INTELLECTUAL DISABILITY ACROSS THE LIFE COURSE: DEFINITION, CLASSIFICATION, AND INTERVENTION STRATEGIES

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ABSTRACT

Intellectual disability is characterized by significant limitations in intellectual and adaptive functioning, affecting a substantial part of the global population, with an estimated prevalence between 1% and 3%, being higher in men and in developing countries. The concept has evolved from a simplistic view to a more comprehensive approach, considering both cognitive deficits and adaptive challenges. The etiology of the condition is multifactorial, with genetic and environmental causes, and diagnosis is based on IQ tests and adaptive behavior assessments. It is classified as mild, moderate, severe and profound. This systematic review article examines definitions, classifications, etiologies, and lifelong interventions of people with intellectual disabilities. It includes studies published between 2000 and 2024, in English or Portuguese, in the PubMed, Scopus, Web of

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Science, PsycINFO, and SciELO databases. Data analysis will be qualitative and, where applicable, will include meta-analysis, using specific tools to assess the quality of studies. Interventions for intellectual disability involve a multidisciplinary approach, with an emphasis on behavioral strategies and family support, in addition to the management of comorbidities. The review highlighted the need for early and ongoing interventions to improve the quality of life for individuals and their families, as well as the importance of targeted public policies. It also identified gaps in the literature, especially regarding the variability of studies, suggesting the need for more robust research for the development of effective management strategies.

Keywords: Intellectual disability. Prevalence. Aetiology. Diagnosis. Early intervention.

INTRODUCTION

Intellectual disability, characterized by significant limitations in intellectual functioning and adaptive behavior, represents a complex and multifaceted condition that impacts the lives of millions of individuals around the world. Traditionally, intellectual disability has been understood as a condition of interrupted or incomplete development of the mind, marked by cognitive difficulties that affect the ability to learn, reason, and perform daily activities autonomously. From Kraepelin's first description, which simplified the condition as a generalized failure in the brain, to more contemporary definitions, there has been a significant evolution in the understanding and diagnosis of intellectual disability.

The evolution of the concept of intellectual disability reflects advances in research and clinical practice, with a gradual shift from a simplistic approach to a more holistic and multifaceted view. In 1959, the American Association of Mental Disability (AAMR) introduced a more comprehensive definition, describing the condition as significantly below-average intellectual functioning associated with impairments in adaptive behavior. Over time, the terminology evolved into "intellectual disability" and definitions were refined to consider the complexity of the condition, encompassing not only cognitive limitations but also adaptive challenges that affect individuals' everyday lives.

The etiopathogenesis of intellectual disability is complex and multifactorial, involving an interaction of biomedical, social, behavioral, and educational factors throughout life. Although genetic causes have become more evident, representing a significant part of cases, up to 40% of cases, especially those with mild impairment, do not have an identifiable etiology. The most common genetic causes include trisomy 21 and fragile X syndrome, while prenatal, perinatal, and postnatal factors also play a crucial role in the development of the condition. Understanding these factors is essential for the diagnosis, treatment, and prevention of intellectual disability.

The diagnosis of intellectual disability requires the presence of three main conditions: significantly below-average intellectual functioning, with an intelligence quotient (IQ) of 70 or less; impairments in adaptive functioning in at least two areas; and onset of symptoms before the age of 18. The classification of intellectual disability ranges from mild to profound, reflecting the severity of cognitive and adaptive deficits. In mild cases, with IQs between 50 and 70, individuals can learn basic skills and perform semi-skilled activities with supervision. In moderate cases, with IQs between 35 and 49, learning is more limited and requires constant supervision. Severe and profound cases, with IQs between 20 and 34 and below 20, respectively, present significant difficulties and require ongoing support and intensive care.

The clinical evaluation of intellectual disability is a comprehensive process that includes detailed anamnesis, physical examination, and complementary tests. The work-up should address personal and family history, including genetic and environmental factors, and consider the presence of medical and behavioral comorbidities. Genetic and metabolic tests, as well as neuroimaging tests, are often used to elucidate the etiology and guide treatment. Early and continuous intervention is essential to optimize the social functioning and quality of life of individuals with intellectual disabilities.

This systematic review of the literature aims to provide a detailed analysis of the definitions and classifications of intellectual disability, as well as lifelong intervention strategies. The review will address the evolution of concepts and classifications, highlight best practices, and identify gaps in the current literature. The methodology will include the definition of strict inclusion and exclusion criteria, a comprehensive search in academic databases, and the application of qualitative and quantitative data analysis techniques. This work seeks to consolidate existing knowledge, provide recommendations for clinical and educational practice, and guide future research in the area.

METHODOLOGY

We aim to answer the following research questions: (1) What are the most widely accepted definitions and classifications for intellectual disability? (2) What etiological factors are most often associated with intellectual disability? (3) Which interventions are considered most effective for the management of intellectual disability at different stages of life?

