetic Counselling on Parental Empowerment

Genetic counselling sessions designed to provide detailed explanations about the etiology, implications, and management of FXS demonstrated measurable improvements in parental empowerment. The average increase in GCOS-24 scores of 18.32 points after-counselling not only exceeds the Minimum Clinically Important Difference (MCID) threshold of 10.3 points but also signifies a transformative impact on parents' perceptions and coping strategies. These findings align with recent studies, including Fitrianingrum et al. (2021), which emphasize that parental empowerment is influenced by various factors, such as early diagnostic processes and psychosocial support.[19]

Parental empowerment plays a vital role in shaping caregiving approaches, educational decisions, and family dynamics. As observed in this study, counselling facilitated a shift from initial feelings of rejection or helplessness toward a more constructive and informed empowerment of their child's condition.

Educational Level as a Modifier of Outcomes This study also reveals that parental educational levels significantly influence the magnitude of GCOS-24 score improvements. Parents with high school education showed greater improvements compared to those with only elementary education. This finding aligns with Fitrianingrum et al. (2021), which suggests that parents with lower educational levels tend to hold stronger fatalistic beliefs, potentially affecting how they accept their child's condition.[20]

Barriers to Effective Genetic Counselling in Indonesia Despite these positive findings, several systemic barriers to effective genetic counselling and FXS management in Indonesia remain evident. Limited access to advanced molecular diagnostic tools such as advanced molecular diagnostic tools, insufficient trained genetic counselors, and inadequate public awareness hinder timely diagnosis and intervention. Notably, the lack of adequate healthcare infrastructure outside urban centers exacerbates these disparities, leaving many FXS cases undiagnosed until severe learning impairments become evident.[20]

These results align with those reported by Fitrianingrum et al. (2021), logistical constraints and the lack of specialized services also hinder parents' ability to accept their child's condition. Efforts to strengthen diagnostic capabilities and enhance training for genetic counselors are critical steps to address these challenges.[19]

The significant improvement in parental empowerment scores following genetic counselling, as evidenced by the paired t-test ($p < 0.001$), aligns with existing studies emphasizing the critical role of early diagnosis and tailored interventions in enhancing outcomes for families dealing with intellectual disabilities. Furthermore, the one-way ANOVA highlights the influence of educational background, with parents of lower educational levels demonstrating greater score improvements. This finding resonates with reports that underscore the importance of accessible and culturally sensitive genetic counselling, particularly in populations where educational disparities may impact comprehension and empowerment of genetic information. These results advocate for targeted educational strategies within counselling sessions to address the unique needs of diverse parental demographics effectively.[19]

The observed improvements align with findings from prior studies that emphasize the transformative potential of genetic counselling in empowering families. For example, research has consistently shown that genetic counselling enhances parental understanding of their child's condition, improving cognitive and emotional responses.[9,14] Furthermore, emotional regulation and hope for the future, as highlighted by increased scores in related GCOS-24 items, reflect the counselling's ability to instil optimism, consistent with evidence that emphasizes the psychological benefits of counselling.[7,17]

However, persistent challenges, such as residual feelings of guilt and helplessness among some parents, underscore the need for supplementary interventions, including hope-based counselling strategies.[9,17] Additionally, the low baseline awareness of genetic testing, with 96.5% of parents never having undergone such tests, highlights systemic barriers such as cultural stigma and limited access to services, which are also reported in resource-limited settings like Indonesia.[8,15] Addressing these gaps through targeted educational initiatives and infrastructure development could further enhance the effectiveness of genetic counselling and its reach.

Policy and Practice Implications

Efforts to improve genetic counselling in Indonesia require a multi-level approach involving the government, researchers, healthcare providers, and the general public. Increasing the number of trained genetic counselors is a critical step, as the current workforce is insufficient to meet the growing demand for genetic services. Policies should focus on funding and supporting the education and certification of genetic counselors to establish a robust network of professionals.

Raising awareness about genetic conditions and the role of genetic counselling is equally important. Government campaigns, community-based initiatives, and collaboration with healthcare providers can enhance understanding among healthcare professionals, researchers, and the general public. This awareness is essential to promote early diagnosis and timely interventions.

For researchers, more emphasis should be placed on investigating genetic conditions prevalent in Indonesia, including Fragile X Syndrome, to develop locally adapted management and counselling protocols. Community involvement, through education and advocacy programs, can help reduce stigma and improve empowerment of genetic conditions, ensuring families have access to the care and support they need. These collective efforts will pave the way for integrating genetic counselling as a foundation of public health strategy in Indonesia.

Limitations and Future Research Directions

While this study offers valuable insights, certain limitations should be acknowledged. The use of purposive sampling may have introduced selection bias, potentially affecting the generalizability of the results. Additionally, since the research was conducted solely in the Jakarta, it may not fully represent the broader diversity of Indonesia. Constraints in funding and time also impacted the study's scope. Future research should consider employing randomized sampling techniques across various locations to enhance validity. Furthermore , longitudinal studies examining the sustained effect of genetic counselling on parental empowerment and child outcomes could offer deeper insights into its long term benefits.

CONCLUSION

This study underscores the transformative potential of genetic counselling in fostering parental empowerment for managing FXS. By addressing emotional, cognitive, and decisional dimensions, genetic counselling serves as a foundation for comprehensive care. However, realizing its full potential requires concerted efforts to overcome systemic barriers, enhance diagnostic and counselling infrastructure, and integrate these services into broader healthcare frameworks. These findings provide a robust foundation for healthcare practitioners and policymakers to advance genetic counselling practices and improve the quality of life for families affected by FXS in Indonesia.

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