Psychosocial impact of Huntington’s disease on families and spouses from the perspective of the Family Systems Theory

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Huntington’s disease (HD) is a hereditary neurological disorder with a late onset and a long period of progression that is characterized by changes in personality and physical aptitude. Predictive testing for HD was introduced in 1993 but there have been no major advancements toward a cure since. The disorder is accompanied by emotionally charged issues that affect not only the carrier but the family members as well. The current understanding of the psychosocial impact of Huntington’s disease is limited in terms of both the depth and the breadth of the existent research. The present study examines the literature to date from the perspective of the Family Systems Theory, introduced by Murray Bowen in 1978. A description of the implications of the introduction of the disease into family life as well as a discussion of the various psychological aspects of the disorder are presented in light of four postulates of the theory. Findings suggest that Family Systems Theory may be employed as a useful model for implementation of better family and marital therapy practices for the relatives of the affected individuals because it calls for a more individualized and flexible approach, which is ultimately what is required to meet the unique needs of HD patients.

The focus of the present review is to investigate the interrelationships between four major principles of the Family Systems Theory and emotional effects experienced by individuals in response to Huntington’s disease. An examination of the psychosocial impact of the disease and its relevance to the theory itself as well as to its four specific postulates is presented in detail. The importance of an effective family therapy is discussed as one of the major applications of the psychological research in this area.

BIOLOGICAL BACKGROUND

Huntington’s disease (HD) was first described in 1872. It is a rare hereditary neurodegenerative disease with major observable characteristics being chorea, dementia, and emotional disturbances. The frequency of occurrence is about 5 to 8 people per 100,000, with variable prevalence in different countries. In Canada, 1 in 10,000 Canadians is affected with the disorder.

Huntington’s disease is an autosomal dominant trait, which means that every child of a carrier, independent of sex and assuming that the other parent is a noncarrier, will have a 50% chance of developing HD. First symptoms of the disease usually appear between 25 and 50 years of age, with a period between 35 and 45 being the most common time of onset. The juvenile form of HD is much more rare – it occurs in only 10% of the cases. Common symptoms and signs include: involuntary facial expressions, exaggerated hand gestures, fiddling, rapid and jerky movements, inability to keep oneself stable when walking and/or sitting, problems with chewing, swallowing and talking, mood changes, addictions, aggressiveness, depression, slow thought processes, reduced aptitude for learning new things, and inadequate perception of the surrounding environment. Not all the symptoms are present in any one patient at the same time, nor is there a particular order in which they appear. In addition, the rate of progression of HD also shows marked individual differences – some retire from work right away, while others continue working for years. The time of onset as well as the way the disease develops is very specific for each individual. Death usually occurs within 15 to 20 years after the onset of symptoms, assuming the patient is maintained under adequate care. The cause of death is usually a complication of HD rather than the disease itself. Common causes of mortality include infection, physical injuries, and suicide.

There is no current treatment for the disease. Any interventions focus solely on alleviating the symptoms rather than halting the progression or curing the disease. There are no residential care centres specialized for HD because of the unique needs of the patients, extensive training required by staff, broad distribution of patients over large geographical areas, and an overall substantial financial burden on the health care system. The patients are usually in homecare,
with some privileged families being able to afford regular nurse visits to ease the workload. Many families also choose to attend support group meetings. Prescription drugs such as antidepressants may be given depending on the severity of emotional disturbances of the patient. For the most part, the required care deals with management of routine daily activities such as eating, sleeping, and keeping clean. The primary care supporters are for the most part the partners of the affected individuals.

Huntington’s disease has been given a great deal of attention due to the fact that it is one of the genetic conditions that can be diagnosed using a presymptomatic test. In fact, it is the first autosomal dominant inheritable late onset disorder for which such a procedure is accessible.7 A simple blood test provides results with 96%21 to 99% accuracy.53,57 However, presymptomatic testing raises ethical questions because the result has implications not only on the individual being tested, but on his/her relatives as well. The effects of the testing procedure are felt by the whole family in the biological, psychological, and social functioning of its members.23,28,29 Perhaps surprisingly, it has been reported that both positive and negative results can have serious consequences on the family dynamics.

**PSYCHOSOCIAL IMPACT**

Two major lines of research on HD can be identified: one dealing with the pathogenesis of the disease and the other with the psychosocial impact of the disease. Psychosocial studies can be further subdivided into two streams: the impact of HD on affected individuals, and the impact of HD on the families of such individuals. Even though the research on the former has been quite extensive, the latter has received fairly little attention despite the fact that family plays a critical role in the maintenance of the well-being of a patient. The main reasons for research in this area are the following: insufficient knowledge about the psychological troubles and needs of families affected by HD; inability of health care providers to address all the family problems associated with HD in a cohesive manner; and ineffective current family therapy practices. The information and major findings are presented in the context of Family System Theory, which serves as a unifying framework for the research.3,12,13,26,40,41,49