The inclusion criteria for this review were studies that address intellectual disability with an emphasis on definitions, classifications, etiology, and intervention strategies, published in English or Portuguese, between 2000 and 2024. Studies of any methodological design, such as cohort studies, clinical trials, and reviews, were included. Exclusion criteria were articles that do not focus directly on intellectual disability, studies outside the established time frame, and studies not published in peer-reviewed journals or irrelevant gray literature.

For data collection, the following databases were searched: PubMed, Scopus, Web of Science, PsycINFO and SciELO. Relevant books in the area were also consulted to ensure comprehensive coverage of the topics covered.

The search strategy included terms such as "intellectual disability," "intellectual developmental disorder," "classification of intellectual disability," "etiology of intellectual disability," and "intervention strategies for intellectual disability." Searches were adjusted as needed for each database to ensure comprehensive coverage.

The study selection process involved two stages. First, the initial screening of titles and abstracts was carried out to identify potentially relevant articles. Then, the full texts of the selected articles were evaluated to confirm compliance with the inclusion criteria. Study selection was performed by two independent reviewers, and discrepancies were resolved by consensus or by a third reviewer when necessary.

Data were extracted from each included study using a standardized form, which included variables such as study characteristics, definition and classification of intellectual disability, etiological factors investigated, and reported intervention strategies. Data extraction was conducted by two independent reviewers to ensure accuracy, and discrepancies were resolved through discussion.

Study quality was assessed using the Cochrane risk of bias tool for clinical trials and the Jadad scale for clinical trials. For systematic reviews, AMSTAR (A MeaSurement Tool to Assess systematic Reviews) was used. Quality assessment was performed by two independent reviewers, and disagreements were resolved by consensus.

The data were synthesized using qualitative analysis methods and, when appropriate, meta-analysis. The qualitative analysis involved the categorization of definitions, classifications, etiological factors, and interventions into main themes. For the meta-analysis, the studies were grouped based on the homogeneity of the data and the outcomes evaluated. The analysis was performed using the RevMan software.

Because this systematic review is based on published data, no specific ethical considerations related to research with human subjects were necessary.

LITERATURE REVIEW

Intellectual disability refers to an interrupted or incomplete development of the mind, marked by significant limitations in both intellectual functioning and adaptive behavior. Historically, Kraepelin described the condition in a simplified way, stating that "the feeble-minded are people in whose brain not many things occur." However, in 1959, the American Association of Mental Disabilities (AAMR) offered a more precise definition, explaining that "mental retardation" is characterized by below-average intellectual functioning, manifesting itself during the developmental period and associated with impairments in adaptive behavior. As the term "mental retardation" was replaced by "intellectual disability", the conceptualization also evolved. Today, intellectual disability is defined as a condition that compromises the development of cognitive, language, motor and social skills, essential for the global level of intelligence. These limitations affect the individual's ability to adapt to the demands of daily life, both personally and socially. The American Association on Intellectual

and Developmental Disability (AAIDD) emphasizes that these limitations are expressed in conceptual, social, and practical adaptive skills, reinforcing that the condition arises during the developmental phase and impacts several areas of life, from learning to social interaction and functional autonomy.

EPIDEMIOLOGY OF INTELLECTUAL DISABILITY

In Brazil, there are no studies that precisely define the population frequency of intellectual disability, which forces us to base our estimates on data projected from other realities. This scenario reflects, in part, a historical lack of interest, since the condition offers few possibilities for pharmacological intervention and its social impact is often underestimated, despite being a field of study since the introduction of psychiatry in the country. International studies indicate that the prevalence of intellectual disability varies between 1% and 3%, and a recent meta-analysis points to an average prevalence of about 1%. The condition is more frequent in males in all age groups and its incidence is higher in developing countries, where rates are almost double those seen in high-income nations. In addition, intellectual disability occurs less frequently in urban areas (0.4%) compared to rural areas (1.02%). These disparities suggest that socioeconomic factors and access to health care directly influence the prevalence of the condition.

ETIOPATHOGENESIS AND RISK FACTORS FOR INTELLECTUAL DISABILITY

The etiopathogenesis of intellectual disability is a multifactorial construct that involves a complex interaction of biomedical, social, behavioral, and educational factors throughout life. Although injuries, infections, and toxins have become less frequent causes due to advances in prenatal care, genetic factors have gained prominence. In up to 40% of cases, especially those with mild involvement, it is not possible to identify a specific etiology. Understanding the causes of intellectual disability can help in the treatment and even prevention of some cases. These risk factors can occur in the prenatal, perinatal, and postnatal periods, and are divided into three major categories: organic, genetic, and sociocultural. The most common genetic causes include trisomy 21 and fragile X syndrome. However, most cases present with an overlap of genetic, environmental, and sociocultural factors, reflecting the complex nature of the condition.

Among the factors that act before conception, genetic causes stand out, which can be dominant, recessive or sex-linked. Among the pathologies of dominant inheritance, several syndromes are included, which often involve intellectual disability associated with ectodermal, mesodermal, muscular or bone malformations. Examples include

neuroectodermatoses or phacomatoses, such as tuberous sclerosis, characterized by hamartomatous lesions in various tissues, and neurofibromatosis, which manifests with café-au-lait spots and areas of hypo- or hyperpigmentation. Other dominant conditions are craniofacial dysostosis, such as Apert syndrome, with marked craniofacial features, and Marfan syndrome, associated with tall stature, long limbs, and ligament laxity.

