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Original Article

INVESTIGATING THE TRENDS OF PREVIOUS STUDIES ON ATAXIA

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ABSTRACT
The review is concerned with ataxia; a neurological disease that impairs voluntary movements such as walking, speech, and fine motor skills because of muscle coordination loss. The paper examines the genetic factors and outcomes of therapy of the numerous types of ataxia, including vestibular, cerebellar and sensory ataxia and the availability of medical facilities. It examined the influence of the environmental factors, rehabilitation treatments and the genetic tendencies. Previous studies highlighted the role that neurodegenerative mechanisms, autoimmune responses, and genetic defects have in the development of ataxia. To obtain a comprehensive picture of the disease and potential methods of treatment, a systematic literature review of the last 10 years was conducted to identify trends and shortcomings.

I. INTRODUCTION

Ataxia is a neurological condition that is typified as the lack of coordination, balance or voluntary control of the muscles usually as a result of the inability of the cerebellum or the nerve pathways to be effective (Klockgether, 2010). It could appear because of several factors, including genetic mutations, autoimmune diseases, infections, stroke, multiple sclerosis, exposure to toxins (like chronic alcoholism), or a neurodegenerative disease (Duehr, 2010). The symptoms include slurred speech, deficiencies in the fine motor system, unsteady gait (dysarthria), uncontrolled movement of the eye muscles (nystagmus), and tremor (Tada et al., 2016). The most prevalent types of ataxia include Friedreich ataxia and spinocerebellar ataxia in the hereditary category, and the acquired type can be a result of metabolic disorders or other external damages (Manto, 2018). The diagnosis process involves the use of neurological tests and brain scan (MRI or CT) and in rare situations, genetic testing to establish inherited variations (van de Warrenburg et al., 2014). Despite the absence of a known treatment, the majority of the ataxias, physical therapy, occupational therapy, speech therapy, and medication would help a patient to lead a better life (Klockgether et al., 2019).

The term known as ataxia is used to refer to a series of diseases that affect speech, balance and coordination. This is through impairments that are brought about by the complications in the peripheral nerves, spinal cord, or cerebellum (Klockgether, 2010). The ataxia symptoms can be either mildly loss of balance or severe handicap that can either be acquired, inherited, or idiopathic. Hereditary ataxia is commonly caused by a genetic mutation and the others are acquired as a result of infection, poisonous substances or trauma (Harding, 1993). The low-resource

settings continue to undergo numerous studies on ataxia, albeit not clearly comprehended in certain conditions. Scarcely any research has been conducted on environmental and sociocultural influences on its development and treatment. This paper tries to address these gaps since it carries out research on the clinical presentation, diagnostic issues, and therapeutic outcomes of ataxia. It is also an assessment of the promise of assistive technology and therapy in assisting in enhancing the management of ataxia.

Accessibility of healthcare facilities among patients experiencing ataxia varies, and it is geographically variable based on the geographical location, health facilities, and the specialists. Special ataxia clinics containing multidisciplinary teams are necessary to find the diagnosis correctly and care (Kuo et al., 2019). However, this is not equal, particularly in nations with low resources, with the inequalities present when health care providers do not have adequate access to neurologists, genetic testing, and rehabilitative treatments, and, consequently, individuals are frequently diagnosed later and receive a lower quality of treatment (Muller and Jahn, 2019). These gaps have been bridged partially with the assistance of telemedicine, patient advocacy groups, and remote consultations (Ashizawa et al., 2018). Nevertheless, deficiency in care-based protocols, expensive care, and limitations with insurance still affect a patient's result (Manto et al., 2020). The growth of the number of specialized care units, creating digital health technologies, and increasing awareness among professionals about this problem are all opportunities to increase the accessibility of healthcare to people affected by ataxia.

Rehabilitation plays a crucial role in improving the quality of life of people affected with ataxia through increasing their mobility, coordination, and independence. The multidisciplinary rehabilitation programmes include physical therapy, occupational therapy, and speech therapy in a group of those that help to control the symptoms and avoid the secondary complications, including stiffness of the joints and muscle weakness (Ilg et al., 2014). It is shown that the increase in the functional outcome in degenerative cerebellar ataxias can be achieved with the help of gait training, balance training, and task-specific methods of learning motor skills (Fonteyn et al., 2014). Assistive devices also include walkers and adaptive tools used in daily activities to help an individual be independent and reduce the risk of falls (Synofzik & Ilg, 2014). In addition, psychological support and cognitive-behavioural therapy can be used to resolve emotional issues since anxiety and depression are common among cases of progressive ataxia (Schmitz-Huesch et al., 2010).

