

## Life without Pain: Children with Hereditary Sensory and Autonomic Neuropathy

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**Abstract:** Pain is a perceptual experience, which involves multiple integrated systems that act in a coordinated fashion. Pain plays such an important role in our life. Can we imagine a life without this perception of pain? There are certain rare diseases which result in loss or reduction of pain sensation. One such condition is hereditary sensory and autonomic neuropathy. The present study was conducted to assess the living pattern of children from same family who are suffering with this rare disease called hereditary sensory and autonomic neuropathy. 3 participants were included in the study by using purposive sampling technique. Qualitative research design was adopted for the study. Semi-structured interview schedule and observation check list was used to collect the data. The findings reveal that the children have severe cracks in their feet since 2 yrs of age. They can do their day to day activities but they do not experience any pain sensation. They also had reduced sensation to hot and cold. The study concluded that hereditary sensory and autonomic neuropathy is a familial entity. The children suffering with this rare disease have to lead a life without the sensation of pain in remaining part of their life. Health education to family members and children regarding foot care can help to prevent complications in the future.

**Key words:** pain, hereditary sensory and autonomic neuropathy, children, life, experience.

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### I. Introduction

Pain is a perceptual experience, which involves multiple integrated systems that act in a coordinated fashion. "An unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage. Our thinking regarding the nature of pain has shifted over the past four centuries from the linear dualistic concepts of Descartes to the Gate Control Theory of Pain, a more global model that includes affective components of pain. The evolution of scientific research has helped us appreciate that the pain experience is more complex and highly multifaceted from the subjective to the specific. Pain is a subjective experience. It is associated with our perception of the event and influenced by our past experiences. It is important to note that this definition is not a dualistic, either sensory or emotional experience, but a combination of both, as reflected in the Gate Control Theory of Pain (**International association for the study of pain**).

Pain motivates the individual to withdraw from damaging situations, to protect a damaged body part while it heals, and to avoid similar experiences in the future. Pain is the most common reason for physician consultation; it is a major symptom in many medical conditions, and can significantly interfere with a person's quality of life and general functioning.

Pain plays such an important role in our life. Can we imagine a life without this perception of pain?

There are certain rare genetic diseases which affect the human kind where the person's pain mechanism is altered. In such cases, the person cannot receive the pain stimuli when he/she is exposed to harmful circumstances which may endanger his/her life. One such rare condition is hereditary sensory and autonomic neuropathy.

Although genetic disorders affecting the sensory and autonomic nervous systems are rare, their existence provides a means of furthering our knowledge regarding a very complex part of the nervous system. The close relationship between development and survival of the sensory and autonomic nervous system is especially well illustrated in the diversity of a group of genetic disorders known as Hereditary Sensory and Autonomic Neuropathies (HSAN). Classification of the various sensory disorders is unsettled and ongoing.

Hereditary sensory and autonomic neuropathy type I (also known as hereditary sensory neuropathy type I and hereditary sensory radicular neuropathy) is the most common form of HSAN. It is characterized by progressive degeneration of dorsal root ganglion and motor neurons, leading to distal sensory loss and later distal muscle wasting and weakness and variable neural deafness.

In the present research, the researcher has planned to study in-depth about the children who are living with hereditary sensory and autonomic neuropathy.

### Statement of problem

A case study on mother's perception on experience of children with hereditary sensory and autonomic neuropathy

### Objectives

- To assess the mother's perception on children's daily activities /life pattern with hereditary sensory and autonomic neuropathy
- To find out the children's feelings about living with rare hereditary disease.

## II. Methodology

A qualitative research design with case study approach was used for the study. 3 children from same family suffering with hereditary sensory and autonomic neuropathy who were admitted in BLDEA's Shri B.M.Patil Medical College and research center, Bijapur, Karnataka were participated in the study. The sampling technique used was purposive sampling technique. Semi structured interview schedule and unstructured observation method were used for collecting data from the study participants. Data was collected from the children as well as their mother after obtaining informed oral consent. The data was collected for the period of three days. Data was entered in field note as it is been collected which was later on used for data analysis.

