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Impact of Neurofibromatosis type 1 on families

Master Thesis

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Abstract

Objective: The study investigates the impact of Neurofibromatosis type 1 on the families, specially focusing on the socio- demographic factors such as age & gender of NF1 children, total number of children in the household, monthly income, employment status & marital status of parents on the Total family Stress, Distress Scale, Health related quality of life & Coping strategies.

Method: In this cross sectional study design, the research instruments such as Impact on Family Scale (FABEL), SF 12, Distress Thermometer, & CODI were used. Questionnaires were answered by the heterogeneous sample of 66 parents of NF1 children.

Result: Parents reported of higher personal stress and lower problems related to siblings. Majority of parents answered of "No distress" in the Distress Thermometer Scale. Significant difference were found between the "*monthly income*" and the "*PCS (Physical Component Score)* of SF 12, health related quality of life with the significance value of p = *0.04.* "*Wishful thinking"* was commonly used coping strategy while the "*Emotional reaction*" was the least used coping strategy. Significant differences were found between the total number of children in household and the *"Distance"* Strategy with the significance *value of p = 0.02, effect size = -0.28.* No significant differences were found between age & gender of NF1 children, employment status & marital status of parents on the Total family stress, Distress Scale, health related quality of life & Coping strategies

Conclusion: Monthly income had an impact on the Health Related Quality of Life. Families with more number of children in the household used "Distance" as their coping strategy. The study also demonstrates the usefulness of using the research instruments such as FABEL, CODI, SF-12, & Distress Thermometer.

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1. Introduction

NF1 is an inherited neurological disorder that causes tumours of the peripheral nerves known as neurofibroma (Brosius S, 2010). Though Neurofibromatosis type 1 is classified as a rare disorder, but it is common autosomal dominant genetical disorder (Brosius S, 2010). One of the unique genotypic features of NF1 is its high mutation rate which is 100 times higher than the normal mutation process (Upadhyay M, Shen M, Cherryson A, et.al, 1992). The phenotypic feature of NF1 varies from affected person to person (Szudek J, Joe H, Friedman J M, 2002). Neurofibromatosis type 1 has marked effect on the lives of affected patients and families (Benjamin C M, et.al, 1993). In spite of this, only few researches have been conducted so far on the impact of Neurofibromatosis type 1 on families.

Parents are the rock of the family, assigned with the most challenging jobs to provide safe and comfortable environment to the needs and care of their diseased children. Chronic disease in children has been associated with the increase in marital stress, and subsequently ending up in divorce (Eiser C, 1993). Healthy siblings also end up suffering since special care and attention are provided to the diseased children by their parents (Eiser C, 1993). Life threatening disease condition in children brings about change in parental priorities and expectations (Eiser C,1993). Maintaining a profound inter-personal relationship among the family members has emerged a positive effect on the somatic and psychotic functions of patients and families (Fischer L, Weichs K, 2000). The thesis focuses on the Impact of NF1 on families specially the impact of the socio- demographic factors such as age & gender of NF1 children, monthly wages, employment status & marital status of parents, total number of children in the household on the Total family Stress, Distress Scale, Health related quality of Life & Coping strategies.

The first part of the thesis provides a brief overview on the Empirical Background of the research study which focuses on the basis of NF1 such as definition, epidemiology, diagnostic criteria & management of NF1. It also throws some light on the desk research of previous studies conducted on Impact of NF1 on families in terms of different domains such as emotional impact, economic impact, social impact, impact on Sibling & impact on coping. Furthermore a short description is provided about the health related quality of life of NF1 children & parents, since one of the study instrument used in this research determines the health status of parents. I have also described shortly about the effect of chronic diseases and genetical diseases on the families. The second part of thesis describes about the Methodology section which gives an outlook on the type of study, the research instruments used, and the usage of different statistical analysis. Third Part of thesis describes about the Result section which gives an overview on the result obtained after using the statistical analysis. The fourth part of thesis focuses on the evaluation part giving a gist of summary of findings and the valuable explanations confronting the result output as well as the limitation and conclusion of the study.

2. Literature Search

Identifying a research question: Before starting the literature search, it was very important to formulate the research question. Earlier during my internship on coping in NF1 children and adolescents, I had spent time to understand the core of the genetical disease, which gave me a thorough understanding on the disease background as well as other psychological problems experienced by the NF1 children and also on coping strategies used by them. While doing a literature search, I came across that most of the researches on NF1 were focused on children, adolescent, adult groups and its quality of life, very few researches were conducted on the impact of NF1 on families, in short how family is burdened to have a child affected with genetical NF1 ailment. Hence I decided to formulate the research question based on the impact of the disease on the families and by more narrowing the topic I decided to study the impact of socio- demographic factors such as age, gender of NF1 children, marital status, total number of children at home, monthly income & employment status on the total family stress, quality of life, distress scale & coping strategies.

Sources of Information: Journals, Books, Dissertation, clinical research articles, systematic review articles, Weblinks were used as information sources.

Search Engines/Database: Pub med, Ovid Medline, PSYNDEX plus databases, Springer Link, Sage journals, Google Scholar was the Data bases used for the thesis.

Search Terms: Impact of NF1 and families, Family Stress and NF1, Economical Impact and NF1, Social Impact and NF1, Sibling Impact and NF1, Emotional Impact and NF1, Coping and NF1, Health related Quality of Life and NF1, Impact of genetical disease and families.

Search Criteria: Gender - Male & Female include

Language- English

Age Child: 0-18 years, All Adult: 19+ years

Year of publication from 1980 onwards.

Search Results:

Search Terms	Search Results
Impact of NF1 AND Family	20
Family Stress AND NF1	8
Economical Impact AND NF1 Financial burden And NF1	5
Social Impact AND NF1	20
Sibling Impact AND NF1	5
Emotional Impact AND NF1	10
Coping AND NF1	19
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3. Empirical Background of the Study

3.1 Neurofibromatosis type 1

NF1 is the most common autosomal dominant disorder with a prevalence of 1 in 2500 to 3000 individuals (Carey J, Baty B, et.al, 1986). In 1882, Friedrich von Recklinghausen, a German pathologist named the autosomal genetic defect as " Neurofibroma" due to its origin of tumors from neural sheath (Ferner R E, Huson M H, Evans D G, 2011). Though coined as a "rare" disorder, it is startling a common disorder (Korf R B, Rubenstein A E, 2005). All ethnic groups have likely chances to inherit the defective NF1 (North K, 1999). NF1 is not entitled to any one particular sex; both the genders have an equal risk of inheriting the disorder (Pinson S, 2005). The prevalence of NF1 is found to be more in younger children when compared to the adults (Friedmann J, 1999). There is a 50% risk of transferring the defective NF1 gene to the next generation (Pinson S, 2005).

NF1 is labeled as tumour predisposition syndrome due to its tendency to produce benign and malignant tumours in different parts of body (Pasmant E, Vidaud M, et.al, 2012). According to the linkage analysis, the genetic defect responsible for the NF1 (Mendelian disorder) has been plotted to be on the chromosome 17 (Menon A, Ledbetter D, et.al, 1989). The symptoms vary from person to person and its severity in future is unpredictable (Cnossen M, Goede-Bolder A, et.al, 1998). As age increases the frequency and the severity of symptoms also increases (Friedman.J, 1999). More than 50 percent of cases are inherited from parents (Siqviland E, Pond D, 2009). Few clinical manifestation of NF1 is seen at birth, while the tumours and the fibromas formation are age related (Hersh J, 2008). The NF1 gene has a probability of the formation of not only the neural tumors but also the non – neural tumors, which are mostly carcinogenic in nature (Matsui I, Tanimura M, Kaboyashi N, Sawada T, et.al, 1993). NF1 patients have reported of decrease in reproductive fitness and diminishing life expectancy (Castle B, Baser M E, et.al, 2003). Recently in countries like Denmark, rules have been tighten on sperm donation after one of the donor was found to have transmitted NF1 gene to more than 43 babies (Reuters, 2012).

Periodic checkups of NF1 patients and required treatment at the appropriate time may minimize any further complications in the later stages. Genetic Counseling for the NF1 patients, family members and symptomatic medical aid and care to deal with the complication for the NF1 patients can be better way to manage this genetical disorder.

3.2 Epidemiology of Neurofibromatosis type 1

In order to estimate the prevalence of NF1 in Germany, a routine medical examination at elementary school was done in 2000 & 2001. Screening of 152819 children aged 6 in six German states was done, the prevalence rate was found to be 1 in 2996 (Lammert M, Friedman J M, Kluwe L, Mautner V, 2005). The prevalence rates were estimated to me 1 in 5681, during an epidemiological & genetic survey of NF1 children under 16 years in Northern Ireland. The record of NF1 children were obtained from the Department of Medical Genetics of North Ireland (Mckeever K, Shepherd C W, Crawford H, Morrison P J, 2008). During the screening of 374440 young adults, the prevalence rates of NF1 were estimated to be 1.04 per 1000, 0.94/1000 for males and 1.19/1000 for females (Garty B Z, Laor A, Danon Y L, 1994). In South Wales, the prevalence of NF1 were estimated to be 1 in 4950 during a population based study of 668,100 (Huson S M, Compston D A, Clark P, Harper P S, 1989). During a population based study of 113,700 in Dunedin, New Zealand, the prevalence rate was found to be 1 in 2190 ; the prevalence rates were seemed to be higher in the age group between 20 to 29 (Fuller L C, Cox B, Gardner R J, 1989) . During the population based study, the prevalence rates of NF1 were estimated to be 1 in 3500 in the United States (Poyhonen M, Kytola S, Leisti J, 2000).

3.3 Clinical Hallmarks of Neurofibromatosis type 1

NF1 has varied involvement in different system of body such as skin, bones, CNS (Central Nervous System), CVS (Cardio-vascular System), Gastro-Intestinal System as well as Psychological System. In skin, it causes café- au lait macules, freckling, xanthogranulomas, glomal tumors; in bone, it leads to Pseudo-arthrosis, short stature, reduced bone density, scoliosis, vertebral scalloping, non-ossifying neurofibromas ; in central nervous system, it causes neurofibromas, cognitive impairment, brain tumors, optic pathway glioma, multiple sclerosis ; in cardio vascular system it causes hypertension ; in gastro intestinal system it leads to gastro – intestinal stromal tumors, carcinoid tumors, gastro- intestinal neurofibromas ; in Psychological system it leads to anxiety, fear, depression, learning difficulty, low self-esteem, low self-confidence mainly due to cosmetic disfigurement and unpredicted signs and symptoms in the future (Ferner R E, Huson S M, Thomas N, Moss C, et.al, 2007 ; Karl M, Charles W, Hilda C, Patrik J M, 2008)

3.4 Assessment & Management of Neurofibromatosis type 1.

Once NF1 is diagnosed in patient, routine screening investigation will be required. NF1 symptoms are complex and changes as the age increases. Hence annual investigation will play a vital role in preventing any further complication associated with the disease. The academical skills & social skills of a child should be monitored ; optic system should be checked in order to detect any optic deformity such as optic gliomas or glaucomas ; the puberty development (height, weight , precocious/delayed puberty) should be checked ; cardio- vascular system should be checked in order to detect any heart disease ; blood pressure should be monitored ; bone examination should be done in order to detect any bone deformity like scoliosis ; skin examination should be done in order to detect any plexiform neurofibromas, café – au lait macules or freckles (Ferner R E, Huson S M, Thomas N, Moss C, et.al, 2007). Tumors caused in different parts of body can be removed surgically; Spinal fusion can be helpful in case of Scoliosis; Psychological counseling would be beneficial in case of any anxiety, depression or fear caused in NF1 patients & family members (Kaufamn D, 2008). Special coordinator for NF1 children would be beneficial in order to manage the learning difficulties, ADHD symptoms along with the mutual support from parents, teachers & pediatricians (Virginia C, Lucas J, Michael A, David H, et.al, 2009).

4. Impact of Chronic Diseases on Families

4.1 Definition, Risk factors, Burden of Chronic Disease

Chronic disease is defined "*as the illness that is prolonged in duration which does not resolve spontaneously and is rarely cured completely"* (Australian Institute of Health & Welfare, 2012). In 2008, Chronic disease such as Cardio-vascular diseases, Diabetes, Cancer, Chronic Respiratory diseases and other non-communicable disease accounts for 92% of death in Germany (WHO Health Statistics, 2008). The Behavioral factors such as (smoking, alcohol-consumption, poor diet, and physical inactivity) & metabolic factors such as (obesity, raised blood glucose, hypertension, and high cholesterol) are considered to be the major risk factors associated with chronic disease. Chronic Disease ranks the highest among the overall death rate in Europe (Busse R, Blumel M, Kresnen D, et.al, 2010). In 2005, Cardiovascular diseases was responsible for the 52 million of death, with the disease burden of 34.42 million DALYs; Cancer caused 1.86 million of overall death in Europe with the disease burden of 17.03 million DALYs; Chronic Respiratory diseases such as COPD (chronic obstructive pulmonary disease and Asthma lead to 0.42 million of all deaths with the disease burden of 6.84 million DALYs; Disease such as Diabetes caused 0.15 million of all death with the disease burden of 2.32 million DALYs; Musculoskeletal disease were the cause of 0.03 million of death with the disease burden of 5.75 million DALYs (Singh D, 2008).

