CLINICAL CASE

ENFERMEDAD HUÉRFANA CREUTZFELDT-JAKOB; IMPACTOS PSICOSOCIALES PARA FAMILIARES

ORPHAN CREUTZFELDT-JAKOB DISEASE; PSYCHOSOCIAL IMPACTS FOR FAMILY MEMBERS

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RESUMEN

Objetivo: Evaluar el impacto psicosocial en los familiares del caso de la enfermedad huérfana (EH) Creutzfeldt-Jakob (ECJ). **Metodología**: Reporte de caso con diagnosticó de ECJ y entrevista a familiares de los impactos y desafíos psicosociales. **Resultado**: Se realizó cronología de la ECJ y se definió la operacionalización donde se determinó factores psicológicos de la emoción, el malestar psicológico (incluyendo la depresión, la ansiedad y el estrés) y los factores sociales del aislamiento social, el apoyo social a los familiares. **Conclusiones**: La ECJ debe considerarse como un diagnóstico diferencial en pacientes que presentan síntomas psiquiátricos, cambios de personalidad y síntomas neurológicos focales; asimismo, en el momento del diagnóstico de las personas afectadas por una EH, los familiares que experimentaron este proceso de su familiar con ECJ, no solo mostraron una mayor necesidad de atención psicológica durante el tiempo empleado en la búsqueda de un diagnóstico, sino que una vez obtenido este, también sintieron de forma más aguda las implicaciones sociales.

PALABRAS CLAVE: Enfermedad huérfana, Impactos psicosociales, informe de caso, ECJ.

ABSTRACT

Objective: To evaluate the psychosocial impact on the relatives of the case of orphan Creutzfeldt-Jakob disease (CJD). Methodology: Case report with a diagnosis of CJD and interview with family members about the psychosocial impacts and challenges. Result: Chronology of CJD was conducted and the operationalization was defined where psychological factors of emotion, psychological distress (including depression, anxiety, and stress) and social factors of social isolation, social support for family members were determined. Conclusions: CJD should be considered as a differential diagnosis in patients

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presenting with psychiatric symptoms, personality changes, and focal neurological symptoms; Likewise, at the time of diagnosis of people affected by a rare or orphan disease, family members who experienced this process of their relative with CJD not only showed a greater need for psychological care during the time spent searching for a diagnosis, but once this was obtained, they also more acutely felt the social implications.

KEYWORDS: Orphan disease, Psychosocial impacts, case report, ECJ.

INTRODUCCIÓN

The term orphan disease is a general designation used to describe conditions with low prevalence within the population [1]. While there is no universally accepted definition of orphan diseases (ODs), in the Colombian context, Law 1392 of 2010 classifies these conditions as a public health priority, establishing measures to ensure the well-being and social protection of affected individuals and their caregivers [2]. Additionally, these diseases are characterized as severe, chronically disabling, and potentially life-threatening conditions, with a prevalence of fewer than 1 in 5,000 individuals [3]. Although each disorder is rare on its own—amounting to an estimated 6,000 to 8,000 distinct ODs [4]—it is estimated that, collectively, between 6% and 10% of the population is affected by an orphan disease [5].

In 2023, Colombia reported 13,714 cases of orphan diseases (ODs), representing a 3.46% decrease compared to 2022, when 14,206 cases were reported [1]. These figures indicate that a significant number of individuals are affected by an orphan disease—either as patients themselves or as caregivers or parents of those living with such conditions [2].

Although there is considerable variation in the etiology and symptomatology of orphan diseases (ODs) [6], the majority are of genetic origin and tend to be chronic, complex, and debilitating [7]. Unlike individuals with more common conditions, those affected by ODs often face a lack of awareness and understanding from healthcare and social service professionals [8], as well as prolonged and delayed diagnostic processes [9]. On average, it takes more than five years for a person to receive an accurate diagnosis of an OD [10], with some individuals waiting decades and others never receiving a definitive diagnosis [11]. From the onset of symptoms to the final diagnosis—a period often referred to as the "diagnostic odyssey" due to its frequently extended nature—many patients must attend numerous medical appointments, consult with multiple healthcare professionals and specialists, undergo a series of (sometimes invasive) medical investigations, and endure inadequate or inappropriate treatments [12][13]. Even after a diagnosis is made, there is typically a lack of available information about the disease, limited treatment pathways, barriers to accessing optimal therapies, and a scarcity of specialized services and support [9].

