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Discredited legacy: Stigma and familial amyloid polyneuropathy in Northwestern Portugal

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ABSTRACT

Rationale: Genetic inherited conditions may result in feelings of stigmatisation, mainly because of visible physical appearance and its transmissibility to offspring.
Objective: This article reports accounts of stigmatisation from Portuguese patients affected by the inherited neurodegenerative disease, familial amyloid polyneuropathy (FAP), living in the largest cluster of patients worldwide.
Method: We draw on semi-structured interviews conducted with individuals at-risk or affected by FAP, recruited through the national patients' association, about their experiences of stigmatisation related to the illness.
Results: Findings highlight the influence of a discrediting social context in the enactment of stigma. FAP was described as a source of devaluation and social distance and was permeated by beliefs of contagion in the community, especially in the past. The multigenerational nature of the illness within small communities was felt as a source of rejection for courtship and of devalued reproductive worth. Decisions to have (potentially affected) children seemed to be a target of implicit negative judgment. Dealing with stigma entailed restraint in talking about FAP especially outside the family, resistance to being treated as different, and social withdrawal. Some participants referred to recent substantial improvements in their social acceptance and a reduction in the intensity of the stigmatisation to which they are subject.
Conclusion: The pattern of stigma may have changed considerably within the past few decades, as medical information about the disease became more widespread, as new medications have been introduced and as clinical trials of other potential treatments have been established. Our findings report the social consequences of stigma towards this disease group and may help to understand how stigma is experienced in other heritable diseases.

Stigma is a common outcome of many illnesses. Health-related stigma is defined as the social disqualification of individuals and groups who have, or are at-risk of, particular health problems (Weiss et al., 2006). There is abundant research on health- and disability-related stigma, including mental illness (Whitley and Campbell, 2014), HIV (Parker and Aggleton, 2003), obesity (Barlösius and Philips, 2015), and families of children with disabilities (Green, 2003); however, research on stigma as related to inherited genetic diseases, although increasing, is still scarce.

Concerns about potential stigmatisation in genetic disorders have been raised as echoes of the abuses involved in the eugenics of the past (Kerr, 2004). Risks of stigma ensuing from genetic screening programmes are not new (Kenen and Schmidt, 1978). Genetic conditions can result in feelings of social devaluation and discrimination (Cole, 1993; Peters et al., 2005). This is partly because some conditions involve potentially stigmatising visible physical appearance (Gollust et al., 2003; Rickert et al., 1996;

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Rozario, 2007), and these features may be transmitted to offspring, leading to the risk of social prejudice towards an affected child (Clarke, 2013). Discrimination may also be based on the risk of future neurological or behavioural impairment, as is known to occur in the context of Huntington's disease (HD) and some other late-onset neurological disorders (LONDs) (Bombard et al., 2008).

In this study, we report accounts of stigma from people affected by familial amyloid polyneuropathy (FAP), an inherited, LOND leading to severe motor but not cognitive impairments. We draw on semi-structured interviews with inhabitants of the Northwestern region of Portugal where the illness was first identified and which remains the largest known cluster of patients worldwide.

1. Stigma, genes, and geography

Contemporary perspectives on stigma are largely grounded on the work of Goffman (1968), who originally described the relational nature of stigma as it is attached to the spoiled social identity of a person or group. He further distinguished stigma associated with discredited marks easily perceived by an observer, from that associated with potentially discreditable but not readily apparent information. Also, family members of the stigmatised may feel *courtesy* or *affiliate* stigma (Mak and Kwok, 2010) and thus "share some of the discredit of the stigmatised person to whom they are related" (Goffman, 1968, p. 43). Scambler and Hopkins (1986) termed *felt* stigma as the anticipation of stigmatisation that may arise when discrediting events are seen by others (such as epileptic seizures); they also termed *enacted* stigma, as the discriminatory attitudes and behaviour towards those showing discredited conditions. Thus, stigma conveys both the individual's possession of a characteristic viewed by others as devalued and the result of being known by others to possess such a characteristic.

