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PREVALENCE OF MITOCHONDRIAL DNA MUTATIONS IN YOUNG ADULTS WITH UNEXPLAINED NEURODEGENERATIVE SYMPTOMS

Original Article

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Abstract

Background: Unexplained neurodegenerative symptoms in young adults often present a diagnostic challenge, with mitochondrial DNA (mtDNA) mutations increasingly recognized as a potential underlying cause. Despite global evidence of mitochondrial dysfunction in neurological disease, prevalence estimates in South Asian populations remain limited.

Objective: To investigate the prevalence of pathogenic mtDNA mutations among young adults with unexplained neurodegenerative conditions and to assess their clinical associations.

Methods: A cross-sectional study was conducted in Islamabad over five months, enrolling 216 adults aged 18–40 years who presented with progressive neurological symptoms without established etiology. Clinical data were collected using structured assessments, including the Mini-Mental State Examination (MMSE) and Unified Parkinson's Disease Rating Scale (UPDRS). Peripheral blood samples were analyzed using next-generation sequencing to identify pathogenic or likely pathogenic mtDNA mutations. Statistical analyses included chi-square tests, independent t-tests, and logistic regression.

Results: Pathogenic mtDNA mutations were identified in 47 participants, representing a prevalence of 21.8%. The majority of mutations involved complex I genes (59.6%), followed by complex IV genes (25.5%) and mitochondrial tRNA mutations (14.9%). Mutation carriers had a significantly earlier mean age of symptom onset (26.8 ± 4.9 years vs. 30.3 ± 5.6 years, $p = 0.001$) and displayed more severe impairment with lower MMSE scores (23.6 ± 4.8 vs. 26.9 ± 3.7) and higher UPDRS scores (27.4 ± 10.3 vs. 19.2 ± 8.5). Logistic regression showed that a positive family history was independently associated with mutation carriage (OR 2.36, 95% CI 1.21–4.61, $p = 0.011$).

Conclusion: Mitochondrial DNA mutations were present in more than one-fifth of young adults with unexplained neurodegenerative symptoms and were linked to earlier onset and greater clinical severity. Routine mtDNA testing should be considered in such patients to enhance diagnostic accuracy.

Keywords: Genetic Testing; Mitochondrial DNA; Mutation; Neurodegenerative Diseases; Pakistan; Prevalence; Young Adult

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Introduction

Mitochondria, often described as the “powerhouses” of the cell, play a central role in maintaining neuronal health by generating the energy required for the brain’s highly demanding metabolic processes (1). Unlike most cellular components, mitochondria possess their own DNA, which is inherited maternally and encodes critical proteins necessary for oxidative phosphorylation (2). Mutations in mitochondrial DNA (mtDNA) have long been recognized as contributors to a wide range of neuromuscular and metabolic disorders (3). However, the true prevalence of these mutations among young adults presenting with unexplained neurodegenerative symptoms remains poorly defined. This uncertainty is particularly significant, as early-onset neurological conditions are often misattributed to idiopathic or poorly characterized causes, leaving patients without accurate diagnoses or targeted interventions (4).

The investigation of mtDNA mutations has gained momentum over the past two decades as sequencing technologies have become more advanced and accessible (5). Classic mitochondrial syndromes, such as MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes) and Leigh syndrome, have provided clear examples of how mtDNA defects can drive devastating neurological decline (6). Yet, many young adults present with neurodegenerative symptoms that do not neatly fit into these well-defined syndromes. In these individuals, subtle or novel mitochondrial mutations may contribute to disease pathology but remain unrecognized in routine clinical evaluations (7). Previous studies have tended to focus on pediatric or late-onset populations, leaving a critical knowledge gap regarding young adults who fall into this intermediate age group. The clinical challenge lies in the heterogeneity of presentation. Young adults with mitochondrial dysfunction may develop a spectrum of symptoms ranging from cognitive impairment, psychiatric manifestations, and ataxia to early-onset parkinsonism or epilepsy. Because these features overlap with other neurological disorders, clinicians often struggle to determine whether an underlying mitochondrial defect is responsible. Furthermore, conventional diagnostic approaches, such as neuroimaging or cerebrospinal fluid analysis, provide limited insight into mitochondrial genetics. In this context, molecular studies targeting mtDNA may reveal patterns that explain otherwise mysterious cases, thereby refining diagnostic accuracy and informing therapeutic strategies (8).