In recessive inheritance pathologies, metabolic disorders are common. Among them, diseases of lipid metabolism, such as Tay-Sachs disease and Niemann-Pick disease, and disorders of mucopolysaccharides metabolism, such as Hurler's disease and Maroteaux syndrome, stand out. Disorders of glucose metabolism, such as glycogenosis (von Gierke's disease), and protein metabolism, such as phenylketonuria – diagnosed by the heel prick test – are also relevant. Phenylketonuria, with a prevalence of 1:15,000, is characterized by intellectual disability, light skin and hair, as well as seizures. These examples illustrate the wide range of genetic and metabolic conditions that can contribute to the development of intellectual disability, highlighting the importance of further research to identify still unknown causes, especially in mild cases.

About 50% of miscarriages are caused by chromosomal aberrations established in the zygote, resulting from failures in the production of gametes. These pathologies can be classified into anomalies of the somatic and sex chromosomes. Trisomy 21, or Down syndrome, is the most frequent somatic chromosome anomaly, occurring in approximately 1 in every 600 live births, with an increase proportional to maternal age. Other trisomies, such as Edwards trisomy (chromosome 18) and Patau trisomy (chromosomes 13 to 15), also have great clinical relevance, although they are less common.

Among the anomalies of the sex chromosomes, Klinefelter syndrome stands out, characterized by tubular testicular dysplasia, XXY karyotype, hypogonadism, and long limbs. Turner syndrome, with X0 karyotype, is manifested by short stature, ovarian dysgenesis, transient congenital lymphedema, and bone malformations. The condition known as superfemale, with karyotype XXX, can cause mental retardation, hypoplasia of the middle third of the face, and inconstant amenorrhea.

Prenatal factors are essential for the prevalence and prevention of intellectual disability. Congenital infections, such as toxoplasmosis, cause significant impairments, including Sabin's tetrad (intellectual disability, microcephaly, intracranial calcifications, and chorioretinitis). Congenital rubella also causes hearing and visual impairment, while congenital syphilis causes physical malformations such as saber tibia and Hutchinson's teeth. Cytomegalovirus is another relevant viral infection. In addition, prenatal malnutrition contributes to insufficient fetal development, and physical factors, such as radiation

exposure, as well as immunological factors, such as blood incompatibility, can compromise fetal development.

The most common prenatal poisoning is caused by fetal alcohol syndrome, which is manifested by mental retardation, growth deficiency, microcephaly, and craniofacial changes. Endocrinological disorders, such as diabetes and thyroid disease, increase the risk of malformations. Intrauterine hypoxia, resulting from uterine hemorrhage, placental insufficiency or intoxication, also interferes with fetal development.

In Brazil, perinatal factors are particularly important due to deficiencies in maternal and child care. Complications such as neonatal anoxia, hypoxia and tocotraumatism, in addition to prematurity, are some of the main causes of intellectual disability.

In the postnatal period, infections, head trauma, malnutrition, and sensory and family deprivation should also be considered. Infections such as bacterial meningoencephalitis (caused by H. influenzae and S. pneumoniae) and viral meningoencephalitis (caused by H. influenzae and S. pneumoniae) are the main culprits. Deprivation, when associated with other risk factors, can aggravate the condition, but they are rarely isolated causes. Even with diagnostic advances, about 30% of cases of intellectual disability remain without established etiopathogenesis, which highlights the need for further studies in the area.

CLINICAL PICTURE OF INTELLECTUAL DISABILITY

The main manifestations of intellectual disability include delays in cognitive development and deficits in social adaptive functioning. Children with this condition often have significant language delays, with difficulties in both comprehension and expression. The response to external stimuli is usually slow, and there are difficulties in discriminating details such as colors and sizes. Global cognitive performance (analysis, reasoning, comprehension, calculation, and abstraction) is compromised according to the severity of the condition, while the ability to concentrate is reduced, with difficulty in remembering information and the formation of inaccurate memories being common.

Emotionally, these individuals tend to have naïve and immature reactions, and many are shy and withdrawn. Emotional control is limited, resulting in impulsive behaviors. Motor coordination is also impaired, with many patients experiencing clumsy and exaggerated movements. Aggressive, self-destructive, and stereotyped behaviors (such as body shaking, head banging, or hair pulling) are frequent in some cases.

Compared to the general population, children with intellectual disabilities are at higher risk of comorbidities, such as epilepsy (22%), cerebral palsy (20%), anxiety disorders (17%), oppositional defiant disorder (12%), and autism spectrum disorder (10%). Symptoms

such as restlessness, impulsivity, irritability, and frequent crying are common, as well as visual and hearing difficulties, present in 5% to 10% of cases. The use of hearing devices, corrective lenses or surgeries can alleviate these problems, but patients with multiple disabilities remain a major challenge in clinical care and care.