In this context, the burden of the disease and its associated challenges often fall on the individual's family members—particularly parents, siblings, and, in some cases, spouses. In addition to navigating complex and often confusing medical processes, parents are most likely to serve as the primary caregivers of individuals with an orphan disease (OD), frequently providing substantial physical, practical, and emotional care [14]. The care required is often extremely

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demanding, intensive, and tailored to the specific needs of the affected individual. Families of those living with ODs frequently face the added challenges of limited access to support services, expert professionals, and relevant advocacy groups [12]. Although there is variation in the clinical presentation and symptoms across different ODs, there appear to be commonalities in the experiences and challenges faced by family members.

For this reason, prion disease was selected as a case study. It is a lethal, neurodegenerative orphan disease that affects both humans and animals. The condition is caused by the misfolding of prion proteins (PrP). Formerly referred to as transmissible spongiform encephalopathy, the term prion was coined by Stanley B. Prusiner in 1982, derived from “proteinaceous infectious particle.” Although prion diseases in humans are extremely rare—with an incidence of approximately two cases per million—Creutzfeldt-Jakob disease (CJD), a type of prion disease, is the most common form worldwide [15].

Most family members and caregivers of individuals with an orphan disease (OD) are required to confront numerous challenges, and it is not unexpected that they often experience a significant caregiving burden, along with adverse effects on their health and lifestyle [14]. Moreover, due to the impacts on both the patient and their spouse, it is unsurprising that other family members—such as parents and siblings—are also frequently affected by having a relative with an OD. While some family caregivers report positive aspects of supporting a person with an OD, numerous studies have found that these challenges often lead to negative psychosocial outcomes and a reduced quality of life among family members of individuals with ODs [9].

The effects of orphan diseases (ODs) can be understood through a biopsychosocial lens, and recent studies have reported that applying this model can help elucidate the broader impacts of ODs and illness in general [16]. Indeed, the World Health Organization’s International Classification of Functioning, Disability and Health (ICF) is grounded in this framework [17], recognizing the importance of adopting a multifaceted approach to health and well-being. A biopsychosocial approach acknowledges that ODs have a biological basis, implying that accurate biomedical diagnosis and, where possible, medical interventions are likely to benefit both the affected individual and those who support them.

It also recognizes that the specific biological characteristics of a given disorder are strongly associated with outcomes. However, biomedical factors do not fully explain the impacts of orphan diseases (ODs), and it has long been acknowledged that psychological factors (e.g., coping style) and social factors (e.g., access to social support) are also fundamental to understanding the influence and outcomes of ODs. The objective of this research is to focus on the psychological and social (psychosocial) effects of ODs on family members.

Creutzfeldt-Jakob disease (CJD) is a rapidly progressive, rare, transmissible, and universally fatal neurodegenerative condition caused by prion proteins. The disease has a long incubation period [18][19]. CJD was first described in 1920 by Hans Creutzfeldt and subsequently in 1921 and 1923 by Alfons Jakob. Later, Clarence J. Gibbs began using the term “Creutzfeldt-Jakob disease” because the acronym “CJD” more closely matched his initials [20][21]. CJD is caused by transmissible agents known as prions, which replicate within the central nervous

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system (CNS) and produce characteristic neuropathological findings, including spongiosis, gliosis, neuronal loss, and deposition of pathological prion protein (PrP^Sc). Human transmissible spongiform encephalopathies (TSEs) include sporadic, genetic, iatrogenic, and infectious forms [22]. Surveillance of human TSEs began in Italy in 1993, and sporadic Creutzfeldt-Jakob disease (sCJD) is currently considered the predominant human prion disease both in Colombia and worldwide. The identification and mandatory reporting of CJD cases in Colombia have enhanced our understanding of CJD pathogenesis, its variable clinical phenotypes, and possible geographic clusters.

