EHLERS-DANLOS SYNDROME: CURRENT MEDICAL PERSPECTIVE

A RESEARCH PAPER

SUBMITTED TO THE GRADUATE SCHOOL

IN PARTIAL FULFILLMENT OF THE REQUIREMENTS

FOR THE DEGREE

MASTER OF ARTS

BY

AMANI IBRAHIMKHAN

DR. C. ANN BLAKEY, ADVISOR

BALL STATE UNIVERSITY

MUNCIE, INDIANA

DECEMBER 2015
ABSTRACT

RESEARCH PAPER: Ehlers-Danlos Syndrome: Current Medical Perspective

STUDENT: Amani Mohammedwali Ibrahimkhan

DEGREE: Masters of Arts

COLLEGE: Science and Humanities

DATE: December 2015

PAGES: 39

Ehlers-Danlos Syndrome is a disorder of a fault in the collagen tissue structure and production. It is a connective tissue disorder that is inherited in many manifestations that are grouped into the many types of the EDS. The difference in the presentations of the disorder is brought about by mutations in the collagen alpha-1 gene and the collagen alpha-2 gene. Different transcripts have been determined for this gene that presented this mutation. Collagen tissue is produced before tougher tissues, which is made by fibroblasts. It helps the body tissues to fight deformation, and is a very significant help in providing strength and resistance to pressure for the skin, muscle, ligament, and blood passage. The patient of EDS is time and again confused with depression and fatigue. In the primary view, EDS is a poorly known disorder among many medical personnel. There is no known cure for EDS. Raising awareness within the medical community is the most therapeutic means to deal with the Ehlers-Danlos Syndrome so far; this study is a review of the current medical perspective on Ehlers-Danlos Syndrome.
DEDICATION

To my parents whom I owe this achievement without their encouragement and continuous support this paper would not have been realized. To my husband, Mohammad Abdullah Doom who suffered from my being away from home most of my stay at school, and to my son Ajwad and my daughter Deema, who have suffered from my inattention during my study and writing this research. Without Allah’s blessing and their patience and assistance in creating an environment conducive for my reading, writing, and travel, getting this paper would not have been possible.
ACKNOWLEDGEMENTS

My immeasurable thanks to Allah for giving me all strength, health, patience, and courage to finish this paper.

My prayers go to my parents I ask Allah to reward both of them with the highest degree in heaven.

My sincere appreciation goes to all those who made my endeavor successful despite any difficulties I faced in my personal life and health.

Special thanks go to my Father who has been staying with me, mentoring me and caring of my children. Not to forgetting my beloved mother who has been following up on my progress; my sister Tahani, who has been editing my work as I used to go along, without them I would not have been able to finish.

I express my Deepest appreciation and great of thank to my Mentor and Supervisor of this research : Dr. Blakey
# TABLE OF CONTENTS

**ABSTRACT** ........................................................................................................................................... i

**DEDICATION** ......................................................................................................................................... ii

**ACKNOWLEDGEMENTS** .......................................................................................................................... iii

**TABLE OF CONTENTS** ............................................................................................................................... iv

**TABLE OF TABLES** ................................................................................................................................... vii

**CHAPTER ONE** ...................................................................................................................................... 1

   *Introduction* ........................................................................................................................................... 1

**CHAPTER TWO** .................................................................................................................................... 3

   *Research Methods* ................................................................................................................................. 3

**CHAPTER THREE** ................................................................................................................................. 4

   *Results* ................................................................................................................................................... 4

      **I. Information Sources for Data Analysis** ......................................................................................... 4

         A. *Research Sources* ......................................................................................................................... 4

         B. *Types of Ehlers-Danlos Syndrome* .............................................................................................. 5

            1. *Classical EDS* .......................................................................................................................... 5

            2. *Hypermobility EDS* ................................................................................................................ 6

            3. *Vascular EDS* ........................................................................................................................ 6

            4. *Kyphoscoliosis EDS* ................................................................................................................. 7

            5. *Arthrochalasia EDS* .................................................................................................................. 7

            6. *Dermatosparaxis EDS* ............................................................................................................... 7

            7. *Other Forms of EDS* ................................................................................................................. 8

      **II. Interview Data Analysis** ............................................................................................................. 8

**CHAPTER FOUR** ................................................................................................................................. 10

   *Discussion* .......................................................................................................................................... 10

      **I. Information Sources** ................................................................................................................... 10

         A. *Research Sources* ....................................................................................................................... 10

         B. *Types of Ehlers-Danlos Syndrome* ............................................................................................ 10