DIAGNOSIS AND CLASSIFICATION

The diagnosis of intellectual disability, according to the DSM-5 and ICD-10 criteria, requires the presence of three main conditions: intellectual functioning significantly below average, with an intelligence quotient (IQ) equal to or less than 70; concomitant impairments in adaptive functioning in at least two areas, such as communication, self-care, social skills, use of community resources, self-direction, among others; and onset of symptoms before the age of 18, in the developmental period. IQ is assessed by specific tests that measure various skills, such as reading, arithmetic, vocabulary, memory, and abstract reasoning. Although it has a strong hereditary component, environmental factors also exert a great influence on the development of intelligence.

The classifications of intellectual disability vary according to the severity of cognitive deficits and social adaptation. In mild cases, with IQ between 50 and 70, which account for about 80% of cases, there are delays in developmental milestones, such as walking and talking. These children are able to establish proper communication and learn basic skills, but they have difficulty with abstract concepts and complex reasoning. Generally, they reach the seventh grade and can carry out semi-qualified activities with some level of supervision. In moderate cases, with IQs between 35 and 49, which correspond to 12% of cases, development is slower and learning generally does not exceed the third or fourth year of school. They are able to perform simple activities, but they need constant supervision.

In severe cases, with IQs between 20 and 34, which account for 3% to 4% of cases, children face significant developmental delays and have marked difficulties in language and basic self-care skills, requiring ongoing support. In deep cases, with an IQ of less than 20, which comprise 1% to 2% of cases, the limitations are severe, with most patients unable to take care of themselves. These individuals often experience seizures, associated physical disabilities, and reduced life expectancy.

Diagnosing intellectual disability requires a comprehensive assessment of cognitive and behavioral skills. Children with more severe forms are usually identified earlier, due to the significant contrast with typical development. Mild forms, on the other hand, are often only diagnosed during the school phase, when learning difficulties arise. The differential diagnosis should be made with caution, considering conditions such as specific

developmental disorders, learning disabilities, and factors such as anxiety or depression, which can influence academic and cognitive performance without necessarily indicating intellectual disability.

CLINICAL EVALUATION

The clinical evaluation of patients with intellectual disabilities begins with a detailed anamnesis, addressing fundamental aspects of personal and family history. It is essential to collect data on the individual's neuropsychomotor development, behavior, social interaction, and school performance. In addition, the history of previous treatments, including medications, and quality of life should be considered. In family investigation, the aim is to identify relatives with genetic or neurobehavioral disorders and to investigate the consanguinity between the parents, due to the increased risk of genetic diseases. Maternal history of miscarriages, neonatal death, alcohol or drug use during pregnancy, as well as congenital infections and hypoxemia, may provide clues about possible causes of intellectual disability.

Physical examination contributes to etiological elucidation and to the identification of clinical comorbidities. Detailed evaluations of anthropometric measurements (weight, height, head circumference), in addition to growth velocity, are essential. The presence of body dysmorphia may suggest genetic or syndromic etiologies, guiding the choice of genetic testing. Physical evaluation should include careful examination of the ears, nose, oropharynx, as well as a complete ophthalmologic evaluation, including fundus, visual field, and visual acuity. Cardiopulmonary, abdominal, genitourinary, back, extremity, and skin assessment is also important. The neurological examination should be thorough, considering neuropsychomotor development. In addition, the child's behavior, including attention, impulsivity, motor skills, and social interaction, must be carefully observed. Parental interaction also deserves attention, both to investigate physical characteristics suggestive of genetic disorders and to assess disorders related to caregiver stress or burden.

With regard to complementary tests, specific genetic tests may be necessary, especially in cases with suspected conditions such as Down syndrome, fragile X syndrome, Rett syndrome, and muscular dystrophies. If no genetic etiology is suspected, the microarray technique (CMA) can be used, allowing the diagnosis of up to 20% of previously unidentified cases. In addition, metabolic tests are indicated, especially when there are other clinical manifestations, such as seizures or hepatomegaly. The heel prick test, performed on newborns in Brazil, screens for metabolic disorders such as hypothyroidism,

phenylketonuria, and galactosemia, important causes of intellectual disability. In patients with neurological findings, neuroimaging tests, such as magnetic resonance imaging, may be requested, as well as the electroencephalogram, in cases of seizures or developmental regression.

TREATMENT AND MANAGEMENT

Treatment of diseases associated with intellectual disability, such as phenylketonuria, hypothyroidism, and hydrocephalus, should be initiated immediately to avoid long-term damage. However, the focus of this topic is to discuss the ongoing follow-up and management of common clinical disorders in individuals with intellectual disability, regardless of their etiology. Early intervention is crucial, involving family support and strategies to minimize the impact of cognitive difficulties. The main objective is to optimize the social functioning of the individual, adopting a multidisciplinary approach.

Follow-up of these children should follow a similar pattern to that of typically developing children, including regular childcare visits, vaccinations, growth assessment, and accident prevention. However, it is essential to pay special attention to development, school performance, social interaction and quality of life, in order to detect and intervene when necessary. With the proper support, these individuals can achieve significant progress in learning and communication. Early and continued interventions may be needed in areas such as speech therapy, occupational therapy, physical therapy, behavioral interventions, family support, special school assistance, and nutritional assessment, including guidance on appropriate diets.

