

Genetic counseling: Recommendations for the psychological support within an integrated model.

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Abstract

It is commonly assumed that a strong link exists between genetic variability and environment influence on the onset of many human genetic diseases, the majority with features and peculiarities that have not only medical/physical implications but also a psychological and social nature, with a relevant impact on the personal Self of any individual. Protocols of support by a multidisciplinary team of different professional healthcare providers have not been reported as yet, therefore, through a review and analysis of the extent literature, we propose an “integrated” model and some possibly helpful recommendations with specific regard to the psychological care provided by a therapist during the genetic counseling process within a multidisciplinary clinical team.

This paper is specifically focused on the role, interventions and positioning (addressed to the patient and the caregivers as well) of the psychologist and aims to demonstrate that he/she has a pivotal role in genetic counseling and which vocational education and training, skills and approach should the therapist preferentially have to provide the best support to patients affected by genetic diseases.

Keywords: Psychology, Genetic counseling, Psychological implications, Genetic diseases, Biopsychosocial, Multidisciplinary support.

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Introduction

Genetic disorders and tests: Features, peculiarities and implications

Because of their nature and the fact of being strictly linked and attributed to the DNA and the genes, or rather, linked to what is considered the most intimate and personal structure and basis, not only of and for the physical differentiation and characterization, but also of and for the personal identity, genetic disorders, even more than the other types of diseases and disorders, attack the deepest nucleus of the Self and of the personal identity of individuals and because of this, as a result of a diagnosis of positivity to any genetic disorder, it will always be the whole human being to feel and live itself as deficient and defective and it will feel a feeling of unchanged and incurability for their condition and a feeling of permanence and immanence of the disease. This is also due to the fact that the genes are considered parts of the Self present everywhere in our body and therefore as a result of a diagnosis for a disorder it is impossible to find and assign the guilt for the disease to a single organ or circumscribed area, but the feeling of guilt will inevitably be extended to

the whole body and the whole human being and it is for this reason that many defensive mechanisms that are involved to take the distance from the threat of the disease and to lower the levels of anxiety and distress very often have no efficacy and do not work.

In addition, regardless their positivity/negativity result, the outcomes of genetic tests can therefore act as stressors with variable nature and seriousness and they can also bring along even serious psychological issues, for example they can bring feelings of low self-esteem, inferiority, helplessness, frustration and disillusionment or may even lead to catastrophic issues and symptoms with a really significant gravity, such as severe depression, risky behaviors as abuse and addiction to alcohol, drugs and narcotics, up to suicidal tendencies and other psychiatric issues of variable nature. Therefore, feelings of shame, guilt, anger, sadness, hopelessness, apathy, sense of fatalism with the consequent paralysis in the decision-making and paralysis toward an impotent passivity, are among the most common and pervasive emotional responses, not only to the outcomes of the test but also to the narcissistic wound that the disease itself involves, since the disease itself represents

a direct attack to the self-esteem, to the desire and the need to reach own stability and a sense of identity, control and personal omnipotence, therefore the disease involves a strong attack the integrity of the Self and the own Person [1].

May also emerge social problems and issues of various entities, also linked to the Country of origin, such as relational issues, difficulties in regard to the insertion into working environments, difficulties with the provision of goods and services, social discrimination, etc., binded with these issues related to the wider social sphere, there are also all those problems and issues with a more strictly familiar nature, linked to the planning and functioning of the family, as to give up with the idea of procreate or to do it through artificial insemination or other techniques in order to reduce the risk of occurrence or transmission of the disease to the offspring and later generations, otherwise as a result of prenatal diagnosis the family can go through the decisions of interrupt the pregnancy in progress, or can lead to sexual and couple issues, with the consequent attack to the stability of the couple itself, that can may also lead to the possibility of conflicts between the partners, and also lead to separations and divorces.

Obviously all these "physical-psycho-social" issues are even tightly binded with ethical-moral issues, both the ones more closely linked to the morals and the religious beliefs of the family itself and the ones that are linked to their own country's legislations and social precepts.

Between ethical and social issues there are also all those issues strictly related to the informed consent, and to the confidentiality and privacy regarding the communication of the information about the health of the individual, since these tests also arise outcomes not only tied to the health of themselves but also to the health of the entire parental core, and then with the consequent issues about the desire to whether know or not these information.

Dealing with all these issues it is very clear that we must avoid entering into a Deterministic view that involves a linear cause-effect model, but we should move toward a Systemic view, for which behavioral responses of the individual and his family are multi-determinate, and many and different factors affect these responses, such as, *in primis*, the individual personality, their own and personal ways of dealing with the stressors and manage stress, their skills of resilience or the vulnerability to stressful events, the type and characteristics of the disease (severity, duration, prognosis, available therapies, degree of raised invalidity, visibility, etc.) and the status of the patient himself (healthy carrier or positive affected or at risk of susceptibility to certain diseases), the stage of the family's life cycle in which this diagnosis of genetic disease emerges, the quality and type of interpersonal relationships and the type and the presence of social support at both micro and macro levels (from the support of relatives and friends to the support provided by the systems, organizations and professional public or private structures), etc. It is also evident that the individual, the couple or the family use different and

personal strategies trying to restore the order and equilibrium into their system, order shaken by the diagnosis of disease, and trying to reduce to a minimum and to limit the stress in order to promote and maintain a sense of personal value and to maintain the relationships and the support from significant others. However it is clear how the nature of these multiple, different and more or less personal, emotional and behavioral responses is quite always a defensive nature and it is clear that without the correct and necessary help and support it is this precisely defensive nature that inevitably doesn't leave sufficient space to actual exploration and processing of these responses.