4.2 Chronic Diseases Impact on families

Different diseases have different levels of impact on their families. Families having Alzheimer disease patients were found to have increase level of stress when compared to the families without any identical responsibilities, immunity too was found lower in affected families when compared to the normal cohort families (Janice K, Kiecolt G, Ronald G, et.al, 1987). Life threatening disease such as AIDS (Acquired Immune deficiency syndrome) brings stigmatization not only to the affected individual but also to the family members, hence during such cases when family plays a role as care- givers, it helps to increase the quality of life of the affected person by reducing its stress and depression and increasing the self- esteem (WHO, 2005). In diseases such as Cancer , family members suffers differently based on the phases of cancer that is family members undergoes emotional turmoil during initial phase, Uncertainty during the adaptation phase and grief due to loss during the terminal phase (Northaus L, 1984). The partners of Long time Stroke survivors is burdened up with constant worries about the future, uncertainty of survival of their stroke partners and constrained social life (Scholte W J, Hann R J, Ryinder P T, Limburg M, Vanderbos G, 1998).

In chronic disease like asthma in children, parents may have to undergo a period of disturbed or lack of sleep since the asthmatic child will suffer from asthmatic episodes usually at night; parents also need to prepare separate food since the child can be allergic to some food which aggravate the asthmatic attack; parents also need to have deal with extra cleanliness of house, in order to protect the child from asthmatic attacks caused due to particular allergens, in short parents has more physical impact than mental impact in cases of chronic disease like asthma (Travis G, 1976).

Chronic dermatological diseases are known to significantly affect the Health related quality of life of family members (Basra M K, Finlay A Y, 2007). Chronic dermatological diseases such as Psoriasis and Eczema lead to overall impact on Psychological, Social & Financial domains not only of patients, but also on the families and society (Basra M K, Shahrukh M, 2009). One third of the population are affected with the chronic skin disorder known as Psoriasis (Parish L, 2012). Psoriasis is the auto- immune chronic disease leading to psychosomatic disorders in patients, lowering the self-esteem and causing depression associated with the bodily system of itching/pruritis (Gupta M, Gupta A, Ellis C, Voorheis J, 1990). Psoriasis has been associated with stress related disorder; where increase in stress is associated with the increase in symptomatology (Farber E, Nall L, 1993). To identify the quality of life of family members of skin diseases patients, a qualitative interview was conducted, it was evaluated that the families had to sacrifice some of their leisure activities, have restriction in the areas which they enjoy, families undergo with emotional stress; which is rated in a higher scale; Social zone and financial zone of the families were also affected (Basra M, Finlay A, 2007). Cost analysis research were done to identify the extent of financial burden of Psoriasis on patients and families, it was estimated that the treatment being costly; lower quality of life was experienced by the patients and family members (Feldman S R, Fleisher A B, Rebaussin D M, et.al, 1997). An issue such as sexual problems has also come up between the Psoriatic patients and their partners (Sampogna F, Gisondi P, Tabolli S, Abeni D, 2007). Due to the ugly appearance of skin, most of the people think it as an infectious diseases, hence psoriatic patient suffer from fear of rejection in public places like swimming pool and gym, hampering up the social interactive life (Ginsburg I, Link B, 1993). Due to chronic anxiety and depression, suicidal tendency has also been observed in psoriatic patients, hence constant care, encouraging and supportive words from the care givers of the family would be an advantage (Gupta M A, Gupta A K, 1998).

According to the Gale Encyclopaedia of Medicine the Vitiligo is defined as a chronic inherited disorder in which melanocytes responsible to provide skin colour is lost, as a result white patches are seen on the skin. Though vitiligo do not have physical symptoms but psychological symptoms are found more in the vitiligo affected patients. Previous study suggests that Vitiligo causes embarrassment, low self-esteem; also responsible for high psychiatric impact on the patients when an evaluation was done on 130 patients suffering from vitiligo (Dolatshahi M, Ghazi P, Feizy V, et.al , 2008). A person can come across to vitiligo at any age and can affect both the sexes. During a cross sectional study design to study the impact of quality of life among the vitiligo patients in Saudi, it was found a woman has major impact in the quality of life than men. Increase in number of white patches (depigmentation) was directly associated with the impairment in the quality of life (Mubarak L, Mohanna H, Alissa A, Jabak M , et.al, 2011). Since vitiligo can occur at any age, when it occurs after marriage to a female, then it causes disruption in the families leading to increase in marital problems even leading to end of marriage in certain strict orthodox communities (Dolatshahi M, Ghazi P, Feizy V, et.al, 2008). In children, vitiligo can lead to increase in stress level, when they are exposed to new people, new surrounding or new environment like changing of school, or meeting new people or in any social gathering (Manolache L, 2011). During adolescence the psychological impact of vitiligo depends on the increase of white patches, with subsequent anxiety and depression pertaining to the disorder. Visibility factor of vitiligo also played a vital role in affecting the quality of life. When the white patches were seen in head, face & neck, negative impact on the quality of life was seen. Stigmatization was the another issue faced by the vitiligo patients (Manolache L, 2011).

Another dermatological disorder known as Leprosy, a chronic disease which mainly affects the skin, peripheral nerves and the respiratory tract. In skin it causes sores initially and then lumps, which is not eye pleasing, the colour of skin turns light too (WHO, Leprosy Fact Sheet). Leprosy is one of stigmatizing disease specially seen in the developing countries. When person is affected with leprosy, they are either isolated and has to leave home and stay in Lepor colonies. Though it is a curable disease with proper antibiotic regime, but it takes time for the disease to be cured. During the treatment the leprosy affected patients had to manage with the social problems, physical problems, emotional problems due to unhealthy appearance of the skin surface, which has an impact on the quality of life. A study was conducted to assess the impact of Leprosy on the quality of life in India, using the WHO Quality of life Questionnaire; during the study, certain domains such as physical, psychological, social, spiritual, environmental were analysed. It was observed that the economical factor had an impact on the quality of life. Men with increase skin deformities had a negative impact on the quality of life. The environmental condition such as unhealthy surrounding, poverty, low educational status had a negative impact on the quality of life. It was also observed that women scored better in quality of life compared to men, since most of the women used acceptance as their coping strategy which helped them to manage with the skin ailment easily (Joseph G, Rao S, 1999).

Thus impact of Chronic disease in patients and families depends on the type of chronic disease. In cases of chronic disease such as cardiac disease, diabetes, cancer, chronic asthma which has more physical impact , while chronic disease such as Psoriasis, chronic dermatitis, leprosy, vitiligo has more psychological impact such as low self-esteem, low confidence, anxiety , depression, lack of socialization than physical impact.

5. Impact of Genetical Disease on Families

Each Cell in a human body is composed of 23 pair of chromosomes, each chromosome has DNA encoded in it, inside which the gene with all the genetic information are coded, any mutation or alteration in the gene leads to genetical disease (WHO, Genomic Research Center). Hence the genetical diseases are distinguished in three categories namely single gene disease, chromosomal diseases, and multifactorial diseases. The examples for the diseases included in the single gene disorders are cystic fibrosis, Neurofibromatosis, Phenylketonuria, Huntington's diseases, Sickle cell Anaemia. Chromosomal diseases include the diseases such as Down syndrome, Turner Syndrome, William Syndrome, Klinefilter syndrome. The multifactorial genetical diseases which are mostly associated with the environmental factors are Hypothyroidism, Colon Cancer, Breast cancer, Alzheimer's disease (Genetic Science Learning Center, 2012). Most of the genetical diseases have lifelong effect on the human system with the symptoms pertaining to particular diseases, the treatment are not permanent but just a palliative mode for the temporary relief. In the initial phase of detection of any genetical abnormality in an individual, the affected individual as well the member's associated with the individual also goes through a series of emotional turbulences. In this century, the parents (mother & father) & the siblings are the most closest associated of any affected individual, unlike the previous centuries where even the grandparents use to be the part of the family and the burden associated with any illness of individual were being shared and taken care. Child rearing is the challenging job considered by parents, but rearing a child with disabilities or illness is much more challenging task. Most of the genetical disease like Down syndrome or any other chromosomal abnormality syndromes are diagnosed right when the child is born, parents experience a state of shock when they first realize about the disability of child. Some parents in the initial stage go through the stage of denial and slowly by slowly they come to terms and accept the disability of child (Dykens E M, 2000). Parents reaching a stage of adaptation to accept the illness of child require a good bonding between couples, siblings, relatives and friends.

Higher prevalence of psychiatric illness, behavioural problems (ADHD, sleep problems eating problems), and development delays are seen in Down syndrome cases (Bhatia M, Kabra M, Sapra S, 2005). Mental retardation, intellectual disability, slow in learning, delayed cognitive ability, low IQ level is seen in the Down syndrome children. Parental participation especially maternal support during the preschooling time has shown a positive sign in the mental development of the ill child (Goldstein S, Reynold C, 2011). During the growing stage, parents try to train their child born with intellectual disabilities about the daily chores, to teach the independent way of managing the daily activities such as toilet training self-feeding which in long term can be beneficial to parents and the child too. Research suggests that parents are more over-indulgent to teach children about toilet training & feeding habit and neglect the socialization part (Bhatia M, Kabra M, Sapra S, 2005). Parents need to invest extra time in order to teach which can bring about initial stress in parents. Coping during important mile stones in a child life, like joining a school, social and learning expectation can be quite difficult both for families and the affected child. Different kind of worries has been experienced by the family members of Down syndrome patients such as anxiety worries (health of diseased child, frequent visit to the hospitals), sibling worries (how healthy sibling will cope up), family functioning (house managing, work & family balance) worries and social worries (what others with think of them) (Laura M, Holsen M, 1999).

In the Single cell genetical disorder such as cystic fibrosis which produces sticky mucus in the lungs or pancreas, hence causing blockage in the airway tract leading to respiratory disturbances while in pancreas causing damage to the digestive system, the life expectancy of such genetical disorder are less, but with the advanced discovery of new medicine, the life span of the affected individual is a bit prolonged (American Lung Association, Cystic Fibrosis). During the parental interview about the burden of cystic fibrosis ill child in the families, it was found that parents reported more burden in their life when the ill child need to be hospitalized, Due to frequent visit to the hospital, the siblings complained of receiving lack of attention as parents were more involved in taking care of the ill child; problems in marital relationship as well as relationship with the close relatives were also noticed because of receiving less attention (Philip S, Bohannen W E, Gayton W F et.al, 1990). Cystic fibrosis, patients suffered with more anxiety, depression, poor social & emotional outcomes which had more impact on the quality of life of the patients (Ashish A, Shaw M, Mcshane J, et.al, 2012). Single parent taking care of ill child also brings lot of burden to care taker, when the responsibility are not been shared , for example if the mother is not working , she is assigned with the round the clock duty to take care of ill child, such issues can bring about the psychological impact on the care- taker. Hence sharing the responsibility among the family members can be better managerial way to deal with the day to day burden associated with the ill child. Lack of communication among the family members can be one of the reasons responsible for the personal stress & strain while taking care of cystic fibrosis ill child. In diseases like muscular dystrophy, where the muscles are weak, and the children have difficulty in movement needs constant physical help to carry them which puts more pressure on parents related to physical work (Travis G, 1976).

Genetical diseases such as achondroplasia (short stature, dwarfism) also have more psychological impact, but the intelligence level and the academic performance is similar to the normal child without any illness (Trotter T, Hall J, et.al 2005). Dwarfism patients suffer from day to day challenges because of their height issues such as while shopping they cannot pick up the things which are kept in higher place, hence always need someone to accompany them, difficulty in buying clothes for themselves, the ways how society has a look on them, or labelled as “little” which has an impact on the self-esteem. Lifelong health issues are also seen in dwarfism patients which are mainly orthopaedic in nature, where they may have to undergo lot of surgeries related to their disorder.

In hereditary disorder such as Haemophilia where oozing of blood or bleeding occurs anytime when the affected individual is exposed to any injury. In cases of Haemophilia economical burden is more seen among the family members, since any injury may need costly medical intervention such as blood or plasma transfusion; the kids also need to be careful in schools, where any slightest of injury can cause non-stop bleeding. Hence parents are exposed to constant worry about their safety of child in school. Skipping the school due to constant hospitalization can lead to poor performance in school. Children suffering from Haemophilia need to avoid outdoor games, hence this can also bring psychosocial issues such as anger, denial & low self-esteem in them (Psychosocial Implication of Haemophilia, Centre of Disease Control & Prevention). Haemophilia patients even have to be very selective in choosing a job, where they need to avoid any strenuous work such as dealing with machines, working outside the field where chances of injury would be more. Hence constant precaution to avoid any hurt may lead to decrease in work efficiency (Boon R, Robert D, 2008).

Diseases such as Autism which is neurodevelopmental in origin are caused due to genetic and environmental factors. In cases of Autism, children find it difficult for an interactive social relationship, verbal communication becomes difficult as there is delay in talking and understanding the language, lack or limited interest is shown in games or any physical activities. Recently a study was conducted to know whether Autism has an influence on the

Parental fatigue, it was found that fatigue is directly associated with the changing behavioural pattern of the autistic child (Seymour M, Wood C, Giolla R, 2012). Increase in behavioural problems in the autistic child had also been related to the increase in the stress level in mothers (David N, Carter A, 2008). Increase or decrease in stress level to manage the autistic children is also correlated with the coping strategies used to cope with the changing behavioural difficulties. Increase of stress is related when parents use maladaptive coping strategies such as feeling pity about themselves, or blaming themselves for the child disease. Hence coping plays a vital role in managing the anxiety, depression, stress, emotional & physical fatigue experienced by the families due to the sick child in the family. Using correct coping style can help parents to manage with day to day emotional or physical stress.

