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# TRENDS IN DENTAL CARE FOR INDIVIDUALS WITH ECTODERMAL DYSPLASIA

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This is to certify that the thesis prepared by Justin Scott Edwards, B.S., D.M.D., entitled TRENDS IN DENTAL CARE FOR INDIVIDUALS WITH ECTODERMAL DYSPLASIA

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# TRENDS IN DENTAL CARE FOR INDIVIDUALS WITH ECTODERMAL DYSPLASIA

A thesis submitted in partial fulfillment of the requirements for the degree of Masters of Science in Dentistry at Virginia Commonwealth University.

by

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# Abstract

TRENDS IN DENTAL CARE FOR INDIVIDUALS WITH ECTODERMAL DYSPLASIA

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**Purpose:** The specific aim of this study is to evaluate the trends in dental health care for individuals with ectodermal dysplasia.

**Methods:** This was a cross sectional analysis of subjects recruited through the National Foundation of Ectodermal Dysplasia (NFED). From 1997 to 2000, individuals with ectodermal dysplasia or their caregiver (if the individuals were too young to self-report) voluntarily completed questionnaires. The questionnaire consisted of 37 items consisting of demographics, ectodermal dysplasia diagnosis, access to dental care, level of dental utilization, and type of dental services received. Descriptive statistics were used in addition to ANOVA analyses to evaluate the changing trends in oral health care for individuals with ectodermal dysplasia.

**Results:** Preliminary results indicate: 1) individuals with ectodermal dysplasia are being diagnosed earlier than in the past, 2) physicians are primary source of the initial diagnosis of ectodermal dysplasia, 3) children with ectodermal dysplasia are receiving prostheses earlier than in the past, and 4) access to care is problematic.

**Conclusion:** Diagnosis and recognition of treatment needs are occurring at an earlier age and that an access to dental care for individuals with ectodermal dysplasia continues to be an issue.

# INTRODUCTION

Ectodermal dysplasia is a hereditary, clinically diverse, genetically heterogeneous group of conditions, characterized by developmental defects in the tissues of the embryonic ectoderm. Ectodermal dysplasia may be inherited by all Mendelian means of inheritance including spontaneous mutations.[1] The ectoderm is one of the three germinal cell layers that form the early embryo. It eventually develops into the epidermis (surface skin), nails, hair, tooth enamel, sweat glands, sebaceous glands, and nerves. In addition, other derivatives of ectoderm include keratinocytes, melanocytes, endocrine glands, apocrine glands, ears, nipples, mucosa, the lens of the eye, the central nervous system, the anterior pituitary and the adrenal medulla. Any tissue that forms abnormally (dysplastic) may be characterized by the term ectodermal dysplasia. This simply implies that an end product of the ectoderm has not formed properly.[2]

An abnormal derivative of the ectoderm may be the only dysplastic tissue in an individual and occasionally, this may coincide with abnormalities involving tissues originating from other germ layers. The terminology used for ectodermal tissue deformities is straightforward. Trichodysplasia indicates that the hair is inherently abnormal. Onychodysplasia indicates that the nails are abnormal. Enamel dysplasia indicates irregularities in tooth enamel. Any one of these isolated abnormalities may be called an ectodermal dysplasia, however, the syndrome of ectodermal dysplasia is historically characterized by deformities involving multiple tissues derived from ectoderm.[3]

When multiple ectodermal tissues are affected, it may be referred to as a syndrome. A syndrome is a pattern of signs and symptoms that occur together as part of an abnormal developmental process. There are ectodermal dysplasia syndromes such as the Tooth and Nail syndrome, the Trichodental syndrome, and the Christ-Seimens-Tourraine (Hypohidrotic Ectodermal Dysplasia) syndrome. Since only ectodermal derivatives are involved in these syndromes, they may be viewed as pure ectodermal dysplasia syndromes.

The terminology is more complex when derivatives of the ectoderm and derivatives of other tissues mesoderm and endoderm are affected at the same time. For instance, Ectodactyly-Ectodermal Dysplasia-Clefting syndrome exhibits abnormalities of hair, teeth, and sweat glands along with orofacial clefting and anomalies of the hands and feet. Tricho-dento-osseous syndrome in which the hair and teeth are abnormal, also exhibits an unusual radiodensity of the bones. These syndromes cannot be called "pure" ectodermal dysplasia syndromes, because the bone is derived from the embryonic mesoderm. A better approach may be to refer to these latter syndromes as "complex" ectodermal dysplasia syndromes.

Classification of ectodermal dysplasia is constantly evolving and will likely continue to change as further clinical research and advancements in molecular genetics occur.[4] There have been more that 200 forms of ectodermal dysplasias described in the literature, however, the causative gene is known in only 30% of the defined forms of ectoderm dysplasia.[5,6] The most well known and studied form of ectodermal dysplasia is hypohidrotic ectodermal dysplasia (HED). The database of the National Foundation for Ectodermal Dysplasias (NFED) – the North American support group for ectoderm

dysplasia – has registered more than 5,200 individuals with ectodermal dysplasia from all 50 states in the US, and from over 70 countries.[5,7] More than one-third of the registered individuals have HED (>1900 or 36.5%), and more than half of the group (>2600 or 50%) have no specific clinical diagnoses.[5,7] Only a third of the registered individuals have a genetically defined diagnosis.[5]