Family Systems Theory (FST), also known as Bowen Theory, originated from General Systems Theory and was first developed by the psychiatrist Dr. Murray Bowen in 1978 as an effort to bring some guidance with regards to therapeutic interventions used to treat patients.56 His perspective has formed the basis of family and marital therapy as it is today. The essence of the theory holds that a family is one dynamic emotional unit, one system of individuals who are all interrelated, where any changes in the life of one will have an effect on behaviour, thoughts, and feelings of everyone else. The theory argues that family members are interdependent on one another and thus cannot be considered and studied in isolation. A possible justification for the evolution of such interdependence is the potential for strengthening the internal family ties and encouraging common thought and cooperation among its members in order to function more effectively as a whole.56 Therefore, by adopting this strategy of thought, the family can be considered as a separate system with its own characteristics that are distinct from traits of any individual who forms part of the system. FST has eight postulates, but only those that are relevant to the topic of this discussion will be addressed in detail.56,59

There are four basic assumptions of the theory: each family is unique; families are constantly adapting to the changes in the surrounding environment, with strategies differing between families; the growth and development of each member depends on others; members react differently to the major life events that the family faces.57 The last assumption is tightly linked to the notion of the Family Life Cycle – a perception that views a family’s progression through life being divided into six different stages: leaving home, marrying, having children, seeing them go to school, graduating, then marrying and producing grandchildren. The problems tend to arise when the transition between the stages does not go as it should, when there is an interruption of the predetermined “normal” sequence of events. Since, according to the theory, families are (for the most part and to varying degrees) resistant to change, it is during these transition points that the family is very sensitive to any unexpected shifts; the delicate balance is shaken and readjustments of the various aspects of the family structure need to take place in order to maintain stability.56,59 Any disturbance may be extremely stressful because it is unexpected and has the potential to alter family’s structure and identity to such an extent that the family just does not know how to deal with it.51

A good example of a change that throws a family off track is an onset of a chronic illness. The introduction of HD (either through genetic testing or by a direct onset of symptoms) is a change that perturbs the normal Family Life Cycle. The way in which the family accepts the information is subject to much variability from case to case and depends on many factors. Should the influence of any one individual be too great, such that it threatens the strength of togetherness (in this case, a different health status is this influence), the family adopts appropriate coping strategies to ease the tension.58 This adaptation to the situation is a manifestation of the family’s efforts to sustain its function by providing a response to the situation that does not interfere with family structure. Since family life is a cycle and at any given point there are usually members representing different age groups, the effects of HD onset are multigenerational. For instance, an affected child may die before the parent if the symptoms appear early in life.5 Another common scenario would be to see a healthy child caring for an
affected parent, then for an older sibling, and finally getting HD him/herself.24

FAMILY SYSTEMS THEORY AND HD

To take a Bowenian approach to family therapy means to carry out discussions via a circle process. The main advantage of this method is the absence of personal counterattacks, thus allowing the individual who is talking to fully acknowledge his/her sense of insecurity as well as bring out personal worries, unresolved issues, feelings of abandonment and the like. Other family members are given an opportunity to actively listen to the fellow member in an attempt to empathize and see his/her point of view. Reaching a point of mutual understanding allows the family to enter a receptive frame of mind and focus on the role played by everyone in controlling each other’s individuality. From the FST perspective, several aspects of the family structure are potential indicators of the psychosocial impact that might be expected at the introduction of HD. Accordingly, this document addresses the potential improvements to family therapy practices that might arise as a result of a more complete understanding of how the incoming presence of HD affects family’s functioning in light of the following four postulates of the FST: nuclear family emotional system, differentiation of self, family projection process, and emotional cutoff.

DIFFERENTIATION OF SELF

A differentiated self, as defined by Bowen, is the “one who can maintain emotional objectivity while in the midst of an emotional system in turmoil, yet at the same time actively relate to key people in the system” (p. 485). “Self” is a part of one’s character that cannot ever be lost or traded; it is one’s specific autonomy that distinguishes him/her from the rest. In other words, “self” is the level of one’s uniqueness and individuality relative to other family members – the extent to which each member is linked to everyone else and how much information gets shared. In the case of a marital relationship, the level of differentiation of self in each partner determines the degree of common fusion with each other. However, the sense of differentiation experienced by an individual is not only defined by him/herself, but also by the family’s internal structure and patterns of communication. The more undifferentiation there is in the family, the less developed is one’s self, and the more vulnerable he/she is to the external factors and influences by other people. One manifestation of an undifferentiated self is emotional instability in the reaction to a life changing event.