6. Impact of Neurofibromatosis type 1 on families

Family is a main pillar in a dynamics of human relationship (M.D Robert, 1957). According to the reports of Current Population Survey (CPS) issued by U.S Census Bureau, family is defined as *" a group of two people or more related by birth, marriage, or adoption & residing together, all such people (including related sub – family members) are considered as members of one family"*. Family plays a vital role in upbringing the child's overall development in terms of learning & behavior in social and emotional terms. If one family member is affected by illness or any disability, its impact is felt in the whole family. It changes the normal routine of the family. Family members need to go for adjustments depending upon the situation they come across, due to illness of child. The coping strategies adapted by the family plays a magnificent role in the management of the diseased child. Some family members cope it well while some family members experiences struggles all through their way. Hence health among the family members plays an important role for the smooth family functioning. Family health is defined as "*encompassing a family's quality of life, the health of each family members, family interaction, spirituality, nutrition, coping environments, recreation and routines, sleep & sexuality*" (Perri B, 2004). Person's individual character & behavior, Physical Environment, Social & Economical Environments are the fundamental determinants of Health (WHO, Health Impact Assessment).

Reismann & Stein (1980) defined impact as the consequences of the children illness on the family. Family Impact focuses on two fundamental issues; First and foremost on what ways are the families affected by the issues (illness, disability) in terms of personal life, social life, economically, emotionally & adjustment by the siblings. Secondly in which ways the families deals with the issues (Bogenschneider K, Little O, Ooms T, Benning S, Cadigan K, 2012). Family Impact depends on nature of the family types. Family types depends on the *structure of the family* (marital status, single parent managing the family, both the parents managing the family, nuclear family, joint family, Foster family, Adoptive family), *socio- economic characteristics* (income, educational status), *family life cycle stages* (families with an infant, school aged children, teenager, adolescent, adulthood, families having member with illness & disabilities) (Family Impact statement Guidelines, 2009).

6.1Emotional Impact

As soon as any life –threatening disease is diagnosed, patient and the family starts with the emotional influence, the way the family react, strategies to fight against the disease and finding way to manage the disease. Out of all the emotions, fear and the grief are mostly seen in the ill patient and the family members. Every individual deals differently when encountered with the disease and expresses its emotions differently, for e.g. Children when affected with any dermatological illness will experience fear to meet when they are in public, adults may feel embarrassed when they are in public. Depending upon the type of illness, the emotional outburst happens. Illness affects the patient emotionally as well as physically. Emotional burden hinders the person self- esteem, self- confidence, mental well-being its work potentiality & general well-being. Furthermore the emotional burden can make the disease person think about the negative side of life and may consider thinking that life is a meaningless journey. Hence when provided with the emotional support; emotional transition of feelings may occur. Hence psychological way of managing the disease becomes easy. The emotional burden may pile up when the ill patient are unable to do their task because of the disease. In case of NF1 children, parents themselves will realize and think about the difficulties in the future, the ill child may have to deal it. Every parent aspire their child to be a good performer in school, but when the reality strikes about the illness of child which deals with the poor learning abilities, poor cognitive functions, may worsen the condtion. Hence what matters is the acceptance of the disease or coping with the illness, which cannot be obtained in a single day, but constant step by step strategies which may take longer time to come in terms. Anger is the another outburst of emotions which the ill person come up with. Unable to accept the illness may provoke the anger in the person. When engulfed with anger or fear, the ill person may lose control on its own actions and land up making wrong decisions and give a pave for wrong thoughts in their mind. In case of chronic illness, isolation can be another strategy adopted by the sick child, In case of NF1, children or adolescents will be facing problems in public places, where the appearance of nodules and ugly appearance of the skin may make them feel awkward to face the people in the public. Episodes of depression are mostly seen in cases of Psychiatric disease and long term chronic disease which is mostly accompanied by the anxious thoughts ,worry, guilt, helplessness, restlessness, sadness, lack of sleep, feeling low, or having lack of strength.

Emotional turmoil among the parents such as anger, fear, sadness, grief, guilt, depression would start immediately after the diagnosis of NF1 in children (Korf R, Rubenstein A, 2005). During the quantitative study to investigate the emotional effects associated with the disease on families, it was found that parents were more anxious about chances of inheriting the disease among the offspring's and were more anxious about their children's health rather than about their own health (McAllister M, Davies L, Payne K, Nicholls S, 2007). Parents reported of poor emotional outcomes among NF1 children when compared with their unaffected siblings (Barton's, North K, 2004). During the assessment of quality of life among the NF1 children, it was found that a good bonding among the family members had a positive impact on quality of life (Grof A, Landolt M, Mori A, Boltshauser E, 2005). Parents who knew about NF1 inheriting in their family, found it difficult to decide whether to have a baby or not. On the other hand parents who wished to have a baby emphasized on the ante natal diagnosis, and in case of detection of any chromosomal abnormality made a decision on the termination of pregnancy (Benjamin C, Colley A, Donnai D, 1993). Cosmetic concern & visibility factor also lead to provoke emotional effects such as anxiety & depression among the parents and adolescents (Wolkenstein P, Zeller J, Reviz J, et.al, 2001).

During an assessment of CHQ (Child Health Questionnaire) to investigate the Health Related Quality of Life among the NF1 children, it was observed that parents had a significant impact on emotional & behavioural domain (Krab L, Oostenbrink R, Bolder A, Aarren F, et.al. 2009). NF1 children were observed to undergo a series of psychological symptoms which mainly included depression (Prinzie P, Descheemaeker M, Vogels A, Cleymans T, 2003). A sense of shock is experienced by the parents when they come across with the first diagnosis of NF1 in their own children, parents seldom pass through the denial stage in order to positively cope up with the situations, later on through an Anger stage, about *"why their children are affected with NF1"* (Korf R, Rubenstein A, 2005). Counseling to parents and children can be remedial recommendation in order to deal with different emotional situation arising because of this genetical ailment (Ferner R, Huson S, Thomas N, Moss C, et.al, 2007). An optimistic attitude in a chronically ill patient will help to manage the disease . Good communication skills between the parents and the children will help to manage with the disease.

6.2 Social Impact

Social Impact is defined as *"the effect of other people on an individual"* (Bibb L, 1981). Social Skill is defined as "*the ability to induce co-operation in others"* (Fligstein N, 2002). Social Skills such as "communication skills", "co-operation skills", "maintainance skills", "friendship skills", "conflict resolutions skills" needs to be developed right from the younger age to enhance the social competence (McGrath H, Francy S, 1991). In case of Neurofibromatosis, the cosmetic deformity can be one of the reason to face the people or community (Ablon J, 1999). Moreover delay in learning, speech & motar abilities in NF1 children can be one of the obstacles they face in school while coping up with other children of the same age, hence leading to low academic performance (Krab L, Oostenbrink R, Bolder A, Aarren F, et.al. 2009). Adolescent with NF1 may have to face challenges while making friends, due to their appearance and slow activity and seldom feels dejected out from a group (Church C, 1992).

Parents may find difficulty to mingle up with their friends or relatives. NF1 children were found to be less active and had decrease stamina and skills for sports when compared to their siblings (Barton B, North K, 2004). NF1 children develop negative self-concept about themselves related to physical activities like sports (Barton B, North K, 2007). Research has been conducted to examine the social domain of NF1 children and was reported to suffer from more social problems compared to their unaffected siblings (Johnson N, Saal H, Lovell A, et.al, 1999). Poorer social perceptions were seen in NF1 children, finding difficult to understand the gestures of their colleagues or family members due to the visual perception disability (Eliason M, 1986). During the social skill rating system of NF1 children, it was found that the NF1 children having ADHD (Attention deficit hyper active disorder) were more liable for poorer social domain (Barton B, North K, 2004). During a research study conducted to analyse the social, emotional & behaviour functioning of children with NF1, it was evaluated that the NF1 children have high rating of psycho-social problems; low levels in leadership qualities; less mingling up with friends; problems with CNS (Central Nervous System) was known to be the reason of decrease in social skills (Noll R, Purtill J, Moore B, et.al, 2007).

6.3 Economical Impact

Economical burden of the illness hugely depends on the employment status of the family. Also it depends whether the respective states funds for the illness of any member in their family. During the sick child at home, mostly the situations arise where either of parents quit the job in order to take care of the sick child, which has an impact on the monthly income which reduces accordingly. Hence financial burden gets piled up in such cases. Increase hospitalization due to illness may also add up into economical problems in the family. Economical impact also depends on the number of children in the family, also depends on whether it's a joint or nuclear family, depends on total number of earning members in the family. Costly medical intervention and the severity disease may lead to increase in financial problems. Here in case of NF1 the formation of nodules on the body may require persistent incision, since it can recur again and again.

Due to genetical involvement, there is no permanent cure of NF1, signs and symptoms prevail in the patients for life long. Appropriate treatment during the emerging of any symptoms is the only palliative way to deal with the disease. Medical and surgical intervention are required depending upon the severity of symptoms ; during the cost evaluation analysis for the management of NF1 was done , it was observed that the hospital & the treatment cost is higher for the moderate and the severe cases while lower cost for the milder cases (Wolkenstein P, Zaleski D, Morina J, et.al, 2000). Parents may undergo emotional effects such as depression which may affect the productivity of work & impair the work performance; frequent absentee at work place due to illness of child can also create an economical burden on parents (Wang S, Simon G, Kessler R, 2006). In order to tackle with the motar (fine, gross) skill problems, hearing difficulty, speech difficulty, learning difficulties of NF1 children , a special coordinator may be required which can also add up as an economical burden to the families (Virginia C, Lucas J, Michael A, David H, et.al, 2009).

Hence economical burden emerge up with the start of disease depending upon the financial status of person. If the genetical disease is caused to high class people of high income group, the burden is not so much felt, financial problems aggravates to the middle & poorer section of society when caught up with any illness. Health Insurance also plays an important role. If the family has health insurance for all the members, financial burden is not much felt because of the illness, since all the expenditure pertaining to disease is being take care of by the insurance. In countries like Germany where social insurance is an added advantage, the developing countries are burdened up with economic turmoil, when family faces with any illness. Hence economical impact depends on the several socio- demo graphic factors of the person.

6.4 Impact on Sibling

Special care and focus is needed by the parents to take care of the NF1 children, hence sometimes parents had to sacrifice quality time with their children who are unaffected by NF1. Health siblings had to undergo some adjustments in their daily routine. If the economic situation of the family is worse and if the cost is high for the treatment of NF1 affected sibling, the healthy siblings have to sacrifice its wants or desire. Social interaction can also be hampered of the healthy siblings, when they find it difficult to participate outdoor activities with friends due to the illness of any members in the family (William P, 1997). If the sibling is of adolescent age, he/she need to even spend time with their affected sibling on their parental absence (Eiser C, 1997). Hence sibling needs to undergo lot of adjustments due to illness of brothers or sisters.

Healthy siblings undergo a risk of negative psychological effects (Sharpe D, Rosseter R, 2002). Mayer & Vadasy (2007) who conducts siblings workshops suggest some important strategies in order to help healthy siblings to cope up with the stress associated with the illness; they recommend the following things such as discussing with the healthy sibling about the illness of their brothers/sisters, answering the queries, providing them with the vital information about the disease severity and ways to handle in emergency & to include them during the hospital visits with their parents.

Siblings may also suffer from attention deficit syndrome, when they find their parents giving special attention to the care of the sick child. Such syndrome can bring about detachments in the long run. The age of sibling also plays an important role in the way they deal with the situations. Normally younger children may not understand initially about the cares and responsibility required in order to deal with the sick child. As age increase, the siblings can also be helpful in helping their parents to manage the sick child. Impact on the sibling also depend whether the healthy sibling is the first born or second born. Previous studies suggest that the good sibling relationship with the sick child can enhance the growth of sick child in terms of emotionally, socially & morally (Howe N, Reccha H, 2006).

6.5 Impact on coping

Patients diagnosed with NF1 needs to cope up with unpredictable deterioting health, changing social and intimate relationship & increasing social needs. The general well-being depends upon how they adapt to the changing circumstances. The study of coping is fundamental to an understanding of how NF1 affects children for better or worse as well as how parents cope up with such unexpected situations. Some parents adopted a coping strategy of living one day at a time and do not allow the future worries to torment them, such adaptative goals helped them to cope with the illness of their children (Alkin K, Ahmad W, 2000). Only few researches have been done on the nature of the coping strategies adopted by the parents upon their children being diagnosed with NF1. During a comparative study of parental responses on family functioning; a significant parental gender differences were noticed in all coping subscales (Dylis A, 2005). Coping provides a supportive framework in order to tackle with the unpleasant situations aroused because of the disease. Coping also provides the structural framework for the development of psychosocial care programs or interventions for the chronically ill patients (Ridder D, Schreurs K, 2001). The individual attitude and the corresponding coping strategies influence the course of the disease (Schussler G, 1992). Hence, by recognizing the specific coping strategies adopted by the parents & children to deal with the psychosocial stress, we may be able to develop interventions to improve the quality of life.