**Signs and symptoms**

Individuals with ectodermal dysplasia have variable expressivity of the characteristics, which may make diagnosis difficult. The four cited classical structures are affected in the following decreasing order of frequency: hair, teeth, nails, and sweat glands. These may or may not be associated with alterations in other ectodermal structures.[6] Oral findings can be significant and may include multiple tooth abnormalities including hypodontia with the associated lack of normal alveolar ridge development.[8,9] Hypodontia of the primary and permanent dentition is the second most frequently occurring finding of the four major system findings.[10] Hypodontia is relatively common in the general population, with a reported prevalence of 2.2 -10.1 percent.[11] Hypodontia is a frequent sign (80%) of ectodermal dysplasia, and may be underreported.[12] Severe hypodontia, defined as the absence of 6 or more teeth (excluding third molars), has a much lower reported prevalence of 0.08-0.5 percent.[12] Diagnosis is often delayed until after the first year of life. Ectodermal dysplasia diagnosis is often made after frequent bouts of high fever and failure of tooth eruption. Other physical signs may involve anomalies of the sweat glands, scalp hair, nails, skin pigmentation, and abnormal or underdevelopment of craniofacial structures.

In addition to the classic ectodermal signs other structures derived from the

embryonic ectoderm may be affected. An individual with ectodermal dysplasia may also have absence or hypoplasia of mucous glands resulting in abnormal functioning of the mucous membranes in the nose, sinuses, Eustachian tube, oropharynx, larynx and lungs.[13] The mammary glands, thyroid gland, thymus, cornea, conjunctiva, lacrimal gland, lacrimal duct, and Meibomian gland may also be affected.[5,14]

Oligodontia is also associated with reduced salivary secretion rates.[5] Nordgarden et al. (2001) found that 22% of individuals with oligodontia had salivary flow rates below 0.1ml/min and 36.8% had chewing-stimulated salivary flow rates below 0.7 ml/min.[15] Bergendal reported (2010) the second most common sign aside from oligodontia was low salivary secretion, while only 11% reported abnormal hair, nails, or sweat glands.[5] Bergendal tested salivary secretion in 116 individuals, using the same flow rates from Nordgarden et al., thirty-five (30.2%) had low salivary secretion according to the criteria.[5] One in three individuals with oligodontia had low salivary secretion in Bergendal's study.

In older literature, the terms hypodontia, anodontia, partial anodontia and oligdontia were used to interchangeably describe various conditions of missing teeth. This ambiguity in the definition of these terms may have been partially responsible for the under reporting of ectodermal dysplasia in past publications. In its strictest sense, any congenitally missing tooth, whether it is a single tooth or the entire dentition, is defined as hypodontia.

Current literature on ectodermal dysplasia has specifically defined oligodontia to avoid any further confusion. Hobkirk and Brook considered "severe hypodontia" to be synonymous with oligodontia and defined it as "six or more congenitally missing

teeth."[5,16] Van der Weide proposed that an individual who is missing six or more permanent teeth, excluding third molars, is defined as having oligodontia.[17] Oligodontia can occur in isolation (I) or as part of a syndrome (S); classifying oligodontia into oligodontia/I and oligodontia/S has been suggested.[5]

Oral rehabilitation and preventive dental care of patients with ectodermal dysplasia requires multidisciplinary treatment by various specialities.[5,18] In an effort to minimize the number of missing teeth that need replacement, early diagnosis is critical to allow for adequate measures and treatment planning to occur.[5] Limited evidence is available concerning age of diagnosis or when is it appropriate to initiate a removable partial denture or complete denture for an individual with ectodermal dysplasia.

Many specialists may be involved in the dental treatment of the individual with ectodermal dysplasia. Frequently, orthodontic treatment is used to properly align teeth into favorable positions.[5] Oesterle stated that a prudent clinician should always attempt to use a conventional prosthesis to gather functional and esthetic information to aid in the design of the final prosthesis. This method allows for as much growth as possible before initiating the implant-assisted phase of treatment.[19,20]

As the advancements in dental implants progress, the success rates of implants are increasing and are the preferred option to restore an edentulous area. Bergendal et al. (2005) reviewed 61 patients with oligodontia whose treatment had been planned and finalized by a multidisciplinary team.[5] The case studies revealed that prosthetic restorations replaced only 42% of teeth absent due to agenesis and 66% of the restorations used were implants.[5] It should be noted that the study was done in Sweden where implant treatment for ectodermal dysplasia is covered by the government health

care system. It is reasonable to think that implants are the preferred option and is more utilized in Scandinavian countries than in the US.

Ectodermal dysplasia is a challenging condition not only to the structure and function of the child's physical being, but also to the entire psychological condition of the child and family.[18] Multi-disciplinary treatment planning teams are needed to best serve the wide range of concerns and needs for a child with ectodermal dysplasia and family.[5,18] Due to the fact that ectodermal dysplasia is a challenging condition to manage and treat, little is known about patients' attitudes toward treatment and expectations of treatment.[5] A British study of young individuals with hypodontia revealed that 40% of 451 referred young individuals had "no complaints," while only 14.6% considered "appearance" to be their most important problem.[14] A study in Hong Kong on oral health-related quality of life (OHRQol) examined individuals with severe hypodontia.[21] Twenty-five children aged 11-15 years were missing a mean of 8.9 teeth (range 4-20) and reported considerable OHRQoL impact. A majority (88%) of these children reported functional limitations and impacts on emotional well-being.[5] A similar study by Locker et al. (2010) studied OHRQoL in 36 Canadian children with hypodontia.[22] The children were missing a mean of 6.8 teeth (range 1-14), and 75% reported functional and psychosocial impacts "Often" or "Every day/almost every day."[5]