Despite these aspects and implications are really various and they have variable nature, they can be summarized in the concept of human suffering and the need to find the personal welfare, therefore it is clear that it is desirable, in order to be able to cope better with and facing disease (and all the consequences and implications that derive from it), the need for a support of the individual and the family in a psychological counseling, or in the most serious cases in psychotherapy, obviously taking into account the needs, wishes and consent of the person itself [1].

The genetic landscape in psychosis and mental diseases

The basis of onset of psychosis can be explained only with a multifactorial model, which includes both genetic and environmental factors. The genetic component has a direct role on the psychological traits associated with psychosis (specifically the personality development and neuro-cognitive aspects) influencing trends and final outcomes. In fact, there is strong evidence on a high-level of familiarity on psychosis, confirming a key role of the genetic component. Moreover, the average risk to develop psychosis is 1% but for consanguineous individuals such risk increases in proportion with the degree of inter-familiar relationship. The most-believed hypothesis is that stress (environment) and epigenetic factors affect the likelihood to develop a mental disorder in genetically susceptible individuals only [2,3]. It is also possible that individuals with a mild phenotype have a low level of deleterious DNA mutations in only few genes, so they are less susceptible to the environmental stress than those who have accumulated more variants in several genes and are suffering from a more severe form of mental disturbance. Recently one of the largest genetic studies on mental disorders has been published: according with the authors, the major mental disorders share about the same genetic risk factors. Researchers at the National Institute of Mental Health (NIMH, USA) have screened the genomes of 33,332 patients (affected by autism, ADHD, bipolar disorder, major depression or schizophrenia) and 27,888 controls discovering the same common genetic variants in the group of patients [4]. Moreover, specific variants commonly associated with all five disorders were identified in two genes only coding for molecules involved in the regulation of the metabolism of calcium in neurons. One of these genes, CACNA1C, was already known to have an impact on brain circuits involved in emotion, attention and memory - functions that are also reduced in mental diseases. Many other

mutations were been disclosed in another calcium-channel gene, called CACNB2, which was also correlated with mental illness, especially with bipolar disorder and schizophrenia. These results had contributed to decipher the etiological basis of schizophrenia, among the top ten diseases with a higher social disability impact that still remains very complicated. However, some recent studies employing next generation sequencing technologies have allowed the identification of new important genetic hotspots, some with multiple and unexpected functions [5], while other in genes involved in the same biological process [6]. In addition, 15 new chromosomal regions have also been recently reported in Nature Genetics as linked to the development of depression, especially in Caucasian people, confirming again the evidence of hereditary basis for mental disorders [2]. Profiling the individual genetic risk for psychosis and the discovery of molecular biomarkers may soon lead to the development of new diagnostic tools and innovative therapeutic applications for those forms of psychosis that currently are still missing of effective treatments.

Methods

*Methodology, data collection and selection of the research materials*¹

For the research, collection and selection of the research material, that is academic articles and researches, textbooks and books about Genetics and Genetic Counseling, it was conducted a randomized research using the keywords *Psychology and Genetic Counseling, psychological implications in/and Genetic Counseling, Psychiatry in/and Genetic Counseling, Integrated Genetic Counseling, Genetics, genetic diseases* (and different combinations of the terms so that it could imply simultaneously Genetic Counseling and features with a psychological and/or psychosocial nature) using all the different databases available for the students at Università degli Studi di Bergamo, Human and Social Department, in particular the ones for Clinical Psychology courses, that is PsycInfo, Psychological & Behavioral, PubPsych and further more databases and websites for academic and scientific research, that is PubMed, Nature.com, Springer.com and the academic section from Google Scholar browser.

In particular the articles, researches, studies and books that were taken in account were the ones with these characteristics:

- With better and more actual research matching;
- Theme and argument of Genetics and genetic diseases treated and debated at a scientific and academic level, with a propensity and prevalence for the ones that have not only a clinician and medical point of view, taking in account the difficulty of this, mainly due to the fact that this is a quite recent argument and themes in the psychological fields and ways;
- Studies, researches, and books that treated and debated

the psychological implications caused by genetic diseases and by the Genetic Counseling process itself;

- Trying to mainly report the most recent and updated also in in the field of institution and legislation of and about the Genetic Counseling;
- Easy and free finding and access to the complete text.

It must be highlighted that the Italian bibliography and references are quite scarce and lacking, therefore it was opted for a research of international articles, studies and researches, also due to the more academic and scientific fact of finding and using material and data that were not only specific and related to a single country (in our case Italy) or single cases/diseases, but material and data that were more general, and that especially had more significance, and a scientific and professional approval at international level.

It must also be highlighted that the used literature treated and debated about different diseases as for adult people, without gender distinctions and specifications, so it was excluded all the branches and diseases with the reference to prenatal genetic.