Creutzfeldt-Jakob disease (CJD) primarily affects the central nervous system (CNS). The main functional unit of the CNS is the neuron, a unique type of cell capable of receiving, storing, and transmitting information. Neurons in the CNS do not regenerate, although certain brain regions may exhibit limited healing due to the presence of stem cells. The features that make the CNS distinct from other organ systems include the following: autoregulation of cerebral blood flow; protection by the bony skull; special metabolic substrate requirements; absence of a true lymphatic system; circulation of cerebrospinal fluid (CSF); limited immune surveillance; and distinct mechanisms for injury response and tissue repair.

A prion protein (PrP) is a normal neuronal protein predominantly composed of α-helices and random coils. Infectious particles known as "prions" are self-propagating proteins that lack nucleic acids and primarily consist of proteinase K-resistant β-sheet-rich aggregates. Prions replicate by interacting with normal cellular isoforms of PrP, converting α-helices into indigestible β-sheet conformations. These particles cause Creutzfeldt-Jakob disease (CJD) and other transmissible spongiform encephalopathies such as bovine spongiform encephalopathy (mad cow disease), kuru, and scrapie.

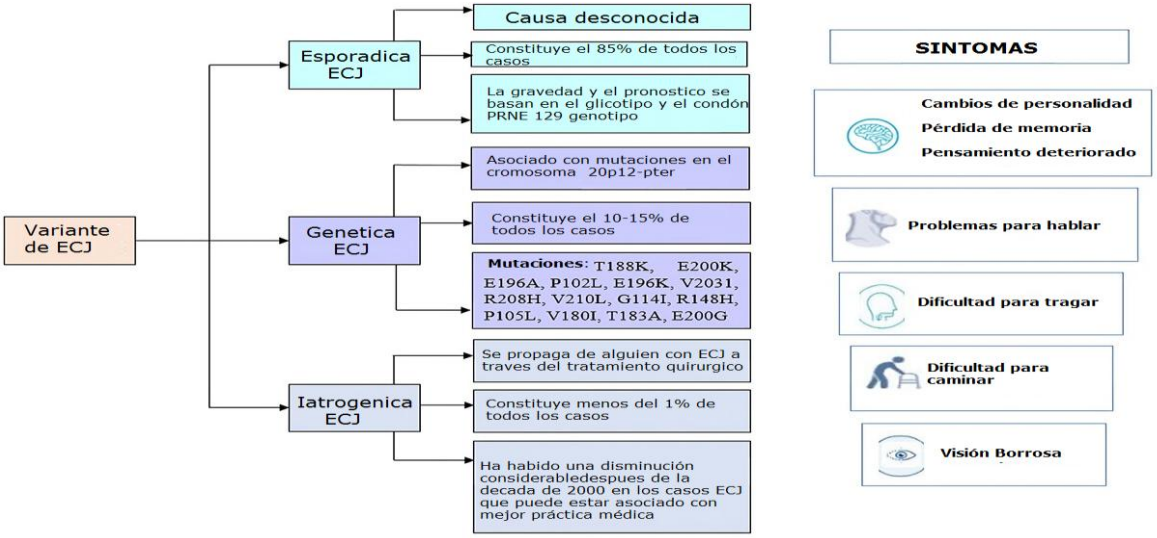
Creutzfeldt-Jakob disease (CJD) can be classified according to its mode of transmission. Sporadic CJD, the most common form (approximately 85%), results from the misfolding of normal PrP isoforms without apparent triggers. Subtypes of sporadic CJD include sporadic fatal insomnia and protease-sensitive prionopathy. Genetic CJD, the second most common form (approximately 10–15%), arises from inherited genetic mutations. Subtypes of this condition include familial CJD, familial fatal insomnia, and Gerstmann-Sträussler-Scheinker syndrome.

Infectious Creutzfeldt-Jakob disease (CJD) accounts for less than 1% of cases and results from the transmission of prions from an external source. Subtypes of infectious CJD include kuru, iatrogenic CJD, and variant CJD. Kuru is a disease of the Fore people of Papua New Guinea, who consumed the brains of deceased relatives as part of ritualistic cannibalism before the practice was banned in the 1950s. Iatrogenic CJD arises from the inadvertent inoculation of prions during medical procedures. Variant CJD is associated with the ingestion of contaminated beef, a mechanism similar to bovine spongiform encephalopathy. The majority of documented variant CJD cases originate from the United Kingdom and France [23][24].