Studies show that the psychosocial benefits of early intervention are as important as the dental benefits.[23] It was reported that children with disabilities become aware of differences between themselves and other children by the age of nine.[24] This may result in a state of depression.[25] Consequently, the dentist, orthodontist or the oral and

maxillofacial surgeon are likely the first medical professionals to be confronted with complaints of oligodontia by individuals with ectodermal dysplasia.[20]

The chance that an individual with a rare disorder will meet a health professional experienced in management of that particular rare disorder is low. This may be troublesome to the individual and the family.[5] Patient organizations and support groups assume vital roles in data gathering about similar disorders and opportunities to meet similarly affected individuals are available.[5] NFED has more than 5,000 individuals in it's membership records and is the largest ectodermal dysplasia support group in the world.[7] Questionnaires have been administered through the NFED registry. The goal of the registry is to collect information that can hopefully provide data that can lead to insightful information about ectodermal dysplasia.

The purpose of this study was to evaluate the trends in dental health care for individuals with ectodermal dysplasia.

The specific aims were:

- To assess the frequency of which dental specialties originally diagnosed the individual with ectodermal dysplasia and how that has changed over time.
- To determine the changes in frequency of what dental specialties provided treatment for patients with ectodermal dysplasia and how that has changed over time.
- To assess the difficulty in locating a dental provider who would provide dental treatment and how that has changed over time.
- Evaluate when a prosthesis was first placed for a patient with ectodermal dysplasia and if that has changed over time.

## MATERIALS AND METHODS

### Sample and data collection

The NFED is a non-profit organization formed in 1981 as a support and advocacy group for individuals and families with ectodermal dysplasia. In its first years, the NFED gathered and published accurate information on the conditions and established a support network of affected families. In 1985, the Foundation expanded its mission to funding seed grants for ectodermal dysplasia research and providing researchers with access to contacting individuals with ectodermal dysplasia. In an on-going basis the NFED solicited participation from the foundation membership and referred interested individuals or their caregiver (if the individuals were too young to self report) to voluntarily participate in answering a questionnaire. In addition to contacting individuals via mail, individuals were also surveyed at annual NFED conferences.

The data in this study is based on the NFED sponsored questionnaires, from a cross-sectional survey of members beginning September 1997 through January 2000. The purpose of the questionnaire was to produce national prevalence estimates of oral health indicators and individual's experiences with the dental health care system.

Each enrolled individual completed the questionnaire that contained structured questions that were used to analyze a wide variety of content. The survey consisted of 37 items. Some questions were dichotomous questions, many questions were contingency questions with closed-ended questions, and a comment section was posted after several questions asking for further detailed information. The analytical data set contained no information on the personal identity of participants and the study was approved as

exempt by the Virginia Commonwealth University Institutional Review Board for Investigations involving Human Subjects.

*Oral Symptoms*

Oral symptoms were assessed through questions 6-13 in the questionnaire on frequent symptoms with ectodermal dysplasia; the following questions were asked:

- Chewing difficulty – Yes/No
- Speech problems – Yes/No
- Was speech therapy recommended – Yes/No
- Speech therapy obtained – Yes/No
- Sucking Habits – Yes/No……..Kind of Habit – Thumb/finger, pacifier, object
- Habit stopped – Yes/No ……...Age when habit stopped (yrs.)
- Salivary problems – Excessive, to little, no problem

*Diagnosis and Treatment*

Measures of diagnosis and treatment were examined in questions 14-20 in the questionnaire; the following questions were asked:

- Who was the first to diagnose ectodermal dysplasia: Physician, dentist, other...what type of MD, DDS or other
- Age of diagnosis (yrs.)…….type of ectodermal dysplasia (if known)
- Age (yrs.) first sought dental care
- Was it difficult finding a dentist to provide care – Yes/No
- Primary dental care is provided by: GP, pediatric dentist, prosthodontist, other

- Type of dental care provided (check all that apply): Examination, sealants, crowns (caps), preventive care (cleaning, fluoride treatments), fillings, orthodontics (braces)
- Was treatment delayed because of: Financial limitations, patient behavior, dentists reluctance to treatment, parents concerns, because of age – yes/no

Dental services and utilization were assessed through questions 21-33 on the questionnaire; the following 8 questions were asked:

- Age (yrs.) when individual received first tooth replacement appliance…….Type: Removable denture, fixed denture, implant denture
- Ages (yrs.) when dentures had to be remade or refitted
- Did individual successfully wear the denture: Upper – yes/no…Lower – yes/no
- Where were the dentures provided – private office, dental school, hospital/medical center, NIH/NIDR, other
- Does patient have implants – yes/no (if no skip to question number 34)
- Age when first implants placed……..No. of implants: upper, lower
- Did any implants fail – yes/no……....No. of implants that failed: upper, lower
- Type of denture placed over implants – Removable denture – Upper and/ or lower; Fixed denture – Upper and/or Lower
- Was implant treatment satisfactory – yes/no
- Who performed dental implant surgery – General dentist, oral surgeon, peridodontist, prosthodontist, don't know
- Where were implants placed: Dental school, private office, NIH/NIDR, SIU, UNC, UW

- Was treatment well coordinated – yes/no
- Rate your satisfaction of the implant procedures: Least 1 2 3 4 5 Most

*Costs of Treatment*

Lastly, the last 2 queries on the questionnaire pertained to cost and insurance coverage; the following questions were asked:

- Portion of bill public or private insurance covered – less than 25%, 25-49%, 50-74%, 75-99%, 100%
- Difficulty dealing with insurance – yes/no