A useful way to examine the process of self redefinition and self reconstruction the person may go through is to consider the spouse of the individual who is either tested for HD or is experiencing early stages of onset. The level of differentiation in the spouse greatly influences how he/she adapts and deals with the situation. The nature of the result is of course another important factor in the impact of the test on the couple. A positive test result leads to more adjustments in the relationship compared to the negative one.6 Regardless of the fact that spouses are not the ones who will be affected by the disease, they take in the results of the test just as closely and as seriously as the at-risk individuals themselves.7,20,35,36

Shortly after the disclosure of the result, the partners experience a range of feelings such as grief, anger, despair, and neglect from relatives and friends since all the attention is focused on the diagnosed carrier.19,22 A lot of the mentioned emotions might arise simply because of the unpreparedness of the partner to face the reality of HD – a good proportion of partners cannot predict the possible impact of HD on their future (30% as reported by Tibben et al.).7,20

Tibben also found that half of carrier partners did not have anyone with whom they could share their feelings about the test result. Interest in the actual disease was observed in less than half. It is common for partners not to show their initial stressful reaction to the carrier for fear of hurting their feelings.20,23 Also, since many usually do not have a support person to listen to them, partners do not have a choice but to keep everything inside, resulting in increased emotional tension and even deeper level of internal stress regarding the result. Spouses often face the challenge of finding a balance between their own autonomy and personal needs, feelings of responsibility towards family members, social perceptions, and dependence on external support. There is constant pressure that everything depends on them, they can’t go away, get sick/injured, or even just relax.13

To manage their emotional functionality, spouses use a variety of different coping strategies. Some partners exhibit a very passive coping strategy by becoming withdrawn and harbouring their emotions inside in order to comply with social perceptions that they have no valid reason to be distressed since they are not the ones who are actually affected.14 On the opposite end of the spectrum, others actively research information on HD, keep up to date on the current research, attend support group meetings, and seek help from other people in similar situation. A correlation exists between the coping strategy used by the spouse and their quality of life – specifically, the more the spouses limited themselves from any activities other than primary care, the more they felt the negative impact of HD on their lives.2,20 Nevertheless, the spouses need to learn to deal with the feelings of guilt that accompany any recreational outing that does not include the patient because those psychological breaks are absolutely essential to relieve the tension created by caregiving.47 The higher the spouse’s differentiation of self, the more capable they are to take control and successfully address the issues pertaining both to themselves as well as to the affected partners.
Examining the family of origin of the tested individual, in addition to the spouse, allows a more complete examination of the impact of a predictive test result on the level of differentiation in close relatives. Usually, HD would remain in the background until an apparent onset of symptoms in one of its members or until age brings an anticipation of symptoms every day – at this point, HD becomes part of daily life and starts to get more attention. With the introduction of predictive testing, the situation has changed and nowadays the HD can come in to the family at any point of the life cycle, depending on which family members and at what time decide to undergo the procedure. Interestingly enough, however, the test does not turn out to be as personal as it might seem at first; the rest of family members report feeling as if they are being tested as well when one member decides to undergo the procedure. Some of responses mentioned by different members include feeling confused and challenged on the family’s structure, rituals, beliefs, and values. There is an observed shift in the family’s identity where everyone becomes more independent from each other. In the study by Sobel and Cowan, over half of participants experienced changes in membership, patterns of communication, and roles. In some families, unaffected siblings feel the pressure of taking on the caregiving role for the affected sibling(s). Where there are multiple potential caregivers in the family, conflict may arise on the grounds of who will take on what responsibilities and for how long. In summary, the introduction of HD into the family forces each member to take on a certain attitude and respond in a way that corresponds to his/her level of differentiation in relation to other members.

In addition, not all the members of the family get a chance to find out about the test results and among those who do, not all receive the same information. Each tested individual has a particular set of criteria used in the process of deciding how, when, and to whom the results will be told. Some report having made the decision even prior to the test. This pre-test preparation of plan of disclosure allows the members undergoing testing to decide the level of differentiation they would like to maintain with the family. Selective disclosure to certain family members is based on their individual vulnerability, which is in turn governed by the current stage in the Family Life Cycle, level of closeness, current occupation, and maturity level. Patients can also choose not to tell specific individuals for fear of receiving inappropriate judgement. The time of disclosure is usually carefully picked out and well planned ahead, taking into account major life events such as graduation and marriage. The “right” time is judged as a proximity of an important life event whose outcome might be influenced by the knowledge of the risk of HD – for example, when offspring decide to become parents themselves. The amount of detail in the disclosed information also varies. Some release what they know right away, while others prefer to disclose information in doses over an extended period of time. Therefore, variability in the method of delivery or a complete failure to tell is guided by the fear to weaken the familial bond and/or the desire to protect the relatives from potentially painful and/or distressing information.

**EMOTIONAL SYSTEM**

It is important to note that the atmosphere in the HD-affected family is very different from a normal family. The presence of HD might be associated with several types of losses experienced by the family: loss of both the personality and the physical aptitude of the affected parent, loss of the usual family structure, and potential loss through death. The fact that there is not an abundance of knowledge regarding HD forces the families to develop their own mental image of the disorder, create their own meaning of the disease and its potential impact on the family. The continuing social perception of the permanently ill as being useless members of the society can contribute to the reported feelings of family members such as shame, secretiveness (the two most commonly noted), depression, and social isolation. One study notes that there is a decrease in secrecy about HD nowadays as compared to past generations. A plausible explanation might be a more empathetic and accepting social attitude towards the families affected with the disorder due to the increased awareness and promising research developments. Nonetheless, the experiences of the HD-affected family are both unique and complex, which is why it is important for health care professionals and support workers to fully understand their needs.