In this regard, Creutzfeldt-Jakob disease (CJD) occurs in three forms: sporadic, familial, and iatrogenic or acquired (Figure 1). Primarily, approximately 85% of cases are sporadic (sCJD), while around 10–15% are genetic (gCJD), and

fewer than 5% are of iatrogenic origin. Very few cases are acquired through infection following exposure to bovine spongiform encephalopathy (BSE). The number of acquired CJD cases has markedly declined as treatment-related setbacks have decreased over time [25][26].

Figure 1. Self-developed (2024). Variants and symptoms of Creutzfeldt-Jakob disease (CJD): This figure provides a concise overview of the different CJD variants—sporadic, genetic, and iatrogenic—alongside their common clinical manifestations.



Creutzfeldt-Jakob disease (CJD) affects approximately one person per million annually worldwide. Approximately 350 cases are diagnosed each year in the United States. Sporadic CJD is the most common form of human prion disease. The disease has a median age of onset of 62 years, although cases have also been reported in younger and older age groups [27][28]. Sporadic CJD occurs with an approximately equal male-to-female ratio. Approximately 1 to 2 new sporadic CJD cases arise annually per 1,000,000 individuals worldwide [29]. Death occurs in nearly 70% of patients within one year of symptom onset. The median survival time for sporadic CJD is 4 to 8 months, with 90% of patients dying within one year.

The normal cellular prion protein (PrP^C) converts into the disease-causing form, scrapie prion protein (PrP^Sc), either spontaneously or as a result of infection by PrP^Sc. PrP^Sc self-propagates and accumulates throughout the brain. Chemically stable β-sheet-rich aggregates disrupt protein folding, ubiquitination, and intracellular protein trafficking in affected neurons. Additionally, astrocytes may swell and degrade in response to prion-induced injury. Neurodegeneration results from these pathological changes [30].

The macroscopic examination of affected brains may not reveal any abnormalities. However, the following features are commonly observed under light microscopy (see Image: Neuropathological Confirmation of Creutzfeldt-Jakob Disease): vacuolization

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or spongiform degeneration, most notably in the cerebral cortex, caudate nucleus, thalamus, putamen, and molecular layer of the cerebellum; neuronal loss; astrocytic gliosis or fibrous proliferation of astrocytes, primarily in the gray matter; and in some individuals, the presence of amyloid plaques.

CASO CLINICO

A 62-year-old male presented with sleep disturbances and nonspecific pain in the upper limbs and cervical region, persistent neck pain, the onset of ataxic gait, and a six-month history of memory loss, behavioral abnormalities, and difficulty walking. The initial symptom was the gradual onset of forgetfulness regarding recent events, which progressively worsened to impair the ability to perform professional tasks.

He developed behavioral changes characterized by decreased interest, irritability, and agitation. Over the following two months, his family observed progressive gait instability. In the subsequent three months, he developed sudden, spasm-like movements resembling electric shocks, initially affecting the upper limbs and gradually progressing to involve the lower limbs as well. These spasmodic movements were triggered by sudden noises. Throughout the disease course, he exhibited signs of bulbar dysfunction and emotional lability. A neurodegenerative disorder was suspected, with a probable diagnosis of Creutzfeldt-Jakob disease (CJD).