The analysis was divided into 4 parts. First, the descriptive variables were examined. Second, the age of diagnosis was examined using linear logistic frequency and prevalence of who originally diagnosed the individual with ectodermal dysplasia and how the diagnosis of ectodermal dysplasia has changed over the decades. Using logistic regression, factors that are most strongly associated with ectodermal dysplasia and diagnosis of ectodermal dysplasia were examined. Third, the associations between age of diagnosis and the medical/dental care providing treatment were examined. Fourth, the association of age of diagnosis and dental utilization of dental services were examined. Statistical measures were calculated using SAS 9.2.[26]

## RESULTS

The data reported here reflect trends for the majority of individuals with ectodermal dysplasia in the US. In this study, the study group comprised of 469 individuals. The prevalence of ectodermal dysplasia in the study was 65% (306) in males and 35% (163) in females (Table 1). Using the date of birth of each individual, the data set was categorized over four decades creating four study groups. Out of the 469 surveys completed: 16% (75) individuals with ectodermal dysplasia were born before 1970. The frequency of response following 1970 to 1979, was 12% (54), from 1980 to 1989 was 40% (186), and between 1990 to 1999 was 33% (154).

*Clinical Conditions*

More than half of the individuals identified in each decade responded that they had difficulty chewing (Table 2). The prevalence of chewing difficulty ranged from 58% to 71%. Over the decades, 61-77% reported that speech was not a problem. Twenty-three to 39% were identified as having a speech problem. Of that 23-39%, 70-80% of those cases were referred for speech therapy. Sixty-three to 82% of individuals referred obtained speech therapy.

*First to Diagnose Ectodermal Dysplasia*

The reported prevalence of which particular caregiver diagnosed ectodermal dysplasia was relatively similar over the decades (Table 3). The three options were physician, dentist and other. The data shows that physicians are more likely to diagnose ectodermal dysplasia before a dentist or another caregiver. Before 1970, 69% of people were diagnosed by a physician, 21% by a dentist and 10% other. Between 1970-1979, 65% of the individuals were diagnosed by a physician, 29% by dentists and 6% by other.

Between 1980-1989, 53% were diagnosed by a physician, 34% by a dentist and 13% by other. Between 1990-1999, 59% were diagnosed by a physician, 29% by dentists, and 11% by other.

*Age of Diagnosis*

The age of diagnosis was the most significant between individuals born before 1970 and the group born from 1970 to 1979 (Table 4, Figure 1). The mean age of diagnosis before 1970 was 13 years while the mean age of diagnosis for the group between 1970 and 1979 was 3 years. The following decades did not show a substantial difference in age of diagnosis and maintained a young age of diagnosis between 1 and 2 years.

*Difficulty Finding a Dentist to Provide Care*

The questionnaire results indicated that no significant changes have taken place over the decades with regards to finding a dentist to provide care (Table 3a). Twenty-nine to 33% of the study group had difficulty in finding a dentist to provide care.

*Was Treatment Delayed*

The highest prevalence in why treatment was delayed was due to financial reasons for the first three decades of the study (Table 3a). The exception was 1990-1999, when 40% experienced delayed treatment because the dentist was reluctant to treat.

*Age when received first tooth prosthesis*

The age variable was categorized into several ranges to evaluate the change of age when an individual would receive their first tooth prosthesis (Table 3b, Figure 2). Before 1970, a high percentage of individuals received a prosthesis during the teenage years or later. The information gathered shows that children are receiving a dental prosthesis at

an earlier age. The prominent group is between 4-6 years of age, followed by 0-3 year-old age group.

*Successfulness wearing a denture*

The prevalence of successfully wearing a denture maintained a high frequency over the decades for both upper and lower prostheses (Table 3b). The rate of those individuals successfully wearing a maxillary denture ranged from 83% to 98%. The rate of those individual successfully wearing a mandibular denture distribution was 74% to 85%.

*Dentures Provided*

Regarding the location where dentures were provided, the results indicated that the location selected most often in the questionnaire was private practice (55-66%). Dental schools were the next highest (23-29%) group. While academic medical centers were one of the least utilized locations prior to 1970 (2%). They gradually increased to 21% by the late 1990s.

## DISCUSSION

This questionnaire gives insight into changes in ectodermal dysplasia over time. In past years, the chance that individuals with ectodermal dysplasia would meet a health professional with experience of their diagnosis is low.[5] In recent years, non-profit organizations, such as the NFED and support groups have been established in many countries to provide individuals and families with the same diagnosis the opportunity to meet. Family meetings in such organizations provide unique opportunities to share experiences on symptoms, treatment, and strategies for mastering everyday life.[5] The amount of continuing education courses focusing on the recognition and treatment of ectodermal dysplasia and the other craniofacial defects has increased greatly and treatment of ectodermal dysplasia has become more of an issue in the dental profession.

Children who are missing permanent teeth are affected physically and psychosocially to varying degrees and present a challenge to both medical and dental professionals. Most of the older literature about ectodermal dysplasia lacks high quality studies and is limited to case reports with low levels of scientific evidence.[5]

The oral functions of chewing and speech continue to be a challenge for individuals with ectodermal dysplasia. The results of the study show that individuals do have a predisposition to having chewing difficulties as well as speech difficulties. A high percentage of the population with speech difficulties (23-39%) were recommended for speech therapy and a high percentage of people obtained speech therapy (63-83%) (Table 2).