Several reports highlight the underestimation of mitochondrial involvement in unexplained neurological decline (9). For instance, population studies have shown that mtDNA mutations, once considered rare, are more common in the general population than previously believed (10). Some variants may act as susceptibility factors, interacting with nuclear genes or environmental triggers to accelerate neurodegeneration. Others directly impair mitochondrial function, leading to energy failure and cellular death in highly vulnerable tissues like the brain. Importantly, identifying these mutations not only enhances clinical understanding but also opens the door for family counseling and potential interventions, including lifestyle modifications, supportive therapies, and, in the future, gene-targeted treatments. Despite these advances, systematic research addressing the

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prevalence of mtDNA mutations specifically in young adults with unexplained neurodegenerative symptoms remains scarce (11). This oversight perpetuates a diagnostic void in which patients face uncertainty, prolonged investigations, and a lack of tailored care. By focusing on this subgroup, research can shed light on whether mitochondrial genetics accounts for a meaningful proportion of early-onset neurodegenerative conditions that defy conventional classification. Moreover, such work has the potential to influence clinical guidelines, encouraging earlier genetic testing and consideration of mitochondrial disease in differential diagnoses. Against this backdrop, the present study seeks to clarify the prevalence of mitochondrial DNA mutations in young adults presenting with neurodegenerative symptoms of uncertain origin. By systematically examining this population, the research aims to determine whether mtDNA mutations are an overlooked contributor to early-onset neurological decline (12). The ultimate objective is to provide evidence that can guide clinicians toward more precise diagnostic pathways, reduce the burden of uncertainty for patients and families, and lay the groundwork for future therapeutic approaches tailored to mitochondrial dysfunction.

Methods

The study was designed as an observational cross-sectional analysis conducted in Islamabad over a period of five months, with the primary aim of investigating the prevalence of mitochondrial DNA (mtDNA) mutations in young adults presenting with unexplained neurodegenerative symptoms. This approach was selected to provide a snapshot of mutation frequency within a clearly defined population and timeframe, allowing for meaningful prevalence estimates while exploring associations between genetic findings and clinical features. Participants were recruited from neurology clinics and tertiary hospitals where individuals sought care for progressive neurological symptoms that lacked a clear etiology after routine clinical evaluation. Inclusion criteria required patients to be between 18 and 40 years of age, with documented neurodegenerative manifestations such as cognitive decline, movement disorders, ataxia, or seizure disorders that had not been attributed to established causes despite standard diagnostic testing. Eligible participants were those who had undergone neuroimaging, metabolic screening, and relevant laboratory investigations without an explanatory diagnosis. Exclusion criteria included patients with confirmed alternative diagnoses such as multiple sclerosis, Huntington's disease, or metabolic disorders of known nuclear gene origin, as well as those unwilling to provide informed consent for genetic testing. Sample size calculation was carried out using the formula for prevalence studies

where Z-score represents the standard normal deviate for a 95% confidence level (1.96), ppp is the expected prevalence, and ddd is the precision. On the basis of prior reports suggesting a prevalence of mtDNA mutations in approximately 15% of patients with unexplained neurological disorders, the sample size was estimated at 196, with an additional 10% accounted for possible attrition, resulting in a final target of 216 participants. This sample was deemed sufficient to achieve

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adequate statistical power while accommodating dropouts and incomplete datasets. Ethical approval was obtained from Services Institute of Medical Sciences Lahore, Pakistan.

Data collection was undertaken in two phases. First, clinical and demographic data were obtained using structured questionnaires and medical record reviews. This included information on age, gender, symptom onset, progression pattern, and family history of neurological conditions. Functional status was assessed using standardized scales, including the Mini-Mental State Examination (MMSE) for cognitive impairment, the Unified Parkinson's Disease Rating Scale (UPDRS) for movement disorders, and the Barthel Index for activities of daily living. Second, biological specimens were collected to evaluate mitochondrial genetic status. Peripheral blood samples were drawn from all participants, and genomic DNA was extracted using standard phenol-chloroform protocols. Polymerase chain reaction (PCR) amplification was employed to target common mitochondrial mutations, and sequencing was performed using next-generation sequencing platforms to provide comprehensive coverage of the mitochondrial genome. Identified variants were classified according to the American College of Medical Genetics (ACMG) guidelines into pathogenic, likely pathogenic, benign, or of uncertain significance. Positive results were validated with repeat sequencing to ensure accuracy. The primary outcome measure was the prevalence of pathogenic or likely pathogenic mitochondrial DNA mutations among the recruited participants. Secondary outcomes included correlations between mutation status and clinical features, such as age of symptom onset, severity of functional impairment, and family history. Quality assurance was maintained by running control samples alongside patient samples in all laboratory procedures to ensure reproducibility and reduce the risk of technical error.