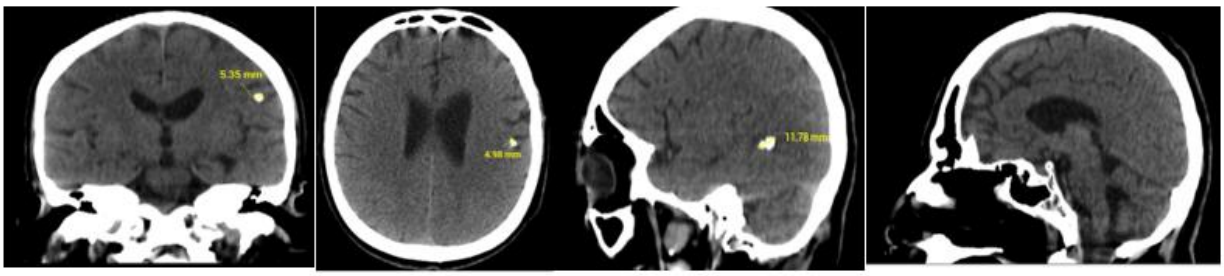

Imagen No.1 de TAC Cerebral de paciente con ECJ

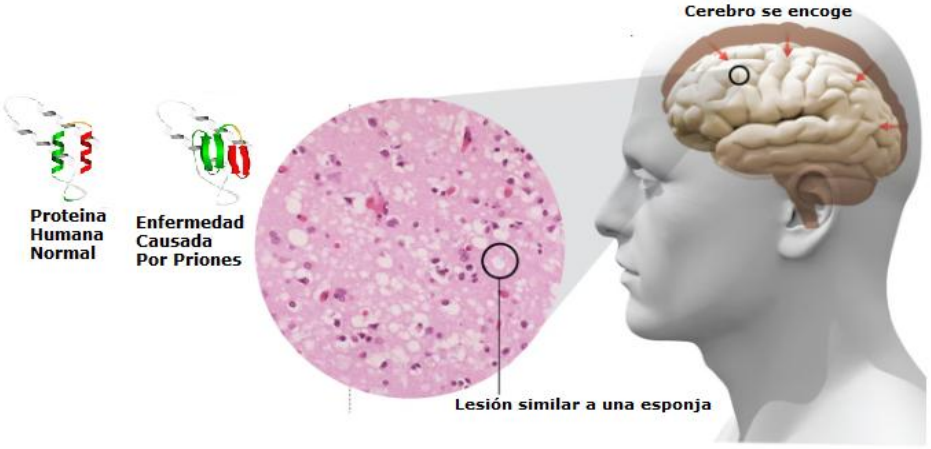

Gráfico No.1, Fuente autores (2024) Caracterización de enfermedad ECJ

In contrast to existing literature, a chronological timeline detailing the progression of CJD is presented for a reported case involving a 62-year-old patient (figure 2).

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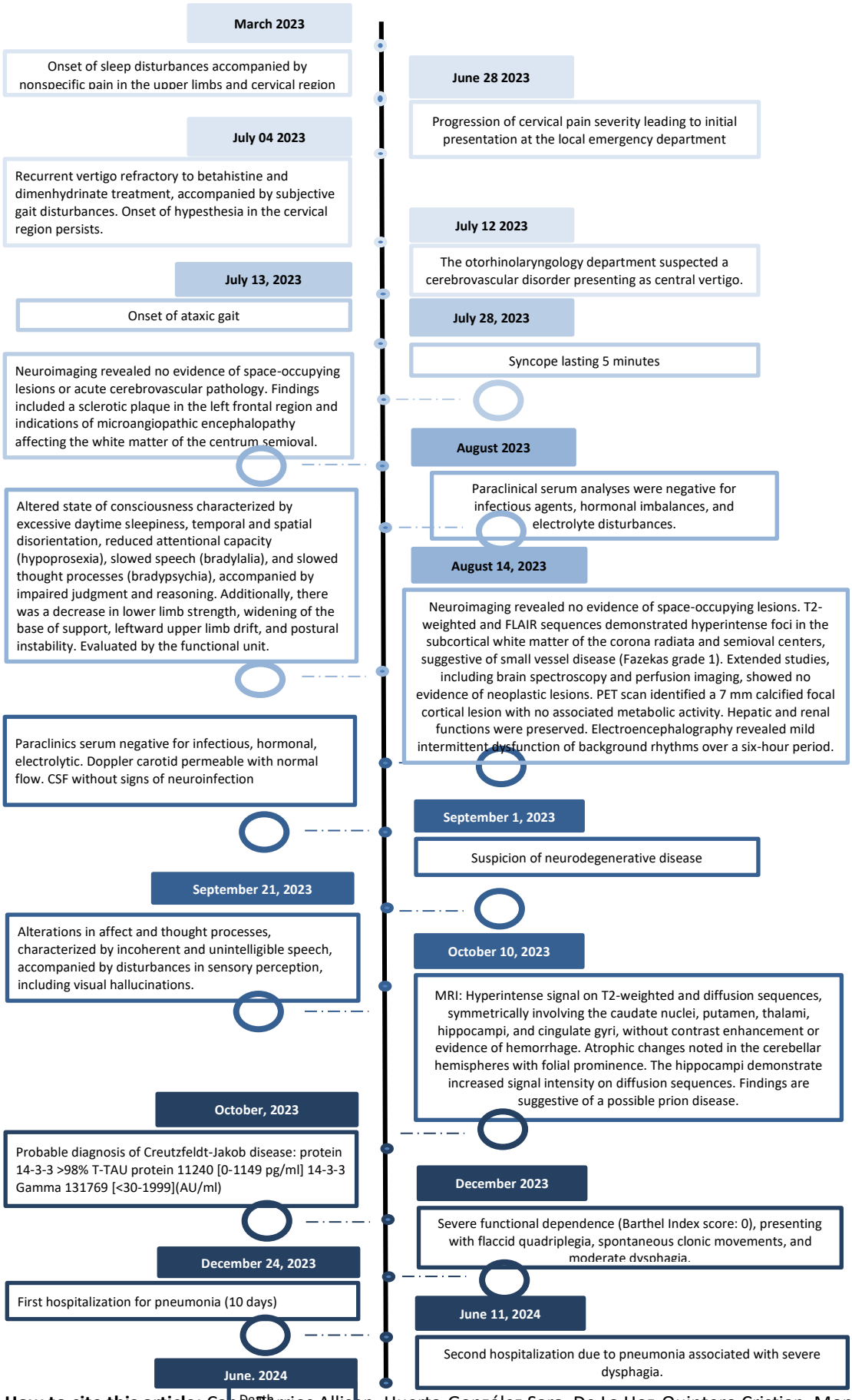