Oral rehabilitation provides improvements in esthetics, speech and masticatory efficiency. Ramos (1995) demonstrated from case studies that early fabrication of

dentures can lead to significant improvements in appearance, speech and masticatory function.[27] Recent clinical studies report fabrication of prostheses as early as three years old if cooperative behavior exists. After observing the large number of individuals with speech difficulties and those who required treatment; it would be advantageous to inquire about speech difficulty before dental treatment and what, if any, improvements occurred after use of a prosthesis.

Nutrition is also a challenge for individuals with ectodermal dysplasia patients. Although dentures are poor alternatives to a healthy dentition, they create conditions for the maintenance of a normal, satisfactory daily diet for individuals with ectodermal dysplasia. This is extremely important, since the establishment of lifelong dietary patterns occurs during childhood.[28]

Since the discovery and heightened awareness of ectodermal dysplasia, as defined by Freire-Maia in 1971, ectodermal dysplasia has encompassed a much larger group than the initial groups first discovered.[6] Historically, little was known about ectodermal dysplasia and how to develop the classification schemes. Modern molecular genetics has increasingly identified the basic defects of the different syndromes and yield more insight into the regulatory mechanisms of embryology. Due to the many forms of ectodermal dysplasia and because some forms may not be diagnosed at birth, the incidence of all forms is undoubtedly higher than was previously reported.[10]

Diagnosis is the first step towards improving the situation for an individual affected with ectodermal dysplasia. Ectodermal dysplasia is now being recognized by all medical and dental specialties early in life. Early recognition of ectodermal dysplasia

allows the families to locate a multi-disciplinary team sooner and in turn will help cover the issues of diagnosis and treatment.

It is critical that physicians and dentists recognize the condition early and direct the patient to the proper health care team. The questionnaire indicates that physician visits are important and may lead to an early diagnosis. Since 1992 and the establishment of the medical home, any child with ectodermal dysplasia characteristics should be recognized earlier by a physician who is capable of managing the condition.[29] In addition to recognizing the importance of a medical home, dentistry developed the concept of a dental home but it was not adopted until after the questionnaire was administered.[30] Both the medical home and dental home concepts may lead to earlier diagnoses.

After 1970, the results indicated that diagnosis of ectodermal dysplasia occurred at a mean age of 2 years (Table 4, Figure 1). The data does have outliers that can skew the data. One individual in the 1970s was diagnosed at 80 years of age. Older generations answering the questionnaire subjects the research to possible recall bias. Self-reporting of data is less accurate than data collection by observation or by dental record abstraction, which potentially limits the usefulness of these data.

Further education on diagnostics and treatment of ectodermal dysplasia has shown improvement for both the medical and dental professions. In addition, databases with up-dated information of rare disorders provide information to health professionals and affected individuals.

Individuals with ectodermal dysplasias continue to have difficulty finding a dentist to provide care. The questionnaire response suggest that individuals have maintained the same difficulty throughout the decades (Table 3a). The prevalence of 29-

33% is significant considering that the 2001 National Survey of Children with Special Health Care Needs found that only approximately 8% of parents reported that their child had unmet dental needs in the past year, which is less than 3 times the rate found in our sample.[31]

*Was Treatment Delayed*

The highest prevalence in why treatment was delayed in the early decades was due to financial reasons for the first three decades of the study. The exception was 1990-1999. In the 1990s the number one reason why dental treatment was delayed was due to the dentist's reluctance to treat. The results of the questionnaire do not give a definitive answer as to why the change occurred, however, in the early decades of the survey removable prostheses were the common standard of treatment for tooth replacement for the general population including individuals with ectodermal dysplasia and were provided mostly for older teens and adults. Currently, the treatment standard in the management of ectodermal dysplasia is the use of prostheses for treatment in young children. Many dentists prefer not to have young children in their practices, especially when the needed treatment is not the usual care for that age group. Also, in some state insurance programs, coverage for the necessary dental treatment for individuals with ectodermal dysplasia now provides coverage for children that may not be available for adults. This has resulted in an increased demand for early treatment.

Before 1970, little information on medical and dental care was available for health care professionals about the placement of removable prostheses. The data from our sample shows that over several generations people are receiving their first removable prosthesis at a younger age and dentures are well accepted at all ages (Table 3b, Figure

2).

Individuals with ectodermal dysplasia are receiving the majority of their denture treatment in private dental practices followed by dental schools. Due to the high frequency of missing responses, it is difficult to make any conclusions concerning preference changes between the different locations. Over the decades, there has been a slight increase in the use of medical centers (Table 3b). Individuals completing the survey may or may not have understood the difference between a medical center, dental program or school associated with a medical center or a freestanding dental school. The NFED is involved in giving financial aid and sponsoring families to receive dental treatment through selected NFED Treatment Centers that may be located in medical centers, dental schools or community teams.

When medical and dental interventions improve the appearance and function of a patient with congenital and craniofacial defects, this can have a profound effect on the individual's happiness and productivity.[32] Prosthetic treatment allows individuals with ectodermal dysplasia to have better self esteem, more opportunities to fulfill their potential socially, and improved employment possibilities.[32] If a young child is cooperative it is beneficial to initiate a dental prosthesis early to allow for normal mastication and function of the oral cavity.

Differential diagnosis is a key prerequisite to planning treatment for individuals affected by ectodermal dysplasia, ectodermal dysplasia syndromes and related disorders. The cause of the observed anomaly and its potential for associated health problems must be addressed before deciding upon a course of action. If an individual has several missing teeth, for example, one should know whether their absence represents a dysplasia

(hypodontia) or is the consequence of advanced periodontal disease. When treatment is planned, the impact of associated health problems must be anticipated. It is also imperative to know that if other health issues exist, such as skin erosion or immune deficiencies, this will complicate treatment.[3]

In addition to completely understanding the diagnosis of the specific ectodermal dysplasia condition and the associated signs and symptoms involved, one should also assess the psychological effects of these deformities. It would be advantageous to measure the change in psychological and psychosocial impacts before and after insertion of the prosthesis.