The literature was reviewed and analyzed in order to try to give answers hypothesis especially to the questions

- Is the psychologist really needed in the integrated process of Genetic Counseling?
- Which vocational education, training and approach could the psychologist take in an integrated process of Genetic Counseling, referring to possible theories, approaches, and models, which could be considered as basis of the process itself?
- Which possible roles, interventions and positioning addressed to the patients and their relatives (and caregivers) and addressed to the MD team/équipe could the psychologist take in an integrated process of Genetic Counseling?

In order to try and be able to draw hypothesis of recommendations for the intervention and vocational education and training for the Clinical Psychologist that will work in an Integrated, and therefore Biopsychosocial, process of Genetic Counseling.

In this paper we specifically focus and analyze the third research question, in order to draw possibly helpful recommendations about the Psychologist's roles, interventions and positioning in an integrated model of Genetic Counseling.

Discussion

Recommendations for the psychologist's role and interventions in the genetic counseling process directed to the patient, the family and the wider social context

From what emerged from the review of the literature it is possible to trace recommendations which might be useful as a base upon which we could establish the psychologist's role and his interventions in aid and support directed to the patient

¹ The study in this paper is a qualitative one, though both qualitative and quantitative researches have been evaluated, considered and analyzed in order to collect *cold data* for the study itself.

and his relatives, bearing in mind the particular attention requested by both the wider social and communitarian net.

The reader must always keep in mind that the psychologist's role and duty within the Genetic Counseling process is not and never, not even remotely, should be a role and duty of psychotherapy, but the latter, if proved necessary, should be requested through the proper procedure to the proper professional figures.

Psychologist's role and interventions directed to the patient

First of all, regarding the work directed to the patient, the duty to assist and support the patient on both psycho-emotional and psycho-social levels should be ascribed to the psychologist, easing, facilitating and encouraging at the same time the support from the various social nets and networks, starting from the nuclear family.

Nevertheless, this psychologist's role and duty should be activated since the very first phases of Counseling in direct and integrated collaboration with the MDs, in first place the geneticist, and their work and roles, and not as a subsequent and additional requested action or as a subsequent referring to the psychologist suggested by the MDs themselves. As a result, the psychologist could connect and create a relationship with the patient since the preliminary phases of the process, phases that would help to avoid and eliminate the social stigma mainly linked and tied to a wrongful cultural vision and perception of the psychologist's profession. This would also facilitate, ease and improve the alliance and relationship psychologist-patient and consequently also the sharing of the information linked not only to the Genetics and genetic diseases but also to the history of both the patient and his family.

An intervention of the psychologist activated since the first phases of the Genetic Counseling process would give the possibility to have an evaluation and a supervision of the relationship and the methods of conducting the sessions used by the geneticist and MDs. There would be particular attention to the answers more or less explicit of the patient, so that it will be possible to evaluate and regulate them, anticipating, predicting and foreseeing possible negative implications, outcomes and answers of the patient, even the behavioural ones, in order to adapt, adjust and reshape the management of the session by the MD, case by case, session by session.

As a result, it would provide benefits to the patient, facilitating the communication, passage and comprehension of the clinical information, and facilitating at the same time the relationship and alliance itself between patient, geneticist and MDs.

The combined intervention of MDs and psychologist might help developing a feeling of acceptance and empathy towards the patient, so helping the latter feel as regarded by the professionals as a person, an individual, and not only

as a mere object of study and testing, besides this, it might enhance his feeling of being treated with respect, perceiving that the professionals are dedicating and devoting their care and time to his cures, with attention and taking all the time necessary.

An important point might also be the drawing up and lay out of a profile regarding the psychological and psycho-social aspects of the patient, giving particular attention to the following aspects:

- Evaluate the baseline levels of anxiety and distress, both specific and generalized, aiming to prevent possible serious psychological implications that might flow into psycho-pathologies of different entities.
- Evaluate which are the valuable and possible resources upon which it is successively possible to focus on and to develop, in order to promote an aid and support to self-esteem, in primis and a support oriented to sustain the required work that ought to be done in the following sessions, since they have themselves a crucial role in adapting and facing stress, anxiety, concerns, etc., through the time.
- Evaluate the vulnerabilities and vulnerability conditions of the patient in relationship and in connection with his own characteristics, the characteristics and features of the disease and the possible answer to different diagnosis of the patient himself.
- Evaluate the sense of self-esteem, mastery, personal power and control, sense of guilt, shame and anything else that might corrode self-esteem itself, the perception of the Self and the own personal identity of the patient himself.
- Evaluate the concept of the Self by the patient and what meaning the latter gives to the term *normality*.
- Evaluate the presence of possible psychopathologies and psychic discomfort already present in the patient, especially if already diagnosed or present within his clinical history. Furthermore, it is necessary to recognize discomforts that might worsen during the course of the sessions and with the advent of news and diagnosis of disease.
- Evaluate emotions, feelings, cognitions, themes, topics and narrations of the patient strongly linked and tied to the concepts, topics, conditions and state of health and sickness/disease, in order to aiming to rectify and improve possible misperceptions and allow to restore the emotional balance and equilibrium. This can also be aimed by modifying the beliefs and cognition of the patient, this would lead to a reevaluation of the concepts of disease, health, normality, test, etc., and the themes and issues linked and tied to them.
- Evaluate, regulate and/or eliminate all possible factors

that might negatively influence on emotions, feelings, cognitions and pro-health behaviours of/by the patient.