Individuals with intellectual disabilities are at increased risk for a range of medical comorbidities, such as cataracts, visual and hearing impairments, congenital heart disease, seizures, and constipation. The identification and treatment of these comorbidities are essential to improve the overall functioning of the individual. The most common comorbidities include seizures, cerebral palsy, gastrointestinal motility disorders, thyroid disease, and behavioral disorders. Neurodevelopmental disorders and mental illnesses, such as autism, attention deficit hyperactivity disorder (ADHD), depression, and anxiety, are also frequent. Additionally, some individuals may be at increased risk for suicidal ideation and substance abuse compared to their typically developing peers. Management of these comorbid conditions should be performed by a multidisciplinary team and include family and educational guidance, specific behavioral interventions, and pharmacologic treatment when necessary. However, the diagnosis and treatment of these conditions often occur late and inadequately.

A common complaint from family members is the presence of behavioral changes, such as repetitive movements, self-aggression and aggressive behaviors. These behaviors can present significant risks to the individual and others, requiring pharmacological intervention and, in some cases, intensive follow-up in a hospital environment. Communication difficulties can complicate the evaluation of behavioral disorders, and it is essential to investigate behavioral changes and explore possible stressors, such as hypoxia, pain, intoxication, infection, trauma, and abuse. A detailed physical examination is necessary to identify potential sources of discomfort, from minor skin lesions to serious conditions such as bone fractures.

Behavioral interventions can be highly beneficial for improving social, behavioral skills, and adaptive functions. Behavioral techniques, such as offering options to choose from and promoting reflections on attitudes and consequences, are helpful. Specific therapies can address problems such as frustrations or needs. For adolescents, group therapy can improve social interaction. Individual guidance on sexuality, transition to adulthood, and preparation for independent living in the community are also important and should be considered.

When behavioral and environmental interventions are not sufficient, pharmacologic therapy may be a necessary option. This treatment is often used in cases with associated comorbidities, although excessive use of multiple drugs should be avoided unless absolutely necessary. It is crucial to consider the potential side effects and drug interactions. Medications for behavioral disorders can negatively impact attention, concentration, learning, and quality of life, as well as cause temporary or permanent motor disorders. Some psychotropic drugs, such as atypical antipsychotics (risperidone, clozapine, quetiapine), tricyclic antidepressants (imipramine, amitriptyline), mood stabilizers, and anticonvulsants, can result in weight gain. Drug intervention, when well adjusted, can improve mental health, functioning, and social interaction, as well as reduce caregiver stress. Children with ADHD may benefit from the use of psychostimulants such as methylphenidate, while those with impulsive behaviors may respond positively to atypical antipsychotics such as risperidone.

Seizure disease has a higher incidence in children with low IQ and in those with cerebral palsy, affecting up to 50% of these patients. This condition is associated with an increased risk of death, and specific guidance should be provided to caregivers. More than one anticonvulsant is often required to control seizures, and serum dosing of these drugs may be useful to maintain therapeutic levels and prevent toxicity.

Cerebral palsy, which refers to a non-progressive motor impairment, affects more than a third of patients with intellectual disabilities. It can be associated with changes such as spasticity, immobility, strabismus, low visual acuity, sphincter dysfunctions, growth changes and malnutrition. Pharmacological treatment for spasticity may include muscle relaxants such as Baclofen®, which has fewer sedative effects. Botulinum toxin can be used to treat dysfunctions in specific muscle groups when medication is not effective. In some cases, orthopedic surgeries may be necessary. Special care should be taken to prevent pressure ulcers and fractures due to bone demineralization in patients with prolonged immobility and nutritional deficiency.

Sleep disorders are common and can impair learning. Initial evaluation should exclude clinical conditions such as obstructive sleep apnea, seizures, and gastroesophageal reflux, and promote good sleep hygiene. Genetic conditions such as Prader-Willi and Down syndromes may be associated with characteristic sleep disorders, and polysomnography may be required for detailed evaluation.

Adults with intellectual disabilities have an increased risk of cognitive decline, and patients with Down syndrome are more likely to develop Alzheimer's disease. However, there are still no standardized criteria to assess memory and cognition in these patients. The diagnosis of dementia must show evidence of worsening cognitive functions relative to previous functioning, excluding treatable causes of cognitive decline such as adverse drug effects, sleep disturbances, social stressors, and metabolic disorders.

Gastrointestinal disorders are common and can include dysphagia and constipation. Dysphagia can be caused by dysmotility of the gastrointestinal tract, esophageal reflux, and gastric emptying disorders. Methods to reduce the risk of bronchial aspiration include modifying the consistency of food and considering the use of gastrostomy or jejunostomy for caloric support. Constipation, often secondary to immobility and lack of physical activity, can also result from medical disorders such as hypothyroidism and use of anticholinergic medications. Treatment should include increased fluid intake, a laxative diet, and, when necessary, the use of laxatives, suppositories, and enemas.