Hence coping strategies purely depends on the type of disease, the age of the first appearance of the disease , duration of the disease, the surrounding environment such as healthy relationship with family members, friends & enrollment of good counseling programs.

7. Health Related Quality of Life

Quality of life is mostly associated with the health status of an individual, but later in 1980, researches started referring the term quality of life as Health related quality of life; which measures the physical & mental health of individual, basically focusing on the subjective health status but more impact on the perceived health status . Health related quality of life is a part of quality of life which is mainly based on important aspects such as physical, emotional & social levels.

HRQoL is defined as " multi-dimensional construct covering physical, emotional, mental, social & behavioural components of well-being & functioning as subjective perceived by a person depending on the cultural context and the value system one is living in" (Von Ruden, 2007; Schlarman J, Metzung S, Schneep W, 2008).

Patrick & Erikson (1993), defined HQRoL "*as the value assigned to the duration of life as modified by the impairments, functional status, perception & social opportunities that are influenced by the disease, injury , treatment or policy"*

In this century, the life expectation of person is more, due to new medical intervention and discovery of new medicines and awareness of the disease in the people, But the important point is based whether in their long life, are the sick person able to enjoy good quality of life? Hence Health related Quality of life measures the quality of life inspite of disease may be pertaining to acute, chronic, genetical illness and also measures whether person is able to enjoy the fullness of life. Unlike Quality of life, Health related quality of life also measures different aspects of life such as the satisfaction level, well-being (physical & mental) and the impact based on the type of work, income, household environment & social relationships. Health related Quality of Life assessments questionnaires has provided useful information on the health surveillance outcomes, determining the risk factors such as (BMI, Smoking and drinking status), determining the burden of disease & disabilities & studying the impact of health on quality of life. Study instruments such as SF 12, SF 36, Quality Well Being Scale, Sickness Impact profile has been useful to assess the perceived health status of an individual (National Center for Chronic Disease Prevention & Health Promotion). The inclusion of health related quality of life questionnaire during the outpatient visit has benefited the patients in the following ways such as the communication between the doctors and the patients has increased. When physician deals with the psychological areas with the patients, a sense of satisfaction is felt by the patients. Moreover the health outcome results obtained from the HRQol survey helps the doctors in the decision making. The feedback from Health related Quality of life survey gives a chance for the physician to improve the way of treatment as well as help the physician to re think the new way of manifesting the treatment which will not only give palliative relief to the patients but also give a hint to deal with psycho –social surrounding too. It has been observed that when patient comes for the treatment , the medicine is given according to the disease symptoms, but there are some chronic diseases which are purely aggravated by the psychiatric & emotional issues such as stress, anxiety & worry. Hence Health Related quality of life assessment questionnaire by the physician helps to measure the areas where it is difficult for a doctor to reach out, for eg. financial issues, emotional issues social issues etc. Assessment of the Health Related quality of life from time to time gives the idea about the disease progression as well as patient’s outcomes and also gives a thorough understanding on the areas to be intervened depending upon the disease condition (Varni J, Burnwinkle T, Lane M, 2005).

During study of impact of socio- demographic factors on the health related quality of life in chronic cases, it was found that the elderly people, with low educational status & single living had impaired health related quality of life; poorer health related quality of life were seen for chronic conditions such as gastric problems, cardiac problems, kidney problems Musculoskeletal problems (Sprangers M, deRegt E, Andries F, 2000). Another pilot study was conducted to determine whether socio-economic status such as educational status & financial accessibility has an impact on Health Related quality of life. It was observed that low educational status lead to decrease in Health Related quality of life and lower social accessibility and low financial resources also had a negative impact on the Health Related Quality of Life (Von-Reuden U, Gosch A, Rajmil L, et.al 2006).

7.1 Health Related Quality of Life in Children with chronic disease conditions.

A comparative study was conducted between paediatric chronic conditions such as (cardiac related problems, childhood diabetes, respiratory problems, rheumatic problems) and the healthy children, it was found that the children suffering from the chronic health condition had a higher impact of disease condition in the quality of life, such as lower in physical functioning, lower in social functional, lower in emotional functioning compared to the healthy children (Varni J, Limber C, Burwinkle T, 2007). In cases of chronic conditions such as brain tumors, the children experience lower health related quality of life, as brain tumors leads to repeated epileptic attacks, hence children had to take leave from school, hence even the functioning level at school is decreased. Normally the survival rate in cases of brain cancerous tumors are five years, if the children survives by the chemotherapy treatment and reaches the adult stage, still the patient will observe the lower health related quality of life due to signs & symptoms of the disease such as blindness caused due to the tumours, bodily perception problems; hence driving would be difficult in such conditions, finding a job will be another problematic task, since because of tumours the efficiency of work will be low, hearing impairment, impairment in perceiving the things, hence in adult stage they may not get the job as per their desire. Hence constant dependent on others has an impact on the health related quality of life (Bhat S, Goodwin T, Burwinkle T, et.al, 2005).

A study was conducted in order the assess the health related quality of life in obese children, it was observed obese children suffers from the lower health related quality of life as they emerge with more psycho social issues such as low self-esteem, difficult in mingling with new people, depression, anxiety due to their obese appearance; bullying in school by their colleagues which has an impairment in social functioning domain of HQRoL. (Schwimmer J, Burnwinkle T, Varni J, 2003). In order to study the impact of the Health Related Quality of Life in Chronic kidney disease in children, a cross sectional study was conducted, it was observed that children experienced lower quality of life, especially during the dialysis treatment , where the children has to opt for absence in school due to regular hospital visit. But one of the interesting facts which were observed during the study was that the adolescents who have a long term kidney illness had a better social, physical & emotional functioning compared to the adolescent who had a short time Kidney illness. It was an interesting fact that as the time passes, the patients both accept their disease condition and try to build their our strategy and personality to cope with the disease condition (Gerson A, Went A, Abraham A, et.al, 2010).

In a recent study the dermatological disorder known as psoriasis was compared with the disease such as diabetis, arthritis, asthma; it was found that psoriasis had a significant impairment on HRQOL with the impact on the emotional domain (Varni J, Globe D, Gandra S, Harrison D, et.al, 2012). Further in the study, in the emotional functioning domain, depression was the psychological issue the psoriatic patient had to manage. During the measurement of the health related quality of life for the patients suffering from Traumatic Brain injury, the physical and social functioning domains were seen to be affected. Followed by the major depressive disorder in the patients with long life disability (Diaza A, Schwarbald M, Thias M, et.al, 2012). In cases of HIV, a long term illness, several episodes of the mood disorder were seen in the ill patients. Such mood disorders which leads to increase in alcohol intake or drug lead to the impairment in the Health related quality of life. Trained counsellors and medically trained profession can play a major role in helping such kind of patients by proper counselling , would progressively help the patients to improve the heath related quality of life (Sherbourne C, Havs R, Fleishmen J, Vitello B, Margrude K, et.al, 2000).

7.2 Health related quality of life in NF1 children & families

An observational study was conducted among parents of 58 children to assess the Health related Quality of life; parents reported a significant impact of NF1 on domains such as physical, mental health, general health perception, self-esteem, behaviour & emotional (Krab L, Oostenbrick R, Bolder A, et.al, 2008). In New South Wales, Australia, a study was conducted in order to assess the social skills of children with NF1. Analysing was done using the Social Skill system, it was found that NF1 children had poorer social outcomes when compared to the siblings who are unaffected by the disorder. Cognitive dysfunction like deficit in visual spacial skills (perception of objects and space), visual perceptual skills (analysis & integration skills), memory (visual & verbal), language skills, learning disabilities were significantly impaired in NF1 children (Hyman S L, Shores A, North K N, 2005).

During the Bone densitometry analysis among NF1 children, it was found that NF1 children were more prone to fractures, due to decrease in bone density especially (hip, femoral & lumbar spine) , hence leading to less physical activities (Stevenson D A, Mayer L, Murray M, et.al, 2007). ADHD (Attention deficit hyperactivity disorder seem to be associated with NF1, hence leading to decrease performance in school due to lack of attention (Kayl A E, Bartlett D, Moore, 2000). NF1 children were known to have more emotional problems when compared to their same aged group, most of the children were not included in social gathering, rejected by their friends which lead to more emotional problems (Noll R B, Purtill J R, Moore B D, Schorrey E K 2007).

Presence of Café –au lait macules & fibromas ; a cosmetic deformity had significant impact on Quality of life especially affecting the emotional aspect of the NF1 patient (Kodra Y, Gustini S, Divona L, et.al, 2009). Cosmetic surgery helps in improving the Quality of life in NF Patients. Malignancy complication in NF1 patients lead to lowering of Health related quality of life especially affecting the emotional domain of the patients & families (Korf R B, 2000). Health related quality of life can be lower among the parents of NF1 children due to constant worry of the future about the health of child. Health related quality of life among NF1 children when compared with children suffering from other chronic disease such as nephrotic syndrome, it was found that the impairment level is found more in children suffering from NF1, the reason can be associated with the dermatological impact of NF1 on patients targeting their mental health compared to physical impairment found in other chronic disease such as nephrotic syndrome (Rueth E, Landelt M, Neuhaus T, et.al, 2004). Disease severity is also related to Health related quality of life where higher the complication associated with NF1; higher is the impact of the disease on quality of life among the families. Lower the complication; lower is the impact of the disease on the families.

8. Research Question

The explanatory variables of socio-demographic characteristic such as marital status, age & gender of the NF1 children, employment status, number of children in the household & the monthly financial income reflects the living condition & overview about the environment of the study participants. Socio- demographic characteristics gives a broader picture of each participant and the environment in which the participant lives , hence to study the impact of having an NF1 affected children in families would be easier by analyzing the socio-demographic characteristics. Analyzing of gender & age differences gives vital information about the impact on themselves as well as impact on the families. It is useful aspect of the research, because physical and psychological responses differ between men and women. Different age categories also uncover different somatic & psychiatric symptoms. Physical & Mental Symptoms varies in childhood, adolescence, adult age and older people. Marital status gives a broder on the relationship aspect, whether it creates a high or low impact of disease on the families. Similarly economical status & employment status of the family has its impact on health. Previous research suggests that the high income status and employment factor have a good accessibility on the dimensions of health also family could provide a better environment for the sick child. The research instruments used for the study such as Total family stress, Health related quality of life, Distress Thermometer, Coping strategies will determine the physical, mental, emotional, social, personal stress, economical burden and appropriate coping methods of the families. Hence keeping all the above points in mind, the research question used for the study which can measure the impact of NF1 on the families were formulated in the following manner.

- Does socio- demographic characteristics such as (age & gender of NF1 children, total number of children in the household, marital status, monthly income, employment status) has an impact on the SF 12 component scores (Mental & Physical), Distress Thermometer (Distress Scale) ,Total Family Stress (FABEL), & CODI Scales ?
- Is there a relationship between the age of parents & SF 12 component scores & Distress Scale?

9. Methodology

9.1 *Participants*:

The Questionnaire was given to the Parents of children who were being treated in OPD (Outpatient Department) at UKE (University Klinikum Eppendoprf, Hamburg). The parents were intimated about the purpose of study and were assured of confidentiality and consented to participate.

9.2 *Study Design*:

It was a Cross- sectional study design. The inclusion criteria were a) Parents of NF1 children whose age is between 8 to 17 years b) NF1 diagnosed patients certified by the Doctors c) Signed consent form. The exclusion criteria were a) Parents of NF1 children above 18 years b) Lack of German Language.

9.3 *Procedure:*

Eligible participants were invited with a letter to participate in the study. Parental Consent was taken before the study. Questionnaire was mailed to the parents who agreed to participate and they answered the questions at home and returned back the filled Questionnaire.

9.4 *Instruments:*

Study Instruments such as FABEL, SF-12, Distress Thermometer, and CODI were included for the study.

FABEL (Impact on Family Scale) is a self-report questionnaire developed to evaluate the outcome of pediatric illness & its impact on family. It consist of total 33 Questionnaires , response of the questionnaire is in the form of 4 point Likert scale from strongly agree to strongly disagree. The five sub scales of Impact on family scale are “*Daily social stress*”, “*Personal Stress*”, “*Financial Stress*”, “*Impact on Siblings*” & “*Problems on coping*” (Ravens U, Stein R, Morfeld M, et.al, 2001). Total family stress were calculated using the scale score calculation of FABEL by computing the variables. Higher score indicates higher impact while lower score indicates lower impact.

CODI instrument, a 29 item questionnaire was used to assess the coping strategies. The response format of the questionnaire was in the form of 5 point likert – scale ranging from never to always. The questionnaire contained six coping strategies such as Avoidance, Acceptance, Cognitive – Palliative, Distance, Emotional Reaction and Wishful thinking (Petersen C, Schmidt S, Bullinger M, 2004).

SF-12 Health Survey is a self – reported Questionnaire designed to produce Physical Component Summary Score & Mental Component Summary Scale. It includes 12 questions from the SF 36 Health Survey Questionnaire. SF12 Health Survey was developed by John. E.Ware in 1996. Total time taken to complete the Questionnaire is 2 minutes. The SF 12 Health Survey has been used for clinical research studies in cases of Arthritis (Gandhi, Salmon, et.al, 2001), Diabetes (Siddique, Rici, et.al, 2002), Heart & Stroke (Lim, Kischer, 1999). Myocardial Infarction (Mc Burney, Eagle, et.al, 2002), Mental Health Disorders (Sanderson, Andrews, 2002).