## CONCLUSION

Valuable information regarding many issues in the understanding and management of the ectodermal dysplasias has been obtained from the evidence presented in this project. It has provided insight into the positive progression and improved treatments for individuals with ectodermal dysplasia.

Cooperation between medical and dental specialists is important so that dental care is initiated in a timely fashion for individuals with ectodermal dysplasia. This is to assure early initial treatment, ongoing treatment during growth and complex treatment needed during the life span. The increasing financial cost of care is still a barrier to individuals with ectodermal dysplasia who are receiving optimal lifetime oral health care. The NFED questionnaire has helped explain the dental challenges that lie ahead for improving treatment needs for individuals with ectodermal dysplasia and their families.

From this study we can deduce the following conclusions/findings:

- The age of diagnosis is occurring earlier in life than previous decades.
- Physicians are the primary source of the first diagnosis of the ectodermal dysplasia.
- The first time use of a dental prosthesis is being initiated earlier in life.
- Dentures are well accepted in all age groups.
- Individuals with ectodermal dysplasia expressed chewing and speech difficulties over the decades.
- Parents and caregivers may seek early dental treatment to help remedy speech and chewing problems.
- Implant prosthetic care is being initiated in the early teenage years.

- The use of medical centers is increasing as the initial source of dental care for individuals with ectodermal dysplasias.
- Access to dental care for individuals with ectodermal dysplasias is a continuing problem with a third of survey respondents listing this as an issue.

**TABLES**

Table 1: Demographic Characteristics

| ***Gender*** | n | % |
|---|---|---|
| *Female* | 163 | 35 |
| *Male* | 306 | 65 |
| ***Generation*** | n | % |
| *< 1970* | 75 | 16 |
| *1970 - 1979* | 54 | 12 |
| *1980 - 1989* | 186 | 40 |
| *1990 - 1999* | 154 | 33 |

Table 2: Clinical Conditions Reported

| *Variable* | *Generations* | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | *<1970* | | *1970-1979* | | *1980-1989* | | *1990-1999* | |
| | *n* | *%* | *n* | *%* | *n* | *%* | *n* | *%* |
| **Chewing difficulty** | | | | | | | | |
| *No* | 30 | 42 | 15 | 29 | 62 | 34 | 47 | 32 |
| *Yes* | 42 | 58 | 37 | 71 | 118 | 66 | 100 | 68 |
| *Frequency missing* | 3 | | 2 | | 6 | | 7 | |
| **Speech problems** | | | | | | | | |
| *No* | 56 | 77 | 35 | 65 | 113 | 62 | 92 | 61 |
| *Yes* | 17 | 23 | 19 | 35 | 70 | 38 | 59 | 39 |
| *Frequency missing* | 2 | | 0 | | 3 | | 3 | |
| **Speech therapy recommended** | | | | | | | | |
| *No* | 4 | 25 | 6 | 30 | 9 | 12 | 11 | 18 |
| *Yes* | 12 | 75 | 14 | 70 | 68 | 88 | 51 | 82 |
| *Frequency missing* | 59 | | 34 | | 109 | | 92 | |
| **Speech therapy obtained** | | | | | | | | |
| *No* | 6 | 38 | 6 | 29 | 14 | 18 | 15 | 25 |
| *Yes* | 10 | 63 | 15 | 71 | 62 | 82 | 46 | 75 |
| *Frequency missing* | 59 | | 33 | | 110 | | 93 | |
| **Sucking habits** | | | | | | | | |
| *No* | 52 | 79 | 31 | 63 | 99 | 54 | 84 | 56 |
| *Yes* | 14 | 21 | 18 | 37 | 83 | 46 | 67 | 44 |
| *Frequency missing* | 9 | | 5 | | 4 | | 3 | |

Table 3a: Dental Experiences Part I

| Variable | Generations | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | <1970 | | 1970-1979 | | 1980-1989 | | 1990-1999 | |
| | n | % | n | % | n | % | n | % |
| **Who first diagnosed ectodermal dysplasia?** | | | | | | | | |
| *Dentist* | 15 | 21 | 15 | 29 | 61 | 34 | 44 | 29 |
| *Physician* | 48 | 69 | 34 | 65 | 96 | 53 | 89 | 59 |
| *Other* | 7 | 10 | 3 | 6 | 24 | 13 | 17 | 11 |
| *Frequency missing* | 5 | | 2 | | 5 | | 4 | |
| **Was it difficult finding a DDS to provide care?** | | | | | | | | |
| *No* | 45 | 67 | 36 | 71 | 123 | 69 | 100 | 74 |
| *Yes* | 22 | 33 | 15 | 29 | 54 | 31 | 35 | 26 |
| *Frequency missing* | 8 | | 3 | | 9 | | 19 | |
| **Was treatment delayed because of:** | | | | | | | | |
| *Financial Reasons* | 21 | 66 | 20 | 61 | 37 | 43 | 28 | 31 |
| *Parental Concerns* | 2 | 6 | 3 | 9 | 3 | 3 | 5 | 6 |
| *Patient Behavior* | 1 | 3 | | | 12 | 14 | 20 | 22 |
| *Dentist's Reluctance to Treat* | 8 | 25 | 10 | 30 | 34 | 40 | 36 | 40 |
| *Frequency missing* | 43 | | 21 | | 100 | | 65 | |