Bowen sees emotions as instincts that constitute a drive for survival for both oneself and the members of one’s community. As such, the nuclear family emotional system can be perceived as a structural framework that the family continuously builds and remodels in order to ensure its function in the changing environment. It is an attempt to manage anxiety and feelings associated with stress that accompanies changes. Bowen proposes four basic relationship patterns adopted by each family to varying degrees that ultimately determine the family’s ability to function, but only two of them are highly relevant to the topic of HD: underfunction/overfunction and marital conflict. More specifically, the first pattern describes the maintenance of the function of some members at the expense of others, which occurs in cases when there is an unequal distribution of anxiety among members – some experience more, some less. This unbalance stems from a reduced ability to function (either mentally or physically or both) by one individual thus indirectly forcing others to compensate for the unfulfilled role and take on extra responsibilities, in other words, overfunction, with a purpose of maintaining stability.

One of the most common observations in the HD families noted by several studies is the phenomenon of preselection. Preselection refers to a certain belief held by the family whereby there exists an unconscious
classification of each member’s disease status as either a carrier or a noncarrier. The preselection takes place when the individual is still a child and the belief is supported throughout life via certain behaviour toward the individual by the relatives.\(^{30}\) For example, if the family believes that a child is unaffected, he/she is encouraged to pursue post-secondary education, look for a promising career, have children, etc., whereas there are not as many expectations for a child who is perceived as a carrier. In addition, there might be an unequal distribution of love, attention, and financial support among offspring.\(^{40}\) The children might be sent to different schools and given different advice with regards to important life decisions according to the assigned HD status. To worsen the scenario, preselected “carriers” are inclined to fully accept the assigned status and make it a part of their identity, their sense of “self.” Moreover, they tend to exhibit mimicry of movements and verbal acceptance that they indeed have HD, thus further solidifying the family’s beliefs.\(^{40}\)

The preselection coping strategy serves as one of the main defence mechanisms of the families who have a history of HD to combat the stress and anxiety associated with uncertainty of the future. It is a way that the family can “fool” nature by artificially making the diagnosis themselves, eliminating the uncertainty of someone’s status, and providing an illusion of having control over something that is essentially uncontrollable. By abstractly determining the status of each member the family can then move on and organize itself according to the expected roles of carriers and noncarriers – this brings more structure and stability into the family system. More specifically, “carrier(s)” would be considered as dysfunctional member(s) whereas “noncarrier(s)” would take on the role of overfunctional member(s). Consequently, a family-specific pattern of emotional functioning develops and becomes the major force that holds the family together.

Once the preselected offspring grow up and enter a new life stage, the dysfunction/overfunction model becomes very relevant in a marital relationship, where the deficiencies in the HD – affected partner are compensated by the healthy partner. Caregiving is a full time occupation that requires dedication and commitment and is certainly not an easy role to take on; caring for an HD-affected individual can prove to be very problematic due to a myriad of disease complications, a wide variety of symptoms, and the lengthy course of the disease. It also requires assumption of additional tasks that were previously carried out by the patient, such as earning money, caring for the children, housework, etc. It has been reported by several studies that spouses of chronically ill patients have a greater chance of having physical, social, and mental problems during their lifetime as compared to the average population.\(^{2,24,46}\)

Greater impact is experienced by those caring for patients with adverse behaviour, severe stages of the disease, and prolonged duration of illness.\(^{30}\) One study even established caregiving as an independent mortality factor,\(^{46}\) possibly due to insufficient amount of time devoted to self care. Common reported feelings include being overburdened,\(^{24}\) stressed, tired, and unable to successfully cope with the disease. A positive correlation has been reported between the duration and the severity of HD in the patient and the negative impact on the daily life of the spouse.\(^{1}\) This is certainly true for the later stages of the disease, when the increasing demands of the patient drain a lot of energy from the spouse, eventually making it impossible for the patient to remain at home under the sole caregiving of one person.\(^{24}\) Indeed, the demands on some spouses are just so great that they end up “joining” their partners in illness.\(^{24}\) One might ask why would someone take on such a burden and sacrifice his/her own mental and physical health. Possible reasons presented by the literature include a sense of commitment, guilt, loyalty, and compassion towards the dysfunctional individual as well as feelings of obligation to oneself and to other family members, in addition to fear of judgement by the society.\(^{2,24,44}\) Due to the possibility of such devastating consequences, it is essential to encourage caregivers to maintain their own health and well being in order to be able to fulfill their role successfully.\(^{13}\)