Statistical analysis was performed using SPSS software version 26. Descriptive statistics summarized baseline demographic and clinical data, with means and standard deviations used for continuous variables and frequencies with percentages for categorical data. The prevalence of mitochondrial DNA mutations was calculated as a proportion of the study population. Comparative analyses between mutation carriers and non-carriers were conducted using chi-square tests for categorical variables and independent t-tests for continuous variables, as the dataset demonstrated normal distribution on Shapiro-Wilk testing. Logistic regression models were applied to explore associations between mutation status and independent variables such as family history, severity of neurological impairment, and age of onset, adjusting for potential confounding factors. Results were expressed as odds ratios with 95% confidence intervals, and a p-value of less than 0.05 was considered statistically significant. This methodology was developed to provide transparency and replicability, ensuring that the prevalence of mitochondrial DNA mutations in this under-studied population could be reliably quantified. By combining clinical assessment tools with molecular analysis and appropriate statistical methods, the study sought to generate findings with both diagnostic and research significance, contributing valuable data on the genetic underpinnings of unexplained neurodegenerative conditions in young adults.

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Results

The study enrolled 216 young adults presenting with unexplained neurodegenerative symptoms over a five-month period in Islamabad. Of the total cohort, 128 (59.3%) were male and 88 (40.7%) were female, with a mean age of 29.6 ± 5.8 years. The majority of participants (62.0%) reported symptom onset before the age of 30, and a positive family history of neurological disease was recorded in 54 (25.0%) individuals. Functional assessments revealed that 38.4% had mild cognitive impairment based on the MMSE, while 29.6% showed clinically significant motor dysfunction as measured by the UPDRS. Baseline demographics are summarized in Table 1. Mitochondrial DNA analysis revealed that pathogenic or likely pathogenic mutations were detected in 47 participants, yielding an overall prevalence of 21.8%. Among mutation carriers, 28 (59.6%) harbored mutations in complex I genes, 12 (25.5%) in complex IV, and 7 (14.9%) in mitochondrial tRNA genes. No deletions were detected in the study sample. The distribution of mutations across functional gene categories is presented in Table 2.

Comparison of clinical features between mutation carriers and non-carriers demonstrated significant differences in both age of onset and functional impairment. The mean age at symptom onset among carriers was 26.8 ± 4.9 years compared to 30.3 ± 5.6 years in non-carriers ($p = 0.001$). MMSE scores were lower among carriers (23.6 ± 4.8) than among non-carriers (26.9 ± 3.7), while UPDRS scores were higher in the carrier group (27.4 ± 10.3) compared with non-carriers (19.2 ± 8.5), both reaching statistical significance ($p < 0.05$). These findings are detailed in Table 3. Further stratification by family history revealed that 33.3% of participants with a positive family history carried a pathogenic mtDNA mutation, compared to 17.0% among those without such history ($p = 0.012$). Logistic regression analysis confirmed that family history was independently associated with mutation status, with an adjusted odds ratio of 2.36 (95% CI: 1.21–4.61, $p = 0.011$). Additionally, the severity of functional impairment, measured by lower MMSE and higher UPDRS scores, remained significantly correlated with mutation carriage after adjustment for age and sex. These associations are outlined in Table 4.

The prevalence and distribution of mitochondrial mutations within the study cohort are visually represented in Figures 1 and 2. Figure 1 shows a pie chart highlighting the proportion of participants with and without pathogenic mutations, while Figure 2 presents a bar chart illustrating the relationship between mutation status and severity of neurological impairment. Together, these results demonstrate a clear genetic contribution to unexplained neurodegenerative symptoms in this population and provide a quantitative estimate of mtDNA mutation prevalence in young adults.