Table 3b: Dental Experiences Part II

| Variable | Generations | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | <1970 | | 1970-1979 | | 1980-1989 | | 1990-1999 | |
| | n | % | n | % | n | % | n | % |
| **Age when received first tooth prosthesis:** | | | | | | | | |
| *0-3 yrs* | 6 | 10 | 12 | 26 | 39 | 33 | 25 | 45 |
| *4-6 yrs* | 19 | 33 | 15 | 33 | 51 | 43 | 30 | 54 |
| *7-10 yrs* | 12 | 21 | 3 | 7 | 18 | 15 | | |
| *11+ yrs* | 21 | 36 | 16 | 35 | 10 | 8 | 1 | 2 |
| *Frequency missing* | 17 | | 8 | | 68 | | 98 | |
| **Successfully wear the denture?** | | | | | | | | |
| *Unsuccessful Upper Dent* | 1 | 2 | 3 | 7 | 19 | 17 | 9 | 17 |
| *Successful Upper Dent* | 50 | 98 | 39 | 93 | 92 | 83 | 43 | 83 |
| *Frequency missing* | 24 | | 12 | | 75 | | 102 | |
| *Unsuccessful Lower Dent* | 6 | 15 | 6 | 16 | 23 | 26 | 7 | 18 |
| *Successful Lower Dent* | 35 | 85 | 32 | 84 | 65 | 74 | 31 | 82 |
| *Frequency missing* | 34 | | 16 | | 98 | | 16 | |
| **Where were the dentures provided?** | | | | | | | | |
| *Dental School* | 17 | 29 | 11 | 27 | 28 | 26 | 12 | 23 |
| *Medical Center* | 2 | 3 | 4 | 10 | 15 | 14 | 11 | 21 |
| *NIH/NIDR* | 1 | 2 | 2 | 5 | 2 | 2 | 1 | 2 |
| *Private Office* | 38 | 66 | 24 | 59 | 64 | 59 | 29 | 55 |
| *Other* | | | | | | | | |
| *Frequency missing* | 17 | | 13 | | 77 | | 101 | |
| **Does patient have implants?** | | | | | | | | |
| *No* | 60 | 88 | 33 | 63 | 166 | 92 | 133 | 99 |
| *Yes* | 8 | 12 | 19 | 37 | 15 | 8 | 2 | 1 |
| *Frequency missing* | 7 | | 2 | | 5 | | 19 | |
| **Did any implants fail?** | | | | | | | | |
| *No* | 6 | 86 | 14 | 82 | 12 | 86 | 2 | 100 |
| *Yes* | 1 | 14 | 3 | 18 | 2 | 14 | | |
| *Frequency missing* | 68 | | 37 | | 172 | | 152 | |

Table 4: Treatment Descriptives

| ***Generation Prior to 1970*** | n | Mean | Std Dev | Minimum | Maximum |
|---|---|---|---|---|---|
| *Age of Diagnosis* | 65 | 13 | 17 | 0 | 80 |
| *Age First Placed* | 8 | 39 | 15 | 21 | 65 |
| *Impl Mand* | 5 | 4 | 2 | 1 | 5 |
| *Impl Max* | 4 | 3 | 2 | 0 | 4 |
| ***Generation 1970-1979*** | n | Mean | Std Dev | Minimum | Maximum |
| *Age of Diagnosis* | 53 | 3 | 4 | 0 | 16 |
| *Age First Placed* | 19 | 16 | 4 | 6 | 26 |
| *Impl Mand* | 18 | 4 | 1 | 1 | 5 |
| *Impl Max* | 8 | 1 | 2 | 0 | 4 |
| ***Generation 1980-1989*** | n | Mean | Std Dev | Minimum | Maximum |
| *Age of Diagnosis* | 171 | 2 | 3 | 0 | 12 |
| *Age First Placed* | 15 | 11 | 4 | 5 | 17 |
| *Impl Mand* | 15 | 4 | 1 | 0 | 5 |
| *Impl Max* | 9 | 1 | 2 | 0 | 4 |
| ***Generation 1990-1999*** | n | Mean | Std Dev | Minimum | Maximum |
| *Age of Diagnosis* | 148 | 2 | 2 | 0 | 18 |
| *Age First Placed* | 2 | 15 | 6 | 10 | 19 |
| *Impl Mand* | 2 | 5 | 1 | 4 | 5 |
| *Impl Max* | 0 | - | - | - | 0 |