Distress Thermometer is a single item scale with 11 responses from 0 to 10 developed to assess the distress felt by the person on the previous week where 0 suggest "*not distressed*" and 10 suggest "*extremely distressed*". There is Distress thermometer available with problem list, but in our research we followed NCCN Clinical Practice Guidelines which includes Distress thermometer without the problem list (Roth A, Kornbith A, Batelcopel L, et.al, 1998).

9.5 Statistical Analysis

Descriptive Statistics were conducted to assess the socio- demographic characteristics of the study participants mainly focusing on Mean & Standard Deviation. Non parametric test such as *Mann Whitney U Test* & *Kruskel Walleys Test* were conducted to assess the impact of age & gender of NF1 children, monthly wages, employment status & marital status of parents on the Total family Stress, Distress Scale, Health related Quality of Life & Coping strategies. In order to assess the strength of relationship between the age of parents & SF 12, component scales & Distress Scale; *Spearman correlation* were conducted. All the statistical analysis was conducted using the IBM, SPSS Statistics Version 20.

Short Summary of Methodology

Study Design	• Cross Sectional Study Design
Participants	• NF1, OPD at UKE University Klinikum Eppendorf
Inclusion Criteria	• Parents of NF1 children whose age is between 8 to 17 years • NF1 diagnosed patients certified by the Doctors • Signed consent form.
Exclusion Criteria	• Parents of NF1 children above 18 years • Lack of German Language.
Study Instruments	• FABEL(Impact on Family Scale) • CODI (Coping with illness) • SF 12 Health Survey, • Distress Thermometer

10. Result

10.1 Socio- Demographic Characteristics

Table 1 gives the short summary of the Demographic Characteristics of the Participants. A total of 66 parents participated for the study. They ranged in age from 32 to 58 Yrs. (M= 44.50 and SD= 4.79). The majority of the participants who answered the questionnaire were mothers (51%). The majority of participants were in the age categories of 41 to 60 years. Majority of the participants belonged to German Nationality (94 %). The majority of the participants were married (81.8%). The majority of the participants had two children in their family (50 %). The highest educational status of most of participants was Secondary School (40.9%). Most of the participants were employed (72.7%). The majority of participants earned more than 3500 Euros as their monthly salary (19.7%). Among the families, majority of them had males diagnosed with NF1 (54.5%).

Table 1: Demographic Characteristics of the Participants

Characteristics	Participants
AGE CATEGORIES OF PARENTS	
21- 40 Yrs.	10 (15.2%)
41- 60 Yrs.	54 (81.8%)
Missing	2 (3.0 %)
NATIONALITY	
German	62 (94%)
Italian	1 (1.5%)
Korean	1 (1.5%)
Austria	1 (1.5%)
Missing	1 (1.5%)

Characteristics	Participants
FAMILY STATUS	
Single / Widow/Divorced	10 (15.2%)
Married	54 (81.8%)
Missing	2 (3 %)
TOTAL NUMBER OF CHILDREN IN THE FAMILY	
One	10 (15.2%)
Two	33 (50.0%)
Three	17 (25.8%)
Four	2 (3.0%)
Five	3 (4.5%)
Missing	1 (1.5%)
GENDER OF THE NF1 DIAGNOSED CHILDREN	
Female	29 (43.9%)
Male	36 (54.5%)
Missing	1 (1.5%)
AGE CATEGORY (NF1 CHILDREN)	
8 - 12 Yrs.	29 (43.9%)
13- 17 Yrs.	36 (54.5%)
Missing	1 (1.5%)
EDUCATIONAL STATUS OF THE PARENTS	
Elementary School	14 (21.2%)
Secondary School	27 (40.9%)
Polytechnique High School	2 (3.0%)
Applied Science/ Technical School	6 (9.1%)
Matriculation/ Vocational Diploma	15 (22.7%)
Others	1 (1.5%)
Missing	1 (1.5%)

Characteristics	Participants
EMPLOYEMENT STATUS	
Yes (Full Time, Part Time)	48 (72.7%)
No (House wife/ House man Unable to work, Unemployed)	16 (24.2%)
Missing	2 (3.0%)
MONTHLY INCOME (IN EUROS)	
500 – 750	1 (1.5%)
750 – 1000	2 (3.0%)
1250 – 1500	4 (6.1%)
1500 – 1750	2 (3.0%)
1750 – 2000	9 (13.6%)
2000 – 2250	4 (6.1 %)
2250 – 2500	4 (6.1%)
2500 – 2750	4 (6.1%)
2750 – 3000	8 (12.1%)
3000 – 3250	1 (1.5%)
3250 – 3500	3 (4.5%)
Above 3500	13 (19.7%)
Missing	11 (16.7%)

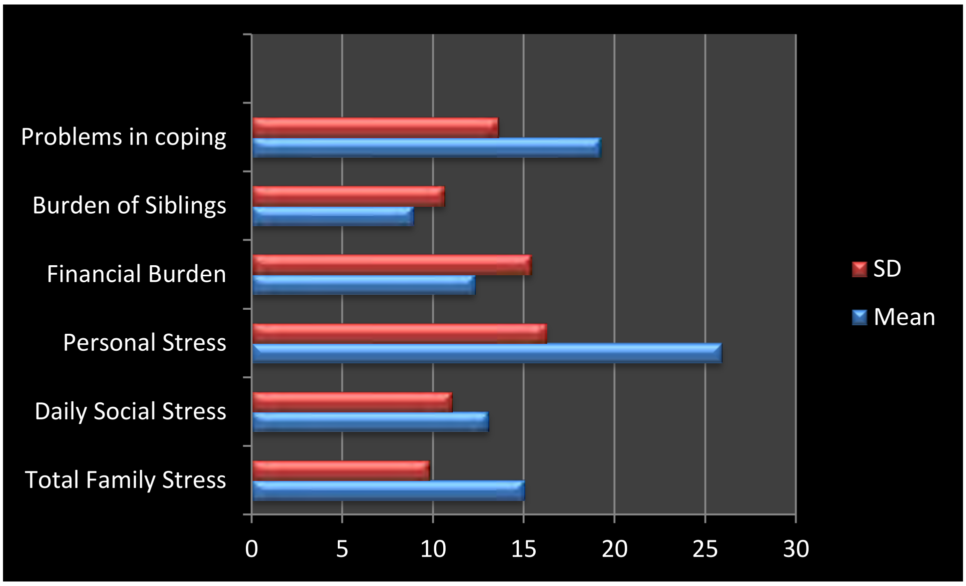
10.2 Descriptive Statistics of Fabel Scales

Table 2. gives a short description of Mean and Standard deviation scored by different subscales of Fabel. Highest mean was seen for the subscale" Personal Stress" (M = 25.89, SD = 16.23) while the lowest mean was seen for the subscale "Burden of Sibling" (M = 8.91, SD = 10.63)

Table 2 : Descriptive Statistics of Fabel Scales

Fabel Scales	N	Mean	SD	Min	Max
Total Family Stress	66	15.03	9.8	2.27	53.03
Daily Social Stress	66	13.05	11.04	1.67	68.33
Personal Stress	66	25.89	16.23	.00	65.00
Financial Stress	64	12.30	15.42	.00	62.50
Burden of Siblings	57	8.91	10.63	.00	37.50
Problems In Coping	66	19.25	13.59	.00	58.33

Figure 1: Descriptive Statistics of Fabel Scales



10.3 Impact of Age & Gender of NF1 children, Total number of children, marital status, Monthly income, Employment status on the Total Family Stress

Gender of NF1 children & Total Family Stress

Mann Whitney U Test was conducted to know whether male NF1 child and female NF1 child differ in terms of the Total Family Stress experienced by the parents. Table 3 shows that the female NF1 child had an average rank of 35.48 & male NF1 child had an average rank of 31.00. The z value is -.951 with the significance level of p = .341. There is no statistical significance in the total family stress scores of males and females.

Table 3 : Gender of NF1children & Total family Stress

Variable	Gender	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Total Family Stress	Female Male	29 36	35.48 31.00	1029.00 1116.00	450.000	1116.000	-.951	.341	-0.11

Age of NF1 children (8-12), (13-17) & Total Family Stress

Mann Whitney U test was conducted to know whether there is significant age group differences (8-12) & (13 -17) for the Total family stress. Table 4 shows that NF1 children with the age group between 8 -12 Yrs., had an average rank of 35.24 & the NF1 children with the age group between 13-17 Yrs. had an average rank of 31.19. The z value is -.859 with the significance level of p = .390. There is no statistical significance in the total family stress score of two age groups (8-12) & (13-17) Yrs.

Table 4: Age groups of NF1 children (8-12) & (13-17)

Variable	Age Group (Yrs)	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Total Family Stress	8-12 13-17	29 36	35.24 31.19	1022.00 1123.00	457.000	1123.000	-.859	.390	-0.10

Total Number of children & Total Family Stress

Mann Whitney U test was conducted to know whether having more number of children differ in terms of Total Family Stress. Table 5. shows that family having less that 2 children had an average rank of 31.93 & the family having more than 3 children had an average rank of 35.09. The z value is -.639 with the significance level of p = .523. There is no statistical significance in the total family stress score & the number of children in the household.

Table 5: Total Number of children in the household & Total family Stress

Variable	No. of children	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Total Family Stress	Less than 2 More than 3	43 22	31.93 35.09	1373.00 772.00	427.000	1373.000	-.639	.523	-0.08

Marital Status & Total Family Stress.

Mann Whitney U test was conducted to know whether there is significant difference across the marital status and Total Family Stress. Table 6. shows that marital status (single, widow,divorced) children had an average rank of 35.90 and married status had an average rank of 31.87. The z value is -.639 with the significance level of p = .529. There is no statistical significance in the total family stress score & marital status.

Table 6: Marital Status & Total Family Stress

Variable	Marital Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Total Family Stress	Single/Widow /Divorce Married	10 54	35.90 31.87	359.00 1721.00	236.00	1721.0	-.630	.529	-0.079

Monthly Income (in euros) & Total Family Stress

Kruskall- Wallis test were conducted to know if there is a significant difference across the different levels of monthly income & Total Family Stress. Table 7, shows that the significance level was .463, this is more than alpha level of 0.05, hence suggesting that there is no significant difference across the monthly income and the Total Family Stress.

Table 7 : Monthly Income & Total Family Stress

Variable	Monthly Income (in euros)	N	Mean Rank	Chi square	df	Sig.
Total Family Stress	500-2000 2100-3400 3500 & above	18 24 13	26.64 26.42 32.81	1.541	2	.463

Employment Status & Total family Stress

Mann Whitney U test was conducted to know whether there is significant difference across the employment status and Total Family Stress. Table 8. shows that family with employment had an average rank of 31.78 & the family with no employment had an average rank of 34.66. The z value is -.536 with the significance level of p = .592. There is no statistical significance in the total family stress score & employment status

Table 8 : Employement Status & Total Family Stress

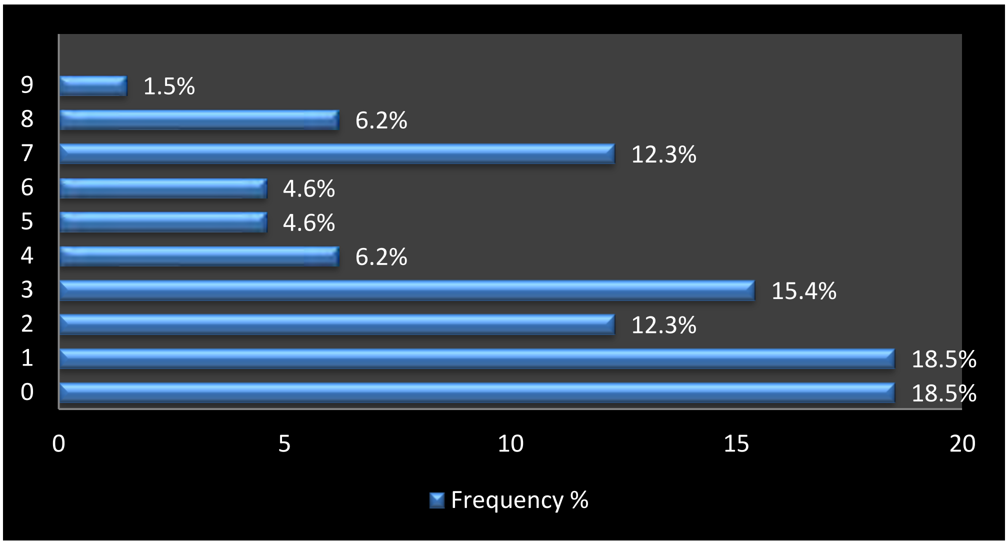
Variable	Employment Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Total Family Stress	Yes(Full/Part Time) No (House Wife, Houseman, Unable to work, Unemployed	48 16	31.78 34.66	1525.50 554.50	349.50	1525.50	-.536	.592	0.06

10.4 Distress Thermometer

Frequency Distribution of Distress Thermometer

A short look on the frequency distribution of the Distress Thermometer scale is seen in Fig. 2 Most of the participants answered of “No distress” (18.5%)

Figure 2:Frequency Distribution of Distress Thermometer



Descriptive Statistics of Distress Thermometer

Table 9, gives an overview of Mean and SD of the Distress thermometer scale (M = 3.14, SD = 2.709)

Table 9 : Descriptive Statistics of Distress Thermometer

Scale	N	M	SD	Min	Max
How much distress you have been experiencing in the past week?	65	3.14	2.709	0	9

10.5 Impact of Age& Gender of NF1 children, Total number of children, marital status, Monthly income, Employment status on the Distress Thermometer

Gender of NF1 children & Distress Thermometer

Mann Whitney U Test was conducted to know whether male NF1 child and female NF1 child differ in terms of the Distress experienced by the parents instrumented by the Distress thermometer. Table 10, shows that the female NF1 child had an average rank of 31.05 &

male NF1 child had an average rank of 33.70. The z value is -.572 with the significance level of p = .567. There is no statistical significance in the distress scale of males and females.