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Figure 2. Source: Authors. Timeline of Disease Progression in a Case of CJD.

The postmortem CentoXome® MOx 1.0 Solo test reported the following findings:
Altered thalamic signal intensity on MRI; thalamic morphological abnormalities; anarthria; caudate nucleus abnormalities; headache; dysarthria; hyperreflexia; hypertonia; abnormal cerebral imaging; history of myocardial infarction; focal T2 hyperintense lesion in the basal ganglia; abnormal morphology of the putamen; neurodegeneration; loss of ambulation; loss of speech; syncope; tremor; and vertigo.
These clinical features were documented in accordance with Human Phenotype Ontology (HPO) terminology.

Inconclusive Result – Variant of Uncertain Significance (VUS) Identified a potentially relevant finding has been identified: a heterozygous variant of uncertain significance (VUS) in the PSEN1 gene. Pathogenic variants in PSEN1 are known to be associated with Alzheimer's disease type 3 with spastic paraparesis, as well as autosomal dominant apraxia. However, based on currently available evidence, the clinical significance of this specific variant remains uncertain.
The variant has been included in the section on potentially relevant findings. No additional clinically significant variants related to the described phenotype were detected.

Tabla 1. Hallazgos principales:

SEQUENCE VARIANTS							
GENE	***COORDINATES OF THE VARIANT***	***AMINO ACID CHANGE***	***SNP IDENTIFIER***	***ZYGOSITY***	***PARÁMETERS IN SILICO****	***ALLELIC FREQUENCIES****	***TYPE AND CLASSIFICATION*****
PSEN1	NM_000021.3:c.1120C>T	p.(Pro374Ser)	N/A	heterozygote	PolyPhen: - Align-GVDG: N/A SIFT: Tolerated MutationTaster: Pathogenic Conservation t:	gnomAD: - ESP: - 1000 G: - CentoMD: -	Missense undetermined significance (clase 3)

The PSEN1 c.1120C>T (p.Pro374Ser) variant results in an amino acid substitution of proline to serine at position 374, located in exon 10 of 12. This variant has not been previously reported in the literature reviewed. According to CENTOGENE's application of the ACMG/AMP/ClinGen SVI guidelines, it is currently classified as a variant of uncertain significance.