      **II. Comments on Interviews** .......................................................................................................... 11

      **III. Summary of Knowledge on EDS** ........................................................................................... 12

         A. *Causes of the Ehlers-Danlos Syndrome* ...................................................................................... 12

         B. *What are the signs and symptoms of Ehlers-Danlos Syndrome?* ........................................... 13

         C. *People Living with Ehlers-Danlos Syndrome* .......................................................................... 14

         D. *Diagnosis of Ehlers-Danlos Syndrome* ...................................................................................... 17

         E. *Management of Ehlers-Danlos Syndrome* ................................................................................. 18

**CHAPTER FIVE** .................................................................................................................................... 21
TABLE OF TABLES

Table 1: Key websites for information on Ehlers-Danlos Syndrome.......................... 4

Table 2: Six major types of Ehlers-Danlos Syndrome based on 1997 re-classifications. 5

Table 3: The effects of Ehlers-Danlos Syndrome (EDS) on patients diagnoses. .......... 9
CHAPTER ONE

Introduction

Ehlers-Danlos Syndrome refers to a group of numerous disorders, adding up to about twenty to twenty six, that are differently inherited with an adverse effect which weakens connective tissues. It is similar to a rare disorder referred to as Cutis Laxa. Ehlers-Danlose Syndrome is abbreviated as EDS. They all involve a defect of the genetic arrangement in the collagen protein sequence and the synthesis of the connective tissue, as well as the connective structures (OMIM 130000, 2015).

One may have concerns over the cause of the EDS. The syndrome is clinically diverse with the major variations occurring by virtue of the differences in abnormalities of collagen lying beneath its occurrence. EDS symptoms usually include loose joints, soft and stretchy skin that easily get bruises, fragile and small blood passages, and poor wound healing after abnormal scar formation. People with the syndrome usually have excessively flexible joints and fragile skin (Ensor, 2013; Mayo Clinic Staff, 2012). In cases where stitches are required due to lacerations of the skin or injuries, the skin cannot hold the stitches. It is very important to be on familiar terms with the clinical, scientific, and medical aspects of the EDS when one is dealing with or is suffering from the disorder.

This work is a detailed review of past research as well as a perspective of current research to identify the arguments and the support regarding EDS. An examination of
different causal agents of the disorder and analysis of the problems associated with possible suggestions of cures and solutions will be discussed. This research will give details on the exploration and results about the developments made on the disorder so far. This research includes the following: historical information, who is the affected, where is the effect experienced, previous research and current research as well as the mechanisms of the disorder (Ensor, 2013). The research was developed to serve as a teaching tool to improve learner knowledge on how to help individuals who suffer from the Ehlers-Danlos Syndrome. This report provides details on the mechanism of the disorder and the therapies at the molecular level of how they work. Readers will receive the basic technical know-how that will enable them deal with the disorder without posing a danger or any form of harm to the affected individual suffering from the syndrome.
CHAPTER TWO

Research Methods

The research involved several methods of gathering information. There were many ways of gathering information at our disposal. This research included both primary and secondary sources of information. The use of various options and alternatives allowed for the best possible ways of getting information about Ehlers-Danlos Syndrome. The majority of previous methods relied heavily on literature searches or utilization of surveys similar to those offered by websites such as StatPac (2014) or SurveyMonkey (2015) from medical websites dealing with the syndrome. Expansion of the research with additional information was obtained from direct interviews with medical doctors. Through interviews with two doctors, information was gathered on the medical profession and provided a good way of acquiring information about the crucial stages of the research and treatment. These interviews also allowed for a review of medical practice information that is not typically available to the public or in other published materials.
CHAPTER THREE

Results

From all the sources of information used and collected, and from the interview data analyzed, the following results were compiled.

I. Information Sources for Data Analysis

A. Research Sources

Information acquired from four key websites were determined as recommended after close scrutiny of different related websites. These websites include: the National Center for Biotechnology Information (NCBI), the Online Mendelian Inheritance in Man (OMIM), PubMed, and PubMed Central (PMC) (see Table 1). From the most recent publications on these websites, information was obtained on over twenty different types and subtypes of the disorder.

Table 1: Key websites for information on Ehlers-Danlos Syndrome.