Table 10 : Gender of NF1children & Distress Thermometer

Variable	Gender	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distress Scale	Female Male	29 35	31.05 33.70	900.50 1179.50	465.500	900.500	-.572	.567	-0.07

Age of NF1 children (8-12), (13-17) & Distress Thermometer

Mann Whitney U test was conducted to know whether there is significant age group differences (8-12) & (13 -17) for the Distress Thermometer. Table 11, shows that NF1 children with the age group between 8 -12 Yrs., had an average rank of 35.09 & the NF1 children with the age group between 13-17 Yrs. had an average rank of 30.49. The z value is -.991 with the significance level of p = .321. There is no statistical significance in the Distress Thermometer score of two age groups (8-12) & (13-17) Yrs.

Table 11 : Age groups of NF1 children (8-12) & (13-17)

Variable	Age Group (Yrs)	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distress Scale	8-12 13-17	28 36	35.09 30.49	982.50 1097.50	431.500	1097.500	-.991	.321	-0.12

Total Number of children & Distress Thermometer

Mann Whitney U test was conducted to know whether having more number of children differ in terms of Distress Thermometer Table 12, shows that family having less that 2 children had an average rank of 31.21 & the family having more than 3 children had an average rank of 34.95. The z value is -.771 with the significance level of p = .441. There is no statistical significance in the Distress Thermometer & the number of children in the household.

Table 12: Total no. of children in the household & Distress Thermometer.

Variable	No. of children	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distress Scale	Less than 2 More than 3	42 22	31.21 34.95	1311.00 769.00	408.00	1311.00	-.771	.441	-0.09

Marital Status & Distress Thermometer

Mann Whitney U test was conducted to know whether there is significant difference across the marital status & Distress Thermometer. Table 13, shows that the marital status (Single, widow, divorced) had the average rank of 33.50 & the Married had the average rank of 31.72. The z value is -.285 with the significance level of p = .776. There is no statistical significance in the Marital Status & Distress Thermometer.

Table 13: Marital Status & Distress Thermometer

Variable	Marital Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distress Scale	Single/Widow /Divorce Married	10 53	33.50 31.72	335.0 1681.0	250.0	1681.0	-.285	.776	-0.03

Monthly Income (in euros) & Distress Thermometer

Kruskall- Wallis test were conducted to know if there is a significant difference across the different levels of monthly income & Distress Thermometer. Table 14, shows that the significance level was .916, this is more than alpha level of 0.05, hence suggesting that there is no significant difference across the monthly income and the Distress Thermometer.

Table 14: Monthly Income & Distress Thermometer

Variable	Monthly Income (in euros)	N	Mean Rank	Chi square	df	Sig.
Distress Scale	500-2000 2100-3400 3500 & above	18 24 13	26.72 28.69 28.50	17.5	2	.916

Employment Status & Distress Thermometer

Mann Whitney U test was conducted to know whether there is significant difference across the employment status & Distress Thermometer. Table 15, shows that family who had the employment had the average rank of 31.16 & the family having no employment had the average rank of 34.47. The z value is -.630 with the significance level of p = .529. There is no statistical significance in the Distress Thermometer & the employment status

Table 15: Employment Status & Distress Thermometer

Variable	Employment Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distress Scale	Yes(Full/Part Time)	47	31.16	1464.50	336.5	1465.50	-.630	.529	-0.07
	No (House Wife, Houseman, Unable to work, Unemployed)	16	34.47	551.50					

10.6 Descriptive Statistics of SF – 12 Summary Scores (Mental Component Score & Physical Component Score)

The descriptive statistics of Physical Component Score and the Mental Component Score is presented in Table 16. The physical Component Score has M = 49.38 and SD = 7.75, while the Mental Component Score has M = 46.40 and SD = 11.73

Table 16: Descriptive Statistics of SF 12 Summary Scores

Summary Scores	N	M	SD	Min	Max
PCS	65	49.38	7.75	24.14	64.84
MCS	65	46.40	11.73	17.87	62.27

10.7 Impact of Age& Gender of NF1 children, Total number of children, Marital status, Monthly income, Employment status on the Physical & Mental Component Scores (SF 12)

Gender of NF1 children & MCS, PCS

Mann Whitney U Test was conducted to know whether male NF1 child and female NF1 child differ in terms on the Physical & Mental Component scores. Table 17, shows that the z value for PCS is -.1.288 with the significance level of p = .198. The z value for MCS is -.546 with the significance level of p = .585. There is no statistical significance in the MCS & PCS scores of males and females.

Table 17: Gender of NF1children & MCS, PCS

Variable	Gender	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
PCS	Female Male	29 35	35.79 29.77	1038.00 1042.00	412.00	1042.00	-1.288	.198	-0.161
MCS	Female Male	29 35	31.00 33.66	902.00 1178.00	467.00	902.00	-.546	.585	-0.06

Age of NF1 children (8-12), (13-17) & MCS, PCS

Mann Whitney U test was conducted to know whether there is significant age group differences (8-12) & (13 -17) for the MCS & PCS. Table 18, shows that the PCS has a Z value of -.189 with the significance level of p = .850. The MCS has a z value of -.433 with the significance level of p = .665. There is no statistical significance in the MCS & PCS score of two age groups (8-12) & (13-17) Yrs.

Table 18: Age groups of NF1 children (8-12), (13-17) & MCS, PCS

Variable	Age Group (Yrs)	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect size
PCS	8-12 13-17	28 36	33.00 32.11	924.00 1156.00	490.00	1156.000	-.189	.850	-.0.02
MCS	8-12 13-17	28 36	33.64 31.61	942.00 1138.00	472.00	1138.00	-.433	.665	-0.05

Total Number of children in the household & MCS, PCS

Mann Whitney U test was conducted to know whether having more number of children differ in terms of MCS & PCS. Table 19, shows that the PCS had the z value of -.779 with the significance level of p = .436. The MCS had the z value of -.722 with the significance level of p = .470. There is no statistical significance in the PCS, MCS score & the number of children in the household.

Table 19:Total Number of children in the household & MCS, PCS

Variable	No. of children	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
PCS	Less than 2 More than 3	43 21	33.77 29.90	1452.00 628.00	397.00	628.00	-.779	.436	-0.097
MCS	Less than 2 More than 3	43 21	33.67 30.10	1448.00 632.00	401.00	632.00	-.722	.470	-0.090

Marital Status & PCS, MCS

Mann Whitney U test was conducted to know whether there is significant difference across the marital status of the family (single, married, widow, divorced) and PCS & MCS Table 20, shows that the PCS had the z value of -.1.599 with the significance level of p = .110. The

MCS had the z value of -.395 with the significance level of p = .693. There is no statistical significance in the PCS, MCS score & marital status.

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Table 20: Marital Status & PCS, MCS

Variable	Marital Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
PCS	Single/Widow /Divorce Married	10 53	23.50 33.60	235.00 1781.0	180.0	235.00	-1.599	.110	-0.20
MCS	Single/Widow /Divorce Married	10 53	34.10 31.60	341.0 1675.0	244.0	1675.0	-.395	.693	-0.04

Monthly Income (in euros) & PCS, MCS

Kruskall- Wallis test were conducted to know if there is a significant difference across the different levels of monthly income & PCS, MCS. Table 21, shows that the PCS has a significance level of 0.04, this is less than alpha level of 0.05,while MCS has a significance level of .577, this is more than alpha level of 0.05. Hence suggesting that there is significant difference across the monthly income and the PCS while no significant difference across monthly income & MCS.

Table 21: Monthly Income & PCS, MCS

Variable	Monthly Income (in euros)	N	Mean Rank	Chi square	df	Sig.
PCS	500-2000 2100-3400 3500 & above	17 24 13	25.03 22.52 39.92	10.933	2	0.04
MCS	500-2000 2100-3400 3500 & above	17 24 13	27.79 29.35 23.69	1.101	2	.577

Employment Status & PCS, MCS

Mann Whitney U test was conducted to know if there is a significant difference across the employment & MCS & PCS. Table 22, shows that the PCS had the z value of -.492 with the significance level of p = .623. The MCS had the z value of -.073 with the significance level of p = .942. There is no statistical significance in the PCS, MCS score & employment status

Table 22: Employment Status & PCS, MCS

Variable	Employment Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
PCS	Yes(Full/Part Time)	48	32.64	1566.5	329.50	449.5	-.492	.623	-0.06
	No (House Wife, Houseman, Unable to work, Unemployed)	15	29.97	449.50					
MCS	Yes(Full/Part Time)	48	31.91	1531.5	355.50	1531.50	-.073	.942	-0.01
	No (House Wife, Houseman, Unable to work, Unemployed)	15	32.30	484.50					

10.8 Descriptive Statistics of CODI Scales

The descriptive statistics of CODI scales has been presented in the table 23, Highest mean is seen in the Wishful thinking scale M = 66.15and SD = 20.65 while the lowest mean is seen in the Emotional Reaction Scale M = 20.34 and SD = 15.87.

Table 23: Descriptive Statistics of CODI scales

CODI Scales	N	M	SD	Min	Max
Avoidance	62	48.52	27.20	0.00	100
Cognitive	60	42.59	18.75	10.00	85.00
Emotional Reaction	61	20.34	15.87	0.00	62.50
Wishful Thinking	59	66.15	20.65	4.17	95.83
Distance	59	30.03	18.10	0.00	66.67
Acceptance	65	62.30	25.04	0.00	100

10.9 Impact of Age& Gender of NF1 children, Total number of children, Marital status, Monthly income, Employment status on the CODI Scales.

Gender of NF1 children & CODI Scales

Mann Whitney U Test was conducted to know whether male NF1 child and female NF1 child differ in terms on the CODI Scales. Table 24, shows that the z value for Distance is -.371. with the significance level of p = .711 The z value for Wish full thinking is -.518 with the significance level of p = .605. The z value for Acceptance is -.801 with the significance level of p = .423. The z value for Emotional Reaction is -.299 with the significance level of p = .765. The z value for Cognitive Palliative is -.857 with the significance level of p = .391. The z value for Avoidance is -1.485 with the significance level of p = .138. There is no statistical significance in the CODI scale scores of males and females.

Table 24: Gender of NF1children & CODI Scales

Variable	Gender	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distance	Female Male	28 36	33.46 31.75	937.00 1143.00	477.00	1143.00	-.371	.711	-0.04
Wishful Thinking	Female Male	29 35	33.86 31.44	948.00 1132.00	466.00	1132.00	-.518	.605	-0.06
Acceptance	Female	28	30.39	851.00	445.00	851.00	-.801	.423	-0.10

	Male	36	34.14	1229.00					
Emotional Reaction	Female Male	28 36	33.29 31.89	932.00 1148.00	482.00	1148.00	-.299	.765	-0.03
Cognitive Palliative	Female Male	27 36	34.28 30.29	925.00 1090.00	424.00	1090.00	-.857	.391	-0.10
Avoidance	Female Male	28 36	36.39 29.47	1019.00 1061.00	395.00	1061.00	-1.485	.138	-0.18

Age of NF1 children (8-12), (13-17) & CODI Scales

Mann Whitney U test was conducted to know whether there is significant age group differences (8-12) & (13 -17) for the CODI Scales. Table 25, shows that the z value for Distance is -.364. with the significance level of p = .716 The z value for Wish full thinking is -.511 with the significance level of p = .609. The z value for Acceptance is .462 with the significance level of p = .644. The z value for Emotional Reaction is -1.923 with the significance level of p = .054. The z value for Cognitive Palliative is -.784 with the significance level of p = .433. The z value for Avoidance is -654 with the significance level of p = .513. There is no statistical significance in the CODI scale scores of two age groups (8-11) & (12-17).