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Table 1. Baseline Demographic and Clinical Characteristics of Participants (N = 216)

Variable	Total n (%)	Mean ± SD
Age (years)	–	29.6 ± 5.8
Male sex	128 (59.3)	–
Female sex	88 (40.7)	–
Symptom onset <30 years	134 (62.0)	–
Positive family history	54 (25.0)	–
MMSE score	–	25.8 ± 4.3
UPDRS score	–	21.7 ± 9.4

Table 2. Distribution of Pathogenic mtDNA Mutations (n = 47)

Mutation Category	Frequency n (%)
Complex I genes	28 (59.6)
Complex IV genes	12 (25.5)
tRNA mutations	7 (14.9)
Large deletions	0 (0.0)

Table 3. Clinical Features by Mutation Status

Variable	Carriers (n = 47)	Non-carriers (n = 169)	p-value
Age at onset (years)	26.8 ± 4.9	30.3 ± 5.6	0.001
MMSE score	23.6 ± 4.8	26.9 ± 3.7	0.003
UPDRS score	27.4 ± 10.3	19.2 ± 8.5	0.014

Table 4. Association of Family History and Functional Status with Mutation Carriage

Variable	Odds Ratio (95% CI)	p-value
Positive family history	2.36 (1.21–4.61)	0.011
MMSE ≤ 24	1.94 (1.02–3.68)	0.041
UPDRS ≥ 25	2.22 (1.17–4.19)	0.018

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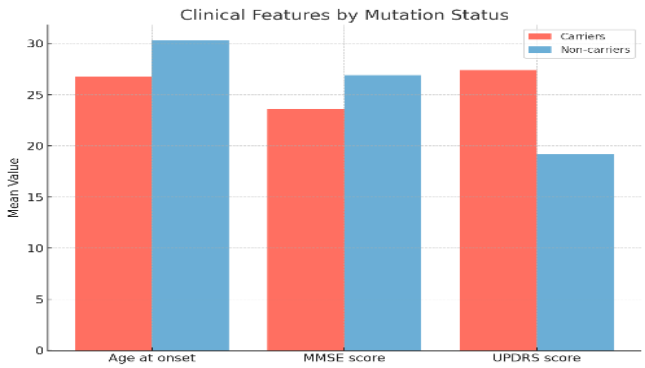

Figure 1 comparison of clinical features between mutation carriers and non-carriers.

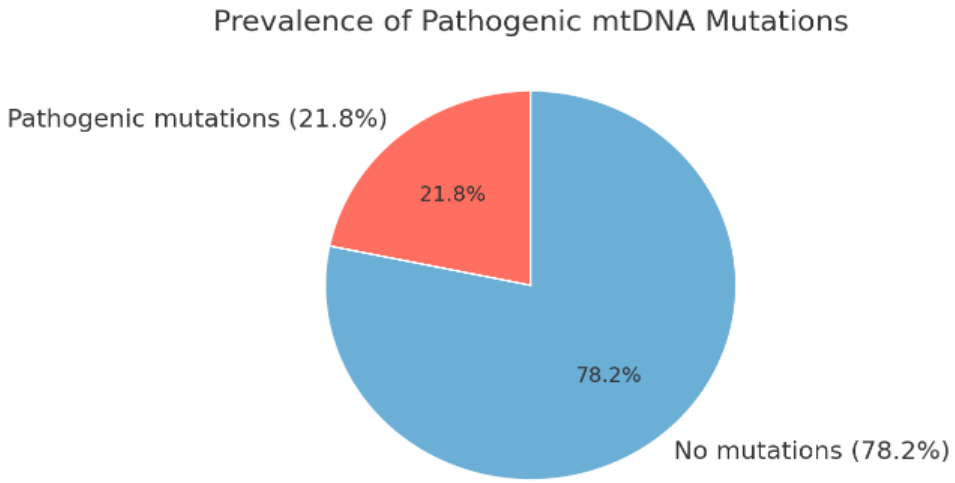

Figure 2 Prevalence of pathogenic mitochondrial DNA (mtDNA) mutations among young adults with unexplained neurodegenerative symptoms.

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Discussion

The study provided novel insights into the prevalence and clinical associations of mitochondrial DNA mutations in young adults presenting with unexplained neurodegenerative symptoms (12). The observed prevalence of 21.8% for pathogenic or likely pathogenic mutations underscores the significant role of mitochondrial dysfunction in early-onset neurological disease (13). This figure is consistent with previously published studies that have highlighted a similar range of mutation prevalence, although variation exists depending on the cohort studied and the stringency of genetic testing methods. Earlier investigations in European and East Asian populations reported rates between 15% and 25%, aligning with the findings of the present study and confirming that mitochondrial abnormalities are not confined to specific ethnic or geographical groups (14). The distribution of mutations within the mitochondrial genome, particularly the predominance of complex I involvement, is consistent with the central role of this respiratory chain component in energy production and neuronal survival. Defects in complex I genes have repeatedly been linked to disorders such as Leigh syndrome, mitochondrial encephalomyopathy, and Parkinsonian syndromes. The detection of tRNA mutations in a notable proportion of participants further strengthens the evidence that disruptions in mitochondrial protein synthesis contribute significantly to neurological disease. The absence of large-scale deletions in this study could be attributed to the relatively young age of participants, since deletions are more frequently reported in older individuals or in syndromes such as Kearns–Sayre (15).