Oral hygiene is an important aspect, with periodontal disease being common. Light sedation with lorazepam may be required for dental treatments in uncooperative children, while deeper sedation, which requires cardiorespiratory monitoring, may be required in extreme cases.

Obesity is prevalent among people with intellectual disabilities, due to factors such as inadequate eating habits, less physical activity, associated chronic diseases, and the use of psychotropic drugs. Monitoring weight, height, and body mass index is crucial, as is

promoting healthy lifestyle habits, including regular exercise and a balanced diet. The potential weight gain associated with some psychotropic drugs should also be considered when choosing drug treatment.

RESULTS

In the systematic review, several studies on intellectual disability were analyzed, revealing a rich diversity in the methodologies used and in the populations investigated. The included studies encompassed a variety of research designs, with a predominance of observational approaches and cohort studies, reflecting different aspects and perspectives of the condition. Most of the research focused on children, but studies with adult populations were also considered, providing a comprehensive view of intellectual disability throughout the life cycle.

The data collected indicate that the prevalence of intellectual disability globally varies between 1% and 3% of the population, with an approximate average of 1%. It was noted that the condition is more common in developing countries, where the prevalence is significantly higher than in high-income countries. This discrepancy is accentuated by socioeconomic factors and unequal access to health care, evidencing the influence of these factors on the prevalence of intellectual disability. In addition, the condition is less frequent in urban areas compared to rural regions, suggesting that environmental and socioeconomic factors play a crucial role in the variation in prevalence.

The etiopathogenesis of intellectual disability is multifaceted, involving a complex interaction between genetic, organic, and environmental factors. Among the genetic causes identified, trisomy 21 and fragile X syndrome stand out, which are often associated with significant intellectual deficits. Other genetic conditions, such as hamartomatous lesions and craniofacial dysostoses, have also been identified in several studies, pointing to a diverse range of genetic etiologies. Organic and environmental factors, including congenital infections and exposure to toxins during pregnancy, remain relevant causes, reflecting the importance of a comprehensive approach in understanding the etiology of intellectual disability.

With regard to diagnosis, intellectual disability is characterized by significantly below-average intellectual functioning, with an intelligence quotient (IQ) of 70 or less, combined with significant impairments in adaptive functioning. The classification of intellectual disability cases reveals a wide range of severity, from mild deficits, where individuals can achieve some degree of independence and basic communication, to severe and profound cases, which require ongoing support and intensive intervention. Difficulties in development

and social adaptation vary considerably, with the severity of deficits influencing the need and type of support needed.

Detailed clinical evaluation is crucial for identifying the causes and comorbidities associated with intellectual disability. Thorough investigation of the anamnesis, physical examination, and complementary tests, such as genetic and metabolic tests, are essential to elucidate the etiology and guide treatment. The presence of physical features suggestive of specific genetic conditions and the identification of comorbidities, such as seizures and behavioral disturbances, have direct implications for clinical management. Early treatment and intervention, which include multidisciplinary support and specific therapies, have shown significant benefits, improving quality of life and promoting the social integration of individuals with intellectual disabilities.

DISCUSSION

The results of the systematic review address crucial aspects of intellectual disability, offering a comprehensive view of its prevalence, etiology, diagnosis, and management. This section discusses key findings, comparing them to existing knowledge, and addressing theoretical, practical, and future directions implications for research.

The review reveals that the prevalence of intellectual disability varies globally between 1% and 3%, with greater frequency in developing countries and rural areas. This pattern suggests that socioeconomic factors and unequal access to health care play a significant role in the observed prevalence. In addition, the etiopathogenesis of intellectual disability is multifactorial, involving complex interactions between genetic, organic, and environmental factors, reflecting the diversity and complexity of the condition. The diagnosis, based on significant deficits in intellectual and adaptive functioning, shows a wide range of severity, from mild deficits to severe and profound cases.

The findings corroborate the existing literature on variability in the prevalence of intellectual disability and reinforce the importance of socioeconomic and environmental factors in determining prevalence. The higher frequency of the condition in developing countries and in rural areas is in line with previous studies that highlight inequalities in access to health care and adverse socioeconomic conditions as critical determinants. The diversity in the causes identified, which include genetic and environmental factors, reflects the complexity of the etiology of intellectual disability and underlines the need for an integrated approach to understanding and management.

The results of this review are in line with previous studies that identified trisomy 21 and fragile X syndrome as common genetic causes of intellectual disability. However, the

review also highlights the growing importance of environmental and organic factors, such as congenital infections and toxic exposures, which may have been underestimated in previous studies. The higher prevalence in rural areas corroborated the findings that socioeconomic factors and living conditions significantly impact the prevalence of intellectual disability, in line with evidence indicating that poverty and lack of access to medical care contribute to the higher incidence of the condition in certain populations.

While the review offers a comprehensive view, there are limitations that should be considered. Variability in the quality of the included studies and the lack of consistent data in some regions may have influenced the results. In addition, the predominance of observational studies may have introduced selection biases and limitations in the generalization of findings. The absence of high-quality studies in some geographic areas also limits the ability to provide an accurate estimate of the prevalence and causes of intellectual disability.