- Evaluate and regulate the emotional baseline answers of the patient and evaluate all the emotions that emerge and intervene subsequently, bearing in mind that the emotions themselves might influence the way information are processed and acquired, so influencing the transmission of the latter to the relatives. Besides, the emotions themselves might be foreseers and anticipators of the rise and kind/type of subsequent cognitive and behavioural answers of the patient.
- Evaluate the cognitive structure and the type of emotional answers of the patient meant to prevent the rise of possible mementi of death through the phases, and so avoiding any engagement of behaviours of avoidance and against health.
- Evaluate needs, expectations, hopes and concerns towards and regarding the testing phase and the future life, both strictly connected to the condition of possibility or certainty of disease. It is necessary to always bear in mind how these might also influence both emotional and behavioural answers, and the processing of information gathering itself. Everything was just said above is done in order to restrain the anxiety and distress linked to the results, outcomes and possible confirming bias of concerns and/or previous expectations with the goal to support the emotional adaptation towards the results and outcomes themselves.
- Evaluate the perception of risk and the possible, related and tied emotions, feelings, cognitions, expectations and hopes, both of baseline and in the following phases of testing and consequent result and outcome of the tests and exams, since these factors could be related and tied to psychological decline and degeneration, depression and increase of the level of stress and anxiety.
- Evaluate how the patient feels uncertainty, how he faces it and how the uncertainty has an influence on both the decision-making processes and the patient's life; moreover evaluate how the patient reacts to it at emotional-psychological and behavioural levels.
- Evaluate the "defences" of negation and minimization and verify if they are adaptive and useful to protect from threats and keep self-esteem and self-control, so they could possibly be approvable and supportive, or contrariwise if they might be maladaptive, so they have to be reduced and eliminated. Evaluate maladaptive "defences" meant to negate in order to avoid the following development of depression and higher levels of stress and anxiety.
- Bearing in mind the importance for the self-esteem of the personal experience along with the personal

history of the patient, review together with the patient the history of his past life, with particular and peculiar focus on the hard and suffering times loaded with uncertainty. This aims to move the focus on the patient's resources in order to allow him to reacquire self-esteem and control on both himself and his life. Furthermore, this helps to gain an evaluation on strategies of coping and resilience used in the past or with higher probability to be used in the future.

- For disease cases previously overcome, evaluate the possible existence of feelings of guilt and shame of "*the survivor*", even more if other members of the family ended up affected by the disease or died because of that specific disease.
- Evaluate the levels of stress and anxiety related to testing phases and the return and feedback of their outcomes that must be followed by the appropriate support.

A preliminary evaluation of all emotional, psychological, psycho-social aspects would allow to the multi-interdisciplinary team of health professionals to obtain an estimate and evaluation of the possible number of Genetic Counseling sessions the patient might need, furthermore it would provide information useful to obtain a preliminary estimate and evaluation about the need of additional sessions, specifically for the support and work related and concerning psychological, emotional and/or psychosocial issues. Because of numerous personal differences from a patient to another, it would be important to evaluate their emotional functioning, their cognitive representations, their thoughts and beliefs, their perceptions and their expectations, since all these factors represent, in the last instance, the regulation of the own Self, hence it would be also useful in order to achieve a good and positive overcoming of the disease and a good and positive adaptation to it, with the ultimate aim of achieving and obtaining personal wellbeing.

A preliminary evaluation would also allow considering if a Genetic Counseling support would be immediately and strictly necessary, and especially recommended in the short period, for the patient and his family, or if contrariwise, it might be necessary to postpone the process. In order to do this it is necessary to bear in mind the life condition and the events experienced by the patient and his family at the time of the decision of undertaking the counseling process, that is for example presence and experience of recent grieves, not treated depression or other factors, such as for example psychological characteristics and features or personality traits of asymptomatic patients that could lead to negative psychological outcomes and implications, such as for example feel and see themselves as already sick even only for the fact that they have to undergo some clinical tests and examinations. If any negative factor is observed it could lead to the decision that Genetic Counseling should be discouraged as it might bring only negative outcomes and implications, and not benefits. Furthermore, a preliminary evaluation

allows proceeding with a possible recommendation for a psychotherapy where such issues and problems bring with them or lead to grave and burdensome developments on a psychological level before the beginning of any assistance. Therefore, it is advisable to avoid a process of Genetic Counseling too anticipated since this process itself might be the cause of distress and discomfort and it could also bring with itself more negative than positive outcomes and implications (in these terms, it is necessary to view this themes linked in connection with both TMT, *Terror Management Theory*, and TMHM, *Terror Management Health Model*, models).