Table 25: Age of NF1 children (8-12), (13-17) & CODI Scales

Scales	Age groups	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distance	8-12 13-17	28 36	31.55 33.24	883.50 1196.50	477.50	883.50	-.364	.716	-0.04
Wishful Thinking	8-12 13-17	28 36	33.84 31.46	947.50 1132.50	466.50	1132.50	-.511	.609	-0.06
Acceptance	8-12 13-17	28 36	31.29 33.44	876.00 1204.00	470.00	876.00	-.462	.644	-0.05
Emotional Reaction	8-12 13-17	28 36	37.55 28.57	1051.50 1028.50	362.50	1028.50	-1.923	.054	-0.24
Cognitive Palliative	8-12 13-17	28 35	34.02 30.39	952.50 1063.50	433.50	1063.50	-.784	.433	-0.09
Avoidance	8-11 13-17	28 36	30.79 33.83	862.00 1218.00	456.00	862.00	-.654	.513	-0.08

Total Number of children in the household & CODI Scales

Mann Whitney U test was conducted to know whether there is significant age group differences (8-12) & (13 -17) for the CODI Scales. Table 26 shows that the z value for Distance is -2.318. with the significance level of p = .020 The z value for Wish full thinking is -.384 with the significance level of p = .701. The z value for Acceptance is -1.206 with the significance level of p = .228. The z value for Emotional Reaction is -1.065 with the significance level of p = .287. The z value for Cognitive Palliative is -1.418 with the significance level of p = .156. The z value for Avoidance is -.868 with the significance level of p = .386.

Table 26: Total Number of Children in the household & CODI Scales

Scales	No. of children	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect Size
Distance	Less than 2 More than 3	42 22	28.65 39.84	1203.50 876.50	300.50	1203.50	-2.318	.020	-0.28
Wishful Thinking	Less than 2 More than 3	42 22	33.14 31.27	1392.0 688.00	435.00	688.00	-.384	.701	-0.04
Acceptance	Less than 2 More than 3	42 22	30.48 36.36	1280.0 800.00	377.00	1280.00	-1.206	.228	-0.15
Emotional Reaction	Less than 2 More than 3	42 22	30.71 35.91	1290.0 790.00	387.00	1290.00	-1.065	.287	-0.13
Cognitive Palliative	Less than 2 More than 3	41 22	29.61 36.45	1214.00 802.00	353.00	1214.00	-1.418	.156	-0.17
Avoidance	Less than 2 More than 3	42 22	31.05 35.27	1304.00 776.00	401.00	1304.00	-.868	.386	-0.10

Marital Status & CODI Scales

Mann Whitney U test was conducted to know whether there is significant difference across the marital status of the family (single, married, widow, divorced) & CODI Scales Table 27 shows that the z value for Distance is -1.56. with the significance level of p = .117. The z value for Wish full thinking is -.82 with the significance level of p = .410. The z value for

Acceptance is -.48 with the significance level of p = .630. The z value for Emotional Reaction is -1.15 with the significance level of p = .249. The z value for Cognitive Palliative is -1.14 with the significance level of p = .253. The z value for Avoidance is -1.04 with the significance level of p = .298.

Table 27: Marital Status & CODI Scales

Scales	Marital Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcoxon W	Z	Sig 2 tailed	Effect-Size
Distance	Single/Widow/Divorce	10	23.80	238.00	183.0	238.0	-1.56	.117	-0.19
	Married	53	33.55	1778.0					
Wishful Thinking	Single/Widow/Divorce	10	36.35	363.50	221.5	1652.5	-.82	.410	-0,10
	Married	53	31.18	1652.5					
Acceptance	Single/Widow/Divorce	10	34.55	345.5	239.5	1670.5	-.48	.630	-0.06
	Married	53	31.52	1670.5					
Emotional Reaction	Single/Widow/Divorce	10	25.90	259.0	204.0	259.0	-1.15	.249	-0.14
	Married	53	33.15	1757.0					
Cognitive Palliative	Single/Widow/Divorce	10	25.55	255.50	200.5	255.5	-1.14	.253	-0.14
	Married	52	32.64	1697.5					
Avoidance	Single/Widow/Divorce	10	26.50	265.0	210.0	265.0	-1.04	.298	-0.13
	Married	53	33.04	1751.0					

Monthly Income (in euros) & CODI Scales

Kruskall- Wallis test were conducted to know if there is a significant difference across the different levels of monthly income & CODI Scales. Table 28, shows that the Distance Scale had a significance level of .295, Wishful thinking has a significance level of .730, Acceptance has the significance level of .660, Emotional Reaction has the significance level of .852. Cognitive palliative has the significance level of .306, Avoidance has the significance level of .258. All the significance values of CODI Scales are more than 0.05, Hence suggesting that there is no significant difference across the monthly income and the CODI Scales.

Table 28: Monthly Income & CODI Scales

Scales	Monthly Income	N	Mean Rank	Chi square	df	Sig.
Distance	500-2000 2100-3400 3500 & above	18 24 13	25.17 31.77 24.96	2.439	2	.295
Wishful Thinking	500-2000 2100-3400 3500 & above	18 24 13	28.36 26.31 30.62	.631	2	.730
Acceptance	500-2000 2100-3400 3500 & above	18 24 13	30.72 27.13 25.85	.832	2	.660
Emotional Reaction	500-2000 2100-3400 3500 & above	18 24 13	29.19 26.63 28.88	.319	2	.852
Cognitive Palliative	500-2000 2100-3400 3500 & above	18 24 13	30.12 28.75 21.77	2.367	2	.306
Avoidance	500-2000 2100-3400 3500 & above	18 24 13	22.94 30.46 30.46	2.708	2	.258

Employment Status & CODI Scales

Mann Whitney U test was conducted to know whether there is significant difference across the employement status & CODI Scales Table 29 shows that the z value for Distance is -.836 with the significance level of p = .403 The z value for Wish full thinking is -.926 with the significance level of p = .355. The z value for Acceptance is -.316 with the significance level of p = .752. The z value for Emotional Reaction is -.956 with the significance level of p = .339. The z value for Cognitive Palliative is -.727 with the significance level of p = .467. The z value for Avoidance is -1.462 with the significance level of p = .114.

Table 29: Employment Status & CODI Scales

Scales	Employement Status	N	Mean Rank	Sum Rank	Mann Whitney U Test	Wilcox -on W	Z	Sig 2 tailed	Effect -Size
Distance	Yes(Full/Part Time)	48	30.94	1485.0	309.00	1485.0	-.836	.403	-0.10
	No (House Wife, Houseman, Unable to work, Unemployed)	15	35.40	531.0					
Wishful Thinking	Yes(Full/Part Time)	48	33.19	1593.0	303.00	423.0	-.926	.355	-0.116
	No (House Wife, Houseman, Unable to work, Unemployed)	15	28.20	423.0					
Acceptance	Yes(Full/Part Time)	48	32.41	1555.5	340.50	460.50	-.316	.752	-0.03
	No (House Wife, Houseman, Unable to work, Unemployed)	15	30.70	460.50					

Emotional Reaction	Yes(Full/Part Time)	48	30.77	1477.0	301.00	1477.0	-.956	.339	-0.12
	No (House Wife, Houseman, Unable to work, Unemployed)	15	35.93	539.0					
Cognitive Palliative	Yes(Full/Part Time)	48	30.60	1469.0	293.00	1469.0	-.727	.467	-0.09
	No (House Wife, Houseman, Unable to work, Unemployed)	15	34.57	484.0					
Avoidance	Yes(Full/Part Time)	48	33.88	1626.0	270.00	390.0	-1.462	.144	-0.18
	No (House Wife, Houseman, Unable to work, Unemployed)	15	26.00	390.0					

10.10 Strength of Relationship

Strength of Relationship between Age of Parents & MCS, PCS, Stress Scale.

The relationship between age of parents & MCS, PCS, Stress scale was investigated using Spearman’s Correlation. The relationship did not reach statistical significance as Sig (2 tailed) p was greater 0.05 (Table 30).

Table 30: Spearman Correlation (Age of Parents & MCS, PCS, Stress Scale)

Variables Spearman Correlation	Age of Parents	Stress Scale	PCS	MCS
Age of Parents				
Correlation Coefficient	1.000	-.025	-.025	-.091
Sig (2-tailed)	.	.933	.845	.476
N	64	14	63	63
Stress Scale				
Correlation Coefficient	-.025	1.000	-.148	-.364
Sig (2 tailed)	.933		.599	.182
N	14	15	15	15
PCS				
Correlation Coefficient	-.025	-.148	1.000	.203
Sig (2 tailed)	.845	.599	.	.105
N	63	15	65	65
MCS				
Correlation Coefficient	-.091	-.364	.203	1.000
Sig (2 tailed)	.476	.182	.105	
N	63	15	65	65

11. Discussion

11.1 Summary

The aim of the study was to investigate about the impact of the socio- demographic factors such as age & gender of NF1 children, total number of children in the household, monthly income, employment status & marital status of parents on the Total family Stress, Distress Scale, Health related quality of life & Coping strategies. . Significant difference were found between the "*monthly income*" and the "*PCS (Physical Component Score)* of SF 12, health related quality of life with the significance value of p = *0.04.* "*Wishful thinking"* was commonly used coping strategy while the "*Emotional reaction*" was the least used coping strategy. Significant differences were found between the total number of children in household and the *"Distance"* Strategy with the significance *value of p = 0.02, effect size* = *-0.28*. Majority of the families had the children of age categories between 13-17 years (54.5%), out of which majority of them were males (54.5%). It was found that 81.8% were married while 15.2% were in the category of single, divorced or widow. Concerning the employment status, 72.7% of participants either did part time or full time job, while 16% were unemployed; they were either housewife, houseman, or unable to work due to health issues or were unemployed. Regarding the financial status of the participants, majority of the families earned Euro 2100 to 3400 Euros monthly.

11.2 Interpretation of the results

In the study, the descriptive analysis of the FABEL Questionnaire suggest that majority of families dealt with problems associated with "*personal stress"* such as doubts and confusion associated with the extension of the family, fear whether another child will also suffer from the hereditary disorder. Constant worry of the future of the sick child, for example in the parental absence who will be the care taker of the sick child. Parents also had to deal with the suggestion and the advices from the relatives about the sick child, parents sometimes alone felt the burden of the having a sick child in their families. While minority of the families had to sort out the issues related to "*problems associated with siblings"* such as lack of attention to other unaffected child, constant worry about the health of other members of the house, worry about the grades of healthy siblings. However no significant differences were found between the age and gender of NF1 children, marital status, employement status, financial monthly income, total number of children in the household and the "*Total family stress"* One of the reasons could be the decrease sample size which failed to provide the statistical significance. None of the previous research has used Impact on Family Scale (FABEL) as the research instrument to determine the impact of *NF1* on *Total family Stress*. Previous study suggest that Impact on Family Scale (FABEL) has been used as an instrument to assess the impact of chronic diseases and disabilities in children & adolescent in the families, in cases of Oral Cleft palate. Developmental disorders, epilepsy, autism spectrum disorders. (Ravins-Suberer U, Morfeld M, Stein R, et.al , 2001). Previous research have indicated that the families with chronic diseases and disabled children had more burden in daily life, physical, financial, psychological & future aspects of the sick child (Ausserhofer D, Mantovan F, Pirhofer R, et.al 2009). Previous studies have also indicated about the emotional, social & economic impact of NF1 on families (Ablon J, 1999)

To understand the distress in bringing up the sick child is very important since it provides an overview about the degree of the stress the ill child and the entire families had to go through, hence the useful resources to address the problems will be an added advantage. In our study, most of the participants response about the distress came into the range of 0 & 1 which suggest of “no distress” One of the reason could be majority of the parents had the children of age range between 13 to 17, where they were able to manage the things by themselves and were not in a need of constant help from their parents which could have brought an increase in distress level. The other reason could be majority of the participants were “married”. Hence both mother and father could have mutually shared their responsibility about the household chores. The other aspect could be the number of children in the household, where most of the participants had less than 2 children hence managing the daily things could have been not that difficult for the parents. Also most of the participants were employed and had a good monthly income in order to deal with the financial stress. The distress among the caring parent increases when the age group of the ill child is less and they need to constant depend upon the parents, it also depends upon the number of persons taking care of the ill child, employment and the financial income also plays an important role, but in our study all such parameters such as age of the ill child, number of children in the household, employment status, financial status were in the favor of the parents, in other word it was in the control hence distress among the parents were less seen , inspite of having the presence of genetical disorder patient in their family. However no significant differences were found between the age and gender of NF1 children, marital status, employment status, financial monthly income, total number of children in the household and the “*Distress Scale”* Although one of the strength of our study is the sample size but for the statistical significance the higher sample size is needed to show up the significance hence we were not able to draw definitive conclusion for the study. This is the first research study where distress thermometer has been used to study the impact of NF1 on the families which is strength of this research. Previous researches suggest that the Distress thermometer scale has helped to measure the distress level in patients as well as the caretaker mostly in cases of cancer in order to recognize the psycho – social morbidity (Patel D, Sharpe L, Thewis B, Bell M, et.al, 2011; Bulli F, Miccinesi G, Maruelle A, et.al, 2009).