## III. Results

The findings of the study are divided into 3 sections:

1. Information regarding demographic variables of children
2. Mother's perception on children's life pattern/activities
3. Children's feelings about life without pain

### 1. Information regarding demographic variables of children

The study revealed that the children were in the age group of 7yrs, 5yrs and 3yrs old respectively. All 3 are males. The first child was studying in 2<sup>nd</sup> standard whereas other 2 children are not yet started schooling. Regarding the father's occupation father is doing agriculture work and both the first and second children are helping him in the field for carrying out field activities. Regarding parent's marriage pattern, mother has married to her maternal uncle.

**Table: 1Demographic variables**

S.No.	Demographic variables	Description
1.	Age	7yr, 5yr & 3yr respectively
2.	Gender	All 3 are male children
3.	Religion	Hindu
4.	Parent's marital status	2 <sup>nd</sup> degree consanguineous marriage
5.	Family income	Rs.50,000/-yr
6.	Family history of hereditary disease	No
7.	Father's occupation	Agriculture
8.	Schooling	2 <sup>nd</sup> standard, No schooling respectively

### 2. Mother's perception on children's life pattern/activities

Mother perception regarding children suffering with a rare disease called hereditary sensory and autonomic neuropathy, the data reveals that all three children have severe cracks on soles of both feet. This problem started for all of them when they were at the age of 2yrs. Mother's explanation was supported by an unstructured observation of children. On observation of feet, the cracks were looking like severe cuts and cleavages. Mother says that children neither experienced nor complained pain sensation in those areas when they walk, run, play or while doing work in the field. The sensation to hot cold was present but it is mild. Mother has expressed that the children used to get fever often and been treated by local doctors for the same. But not even a single time the doctors tried to identify the reason for the fever. Other finding includes that the first child has cataract on one eye along with nystagmus. She said that the cause for the crack was not diagnosed before and the treatment initiated previously was merely a temporary relief. The mother also noted that the final diagnosis was made for her children's condition in BLDEA's Shri B.M.Patil Medical College Hospital & RC. The treatment prescribed for the children include antibiotics, white soft paraffin and light liquid paraffin. When asked about the importance of care of feet to prevent infection among children, mother did not have any idea about it.

### 3. Children's feelings about life without pain

On interview with the children and observing them during the same period, children told that they never expressed pain in the foot due to cracks. When they play more or engage in vigorous activity they feel little soreness in the feet. They also said that they have less sensation towards hot and cold in the lower extremities. On observation, the second child has ulceration in the feet. All three children could walk without any difficulty. The feet were not clean and dirt accumulated in between the cracked areas. The children are not using foot wears.

## IV. Discussion

The first description of sporadic and familial cases of a condition that is compatible with HSAN was made in French literature in the 19th century. The main feature of the familial case was ulcers at the sole of the feet. In 1922, **Hicks** described a similar condition in a London family in which 10 persons suffered from perforating ulcers on their feet, lancinating and shooting pains, and deafness. Subsequently, **Jughenn et al.** and **Denny-Brown** demonstrated that the pathological process underlying the clinical features seen in these conditions was a neuropathy, rather than an anatomical disorder as had been previously suggested. Since then, many other familial conditions with similar clinical features have been reported. In the present study, all the three children born to parents of 2<sup>nd</sup> degree marriage have a familial tendency. But no other family members have this entity.

In the present study, the children present with the chief complaints of severe crack, reduced pain sensation in the lower limbs. It is supported by review from other literatures that HSAN I is characterized by marked sensory disturbances mainly as the loss of pain and temperature sensation in the distal parts of the lower limbs. The loss of sensation can also extend to the proximal parts of the lower limbs and the upper limbs as the disease progresses. Some affected individuals do not lose sensation, but instead experience severe shooting, burning, and lancinating pains in the limbs or in the trunk. Autonomic disturbances, if present, manifest as decreased sweating. The degree of motor disturbances is highly variable, even within families, ranging from absent to severe distal muscle weakness and wasting (**Auer-Grumbach, M, 2008**).

The literature review reveals that if the care of feet is not maintained it can lead to severe complications like septicemia, amputation etc. hence health education was given to the mother and children regarding importance of foot care, how to clean the feet, using foot wears, avoiding sharp instruments etc.

## V. Conclusion

From the above findings, the study concludes that hereditary sensory and autonomic neuropathy is a familial entity. The children suffering with this rare disease have to lead a life without the sensation of pain in remaining part of their life. Health education to family members and children regarding foot care can help to prevent complications in the future.

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