Through TMT and TMHM models the psychologist could and should evaluate and help the patient to regulate, face and ease the *Death Anxiety*, particularly thanks to the support and help of the MDs, combined and integrated with the psycho-emotional and psycho-social support, that might give further information about the disease, consequently increasing and developing the knowledge and information of the problem. The psychologist could and should also provide a training for coping, empowerment and resilience skills of a more cognitive, emotional and psychological nature, paying specific attention to the situations that could be a major source of mementi of death (such as the testing and follow-up phases).

Following the TMT, TMHM models and theories, and the ones about "*Epistemophilic Anxiety*" (*Angoscia Epistemofilica*), it might be possible to hypothesize for the psychologist another possible task aimed to the patient, that could then be helping the patient to prevent, face and deal with the *Death Anxiety*, since it is an intrinsic and innate characteristic of the nature of the Human Being and for this reason unavoidable and inevitable to have. Moreover the *Death Anxiety* could be in this case interpreted as a specific anxiety type that would join with the one specific for the disease. In addition, the disease state, the disease itself, and the related and linked outcomes, implications and consequences may lead to find numerous and additional mementi in the course of time, that might continuously make the anxiety "prematurely" emerge again in a maladaptive way, and hence it would need to be faced and treated with the due tools and with the help of adequately trained professional figures.

Aside from this task and action, the psychologist might help the patient to make the jump into the void of uncertainty, trying to reduce his fears and worries, helping him to questioning those that once were his certainties, and at the same time helping him to retrieve the capability to get control and balance back in the patient's life, bearing always in mind the fact and limit that the genetic diseases inevitably damage and modify the delicate balance and equilibrium previously achieved. So, since the nature of these diseases, it is inevitably and objectively impossible to entirely retrieve and re-establish the previous equilibrium, therefore it is necessary give help so that it would be possible to establish a new equilibrium and balance that would consider the new state and condition, at the same time trying to reduce at a minimum any distress and anxiety.

Another task to establish is that to accompany the patient through the process of informed decision-making in order to make conscious and free decisions while easing the process itself. This task is closely and strictly tied to the work on feelings, emotions and cognitions, since us, as human beings, make our decisions on the amount of previous experiences stored in our memory. This is indeed strictly related and made possible thanks to the emotional system, hence the emotions themselves intervene in the process at the same time with a cognitive function and role, giving the possibility to assign and confer values and meanings to people, things and events [7].

Subsequently, throughout the course of the sessions, the psychologist should gradually evaluate and keep under control the emotional responses and reactions and their related cognitions, all these related and connected to the various phases of the process, to the various information that are progressively introduced, and related and connected to the responses and the results of the testing phases and examinations. Paying particular attention to the "approaching" moments to the disease, particularly the testing and follow-up sessions, the psychologist should also evaluate, review and revise both the patient's psychological profile, and responses and reactions time after time, session by session. Whereas the psychologist would observe or suspect a fall to lower levels at the limits of psychopathology, then he would consider and advice/suggest to possibly begin a psychotherapy before proceeding with any testing session, again paying attention to the patient's condition and state, meaning if the patient is affected by the disease (so bearing a higher chance to be predisposed to depression and *Death Anxiety*), or if he is at risk of carrying the disease (so bearing a higher predisposition to both, general and specific to the diseases, anxiety. and worries/concerns). Furthermore, the psychologist should pay particular attention to possible bias of anchorage and adjustment between the assessment and baseline profiles and the possible subsequent proofs and results of tests, especially if the latter are disconfirming. Concurrently with all the other possible positive and optimistic feelings, like hope and self-mastery, the self-esteem plays a fundamental and crucial role against negative feelings, such as the feeling of vulnerability, guilt, labelling, diversity, etc. Besides, positive and optimistic factors have a fundamental role for and in support and help for the resilience, along with consequent positive implications and developments in support to the subsequent adaptation. Therefore, it is fundamental for the psychologist to evaluate the self-esteem and whereas is deemed necessary begin a work to promote and develop its both support and development, and it is also necessary for the patient to always continue to search and to try to find, during the course of the different sessions, for cues and supports for the self-esteem itself, in order that the patient could find every possible benefits to aid his wellbeing.

Another duty and task should be to help the patient develop adaptive strategies of coping, empowerment and resilience through a proper and specific training, that should

be oriented to help the patient to face and deal with the disease, re-acquiring self-esteem, sense of personal value and control power over his own life. In connection and relationship with emotional-coping, all these strategies are also tied to the desire of information, the desire of perception of control, of personal care, so they might help the patient to anticipate the planning, the programming and the organisation of his own life with a consequent feeling of control and well-being.

Whereas surgery is required by practice related to the development of the disease or in order to prevent it, a higher and more intensive psychological support would be advisable and highly recommended since consequences and “changes/alterations” on a physical level, caused by the surgery itself, might bring a whole series of aspects and negative developments and implications on a psychological level, such as feelings of stigma and disgrace, feelings of diversity, so it is clear how the intervention of a professional figure who is both qualified and properly trained results extremely important if not necessary for both adaptation and wellbeing of the patient.