<table>
<thead>
<tr>
<th>Website Name</th>
<th>URL</th>
</tr>
</thead>
<tbody>
<tr>
<td>OMIM</td>
<td><a href="http://www.omim.org">http://www.omim.org</a></td>
</tr>
<tr>
<td>ORPHA</td>
<td><a href="http://www.orpha.net">http://www.orpha.net</a></td>
</tr>
<tr>
<td>PubMed</td>
<td><a href="http://www.pubmed.gov">http://www.pubmed.gov</a></td>
</tr>
<tr>
<td>PubMed Central</td>
<td><a href="http://www.ncbi.nlm.nih.gov/pmc/">http://www.ncbi.nlm.nih.gov/pmc/</a></td>
</tr>
</tbody>
</table>
B. Types of Ehlers-Danlos Syndrome

Information on each of the major types of EDS and any subtypes were easily found and summarized (see Table 2). The official MIM (Online Mendelian Inheritance in Man) accession number as well as the ORPHA.net number was determined for each of the major types information regards the number of subtypes.

Table 2: Six major types of Ehlers-Danlos Syndrome based on 1997 reclassifications.

<table>
<thead>
<tr>
<th>Types of EDS</th>
<th>Type Number/or Form</th>
<th>Number of Sub-types</th>
</tr>
</thead>
<tbody>
<tr>
<td>Classical</td>
<td>Type I &amp;II</td>
<td>0</td>
</tr>
<tr>
<td>Hypermobility</td>
<td>Type III</td>
<td>0</td>
</tr>
<tr>
<td>Vascular</td>
<td>Type IV</td>
<td>0</td>
</tr>
<tr>
<td>Kyphoscoliosis</td>
<td>Type VI</td>
<td>0</td>
</tr>
<tr>
<td>Arthrochalasia</td>
<td>Type VII-A&amp;B</td>
<td>2 (sub-types of VII)</td>
</tr>
<tr>
<td>Dermatosparaxis</td>
<td>Type VII-C</td>
<td>1 (sub-types of VII)</td>
</tr>
<tr>
<td>Other Forms</td>
<td>Types V, VIII, X; Form: Progeroid, Beasley-Cohen</td>
<td>10</td>
</tr>
</tbody>
</table>

1. Classical EDS

The classical type of EDS is named Types I (MIM#130000, ORPHA#90309) & II (MIM#130010, ORPHA#90318). The expected frequency of this type in the general population is approximately 1 in 50,000 individuals (Genetics Home Reference, 2015). This form of EDS can be inherited through dominant or recessive autosomal genes. Type I severely involves the skin in its manifestation. Type II, skin involvements are varied from mild at times to moderate in other patients. Patients may suffer from joint
dislocations from time to time, with or at times without trauma (Genetics Home Reference, 2015). Pain is a common symptom of this type of EDS.

2. Hypermobility EDS

HyperMobility EDS is Type III (MIM#130020, ORPHA#285) of the six major EDS types. The expected frequency of this type in the general population is approximately 1 in 15,000 individuals. This form of EDS can be inherited through dominant or recessive autosomal genes. It is less noticeable manifestations in the skin, but causes pains in the muscles and the skeleton (Genetics Home Reference, 2015). Affected individuals often suffer from cases of dislocation of the joints.

3. Vascular EDS

Vascular EDS is the Type IV (MIM#130050, ORPHA#286) form. It is inherited as autosomal dominant gene. It affects the production of type-III collagen. The expected frequency of this type in the general population is approximately 1 in 500,000 individuals (Genetics Home Reference, 2015). It is regarded as one of the harsher and most precarious forms of EDS. It can result to the rupture of blood vessels, intestines or the womb in females (Mayo Clinic Staff, 2012). Blood passage organs and the uterus in females are prone to rupture in Vascular EDS.

Many patients will show symptoms primarily through the facial appearance. According to the research reports at Genetic Home Reference (2015), they interviewed patients and observed a characteristic thin nose, outsized eyes, hollow cheeks, small chin and ears without lobes while the lips appeared thin. They also commented that the skin has unusually light, and the body slim with veins clear over the chest and the abdomen.
The doctors in their telephone survey confirmed that at least twenty percent of the individuals with EDS suffered serious health complications by the time they reached twenty years of age (Genetic Home Reference, 2015).

4. Kyphoscoliosis EDS

The Type VI (MIM# 225400, ORPHA# 1900) form is Kyphoscoliosis EDS. It is caused by an autosomal recessive defect. Specifically, the defect is due to a lack of sufficient lysylhydroxylase (PLOD) enzyme (EDNF, 2015; Genetics Home Reference, 2015). It is a rare type, and only very few cases have been reported. It has a characteristic curving of the spine, a condition referred to as scoliosis. The eyes have a thin conjunctiva, and the patient suffers from a severe weakness of the muscles.