The second emotional relationship pattern that can be adopted by the nuclear family is marital conflict, which develops in situations when neither spouse will yield to the other. It is more common to observe this relationship pattern during the early stages of HD, when the healthy spouse is still adjusting to the new reality. The changing behaviour of a patient might cause a lot of grief to both the immediate and the extended family. Some examples include self-centeredness, unrealistic demands, insistent and inappropriate sex remarks.\(^{27}\) Initially, the spouses might be annoyed by the lack of activity coming from the patient when they are fully aware that the patient is still physically capable of performing those activities, especially with regards to chores around the house.\(^{47}\) However, any attempts by the spouse to discuss household issues with the patient, such as the latter’s discontinued contribution to the family’s functioning, often turn the attempted conversations into useless arguments due to the patient’s quick temper and poor judgement of the situation.\(^{27}\) In addition to more household work, there arise financial difficulties due to disease-related expenses and decreased overall family income. Two most difficult things to deal with were identified as mental deterioration of patients and fear of having passed the HD gene to the children.

In the long run, it is inevitable that the nature of the intimate relationship of a couple will change. The actual impact on the relationship cannot be generalized since it largely depends on the time spent together, strength of the bond between the two people, personality and competence of each spouse, presence of any defects in the noncarrier partner,\(^{5}\) presence of any children in the family, age of onset of HD, and whether or not the spouse knew about the HD.
risk prior to marriage.\textsuperscript{24,44,47,49} Partners of those who knew about the recurrence of HD in the family before marriage tend to have less difficulties. In an ideal situation, there would be prior knowledge of HD, late onset, well informed and caring spouse, regular nurse visits, long and happy marriage, and grown up children who have already moved out of the house.\textsuperscript{47} Spouses who have spent together many years feel an obligation to care;\textsuperscript{50} they report loss of someone who knew them the best, loss of their main supporter – overall, they feel as if they lost a part of themselves.\textsuperscript{44}

One of possible unfortunate outcomes of the presence of HD is the subsequent separation of the partners, which often occurs within 2-3 years.\textsuperscript{50} Divorce is observed to be more common in younger couples, specifically in those under 40 years of age or in those who have been together for a relatively short period of time (few years).\textsuperscript{5} The spouses might directly justify their demand for divorce by the need to take care of the children vs. the actual carrier as well as the desire to minimize the children’s exposure to the sick parent.\textsuperscript{44} Other possible reasons (that remain unspoken for the most part) suggested by several studies with regards to divorce include the degree of unexpectedness of the genetic test result, presence of marital problems prior to the test, unequal contribution to the relationship, emotional distancing, irrational arguments, and anxiety.\textsuperscript{5,12,32} Also, failure of the at-risk individual to inform their partner prior to marriage may become the rationale for separation once the spouse finds out and feels hurt and deceived.\textsuperscript{24} It is interesting to note that according to a large study done in 1983 that included 92 families, the divorce rate among HD affected couples was not significantly higher than in the general population.\textsuperscript{50} It would be useful to find out whether and how this statistic has changed in the past two decades.

A strong ground for marital conflict comes from the different reactions of the partners to the predictive testing. Even though there is still some time prior to onset of HD, predictive testing nevertheless brings HD into the foreground and stirs new emotions. It might come as a surprise to some that a test result indicating low risk of HD might still have an effect on the couple because even though the partner does not experience any stress related to the disease, the noncarrier still feels the pressure because the test result has an impact on his/her family.\textsuperscript{22,32} A different possibility is that some participants could have linked HD to problematic aspects of their life and a noncarrier status thus no longer justifies the use of HD as a cause for failures.\textsuperscript{32} If the result does not match the status assigned by the family via the preselection process, then the noncarrier might experience feelings of guilt and anxiety,\textsuperscript{22,28,32} which will ultimately have an effect on the spouse. In contradiction to many studies, a large 2003 cohort study actually concluded their paper with the following statement: “Importantly, however, this study clearly showed that for those persons receiving a decreased risk result, improvement in quality of life is significant and sustained for at least 5 years”.\textsuperscript{34} However, this statement might not be as contradictory as it seems at first, after careful examination of the wording: quality of life of the individual might indeed improve based on the fact that they do not need to worry any more about their own future; the emerging stress might in fact be due to the anxiety that the individual might experience towards the family members. In other words, the results can be interpreted more as a shift of focus of the stress from one area (future of oneself) to another (future of the family).