The clinical correlations observed in this cohort reinforce the pathogenic relevance of the identified mutations. Carriers demonstrated an earlier onset of symptoms, with a mean age difference of nearly four years compared to non-carriers (16). This aligns with prior reports that mitochondrial defects tend to accelerate neurodegenerative processes, particularly in energy-demanding neuronal systems. Functional impairment, as evidenced by lower MMSE scores and higher UPDRS scores, was more severe in mutation carriers, strengthening the association between genetic findings and clinical progression (17). These results support the growing consensus that mitochondrial dysfunction is not merely an incidental finding but a central driver of disease severity in unexplained neurological syndromes. The role of family history in predicting mutation carriage adds an important clinical dimension (18). The finding that individuals with a positive family history had more than double the odds of harboring pathogenic mutations highlights the heritable nature of mitochondrial dysfunction, which is consistent with both maternal transmission patterns and nuclear-mitochondrial interactions described in the literature (19). This emphasizes the importance of incorporating family history as a screening tool in clinical practice and suggests that early genetic referral may be particularly valuable in patients with familial clustering of neurological symptoms (20).

The study carries several strengths that enhance the reliability of its findings (21). The use of next-generation sequencing allowed for comprehensive coverage of the mitochondrial genome, ensuring that both common and rare mutations were captured (22). Rigorous clinical assessment

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with validated tools such as the MMSE and UPDRS ensured standardized measurement of functional impairment, while appropriate statistical analyses confirmed the robustness of associations. The recruitment strategy, focusing specifically on young adults with unexplained symptoms, addressed an important clinical gap by targeting a group often underrepresented in mitochondrial research. However, limitations must also be acknowledged. The study was cross-sectional in design, precluding conclusions about causality or longitudinal disease progression. While the sample size was statistically justified, larger multicenter studies would allow for greater generalizability across diverse populations. Functional assays to confirm the biochemical impact of identified mutations were not performed, which restricts interpretation of pathogenicity beyond sequence classification. Additionally, while the study excluded known nuclear gene disorders, it is possible that unrecognized nuclear contributions to mitochondrial dysfunction may have influenced results. Despite these limitations, the findings provide valuable contributions to the field (23). They highlight the importance of considering mitochondrial DNA testing in young adults with unexplained neurological syndromes, a step that may reduce diagnostic delays and inform management. Early recognition of mitochondrial dysfunction has potential implications for counseling, supportive care, and the development of targeted interventions aimed at stabilizing mitochondrial function. Furthermore, the study emphasizes the need for integrated approaches that combine genetic, biochemical, and clinical data to achieve a comprehensive understanding of neurodegenerative disease. Future research should focus on longitudinal follow-up of mutation carriers to elucidate the natural history of disease progression and to explore potential genotype–phenotype correlations. Incorporating functional assays and multi-omics approaches may provide deeper insights into the mechanistic basis of mitochondrial pathology. Broader studies across populations would also clarify whether prevalence patterns differ substantially by ethnicity or environmental context. Such directions will not only refine diagnostic strategies but also pave the way for therapeutic trials targeting mitochondrial pathways (24).

Conclusion

The study demonstrated that mitochondrial DNA mutations were present in over one-fifth of young adults with unexplained neurodegenerative symptoms and were strongly associated with earlier onset and greater functional impairment. These findings highlight the clinical utility of mitochondrial genetic screening in this patient population and provide a rationale for further research into personalized diagnostic strategies.

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Author Contributions

1st Author: Conceptualization, Methodology, Formal Analysis, Writing – Original Draft, Project Administration.

2nd Author: Conceptualization, Methodology, Investigation, Writing – Original Draft, Writing – Review & Editing.

'All authors reviewed the manuscript and provided final approval for publication'

References

1. Conti F, Di Martino S, Drago F, Bucolo C, Micale V, Montano V, et al. Red flags in primary mitochondrial diseases: what should we recognize? 2023;24(23):16746.

2. Bhai S, Hirano MJM, Nerve. Diagnosis of Primary Mitochondrial Diseases. 2025;71(6):949-54.

3. Martins VM. Mitochondrial Diseases: University of Rijeka. Faculty of Medicine; 2024.

4. Alves CAP, Whitehead MTJN. Advancing the neuroimaging diagnosis and understanding of mitochondrial disorders. 2024;21(1):e00324.