In order to determine the health related Quality of life of parents, the SF 12 was used as an instrument in the research study. It is quick validating instrument to measure the health related quality of life. In our study the descriptive analysis showed that the PCS (Physical Component Score) had the higher mean (M = 49.38, SD = 7.75) compared to MCS (Mental component Score (M = 46.40, SD = 11.73). Previous researches on the interpretation of mean summary score suggest that, when the value of the Mean is above 50, the participants come in the category of average health quality; the mean values of the summary score below 30 suggest lower quality of life. In our study, the mean value of both the mental and physical component score is closer to 50; hence we can say that the participants experienced an average quality of life. No significant differences were found between the age and gender of NF1 children, marital status employment status, total number of children in the household and the Physical & Mental Component score of SF 12 health survey. But significant difference was found between the "*monthly income*" and the "*PCS (Physical Component Score)*" with the significance value of p = *0.04.* Highest mean was seen in the monthly income range of above Euros 3500 (M = 39.92) which suggest that increase in financial income has an association with the better quality of life and lowest mean was seen in the monthly income range of Euro 2100 to 3400 (M = 22.52) which suggest that decrease in the monthly wages had an lower impact on the quality of life. Previous research has suggested that parents reported of higher impact of NF1, the socio- demographic factors such as gender, economical status & visibility factor of NF1 had a profound impact on the behavioral aspect in NF1 children (Krab L, Ostenbrink R, 2009). Previous studies also states that the NF1 children with more neurological problems brought about greater stress in parents , more family problems, as well as less involvement of parents towards the social life (Reiter P, Schorrey E, et.al, 2008). In other words, increase in severity of symptoms or disease, negative was the impact seen in the quality of life of the family. In case of developmental disabilities, parents have reported increase level of stress, as well as increase in problems associated with mental functioning and physical function (Arafa M, Zaher S, et.al, 2008). When children were affected by the chronic diseases like cancer, it was analyzed that mostly mother's quality of life were impaired more than fathers quality of life, one of the reason could be mothers being the care taker, spends more time with the sick child. Other reason could be the most of the finances were being managed by the fathers than the mothers (Arafa M, Zaher S, et.al, 2008). Previous research also suggests that while managing the sick child, the female healthy sibling took much care than the male healthy siblings (Sharpe D, Rossiter L, 2002).

In order to determine the coping strategies adopted by the parents in order to manage with the children affected with NF1, CODI instrument was used. *"Wishful thinking"* was the commonly used strategy while *"Emotional reaction"* was the least used strategy. Although parents may knew that Neurofibromatosis type 1 will have a lifelong impact on children and families, but sometimes using the coping strategy as *"Wishful Thinking"*, where positive thoughts about the child getting cured from the disease may bring a time being relief to the parents to manage with the disease. One of the reason behind uncommon use of " Emotional strategy" , could be that parents must have experienced seeing the progression of disease that using emotions such as anger , crying , self-pity etc, to manage the child health problems would not make the things better but will only worsened the present condition. Previous research on coping strategies adapted by the patients suffering from chronic disease with life threatening condition suggest that the ill patient used the coping strategy of *"Avoidance"* if the socio economic conditions were low, while the sick patients who had low chances of recovery , used the coping strategy of *"Acceptance"* (Fiefer H, Strack S, et.al, 1987). Previous studies also reveal that the socio demographic factors such as income and educational status have an impact on the coping strategies used by ill patients (Schmidt S, Nachtigall C, Strauss B, et.al, 2002). In this study significant differences were found between the total number of children in household and the *"Distance"* Strategy, with the higher mean found in more than 3 children which suggest that increase in number of children in the household, parents find it appropriate to forget that the sick child is suffering from genetical disease, this may also help them to focus and care the other healthy siblings. Previous studies also reveal the use of "*passive coping strategies"* such as withdrawing from the situation, self-isolation, avoiding to meet people in cases of diseases with extreme pain like cancer, bodily or other depression period (Simons L, Lewis R, Logan D, 2008;

Kaminsky et.al 2006). In cases of disability disorders "*active coping strategies*" such as trying to keep oneself busy with activities, has been used which help them to cope up with the stressful circumstances (Compas B, Saltzman H, 2001). Along with adapting certain coping strategies to deal with stress, parents also prefer to take support from family members, relative friends, counselors, psychiatrist to cope up with the changing disease symptoms of their ill child (Simons L, Lewis R, Logan D, 2008). Hence different coping styles are used by the parents depending upon the health condition of their sick child, the type of disease the child is suffering (acute or chronic or genetical) , economical status, educational status , employment status & number of people involved in taking care of sick child.

11.3. Limitation

Neurofibromatosis, genetical disorder has different signs and symptoms pertaining to the age of the affected person, the phenotypic features use to differ in different person, hence the impact of disease on families in terms of quality of life, coping strategies, family burden use to differ depending upon the age and the circumstances provided. For example, coping strategies adopted by parents can change depending upon the type of stress, type of environment (Home / Social) as well as the age of the affected child, hence to come to definite & conclusive findings is difficult .

12. Conclusion

The study confirms that, out of all the socio- demographic factors, only the factor "monthly income" had an impact on the Health Related Quality of Life. Furthermore families with more number of children in the household used "Distance" as their coping strategy. The study highlights the importance of using research instruments such as FABEL, CODI, SF-12, & Distress Thermometer. In future, for the generalization of the research, the study should be conducted in larger sample size with the comparative study with other genetical or chronic diseases. Future research on the impact of NF1 on families would give us an understanding of different strategies to be used in order to provide support to the affected families and helping in improving the quality of life. Parents & teachers should monitor timely progression of their ill child in school & extracurricular activities. Since NF1 has a lifelong impact on patients and families, efforts should be made to prepare a strategy specially focusing on counseling which should be available time to time to the parents as well as the affected child. Awareness campaign of NF1 should be initiated in public places so people are aware of the disease and can help their families and friends who have NF1 affected child in their vicinity. Due to the discovery of new medical technologies and new medicines, the mortality has been decreased. Hence, focus should emphasize on the future research on strategies to improve the quality of life both in terms of physical & mental aspects.

13. References

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14. Appendix

English Translation of Parents Questionnaire for children diagnosed with Neurofibromatosis Type I (Center for Psychological Medicine, University Klinikum, Eppendorf)

Parents Questionnaire

A. Personal Details

1. Gender ☐ Male

   ☐ Female

2. Age: ______Years

3. Nationality:

   ☐ German

   ☐ Others ________

4. Marital Status

   ☐ Single/Widow/Divorce

   ☐ Married
5. How many children do you have?
   ________ No. of Children

6. Which is the gender of your ill child?

   ☐ Girl

   ☐ Boy
7. What is the age of your ill child?
   _______Years.

8. Which is the highest educational status do you have?

   ☐ Elementary School

   ☐ Secondary School

   ☐ Poly-technique High School

   ☐ Applied Science/ Technical School

☐ Matriculation/ Vocational Diploma

☐ Others.__________

9. Are you employed

☐ Yes (Full Time, Part Time)

☐ No (Houseman, Housewife, Unable to work, Unemployed)

10. What is your monthly income?

☐ 500 – 750 €

☐ 750 – 1000 €

☐ 1250 – 1500 €

☐1500 – 1750 €

☐ 1750 – 2000 €

☐ 2000 – 2250 €

☐ 2250 – 2500 €

☐ 2500 – 2750 €

☐ 2750 – 3000 €

☐ 3000 – 3250 €

☐ 3250 – 3500 €

☐ Above 3500 €

B. Questionnaire about health status

1. In general, would you say your health is:

☐ Excellent

☐ Very Good

☐ Good

☐ Fair

☐ Poor

The following two questions are about activities you might do during a typical day. Does *YOUR HEALTH NOW LIMIT YOU* in these activities? If so, how much?

2. *MODERATE ACTIVITIES*, such as moving a table, pushing a vacuum cleaner, bowling, or playing golf:

☐ Yes, Limited a Lot

☐ Yes, Limited a Little

☐ No, Not Limited at all

3. Climbing *SEVERA*L flights of stairs:

☐ Yes, Limited a Lot

☐ Yes, Limited a Little

☐ No, Not Limited at all

During the *PAST 4 WEEKS* have you had any of the following problems with your work or other regular activities *AS A RESULT OF YOUR PHYSICAL HEALTH*?

4. *ACCOMPLISHED LESS* than you would like

☐ Yes

☐ No

5. Were limited in the *KIND* of work or other activities:

☐ Yes

☐ No

During the *PAST 4 WEEKS*, were you limited in the kind of work you do or other regular activities *AS A RESULT OF ANY EMOTIONAL PROBLEMS* (such as feeling depressed or anxious)?

6. *ACCOMPLISHED LESS* than you would like:

☐ Yes

☐ No

7. Didn’t do work or other activities as *CAREFULLY* as usual:

☐ Yes

☐ No

8. During the *PAST 4 WEEKS*, how much did *PAIN* interfere with your normal work (including both work outside the home and housework)?

☐ Not at all

☐ Little Bit

☐ Moderately

☐ Quite a Bit

☐ Extremely

The next three questions are about how you feel and how things have been *DURING THE PAST 4 WEEKS.* For each question, please give the one answer that comes closest to the way you have been feeling. How much of the time during the *PAST 4 WEEKS* –

9. Have you felt calm and peaceful?

- ☐ Always
- ☐ Most of the Time
- ☐ Good Bit of the Time
- ☐ Some of the Time
- ☐ Rarely
- ☐ Never

10. Did you have a lot of energy?

- ☐ Always
- ☐ Most of the Time
- ☐ Good Bit of the Time
- ☐ Some of the Time
- ☐ Rarely
- ☐ Never

11. Have you felt downhearted and blue?

- ☐ Always
- ☐ Most of the Time
- ☐ Good Bit of the Time
- ☐ Some of the Time
- ☐ Rarely

☐ Never

12. During the *PAST 4 WEEKS*, how much of the time has your *PHYSICAL HEALTH OR EMOTIONAL PROBLEMS* interfered with your social activities (like visiting with friends, relatives, etc.)?

☐ Always

☐ Most of the Time

☐ Good Bit of the Time

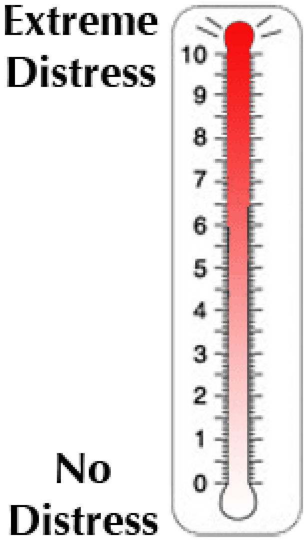
☐ Some of the Time

☐ Rarely

☐ Never

C. Your Stress

Please mark on the Distress Thermometer scale, between 0-10 , during the past week, how distressed have you been?



D.Family Questionnaire

	Totally Agree	Mostly Agree	Somewhat Disagree	Totally Disagree
1.The disease causes the family financial problems.				
2.Due to appointments at the hospital, working hours are lost				
3.I have to shorten my work, because i need to look after the child.				
4.To cover the medical cost, additional income is needed.				
5.I have to quit my job , because of the illness				
6.Due to illness of our child, we cannot take long trips				
7.People in the neighbourhood treat us differently because of the illness of our child.				
8.Due to illness of our child, we have little desire to go.				
9.It is difficult to find a reliable person to take care of the child.				
10. Sometimes we need to change our intention to go out in the last minute due to the illness of our child.				
11. Due to illness of our child , we see our family less often				
12. We are closer, because of the shared experience as a family				
13. I sometimes wonder if my sick child should be treated as normal				

14. My family are very understanding and they help me always.				
15. Because of the illness, i am not thinking about any other child.				
16. My partner and I discuss the problems of the child together.				
17. We try to treat our child as it was a normal child.				
18. The care of my sick child takes so much of time that i barely get time for other family members.				
19. The relatives get involved and think they know better what is good for my sick child.				
20. Our family often has to set things aside due to the illness of my child				
21. Due to illness of my child, I am constantly tired and worn out				
22 .I live from one day to another and do not plan for the future.				
23. No one knows with what tremendous burden, one must be ready.				
24. Going to the hospital is a burden to me.				
25. Learning to deal with my child's illness also enabled me to better get along with myself.				
26 .I am worried what will happen to my child in future.(when he is adult and I am not there)				

27. I sometimes get the feeling that our life is a roller coaster completely on the ground when my child is acutely ill, and on top if his / her health condition is stable				
28. It is difficult to give enough attention to other children because of the sick child				
29. Due to illness of the child, i worry constantly about the health of others				
30. Due to special needs of the sick child, there comes an argument with other child.				
31. The illness of the child brings fear in other children.				
32.My other children seems to be sick more often and more often suffer from pain than other children of their age.				
33. The grades of the children are suffering due to illness of my child				

E. Coping With Illness (CODI)

	Never	Rarely	Sometimes	Often	Always
1.I try to forget the illness of my child.					
2. I pretend to be that my child is alright.					
3. I try to ignore my illness of my child.					
4. I believe that faith in God helps me.					
5. I pray that my child illness will go away.					
6. I learn as much as possible about my illness of my child.					
7. I tell myself that even famous people have illnesses.					
8. I recognize that things could be worse.					
9. I am angry.					
10. I cry.					
11. I am frustrated.					
12. I am ashamed that my child is ill.					
13.I think it is unfair that my child is ill.					
14. I wake up at night fearful or anxious.					
15. I accept the illness of my child					
16. I got used to my child illness.					
17. I am able to manage the illness of my child					
18. I cope well with illness of my child					
19. I face the situation of my child with humor.					

20. I take my child illness easy.					
21. I hope that my child illness disappears.					
22.I want to stop the illness of my child.					
23. I wish my child was healthy.					
24. I don’t think my child illness is serious.					
25. I don’t care about my child illness.					
26. I think my child illness is no big deal.					
27. I forget about my child illness.					
28. Overall, how well do you think you cope with your child illness?					