Another important evaluation is the one over the meaning that the patient gives to the term and concept of *Normality*. This conception should be always evaluated, and if necessary also reviewed and redefined, in the very first sessions, all this in a preventive perspective and in regard to possible negative psychological developments and implications, that in first place could be tied to all those negative aspects linked and related to the feelings of diversity. At any rate, the given meaning to the term and concept should also be reviewed during the entire inter course, connecting it to the various results the patient would receive, aiming to keep or restore balance and wellbeing. Related to this there is also the fact that there is a whole variability and fluidity of boundaries between *Normality* and *Pathology* in the genetic filed. That is because the deterministic link between mutation and pathologic manifestation of the diseases is not at all simple and neither completely known [8], so it must be clarified with the patient himself.

Therefore, the psychologist should keep well in mind and run through all those processes meant to promote and prevent health that are connected to the quality of life and are meant to support the so-called *Life Skills*, typically linked to Psychology of Health and to the main promotion health models, that are *Self-Awareness, Assertiveness, Coping with Emotions, Coping with Stress, Resilience, Critical Thinking, Decision Making, Problem Solving, Creativity, Effective Communication, Empathy, Interaction Capacity and Positive Interpersonal Relationships*. The duty and objective is to instil in both the patients and their relatives the belief and certainty that they own the capability to positively modify their habits by putting into effect appropriate and functional behaviours to improve health and well-being and this means that the patients themselves have power of self and collective efficacy over their own well-being.

Psychologist's role and interventions directed to the family and wider social level

Because of the nature of the genetic diseases and because what was previously viewed and analysed, it could be hypothesized that what has been said above could and should be properly extended, applied and addressed, in first place, also to the relative and/or caregiver whom is elected to accompany the patient through all his session, as it was already suggested “*as protocol*” and secondly, the same practices could and should be extended to the entire close nuclear family. Possibly the psychologist, in agreement with both the *équipe* and the patient, could be also evaluate if it is appropriate to extend those information to a wider range of the family, after considering case by case.

Nevertheless, on familial level there are other interventions that might be made by the psychologist, such as:

- Evaluate the inter/intra-familial relationships.
- Evaluate nature and types of the relationships.
- Evaluate both communicational and relational methods and styles of the family.
- Ease and encourage all the relational dynamics of reciprocal support among every family member, with the aim to attenuate the feelings of guilt, the feelings of shame and all those behaviours of isolation, labelling and stigma that the disease might entail and bring with itself.
- Ease and encourage inter-familial and social relationships in order to mitigate and eliminate social stigma, labelling, etc.
- Obtain a narration of the familial history, both based on the relationship and based on the status of disease.
- Create efficient connections and relationships between the patient-family and the *équipe*.

The objectives of all this are to comprehend their dynamics in order to ease and encourage the inter-familial support, and both communication and comprehension of the clinical information within the family and to avoid that maladaptive or even pathological behaviours, along with related negative developments and implications, that could arise and be destabilizing or undermining for them.

Therefore, it would be important to receive a narration of the family history not only to draft and establish a profile about it and understand how to operate and on which points is needed to press over with force, but also to give patients and their families a chance to reevaluate their own familial history. It is especially important to review all those past hard and burdensome experiences of their life so full of uncertainty, contemporarily evaluating and reviewing together their responses, their successful adaptive strategies, in order to enhance the resources and the strength of cohesion and harmony of the family core and the relationships between its

members. The aim of what was just said is to move the focus of attention only over the positive aspects and resources of the family, and not on the negative ones, in order to re-establish mastery and control over their lives. This could also ease and encourage the psycho-emotional and psychosocial support for the entire nuclear family so to newly calibrate self-esteem, feeling and sense of acceptance, and the feeling and sense of belonging and self-valor.

Therefore it is clear how it is essential and extremely needed a work on the families useful to prevent that internal dynamics that could tear and/or further erase the support needed by the patients and their families.

As ascertained from the literature, there is a strong chance to develop higher and stronger level of stress and anxiety along with strong emotional responses when more members of the same family at the same time have to take part to sessions and phases of testing, and the ones about the discussion of the results. In order to avoid or face this, the psychologist, in co-operation with the MDs, basing on the baseline profiles and on the familial history, should previously intervene preventively through sessions involving all the family members interested by the event.

It is not by chance that from the literature emerged how important is the communication of both the results and the genetic information to the entire family and how the professionals of Health themselves must take responsibility of such communication to not let it rest only in the hands of the patient alone. It is also important to immediately summon the entire family, first of all, because of the diseases' nature and the different types of transmission connected to them, and secondly, in order to offer a better and complete communication and comprehension of such important information. Therefore the privacy of the single family member shifts into a secondary position since such results and information do not influence the single human being and only his own life but rather hit and inevitably and always involve the entire family, so keeping the secrecy of such delicate information might lead to grave health damage for other members of the family. Nevertheless, it is advised to steer towards a general-type communication and no single case-specific, despite the patient's identity might be completely clear. This is meant to avoid a whole series of emotional developments, implications and responses that might lead to another series of exclusion and negative feelings, emotions and behavioural responses, such as also resentment, feelings of hostility, isolation, exclusion, guilt, shame, etc., with the risk of reaching a point that the communication could turn out as problematic, and for example, it might involve the tearing and deterioration of relationships already weak or unstable, along with the reduction of any support from the patient's relatives, etc.