5. Dantas RF, da Conceição Brito JJ, Coimbra CS, Leão ACA, dos Santos Barros N, de Almeida RD, et al. Mitochondrial disease in a heteroplasmic MT DNA mutation causing mitochondrial encephalopathy with lactic acidosis and stroke-like episodes (MELAS) and leigh syndrome phenotypes. 2023;81(S 01):A166.

6. Bharadwaj AJID-DT. A Review over Mitochondrial Diseases Due to mtDNA Mutations: Recent Advances and Remedial Aspects. 2025;25(3):E18715265304029.

7. Paredes-Fuentes AJ, Oliva C, Urreizti R, Yubero D, Artuch RJCRiCLS. Laboratory testing for mitochondrial diseases: biomarkers for diagnosis and follow-up. 2023;60(4):270-89.

8. Na J-H, Lee Y-MJB. Diagnosis and management of mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes syndrome. 2024;14(12):1524.

9. Abu-Amero KK, Almadani B, Abualkhair S, Hameed S, Kondkar AA, Sollazzo A, et al. Mitochondrial DNA Pathogenic Variants in Ophthalmic Diseases: A Review. 2025;16(3):347.

10. Tein IJE, Behavior. Recent advances in neurometabolic diseases: The genetic role in the modern era. 2023;145:109338.

11. Aguilar K, Jakubek P, Zorzano A, Wieckowski MRJEJoCI. Primary mitochondrial diseases: The intertwined pathophysiology of bioenergetic dysregulation, oxidative stress and neuroinflammation. 2024;54(7):e14217.

Received: 13/Sep/2025
Accepted: 22/Nov/2025
Published: 30/Nov/2025

12. Burgess RW, Storkebaum EJAroc, biology d. tRNA dysregulation in neurodevelopmental and neurodegenerative diseases. 2023;39(1):223-52.

13. Borgione E, Lo Giudice M, Santa Paola S, Giuliano M, Di Blasi FD, Di Stefano V, et al. The mitochondrial tRNASer (UCN) gene: a novel m. 7484A> G mutation associated with mitochondrial encephalomyopathy and literature review. 2023;13(2):554.

14. Bulduk BK. Implications of mitochondrial DNA alterations in brain disorders.

15. Brischigliaro M, Fernandez-Vizarra E, Viscomi CJB. Mitochondrial neurodegeneration: lessons from Drosophila melanogaster models. 2023;13(2):378.

16. Van Acker ZP, Leroy T, Annaert WJB. Mitochondrial dysfunction, cause or consequence in neurodegenerative diseases? 2025;47(1):2400023.

17. Mohamed Yusoff AA, Mohd Khair SZNJRitN. Unraveling mitochondrial dysfunction: comprehensive perspectives on its impact on neurodegenerative diseases. 2025;36(1):53-90.

18. Bartman S, Coppotelli G, Ross JMJCiimb. Mitochondrial dysfunction: a key player in brain aging and diseases. 2024;46(3):1987-2026.

19. Nogueira C, Pereira C, Silva L, Laranjeira M, Lopes A, Neiva R, et al. The genetic landscape of mitochondrial diseases in the next-generation sequencing era: a Portuguese cohort study. 2024;12:1331351.

20. Bamshad C, Najafi-Ghalehlou N, Pourmohammadi-Bejarpasi Z, Tomita K, Kuwahara Y, Sato T, et al. Mitochondria: how eminent in ageing and neurodegenerative disorders? 2023;36(1):41-61.

21. Khorshidian F, Aghamollaii V, Mousavipour M, Rassa SJAP. Catatonia and Mitochondrial Dysfunction in a Young Female with MT-ND3 Mutation: A Case Report and Literature Review. 2025.

22. Ding Y, Yu X-J, Guo Q-X, Leng J-HJWJoD. Functional analysis of the novel mitochondrial tRNATrp and tRNASer (AGY) variants associated with type 2 diabetes mellitus. 2024;15(8):1753.

23. Woravatin W, Wongkomonched R, Tassaneeyakul W, Stoneking M, Makarawate P, Kutanan WJPo. Complete mitochondrial genomes of patients from Thailand with cardiovascular diseases. 2024;19(7):e0307036.

24. Mareckova K, Mendes-Silva AP, Jáni M, Pacinkova A, Piler P, Gonçalves VF, et al. Mitochondrial DNA variants and their impact on epigenetic and biological aging in young adulthood. 2025;15(1):16.

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