It is easier for partners of both carrier and noncarrier groups to deal with the test result if they find out at the same time as their spouses. However, this does not eliminate the importance of partners knowing about the history of HD beforehand – previous awareness of the risk of HD smoothens the shocking effect of genetic testing.\textsuperscript{7,22} The ease of acceptance may in fact be due to spouse selection,\textsuperscript{22} where one partner wants to care while the other wants to be cared for – if the test result fits into the scheme, the relationship works out really well.\textsuperscript{5} But in a case of an unexpected decreased risk, partners might feel like they have lost their caregiving job for which they have mentally prepared themselves.\textsuperscript{25} Overall, the relative ease with which the result is taken is determined by whether or not it matches with the expected status.\textsuperscript{8,26,28} In addition, the presence of children in the family greatly increases the level of distress in partners of carriers.\textsuperscript{13,36}

With regards to carrier couples, both individuals go through the same course of emotional fluctuations during the post-test period: an initial high level of stress, which then subsides, but subsequently rises once again.\textsuperscript{7,20,22,23,36} There is an interesting difference between the type of reactions of the partner and the at-risk individual to the test result. In the case of a carrier status, the at-risk individuals were more concerned with the immediate future, whereas their partners worried about the distant future, children, finances, family stability, caregiving, etc.\textsuperscript{7,22,24} Some investigators actually found that partners of carriers were more depressed and had a more pessimistic outlook than carriers themselves.\textsuperscript{5,20,22} They were also more inclined towards adopting passive coping strategies.\textsuperscript{14} Indeed, the prospect of major changes in the intimate relationship, possibility of passing the gene to children and then having to tell them later, as well as potential caretaking responsibilities might not be very optimistic. All these concerns about the future can lay the ground for conflict in the present.\textsuperscript{26}

Couples who decided not to undergo testing were significantly more stressed in the relationship at the baseline.\textsuperscript{23} Thus, individuals and/or couples who refuse to undergo genetic testing when being offered such an opportunity by a health professional, might indeed be the ones that require more intense therapy and more social support. In other words, avoidance of testing may be an indirect sign of the presence of psychological problems and emotional distress in the patient and/or the relationship. As such, not even predictive testing itself but the mere
possibility of it might be too stressful for the couple to endure because the presence of HD brings in more than enough stress into the relationship. This is an important issue that should be taken into consideration by health professionals and/or counselors who provide support to the patients that are eligible for testing.

**FAMILY PROJECTION PROCESS**

Family projection process is closely related to the nuclear family emotional system because it is frequently accompanied by some form of marital conflict and/or some dysfunction in one of the spouses. Family projection process refers to a pattern of interaction whereby parents’ fears and anxieties are transmitted onto children in situations when the former cannot accept all the implications of the situation they are going through. In other words, it is an adopted mechanism for managing excess levels of undifferentiation in the nuclear family emotional system. When the level of anxiety in the couple is high, the high level of undifferentiation that results needs to be absorbed. By subjecting children to their problems, parents are able to diminish the level of internal tension between each other and thus maintain functioning. However, the problems are not erased – they are just passed on to the more vulnerable members of the family. The HD affected parent’s fears and anxieties have a strong impact on a child and thus carry a risk of transmitting a low sense of self to the children.

Since children’s perception of HD is largely affected by how parents behave and deal with the situation at home, then family dynamics, taboos, expectations, loaded issues, and the general attitude toward the disease carry a lot of weight. According to the train of thought developed by Duisterhof et al., every human being has a need early in life to create an intimate bond with a caregiver in order to have someone to turn to in times of trouble – such development of an attachment system enhances an infant’s survival. Proper parenting practices result in a secure bond, which then serves as a model for the child in any future social situations, both intimate and public. Psychological manifestations of an affected parent such as absence of attention, harsh discipline, inconsistent expectations, irritable and demanding attitude and subsequent rage can all have a profound effect on the strength of attachment between the parent and the children, resulting in a weakened relationship and traumatic experiences of childhood. As a result of the family projection, a very interdependent relationship develops between the child and the parents, where the child becomes caught in the parents’ emotional trap that prevents him/her from creating his/her own differentiating position.

The childhood experiences subsequently have an influence on the psychological development, the course of life and decision making process concerning major life events. In one study, 43% of the tested offspring of HD parents had some form of a psychiatric disturbance. In particular, 25% had either conduct disorder or antisocial personality disorder (both of which were strongly related to the breakdown of family structure) and 18% had major depression. Young children living in such families are at considerable psychological and social risk. These findings are consistent with the theory that HD affected parents have a high level of undifferentiation, which is “inherited” by the children.

Fortunately, the transmission process does not seem to be physically oriented nowadays. Prior to 1983, several clinical investigations reported high rates of child abuse by HD affected parents. Since then, however, there were not any studies that specifically focused on child abuse as one of their findings. According to one study, patient’s abuse is more often directed toward the spouse as opposed to children. This may be explained by the relatively larger proportion of time that the patient spends with a spouse as compared to children and the relatively inactive role played by the children in the caregiving responsibilities. Indeed, children are probably the most psychologically vulnerable individuals for whom the acceptance and tolerance of HD is extremely difficult and it is actually not recommended for at-risk offspring to be a support person because of their increased sensitivity to potential emotional damage.

**EMOTIONAL CUTOFF**

Emotional cutoff is a term used to describe the creation of a psychological barrier that makes a person “blind” toward a current issue that is emotionally charged, in an attempt to manage certain aspects of the relationship that have not been resolved in the past. Bowen associates this approach with those who have a low sense of self. Insufficient confidence in one’s ability to deal with the situation results in an emotional closure whereby any thoughts related to the dreaded issue are suppressed. It is a process of one’s separation, isolation or withdrawal – a mechanism adopted to deviate away from the sensitive topic. Emotional cutoff can be seen in family members and spouses of an individual who has been diagnosed with HD prior to the onset as well as during early stages of the disease.