Conceptual analyses of stigma have identified social processes whose effects can be observed within the individual. For example, Pescosolido et al. (2008) view stigma as related to community and individual factors that are set out at the micro (internalised and interactional), meso (local community and social networks), and macro (societal and media) levels. Within this framework, felt (anticipated) stigma contributes to its internalisation, thereby reinforcing its damaging psychological effects. Link and Phelan (2001) conceptualize stigma as the co-occurrence of five components: *labelling*, when people's human differences are given social salience; *stereotyping*, when dominant beliefs link labelled persons to undesirable characteristics; *separation*, whereby labelled persons are categorised as being different from the others; and *loss of status* and *discrimination*, when the individual's ability to participate in the social life of her/his community becomes compromised. This entails a perceived difference in power between the stigmatised and those who stigmatise. Scambler (2009) stresses how this discriminatory environment tends to enact the "vicious circle" of disadvantage, where social exclusion is the end result of the process of stigmatisation.

In the context of inherited disorders, the nature of the disorder and its mode of inheritance have implications for its social impact and potential for stigmatisation. While a genetic cause can ease the potential of stigma because the disorder is not contagious, it can also prompt discredit due to its possible transmissibility and the anticipation of the social prejudice of the risk of having an affected child. The relationship between an identified genetic cause for a disorder and stigma is complex: while perceptions of the stigmatising effect of a hereditary illness may not be directly related to the condition's genetic cause (Sankar et al., 2006), female carriers of sex-linked disorders reported feeling more guilt and being subject to more blame than mutation carriers in autosomal diseases (James et al., 2006).

Some stigmatising diseases have been suggested to represent either tangible or symbolic physical, moral, and health dangers (Major and O'Brien, 2005). If medical information is scarce, people are more likely to rely on myths and beliefs that may contribute to the perpetuation of the stigma attached to their condition. This parallels the influence of the social construction of the illness experience, namely in terms of explanations and causality (Kleinman, 1980; Sachs, 1996). Moreover, stigmatisation is often sustained by everyday social practice and relationships, which make them particularly resistant to change. For example, British Pakistani communities often feel reluctant to disclose information about the family's disease because of the negative impact such information may have on family honour and the marriage prospects of relatives (Shaw and Hurst, 2009).

Studies reporting stigma associated with hereditary diseases in specific geographies are particularly striking in evidencing localised patterns of meaning-making. In Portugal, Boutté (1987) found incest and syphilis as the causal attributions for Machado-Joseph Disease (MJD) in Flores (the Azores), where the disease is highly prevalent. At-risk individuals underwent predictive testing to seek proof of their status as non-carriers as a means of affirmation to the community and so to ensure social integration (Boutté, 1992). Paúl et al. (1999), in a study in central mainland Portugal, showed that people affected with MJD were often labelled as heavy drinkers regardless of their drinking habits. Gait, movements, and speech incoordination would be noticed by neighbours, who would disapprove and gossip about it; this often led to difficulties relating to others and problems in employment. Family dynamics and social life would often become organised around MJD, which would not be spoken about outside the family circle, so that its existence could be concealed.

In the context of HD, Alice Wexler's documentary study traced the route of the disease since the 19th century on the Northeastern coast of the USA. Members of HD families were believed to carry "bad blood," which led to disgrace and social ostracism, as marriage was seen as forbidden to those believed to be tainted with the disease (Wexler, 2008). Stigma relating to reproduction was also reported in the communities living around Lake Maracaibo, Venezuela where HD is highly prevalent (Wexler, 2003). Holguín, in eastern Cuba, has the world's highest prevalence of spinocerebellar ataxia, type 2; the anticipation of social prejudice and fear of stigmatisation were thought to influence the patterns of (non)disclosure of mutation carriers (Cruz-Mariño et al., 2013).

These studies have described a range of negative evaluations towards the discrediting marks of the stigmatised that are generally widely shared within communities and often leading to exclusion and discrimination. Thus, stigma can be culturally dependent, as it is discursively created within a community by a socially negotiated process for identifying and labelling characteristics as atypical (Smith, 2007); there can be a major difference in the social dynamics of stigmatisation, depending on when the condition becomes readily evident to the casual observer and especially whether the condition affects physical appearance from early childhood or neurological functioning from adult life.

2. Familial amyloid polyneuropathy

Familial amyloid polyneuropathy (FAP) type I, also known as paramyloidosis, is a progressive and hereditary autosomal dominant LOND, with onset mainly between the ages of 25 and 35 years (Andrade, 1952). The disease is highly incapacitating and causes dependency and premature death. First described by the Portuguese neurologist Corino de Andrade, FAP has the largest concentration around Póvoa de Varzim and Vila do Conde, just north of Porto, where prevalence is significantly higher than in the country