Chapter 17
Creutzfeldt–Jakob Disease: A Prion-Related Neurodegenerative Disorder

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ABSTRACT

Creutzfeldt-Jakob disease (CJD) is a rare disease associated with neurodegeneration mostly characterized by damage to the neurons. CJD is caused by aggregation of misfolded proteins known as prions; thus, CJD is said to be a prion-related illness. CJD and other prion-related illnesses such as Kuru and Gerstmann-Sträussler-Scheinker disease (GSS) have been reported to have complex mechanisms due to their association with the brain and the nervous system in general. A lot of questions have been raised about the mechanism, diagnosis, and pathogenesis of this disease. The complexity of prion proteins themselves have contributed to more questions about the complications of CJD, whether misfolding of the prions are responsible for neurodegeneration or the misfolding are mere symptoms of the disease. This chapter attempts to explore some details about CJD and answers most related questions about the disease's mechanism. The author finally attempts to explore recent development in pathogenesis, diagnosis, and treatment of CJD.

INTRODUCTION

Human NDs have been one of the most deadly disorders with complex mechanisms and complicated treatment processes (Montie & Durcan, 2013).Particularly, thehumanprion diseases are one of the most rare human neurodegenerative diseases widely studied. These are associated with misfolding of proteins which accumulated and cause destruction of cells of the system (Friedman-Levi *et al.*, 2011). These proteins otherwise called prions, are misfolded leading to their aggregation into opaque amyloid structures. The pathogenesis of these diseases are mostly related to accumulation of the amyloid aggregates which in most cases, is associated with neural damage (Holman *et al.,* 2010). Prion diseases are therefore a group of progressive but fatal neurodegenerative disorders affecting both humans and animals, they are otherwise referred to as transmissible spongiform encephalopathies (TSEs) (Jackson &

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Krost, 2014). Some of these prion diseases including CJD, Alzheimer's disease (AD), and Huntington's disease (HD) are having rare occurrence with complicated pathogenesis (Kovacs & Budka, 2008). CJD is a prion related illness characterized by aggregation of misfolded proteins or prions. It belongs to a family of human diseases associated to and or characterized by neurodegenerative conditions and they include; Gerstmann-Sträussler-Scheinker syndrome (GSS), fatal familial insomnia etc. These diseases are thought to be having similar characteristics with animals spongiform encephalopathies such as ''cow madness'' also known as bovine spongiform encephalopathies (BSEs) (Lang, Heckmann, & Neundörfer, 1998). These could have both sporadic or spontaneous occurrence or could be genetical depending on occurring cases. As such as in BSEs, prions could also be transmitted resulting in the spreading of the disease (States *et al.,* 2005). Most researches in this area are focusing towards establishing a link between variant phenotypes of CJD and with the molecular features studied in wide ranges of CJD conditions.

This chapter attempts to explore details on CJD including the basic description of disease, its pathogenesis, classification, diagnosis, and epidemiology. The author objectively attempts to explain different perspectives of the current researches on Creutzfeldt-Jakob and a future sight as well recommendation on the treatment of the disease.

BACKGROUND

Neurons being the major cells of the nervous system could be damaged progressively by one cause or another leading to neurodegeneration and consequently diseases, such diseases are known as NDs (Aguzzi & Zhu, 2012). NDs are diseases associated with damage in structure and functions of the neurons, one of the important cells of the nervous system (Aguzzi & Zhu, 2012). In neurobiology, neurons form the complex network of communication around the central nervous system. Damage to neurons due to a number of factors has been reported to be lethal and associated with a number of diseases and disorders (Murray & Davis, 2003).

CJD was discovered by the German neurologist, Hans Gerhard Creutzfeldt and Alfons Maria Jakob in 1921 (Cordery *et al.,* 2003). Creutzfeldt was the first to describe the disease characterized with neurodegeneration and Jakob proposed the disease to be associated with prions (Holman *et al.,* 2010). However some of descriptions of the disease made by Jakob and Creutzfeldt did not match the current description of the disease (Creutzfeldt, 2002). This including being transmissible as well as currently being observed to be part of a class of both human and animal diseases called TSEs.

CJD as a TSE is associated with and mostly caused by diseased or misfolded prions (Aguzzi & Zhu, 2012). Prions are proteins that often misfold into amyloid structures and can possibly infect other proteins resulting into spreading or transmission (Jackson & Krost, 2014). In other words, prion proteins affect other neighboring proteins converting them into misfolded prion proteins and initiate the pathogenesis of TSEs including CJD (Lang, Heckmann, & Neundörfer). Spongiform refers to the characteristic appearance of infected brains, which become filled with holes until they resemble sponges under a microscope (Aguzzi & Zhu, 2012) . Transmission of the prion proteins in CJD was not initially established (Belay, 2004). The idea that prion proteins can be transmitted in CJD was established in 1986 after the London epidermic (Garske & Ghani, 2010). The incidence of the epidermic has stimulated interest in study of CJD with major aspects on the molecular mechanisms of its pathogenesis (Wilesmith *et al.,* 1988). Studies on prion proteins in fungi has already established the transmissible property of normal prions in the cell (Anderson *et al.,* 1996). Similar prion studies on vCJD was carried on human patients sample which

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