METHOD

Participants

The literature review entailed the systematic review of the literature over the last 10 years of work in patients having various types of ataxia. The sample consisted of patients of urban and rural neurology clinics. The review conducted by the systematic review is centred on hereditary and acquired ataxia, innovations in treatment, and healthcare accessibility, and is conducted due to the research published in 2010-2020. Key trends are the discoveries of new genetic mutations, advances in the field of gene and neuroprotective therapy, and the effectiveness of stringent rehabilitation regimens.

Measures

The method of conducting a systematic review of the conducted studies over the last decade concerning the subject of ataxia is a productive source of information regarding the genetic, neurodegenerative, and rehabilitative side of the case. The study was conducted regarding the molecular pathogenesis of various forms of ataxia, including Friedreich ataxia and spinocerebellar ataxia, and gene therapies and disease-modifying therapies became the subject of investigation (Matilla-Duenas et al., 2014). The efficacy of rehabilitation processes (such as neuromodulation and intensive coordinative training) in improving the quality of life and motor activity has also been discussed (Ilg et al., 2014).

Moreover, it has been pointed out that, as a matter of fact, there exist vast disparities in healthcare access, specifically with low-resource patients, whose diagnosis is delayed, and access to specialists is scarce, rendering the provision of quality care even more difficult (Kuo et al., 2019). Although the future will require more in-depth clinical research to conclude in the long term on their effectiveness and safety, certain therapeutic interventions are seen as promising, including aminopyridines and gene therapy (Manto, 2018).

Procedure

For the current systematic review to study the patterns of ataxia, the articles published during the previous 10 years (2010-2020) were included. The systematic review's goal was to identify the patterns in ataxia research over the previous ten years (2010–2020). PubMed, Scopus, and Web of Science databases were searched by using the words ataxia, genetic mutation, and rehabilitation. Out of 25 studies included in this review, two were about the causes of ataxia, two were on the access to healthcare of ataxia, and six were about the causes of hereditary ataxia.

II. RESULTS

Based on the systematic review, the present study concluded: 1) Genetic Factors: Hereditary ataxias were closely linked to mutations in genes like ATXN1 (spinocerebellar ataxia type-1) and FXN (Friedreich's ataxia) (D'Silva et al., 2015). 2) Therapeutic Innovations: Rehabilitation programs and pharmaceutical interventions, such as Riluzole, were found to improve motor performance with a moderate degree of success (Milne et al., 2017). 3) Environmental Modifiers: The development and treatment of the illness were significantly impacted by the accessibility of medical facilities (Daker-White et al., 2015).

Table 1: Study Findings

Authors	Type of study	Participants	Outcome Variables	Measure	Key Findings
D'Silva et al.(2015)	Case-Control study	50 patients With FA	Genetic mutation	Genetic screening	Identified FXN mutation as a major cause of Friedreich's ataxia
Harding (1993)	Longitudinal study	120 Inherited Ataxia cases	Neurological progression	Clinical assessment	Classified inherited ataxias and highlight genetic variability
Klockgether (2010a)	Review	Ataxia patients	Ataxia type and causes	MRI	Updated classificationsand causes of degenerative ataxias
Zesiewicz Et al (2018)	Randomized trial	100 Patients on Riluzole	Motor function	Gait Analysis	Demonstrated motor functions improvement with riluzole therapy
Paulson et al (2017)	Cross sectional survey	300 Caregivers	Quality of life	Caregiverss urvey tools	Highlighted the burden on Csregivers and the need for better Interventions
Hadjivassiliou et al. (2017)	Review	1500 ataxia patients	Immune-mediated ataxias	next-generation sequencing	Immune-mediated ataxias are common
Milne et al. (2017)	Review	17 studies on genetic degenerative ataxia	Rehabilitation	randomized controlled trials	Rehabilitation improves function, mobility, ataxia, and balance
Klockgether (2010b)	Interventional study	42 patients with pure cerebellar degeneration	Inpatient physical and occupational therapy	Scale for the Assessment and Rating of Ataxia	Patients clearly benefited in the short term from intensive rehabilitation.
Stanley et al. (2020)	Cross-sectional survey	Cerebellar Ataxia	Healthcare	Scale for Assessment and Rating of Ataxia, Short Form (36) Health	Some areas that should be addressed include employment, community involvement, access to specialized consultants, and

				Survey	informal home care.
Daker-White et al. (2015)	Cross-sectional survey	Ataxia patients	Healthcare	Interviews	Ataxia constrained people's attempts to deal with their conditions