**FIGURES**

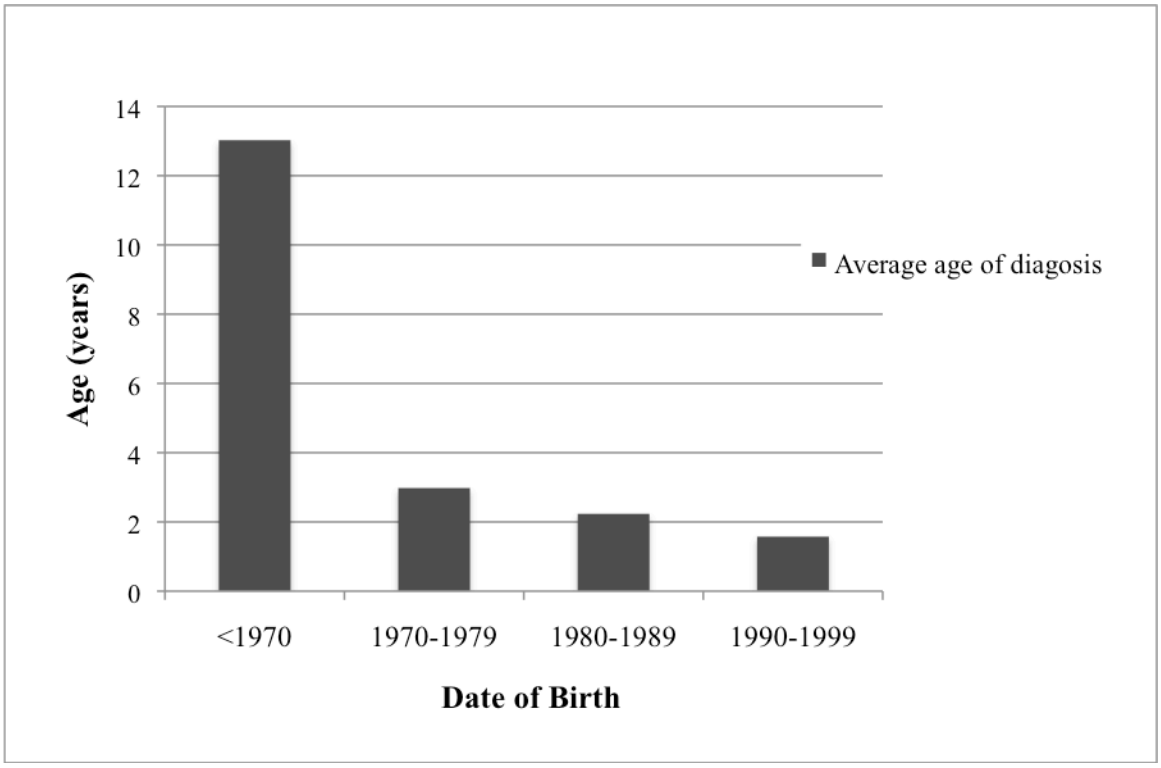


Figure 1: Average Age of Diagnosis

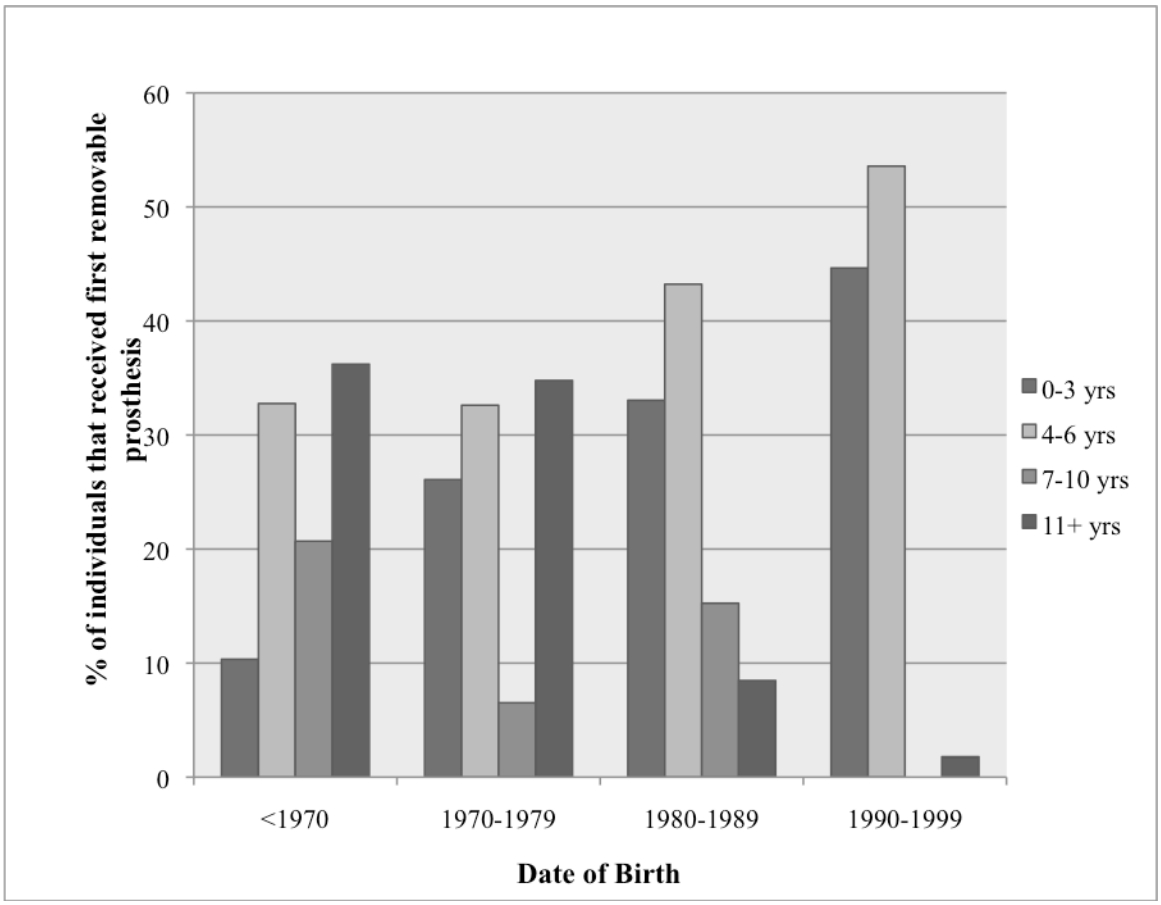


Figure 2: Changes in Age of First Dental Prosthesis

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## VITA

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