The coping strategies of avoidance and denial are so common among partners and other family members that they deserve special attention. While both avenues constitute an effort for self-protection from devastating emotions by bringing relief, comfort, and alleviation of pain, denial is considered as an unconscious defence mechanism whereas avoidance is a conscious act. The onset of the disease is associated with having to face and process potentially burdensome and distressing information, which can thus be considered as a traumatic event with painful emotional consequences such as depression, helplessness, and uncertain future – all of which might be difficult to take in at once. They are usually exhibited during the initial stages of onset.
and demonstrate an adopted attitude of family members and spouses to deal with problems as they come up. Denial of the presence of HD in a carrier masks the fact that the secondarily affected parties are not mentally prepared to face it, that they need time to process the information and adapt to the situation, even if the possibility of risk was known beforehand.\textsuperscript{24,53} However, long term consequences of prolonged denial that extends well into the progressive stage of the disease can be devastating since it does not leave room for adaptation to reality. When the disease becomes serious later on and there is a greater need for a more professional care, the family is not adequately prepared to handle the situation.\textsuperscript{40,53}

Disclosure of a genetic test result can serve as a powerful trigger that initiates emotional cutoff. Revelation of genetic results (either negative or positive), means a loss of a particular type of connection that held the family together in hope. The obtained certainty might expose the reality that might not be wanted.\textsuperscript{9} In a family with a high recurrence of HD, a negative test result can lead to loss of membership of the noncarrier because he/she is no longer concerned with a problem that occupies everyone else.\textsuperscript{23,26} Prior to testing, uncertainty of the future overshadows all members, keeping everyone under the same umbrella and unifying them in a common anxiety; finding out about a decreased HD risk upsets the preexisting equilibrium and breaks this bond.\textsuperscript{23} From the point of view of the family, the noncarriers fall out of the family structure because of the certainty of their future, absence of fear of onset, and no concerns for their children.\textsuperscript{25,51} On the other hand, if the disease occurrence is rare in the family, a positive test result will be very unwelcome because it would bring the problem back to the surface.\textsuperscript{26} The emerging presence of HD sets the carrier apart and introduces new and undesired emotions to everyone else; it therefore has an effect of disrupting the overall balance in the family. In both cases, the individual who had undergone testing feels rejected (to varying extent among different families) and psychologically alienated from the rest. Even though his/her different position might not necessarily be verbally acknowledged, it is nevertheless evident through the post-test communication pattern adopted by the family.

It is important to discuss the situation that involves a decreased risk result a little further in order to specifically illustrate the potential effect of predictive testing on the siblings. It is common for the possibility of developing HD to be seen as a “zero-sum game” in families, meaning that decreased risk of one means increased risk for another.\textsuperscript{9,23} This perception about the risk especially pertains to siblings. They are often not informed about the result due to anxiety about their reaction – the tested individual is well aware that in the minds of siblings, the result directly translates to either increased or decreased chance of developing HD themselves.\textsuperscript{7,16} Broken bonds between siblings might result from the feelings of guilt of behalf of the noncarrier\textsuperscript{3,25,28} and feelings of unfairness of behalf of the sibling.\textsuperscript{17} An apparent contradiction to the proposed behaviour is evident in the 1993 study by Tibben et al., where 90% of noncarriers shared test results with their siblings as compared to 75% of carriers who shared (those who did not feared confrontation and stigmatization).\textsuperscript{20} Even though several studies have addressed the topic of test result disclosure in the family, no statistical evidence was gathered about the pattern of disclosure employed by the tested individuals toward their siblings to support the statements. However, the conflicting information provided by the above mentioned study is not enough to reject the possibility of existence of the “zero-sum” game perception of risk among siblings of tested individuals. Therefore, more research in this area would bring more clarity on the issue.

**FAMILY THERAPY**

The main goal of examining the psychosocial impact of HD on families is to improve current family and marital therapy practices. Lack of residential care facilities specialized for HD patients creates significant financial and emotional burdens on the family. Lack of availability of treatment and the unknown future may cause the family to ignore any professional help; meanwhile, regular visits by a nurse might actually greatly alleviate some of the stress and provide psychological support.\textsuperscript{30} Judging from the reviewed literature, there is insufficient research on specific supportive care needs of families touched by HD.\textsuperscript{13,51,54} Moreover, health care professionals have limited experience and/or knowledge of the psychological aspect of living with HD.\textsuperscript{12,19,50} Many physicians are not adequately prepared to be able to successfully help an affected family because HD spans across many different disciplines, in the fields of both health and social care (i.e. neurology, psychiatry, genetics, physiology), which makes it very difficult to address all the problems at once.\textsuperscript{12,27,53} Therefore, health care professionals need to be better equipped with the knowledge of psychosocial impact of HD on both individuals and the family because the disease has implications for more than one person.\textsuperscript{26,27,29,34}