Pathogenic variants in the *PSEN1* gene are associated with early-onset autosomal dominant Alzheimer's disease (EOAD), a progressive neurodegenerative disorder characterized by cognitive decline. Although EOAD shares the same clinical phenotype as sporadic Alzheimer's disease (AD), it presents at a younger age, typically before 60 years. The initial manifestations of EOAD commonly include episodic memory impairment or behavioral changes. Patients are often anosognosic, making the presence and involvement of a family member crucial for diagnosis. Neurological features that may be associated with EOAD include spastic paraparesis, intracerebral hemorrhages, seizures, extrapyramidal symptoms, and, in rare cases, cerebellar ataxia.
Mode of inheritance: Autosomal dominant (OMIM®: 607822).

Table 1 summarizes variants associated with disorders that show no clear

phenotypic overlap with the clinical presentation and/or exhibit a zygosity pattern inconsistent with the expected mode of inheritance. For instance, this may include a variant of uncertain significance (VUS) in a gene with only partial clinical correlation, or a single heterozygous pathogenic variant in a gene typically associated with an autosomal recessive condition that only partially overlaps with the observed phenotype.

These variants are included in this report due to their potential relevance to the patient's phenotype and their possible role in addressing diagnostic uncertainties. For variants that may have clinical significance, further clinical re-evaluation and/or additional investigations such as family segregation analysis may help clarify their contribution to the observed phenotype.

Table 2. Secondary findings

SEQUENCE VARIANTS							
GENE	***COORDINATES OF THE VARIANT***	***AMINO ACID CHANGE***	***ZYGOSITY***	***PARAMETERS IN SILICO****	***ATTEND ALLELIC NCIASS****	***TYPE AND CLASSIFICATION ******	***RELATED DISORDER (OMIM®) AND INHERITANCE MODE***
COL4A1	NM_001845.4:c.3260T>C	p.(Ile1087Thr)	heterozygote	PolyPhen: Benigna Align-GVDG: N/A SIFT: Tolerated MutationTaster: Polymorphism Conservation_nt: no Conservation_aa:	GnomA D: - ESP: - 1000G: - Centom D:-	Missense Significado uncertain (class 3)	Small brain vessel disease with or without ocular abnormalities (175780), AD

If consent has been provided, and in accordance with ACMG recommendations (ACMG SF v3.2 list for the reporting of secondary findings in clinical exome and genome sequencing; *Genetics in Medicine*, 2023; PMID: 37347242), secondary findings are reported. These include pathogenic and likely pathogenic variants in genes associated with actionable conditions as outlined in the referenced publication. In this case, no clinically relevant variants were detected in the genes recommended for secondary findings.

Carrier State Findings in Table 2 lists sequence variants previously identified or evaluated and classified by CENTOGENE as "pathogenic" or "likely pathogenic" in selected genes associated with severe, early-onset autosomal recessive Mendelian disorders. As only internally classified variants are included, this table should not be considered an exhaustive list of variants in these genes and does not represent a comprehensive overview of all potentially relevant genetic variants in the patient.

No orthogonal validation was performed for these variants. Therefore, if any of these variants are considered for clinical decision-making, confirmation using an alternative method is recommended. Additionally, variant classifications may change over time as new evidence emerges; however, no reclassification reports will be issued for these variants. It is important to note that the identified variants may indicate additional genetic risks or diagnoses relevant to the

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patient and/or their family.

On the other hand, based on existing literature, the definition was operationalized to encompass psychological factors—such as emotional responses and psychological distress (including depression, anxiety, and stress)—as well as social factors, including social isolation and social support. Although the term *psychosocial impact* may occasionally be considered ambiguous, it has been adopted in this context for the sake of consistency with the existing body of research.

Psychosocial impacts, Although the term "psychosocial impacts" is often used in HE literature and research in this area is expanding, it is not well defined what the term precisely encompasses. [31] It appears to have its origin in the definition of psychosocial health by the World Health Organization and consists of psychological, emotional, and social aspects. [32] However, the components of each of these aspects remain ambiguous. Although there is variation in reports of psychosocial impacts, the most commonly reported include psychological distress, emotional experiences, and changes in social and family relationships, as well as educational, leisure, and work activities. In addition, although not fully considered a psychosocial impact due to the inclusion of physical health components [33] and the frequent omission of occupational and leisure aspects, the terms "quality of life" and "caregiver burden" are sometimes used as synonyms for psychosocial impacts in some literature. [33] [34]

Caregiver quality of life and burden measures are therefore sometimes employed to assess how an individual's well-being may be affected by a medical condition [33], with the resulting data often highlighting certain psychosocial impacts experienced by family members. Alternatively, quality of life is occasionally conceptualized as a psychosocial outcome in its own right [35] or as closely linked to psychosocial well-being, where mental health and social factors influence perceived quality of life and, conversely, quality of life may affect these psychosocial dimensions [36]. Given the absence of a precise definition for psychosocial impacts to date, we define them here as a composite of psychological, emotional, social, and educational/occupational factors that directly affect family members of individuals with Huntington's disease, explicitly including quality of life.