The findings have important implications for clinical practice and public policy. Early identification and multidisciplinary management of intellectual disability are essential to optimize the development and quality of life of affected individuals. The evidence that early intervention and ongoing support can significantly improve outcomes suggests that public health policies should emphasize the importance of early screening and intervention in vulnerable populations. In addition, the review underscores the need for targeted strategies to address socioeconomic inequalities and improve access to health care.

The review identifies several gaps that need to be addressed in future research. The need for more robust, high-quality studies to confirm prevalence and identify specific causes in different geographic contexts is evident. In addition, further research on the interaction between genetic and environmental factors may offer a deeper understanding of the etiology of intellectual disability. Longitudinal and intervention studies are needed to assess the effectiveness of management and early intervention strategies, as well as to develop new approaches to treat and support individuals with intellectual disability.

CONCLUSION

This systematic review provides a detailed view of intellectual disability, addressing its epidemiological, etiological, diagnostic, and management dimensions. The analysis of the available data highlights the significant prevalence of the condition and its geographic variations, reflecting the influence of socioeconomic factors and access to health care. The multifactorial complexity of intellectual disability, involving genetic, organic, and

environmental aspects, underlines the need for a holistic and integrated approach to diagnosis and treatment.

The review also reveals the crucial importance of early intervention and ongoing support, emphasizing that appropriate strategies can lead to substantial improvements in the quality of life and development of affected individuals. Recognizing gaps in the existing literature and identifying areas for future research are essential to advancing the understanding and management of intellectual disability.

In summary, an in-depth understanding of the causes and characteristics of intellectual disability is critical for the development of policies and practices that promote equity in access to diagnosis and treatment. Promoting effective preventive and intervention strategies is vital to meeting the needs of people with intellectual disabilities and their families, ensuring a better quality of life and support that is better suited to their diverse needs.

REFERENCES

1. Bouras, N., & Holt, G. (2004). Mental health services for adults with disabilities. *British Journal of Psychiatry, 184*(4), 291–292. Disponível em: https://pubmed.ncbi.nlm.nih.gov/15056571/. Acesso em: 12 ago. 2024.

2. Campos, G. W. S. (2000). Equipes de referência e apoio especializado matricial: um ensaio sobre a reorganização do trabalho em saúde. *Ciência & Saúde Coletiva, 4*(2), 393–403. Disponível em: https://www.scielo.br/j/csc/a/BLy9snvLVLbQRcZCzgFGyyD/?lang=pt. Acesso em: 20 ago. 2024.

3. Cheniaux Jr, E. (2015). *Manual de Psicopatologia* (5ª ed.). Rio de Janeiro: Guanabara Koogan.

4. Cowley, A., et al. (2004). Descriptive psychopathology in people with mental retardation. *Journal of Nervous and Mental Disease, 192*(3), 232–237. Disponível em: https://pubmed.ncbi.nlm.nih.gov/15091305/. Acesso em: 07 set. 2024.

5. Cooper, S., et al. (2007). Mental ill-health in adults with intellectual disabilities: prevalence and associated factors. *British Journal of Psychiatry, 190*, 27–35. Disponível em: https://www.cambridge.org/core/journals/the-british-journal-of-psychiatry/article/mental-illhealth-in-adults-with-intellectual-disabilities-prevalence-and-associated-factors/3916426981C7C20DCC3DA87B5019C36A. Acesso em: 05 set. 2024.

6. Costello, H., & Bouras, N. (2006). Assessment of mental health problems in people with intellectual disabilities. *Israel Journal of Psychiatry and Related Sciences, 43*(4), 241–251. Disponível em: https://pubmed.ncbi.nlm.nih.gov/17338443/. Acesso em: 22 ago. 2024.

7. Dalgalarrondo, P. (2008). *Psicopatologia e semiologia dos transtornos mentais* (2ª ed.). Porto Alegre: Artmed.

8. Deb, S., Thomas, M., & Bright, C. (2001). Mental disorder in adults with intellectual disability. Prevalence of functional psychiatric illness among a community based population aged between 16 and 64 years. *Journal of Intellectual Disability Research, 45*, 495–505. Disponível em: https://onlinelibrary.wiley.com/doi/10.1046/j.1365-2788.2001.00374.x. Acesso em: 02 set. 2024.

9. Fletcher, R. J., et al. (2009). Clinical usefulness of diagnostic manual-intellectual disability for mental disorders in persons with intellectual disability: results from a brief field survey. *Journal of Clinical Psychiatry, 70*(7), 967–974. Disponível em: https://pubmed.ncbi.nlm.nih.gov/19497248/. Acesso em: 15 ago. 2024.

10. Fonseca, S. C., et al. (2022). Formas de avaliação e de intervenção com pessoas com deficiência intelectual nas escolas. *Revista Brasileira de Educação Especial, 28*. Disponível em: https://www.scielo.br/j/rbee/a/gnRr9tJ6CtppZKMvnjM5YBd/?lang=pt. Acesso em: 13 ago. 2024.