On the contrary, privacy is a strong and extremely important topic and issue whereas there is a necessity to share information outside the family since they might be highly influential, with a stigmatizing nature, on social,

insurance, working, etc., levels. Thus, the ultimate decision to communicate outside the close members of the family is up to the patient and his relatives and their free decision.

Despite the possible cons of this approach (such as the possibility that patients perceive to obtain and receive too general responses and not specific their own case) and trying to evaluate in advance its efficacy, validity and appropriate use and application, avoiding any problem of pragmatic arrangement and management (such as wrongful management of time, failed solicitation of interaction and discussion, lack of clarification of the goals, inappropriate number of the participating members and also inappropriate time for the participants due to physical health statuses connected to diseases, etc.), it is possible to hypothesize that it could be possible to ease and encourage the support to the family provided by the wider social community through the organisation of focus groups and inter-families meetings and sessions. This kind of sessions could allow to simultaneously reach and inform a large number of patients (so with also conceivable more advantageous economic and administrative consequences), allowing to ease, encourage and promote the sharing of information, histories and narrations useful to promote, help and encourage the rebuilding of a new history, a new narrative and a new life, in addition to promote and allow the families to receive and perceive additional support, feeling of belonging and social acceptance, with the consequent decrease of the feeling of labelling, stigma and isolation made by the wider social community. Furthermore it could also allow the patients and their families to freely express their emotions and histories without feeling the burden of a judging and stigmatizing social look.

From a socio-cultural perspective it would result important and necessary an evaluation of all the possible aspects and cultural "beliefs" linked to the disease and to the issues related to it and also an evaluation of the knowledge about the diseases themselves, in order to verify if the direct experience of the family with it and the scientific knowledge are actually the same. Such question appears evidently more present for all those genetic diseases with a neurodegenerative and psychiatric nature, that is the conditions that are mostly affected by the stigma and labelling given by the popular culture, that is likewise wrongful and easily could lead to lack of support and isolation of the families. Therefore, it should be chosen to amplify the knowledge of such disorders and diseases on a wide social level in order to reduce as much as possible the stigma that such diseases and most importantly the affected patients and their families, still continue to suffer and carry on their shoulders. Thus, in general, on a wide social level the psychologist duty could be to ease and encourage the spreading, widening and sharing of the information about genetic diseases and enhance the Counseling process through meeting and campaign in collaboration and association with the MD équipe and addressed to the wider social network and not only to restricted groups of professionals and diseased/patients. But that's not all, siding and being sided by other professional figures, the psychologist could contribute to the

study and pragmatic application of informative campaigns, first of all focused on genetic disease with the aim to give them a definition, a conceptualization, and to establish their traits, features and peculiarities, but also to underline the importance and avail of the Genetic Counseling process along with genetic tests. This knowledge could be enhanced in order to provide a higher and better knowledge and comprehension of the “natural”, “innate” and “normal” psychological effects that are possible after a diagnosis, the ones related to the Counseling process and its related testing and/or resulting from the application of genetic technology [8].

In our opinion, the active presence of the psychologist in these cases should be fundamental since it might establish the aim, that is already so much craved in other branches, to bring higher information about his role, shed light on the actual roles, intervention and fields in which he operates, so allowing to reduce the social stigma, sadly still widely present, grasped and related to the profession. This aim, in this case, is worth also in order to promote, ease and encourage pro-health behaviours that would provide further knowledge and information about genetic diseases, and in order to promote the decision making linked to the decision to undergo Counseling processes and especially to take part at the sessions of psycho-emotional and psychosocial support. These are not meant as psychotherapy but as a help and support that is deeply needed in such delicate, thorny and burdensome life conditions and times that put the individual in front of their biggest maximum peak of vulnerability.

Working backwards from the social to the individual sphere, the psychologist should also bring the attention upon the psycho-emotional implications that every single diagnosis has over the individual, a fundamental care unfortunately at risk to be lost as the technology provides more and more diagnostic resources increasingly and mostly connected [9] to wide-scale applications [8].

Lastly, it would be necessary a combined job between the psychologist and all the other professional figures intervening in the Genetic Counseling process in order to discuss and deal all ethical and privacy issues and topics that inevitably might present themselves and that are closely connected to genetic diseases and the process of Counseling itself. Such work would also be necessary to establish efficient, suitable and clear protocols that would keep in consideration all the variables and dimensions that enter in action, with the goal to provide the best information and care for the entire community.

Support to the md staff: education, training & supervision

From what was said above it is therefore deducible how an important role arises for the psychologist figure, not only regarding the patients but also regarding and for the education, training and supervision of the clinicians themselves.