Accordingly, within this manuscript, psychosocial impacts are conceptualized as a broader construct that encompasses, but is not limited to, quality of life. Drawing on existing literature, this definition has been operationalized to include psychological factors such as emotion and psychological distress (including depression, anxiety, and stress), as well as social factors including social isolation, social support, interpersonal relationships, and participation in occupational, educational, and leisure activities. While the term psychosocial impact may at times be ambiguous, it is employed here for the sake of consistency.

Psychological and emotional impacts, regarding the psychological effects of having a family member with Huntington's disease (HD), evidence indicates that caregiving for an individual with HD is associated with a decline in the caregiver's psychological well-being. [14][37] In terms of emotional impact, [38] conducted interviews with relatives of patients with various stages of HD and reported that spouses and parents, in particular, found caregiving to be emotionally exhausting. Family members commonly report experiencing a broad spectrum of emotions, encompassing positive feelings and optimism [39], as well as negative emotions such as guilt, fear, and frustration. [38] Similarly, [40] observed

that brothers of individuals with HD exhibit diverse and complex emotional experiences, often marked by contradictory feelings.

DISCUSSION

This study is among the few that examine the psychosocial impact of receiving a diagnosis of Huntington's disease (HD) from the perspectives of the affected individual as well as their spouse, parents, relatives, or other informants (e.g., family members, guardians, caregivers). Furthermore, the impact was analyzed in relation to whether the patient experienced a delay in receiving their diagnosis. Given that this is a self-reported study, it is common for a greater proportion of respondents to be individuals who have already received a diagnosis.

As noted above, the psychosocial impact is generally more pronounced among individuals with CJD, underscoring the importance of minimizing the time to healthcare engagement (HE). Patients who experienced a delay in obtaining their diagnosis often require psychological support. Those with delayed HD diagnoses report greater challenges within their social environments, including difficulties in explaining symptoms to close friends and family, justifying absences from work or education for medical reasons, and experiencing a lack of psychological support. Likewise, the functional consequences of the disease were more severe in patients with CJD who faced longer diagnostic delays. These individuals were disproportionately affected by loss of independence due to disease progression and by the loss of employment or educational opportunities prior to death.

CJD should be considered in the differential diagnosis of patients presenting with psychiatric symptoms, personality changes, and focal neurological signs. This case is particularly noteworthy as it illustrates that neurological manifestations may follow the onset of psychiatric symptoms in CJD. Therefore, comprehensive assessment of patients with psychiatric presentations should include diagnostic procedures such as lumbar puncture, neuroimaging, and other relevant investigations to exclude organic neurological disorders prior to initiating symptomatic treatment for psychiatric conditions. Adopting this approach can help prevent misdiagnosis of progressive disorders such as CJD and facilitate differentiation from other potentially treatable etiologies.

From the perspective of psychosocial impact, this study examined individuals diagnosed with Huntington's disease (HD) in Ocaña, Colombia, registered in a national patient database, using a purpose-designed instrument that directly captures patients' experiences—one of the few assessments conducted directly with affected individuals. Overall, the psychological impact was found to be significant among people with HD. Family members involved in the diagnostic process not only demonstrated an increased need for psychological support during the often-prolonged period of diagnostic uncertainty but also experienced heightened social challenges (e.g., difficulty explaining symptoms to close friends and family or justifying absences for medical reasons) and functional consequences (e.g., loss of independence and diminished employment or educational opportunities) following diagnosis. Finally, to gain a comprehensive understanding of how various family members are differentially affected—and to elucidate the role of risk and protective factors—further research is warranted. In particular, in-depth

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studies focusing on domains such as employment, education, and economic impact would make a valuable contribution to the currently limited literature.

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