11. Gomes, M. P. C., et al. (2002). Censo dos pacientes internados em uma instituição asilar no Estado do Rio de Janeiro: dados preliminares. *Cadernos de Saúde Pública, 18*(6), 1803–1807. Disponível em: https://pubmed.ncbi.nlm.nih.gov/12488910/. Acesso em: 30 ago. 2024.

12. Kasper, D. L., et al. (2017). *Medicina interna de Harrison* (19ª ed.). Porto Alegre: AMGH Editora.

13. Krahn, G. L., & Hammond, L. (2006). A cascade of disparities: Health and healthcare access for people with intellectual disabilities. *Mental Retardation and Developmental Disabilities Research Reviews, 12*(1), 70–82. Disponível em: https://pubmed.ncbi.nlm.nih.gov/16435327/. Acesso em: 09 set. 2024.

14. Kwork, H., & Cheung, P. W. H. (2007). Co-morbidity of psychiatric disorder and medical illness in people with intellectual disabilities. *Current Opinion in Psychiatry, 20*(5), 443–449. Disponível em: https://pubmed.ncbi.nlm.nih.gov/17762585/. Acesso em: 28 ago. 2024.

15. Martorell, A., Gutierrez-Recacha, P., & Pereda, A. (2008). Identification of personal factors that determine work outcome for adults with intellectual disability. *Journal of Intellectual Disability Research, 52*(12), 1091–1101. Disponível em: https://pubmed.ncbi.nlm.nih.gov/18557967/. Acesso em: 03 set. 2024.

16. Melo, D. G., et al. (2023). Deficiência Intelectual Grave ou Profunda: Investigação Qualitativa de Estratégias Maternas de Enfrentamento. *Psicologia em Estudo, 28*. Disponível em: https://www.scielo.br/j/pe/a/jqM4nsTmhJTXnYVpcvPJBYk/#. Acesso em: 29 ago. 2024.

17. Onocko Campos, R. T., & Furtado, J. P. (2006). Entre a saúde coletiva e a saúde mental: Um instrumental metodológico para avaliação da rede de Centros de Atenção Psicossocial (CAPS) do Sistema Único de Saúde. *Cadernos de Saúde Pública, 22*(5), 1053–1062. Disponível em: https://www.scielo.br/j/csp/a/DgwpmytRqJtNYWFmjfLMtmz/?lang=pt. Acesso em: 25 ago. 2024.

18. Onocko Campos, R. T., et al. (2009). Avaliação da rede de centros de atenção psicossocial: Entre a saúde coletiva e a saúde mental. *Revista de Saúde Pública, 43*(Supl. 1), 16–22. Disponível em: https://pubmed.ncbi.nlm.nih.gov/19669060/. Acesso em: 10 set. 2024.

19. Reiss, S., Levitan, G. W., & Szyszko, J. (1982). Emotional disturbance and mental retardation: Diagnostic overshadowing. *American Journal of Mental Deficiency, 86*(6), 567–574. Disponível em: https://pubmed.ncbi.nlm.nih.gov/7102729/. Acesso em: 01 set. 2024.

20. Salvador-Carulla, L., et al. (2000). Hidden psychiatric morbidity in a vocational programme for people with intellectual disability. *Journal of Intellectual Disability Research, 44*, 147–154. Disponível em: https://pubmed.ncbi.nlm.nih.gov/10898378/. Acesso em: 18 ago. 2024.

21. Schatzberg, A. F., Cole, J. O., & DeBattista, C. (2009). *Manual de psicofarmacologia clínica* (6ª ed.). Porto Alegre: Artmed.

22. Streda, C., & Vasques, C. K. (2022). Síndrome de Down e Deficiência Intelectual: História e Lógica de uma Associação. *Revista Brasileira de Educação Especial, 28*. Disponível em: https://www.scielo.br/j/rbee/a/PVmjj9HscSrG8NY7kXJnSXF#. Acesso em: 30 ago. 2024.

23. Tomaz, R. V. V., et al. (2017). Impacto da deficiência intelectual moderada na dinâmica e na qualidade de vida familiar: Um estudo clínico-qualitativo. *Cadernos de Saúde Pública, 33*(11). Disponível em: https://www.scielo.br/j/csp/a/THGchgJ7SMGKPQK3w4DZ9Xt/#. Acesso em: 28 ago. 2024.

24. Walsh, P. N. (2008). Health indicators and intellectual disability. *Current Opinion in Psychiatry, 21*(5), 474–478. Disponível em: https://pubmed.ncbi.nlm.nih.gov/18650690/. Acesso em: 26 ago. 2024.

25. Zorzi, M., et al. (2023). Avaliação das necessidades e das características da população com deficiência intelectual: Um estudo de caso. *Revista Brasileira de Terapia Ocupacional, 14*(1), 45–53. Disponível em: https://www.scielo.br/j/rbto/a/TbzyWQ8TmLxFtQ8vJxPpYML/?lang=pt. Acesso em: 31 ago. 2024.