III. DISCUSSION

The results accentuated that there are genetic etiologies of ataxia and these are mainly the mutations of genes that involve cerebellar activity alongside neuronal integrity and result in progressive ataxia in terms of deficit of coordination and balance. Autozygous ataxias are either an autosomal dominant (i.e., spinocerebellar ataxias (SCAs)) or an autosomal recessive (i.e., Friedreich's ataxia (FRDA)) one (Duehr, 2010). Polyglutamine extensions in the following genes are the cause of SCA: ATXN1, ATXN2, and ATXN3, which lead to the accretion of toxic proteins and neuronal death (Paulson et al., 2017). FRDA, on the other hand, is a disease brought about by the increase of GAA trinucleotide repeats in the FXN gene that causes a decrease in the amount of frataxin proteins and results in the malfunction of the mitochondria (Pandolfo, 2009). Other genetic ataxias (i.e., episodic ataxias) exist in which ion channel gene (e.g., KCNA1) genes are mutated (Jen, 2008). Genetic testing forms a beneficial part in diagnosis and management, with most treatments being symptomatic.

The treatment of ataxia is symptomatic to a large extent because most of the causes are inherited with no cure. Balance of patients should be improved through physical and occupational therapy (Klockgether et al., 2019). Speech therapy can be used to treat dysarthria, and assistive technology fulfills the work to enhance day-to-day functioning through the use of walkers and orthotic support (Ilg et al., 2014). The treatment is pharmacological (but not exhaustive) and may be applied to the treatment of episodic ataxia and spinocerebellar ataxia due to its effect on cerebellar functioning via the channel-modulation of potassium (Strupp et al., 2011). In cases where ataxia is caused by vitamin deficiencies (e.g., vitamin E or B12), metabolic or immune dysfunction, the underlying condition may be cured to lead to symptom improvements (Manto, 2018). Research on the neuroprotective therapy and the gene therapy in the future is likely to succeed in treating the disease in a disease-modifying manner (Matilla-Duenas et al., 2014). Furthermore, assistive devices used in everyday activities that promote independence and reduce the risk of falls are walkers and adaptive tools (Synofzik & Ilg, 2014). In addition, the emotional issues could be addressed with the assistance of psychological support and cognitive-behavioral therapy since anxiety and depression are common cases of progressive ataxia (Schmitz-Huesch et al., 2010). The best solution to ameliorating the general well-being and life of patients with ataxia is an individualized approach to rehabilitation that is founded on the needs. Some of the multidisciplinary rehabilitation programs include physical therapy, occupational therapy, and speech therapy, which manage the symptoms and prevent secondary complications like joint stiffness and weakness of muscles (Ilg et al., 2014). The gait training, balance training, and task-specific motor learning approaches have been proven to generate functional outcomes in degenerative cerebellar ataxia (Fonteyn et al., 2014).

Some of the factors that inhibit access to healthcare services by patients with ataxia are the geographical location, insufficient specialisation, financial factors and inefficiency in the healthcare system. The inefficiencies linked to the delay in diagnosis in numerous patients, in particular, in rural or low-income areas are because to identify a disease, it is often necessary to implement special neurological and genetic testing that is not available or costly (Kuo et al., 2019). Lack of awareness is another problem that occurs among the general practitioners thus resulting in misdiagnosis and delays in referrals to ataxia specialists (Muller and Jahn, 2019). Moreover, the insurance cover is highly unequal and not offered to the rehabilitative treatment, assistive technology, or new treatment required (Manto et al., 2020). Medicine and patient advocacy organizations ease it because they bridge the gaps between the patient and specialists, and networks, yet differences throughout the world are not disappearing (Ashizawa et al., 2018). The promotion of equitable access to ataxia care may be enhanced through the assistance of the creation of awareness, introduction of special treatment centers, and introduction of digital health solutions.\

V. CONCLUSION

Finally, this review is a close study of ataxia, focusing on its genetic background, treatment factors, access to healthcare, and its predisposition to environmental as well as rehabilitative factors. It graphically outlines the results within the last decade and describes some of the trends of the evolution of the disease, the development of therapeutic methods, and the research gaps. The neural interactions of the neurodegenerative pathways, immune responses, and genetic dispositions determine the development of specific interventions. Future studies need to attempt to fill these gaps through the development of less erroneous diagnostic instruments, tailored therapies, and more comfortable access to special care that will eventually enhance the quality of life of individuals with ataxia.

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