It is conceivable that the psychologist could obtain and have a key role of multidisciplinary support in the education and training of the medical professionals, but also of the

health workers that would get in touch with the patients and their relatives. At Academic level in the various and varying Medicine courses and of other medical, health and health-hospital disciplines, some Psychology courses are provided and included in the curricula, however they are mostly oriented to provide a basic knowledge, mostly historic-theoretical one, of some of the different psychological and psychotherapeutic approaches and theories. However it could be conceivable that with the development of the Genetic Counseling this type of education should be addressed to and focused on the acquisition of relational and psychological abilities, skills and competences by the medical doctors, inclined to promote and improve their communication skills, listening skills and more broadly Counseling skills, essential for a better success in the Counseling work and for a better wellbeing and broad tutelage of the patient. All these skills are essential and fundamental in order to enter the required multi-interdisciplinary approach. Therefore in addition to the courses that are already provided, it could be conceivable as useful and beneficial integrate them with more specific courses, courses with an orientation more typically tied to the psychological Counseling in the Health and Healthcare settings and fields, starting from an education more theoretical regarding the basic and general theories commonly used, regarding the tools, the skills and competences used, which a good counsellor must have and develop, etc., in order to gradually proceed with the pragmatic actualization of what was acquired before through the educational courses and in the specialization schools. This type of education would give the bases on which the MDs would later ceaselessly and continually develop and improve his own skills, tools, etc. and so promoting and fostering a concept of Health and Wellbeing that would influence on the actual medical reality, orienting it towards a perspective that will place as fundamental and essential the centrality of the person towards his own good state of health and/or illness.

Another conceivable role for the psychologist could be to be a trainer for the MDs, that is the psychologist could provide a support to the MD staff in order to allow them to ceaselessly and continually develop their relational and Counseling skills, competences, techniques and tools. If little above it was about a more theoretical education, about the basic theories, tools, techniques, skills, etc. to be conducted during the Academic and/or the specialization schools years, in this case it is indeed meant a support, supervision and training more continuous and directed mostly based on the practice and on the development and improvement of the skills, competences, etc. of the medical staff involved in the Genetic Counseling process.

In our opinion it would be indeed essential the équipe supervision role by the psychologist himself in connection with the medical members of the équipe. In this case, the best efficacy and typology of supervision should be verified, that is if it is considered necessary based on regular and predetermined équipe meeting (for example a predetermined number of meetings per month) or if it is indeed more

useful and effective a supervision directly conducted during the different sessions with the co-participation of the psychologist and the geneticist, with potential other équipe meetings of discussion and supervision conducted with other professionals (biologist, lab technician, nurses, administrative staff, etc.). Independently of its typology, this could also bring a favorable contribution to the work of every member of the équipe, since it could allow to facilitate, encourage and sustain the various équipe dynamics and it could provide a common and shared space and setting of sharing, discussion and reflection about the different themes and issues that easily could involve and touch the feeling and emotional cords of the involved professionals themselves.

Conclusion

Despite possible omissions and gaps, we really hope to have encouraged and stimulated the curiosity of the reader about a subject, themes, issues and problems that are still too little treated and debated, and often too little known (especially in Academics and University settings). With this paper we also hope to have brought some minimum contributions that will be auspicious for foster, encourage and increase the knowing and knowledge about Genetic Counseling, a disciplinary field that is still too little known in Italy, but that is especially, and we would say “unfortunately”, still to be considered not duly taken into consideration yet by Clinical Psychology itself, not only because it would be advanced as a possible and further professional solution for those who are defined as “too enough” psychologists in training. In this case, in our opinion, in a harsh moment of recession like the one we are living in for our profession and in particular for the Healthcare, they could find a new and good field of study and work still fresh and that is still in development, therefore requiring and demanding for professional figures that can take care of it. At the same time this could allow them to show how they are professionally worth, and could also allow Psychology to have a voice in fields that are still too much taken and reserved to and by MD professionals (we highlight how currently in Italy the Genetic Counsellors are quite always MD graduated and majored only in Genetics), even though frequently many aspects and implications are not in their knowledge and competence. All this is important in order to offer the patients and their families an appropriate and adequate support and professionalism in a very difficult and thorny period of their life, thus to be able to help them more and more to achieve a complete state of wellbeing, in the terms expressed by W.H.O. itself, unfortunately still not completely contemplated so, especially in Healthcare fields.

As it was possible to gather from what is found in this paper, the Genetic Counseling field could be a significantly great mean also in the hopeful prospective and objective of reaching a real multidisciplinary approach, that will be such not only at formal and ideological levels but also at the actual and daily practice level in every field in which professional figures, who came from different educations but share the

same and common objectives, such as it is especially the Health and Healthcare fields, have to work.

Dealing with the fact that these disciplinary field, subject, themes and issues are still too little popular and treated in Italy and that they are still too little treated under the psychological perspective or under a really multidisciplinary one also at an international and general levels (see how the international literature quite always debates about these themes under the medical perspective only, though they always recognized the arise, importance and influence of factors also under psychological and social aspects), it is clear how there is still much work to do. For this reason it is also obvious that the recommendations written in this paper, sided by what is already written in the literature, are only to be considered possible hypothesis apt to encourage and promote researches, so that these hypothesis would be verified themselves, in primis and secondly, and mostly, apt to rouse the common opinions and professionalisms of the clinicians and psychologists. In this way it will be common practice to really work with a prospective and in a mood of real and professional Multiinterdisciplinary-équipe approach, especially in the Health and Healthcare fields, not only for the good and best of the disciplines themselves, but above all for the good and best of the patients' health and wellbeing. We must never ever forget that the patients and their wellbeing are exactly the true and only focus and objective on which the mission of our work must be focused on.

If someone may ever forget that this is the mission that every professional of the Healthcare must advance to himself, we really believe that this one should revise his priorities and professional vocations and treat his own professionalism.

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