Many studies concluded their reports by suggesting that more active participation of relatives in the pre and post test discussions might be very helpful in improving family dynamics and encouraging more internal communication about HD and its meaning to the family. One study strongly recommends actively involving family members in the decision making process of whether or not to undergo testing at all.\textsuperscript{51} In fact, pre-test psychological evaluation can serve as a good indicator of the potential strength of psychological impact of the test result on the individuals. In all cases, including pre or post test or during symptomatic stages of the disease, open conversation that involves the whole family can provide the right environment to address problems that otherwise are suppressed.\textsuperscript{3,7,41} It is important for the family to realize what is holding them at a certain point in the Family
Life Cycle and identify the barrier that prevents them from moving on. Encouraging freedom of discussion during therapy sessions on issues relevant to the four discussed postulates of FST would allow family members to talk about HD at a deeper level and bring up issues not previously discussed at home.

It would be fair to say that the most affected person in the family with an individual at risk for or suffering from HD is the spouse. This is because the spouse is the person who spends the most time with the patient, the one who probably knows the patient the best, and the one who serves as the other parent of children (should there be any.) It is the person who would (and in most cases does) take up the role of the primary care provider when the patient becomes affected with the disease. Despite the essential part the partners play in the lives of HD patients, they are not usually given much attention by both health care providers and social workers, resulting in their needs not being met or simply overlooked. Lack of social recognition of the spouse’s burden and lack of support only worsen the internal conflict and increase tension. If proper care is not provided to the person on whom the life of the patient depends to such a high degree, the latter would feel the negative consequences immediately. Thus it is important to consider the impact of the HD on the partners of the affected individuals and include partners in family therapy session and/or invite the couple to take part in marital therapy.

Generally speaking, more social support and psychological help need to be offered in an individualized and flexible manner since there is such variability among families’ responses. The coping mechanisms of the spouse and the family members need to be examined so that the therapy can focus on the characteristics of the family structure that make it functional, with the goal of stabilizing the situation and providing maximal support. Since the resources of the health care system are not sufficient to meet the needs of every HD patient in an out-of-home setting, it is crucial to ensure that the domestic primary care providers are cared for, supported, and well equipped to carry out their roles. In couple therapy, the approach should emphasize the development of appropriate work and organizational skills, increase in self-confidence, and strengthening self-sustainability and independence of the spouses. Consideration of the nature of the intimate relationship allows for a better therapy and increased chance of actually meeting the needs of the affected individuals, not just the carrier. It also highlights the essential nature of the caregiver’s role to the management of the disease. More knowledgeable counselors who are aware of the complexity of psychological responses to HD would be able to provide a therapy that is tailored to the specific needs of each couple/family.

**SUMMARY OF CONCLUSIONS**

As mentioned in the introduction, the objective of this review is to present and analyze the literature available up to date on Huntington’s disease and how it affects family functioning by adopting a theoretical underpinning of the Family Systems Theory. From the above discussion, three main conclusions can be drawn. First, the way families respond to the introduction of HD into the family life is consistent with many aspects of the FST and the observed behaviour supports the main idea of interdependence. Second, due to the lack of public care facilities for the HD patients, it is crucial to ensure that the domestic care providers are emotionally supported and well equipped to carry out their roles. Last, there is a definite need for a more individualized and more flexible approach to couple and family therapies that takes into account specific coping strategies adopted by each family.

In particular, the following important points should be considered. The unique needs required to maintain the stability of the nuclear family emotional system are defined by the family’s history, experiences, perception of the disease, and the current stage in the Family Life Cycle of each member. Both pre- and post-test numerous consultations are essential due to the deep psychological impact of the test result on all family members lasting for various periods of time. The extent of the overall family’s influence on each member’s level of differentiation of self serves as a good indicator of the coping strategy that is/might be used by each individual member. Awareness of the family projection process may prompt counselors to include children in the therapeutic practices in order to alleviate potential emotional damage stemming from the fears and anxieties of the parents. Finally, open conversation that addresses suppressed issues and concerns may decrease the amount of denial and avoidant thoughts in the family that arise as a result of emotional cutoff.

With regards to the future research possibilities, extensive comparisons of divorce rates, physical abuse, and reproduction rates in the families affected by HD as compared to the general population would provide valuable information to health and social care providers. It would be interesting to study the impact on the older parents when their grown up child gets diagnosed because this case presents an example of a complete backwards turn in the Family Life Cycle. In addition, it might prove helpful to deviate away from such a large focus on the issue of predictive testing and examine more closely the impact of HD on members of the family other than the spouse, such as young children or siblings. Concerning young children in particular, a longitudinal study that captures the offspring’s
13. Experiences during childhood, teenage years, and early adulthood has the potential of providing further insight into the psychological impact of presence of HD in one parent on subsequent development of an emotional system in a child.

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