5. Arthrochalasia EDS

Arthrochalasia EDS is Type VII-A (MIM# 130060, ORPHA# 99875) and VII-B (MIM# 130060, ORPHA# 99876) are the first two classifications of Type VII. It is an inherited as a dominant autosomal gene and it is not as common relative to any other types, rather it is a very rare species of the types of EDS (Genetics Home Reference, 2015). Both Type VII-A & VII-B involve mutations with the same exon of thee collagen Type-I protein (Klaassen et al, 2012). It causes twice as dangerous the effects relative to those seen in the Hypermobility type. In this case, the joints are extremely loose and dislocations occur much more frequently.

6. Dermatosparaxis EDS

Dermatosparaxis EDS is an autosomal recessive defect. It is Type VII-C (MIM# 225410, ORPHA# 1901) in the current classification system. It is very rare, and
almost no new cases have been reported since 2013 (Solomons et al, 2013). The most common characteristic is the extremity of the fragile and sagging skin in the patients (Genetics Home Reference, 2015). Here, the doctors revealed that the condition is more prevalent than previously indicated by the ratios, since the most recent clinic experiences show the condition is being under-diagnosed.

7. Other Forms of EDS

Recently, there are many more types of EDS that have been diagnosed. These forms present with soft skin that is mildly stretchable, short bones, and joint dislocation accompanied by extended bouts of persistent diarrhea (Genetics Home Reference, 2015). These patients have been seen with symptoms of rupture of the bladder, poor healing of wounds, and joint hypermobility. The patterns of inheritance observed are autosomal recessive and dominant, as well as X-linked recessive forms (Ensor, 2013). These forms include Type V (MIM#305200, ORPHA#75497), Type VIII (MIM#130080, ORPHA#75392), Type X (MIM#225310, ORPHA#75501), as well as the Progeroid type and the Beasley – Cohen forms of EDS.

II. Interview Data Analysis

Two practitioner Dermatologists were selected and interviewed. Drs. Maha Sulaiman and Talib Al-Abbadi of the Saudi Arabian Airline’s Medical Center, Jeddah, Saudi Arabia were interviewed and were posed questions relating to Ehlers-Danlos Syndrome (EDS). Different questions were posed to each one of them (see Appendix A), with some notable similarities with regards to the symptoms and terminology used by
both medical physicians (see Appendix B). Key diagnostic similarities of the interviews were evaluated and presented in Table 3.

Table 3: The effects of Ehlers-Danlos Syndrome (EDS) on patient diagnoses.

<table>
<thead>
<tr>
<th>Maha Sulaiman, M.D.</th>
<th>Talib Al-Abbadi, M.D.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Major Symptoms:</strong></td>
<td></td>
</tr>
</tbody>
</table>
| Involve a defect of the genetic arrangement of the collagen.  
  • The synthesis of the connective tissue as well as the connective structures.  
  • The syndrome is clinically diverse with the major variations occurring by virtue of the difference in the abnormality of the collagen lying beneath its occurrence. | The signs go around the defect or lack of sufficient levels of collagen in the body. |
| **Tissues involved:** |                      |
| Affect blood vessels, joints and the skin in most cases.  
  • The skin cannot hold stitches in cases where they are required due to skin problems. | It will affect the joints, the blood vessels, and the skin. |
| **Hyperflexibility issues:** |                      |
| People with the syndrome usually have excessively flexible joints and a fragile skin. | The joints are hyper flexible and so unstable that they are easily vulnerable to joint dislocation and prone to sprain.  
  • The patient may be affected by the chronic disease of the joints characterized by a progressive deterioration and loss of function. |
CHAPTER FOUR

Discussion

I. Information Sources

A. Research Sources

The key websites (see Table 1) served as an excellent starting point for information gathering. From these sites, additional sources in the form of articles and websites were easily found for further exploration of EDS. These key sources allow researchers to focus or narrow their searchers to specific types of EDS and their treatments.

B. Types of Ehlers-Danlos Syndrome

In the past, there were only a few known types of the syndrome, approximately ten. Three years shy of the new millennium, in the year 1997, the ten known types were reclassified and brought down to only six major descriptively named forms of EDS (Genetics Home Reference, 2015) (see Table 2). The names of these major types of EDS are 1) Classic type, 2) Hypermobility EDS, 3) Vascular type, 4) Kyphoscoliosis type, 5) Arthrochalasia type, and 6) Dermatosparaxis type. Hypermobility EDS is characterized by an unusual range of joint movement. The most dangerous form is the Vascular type. EDS forms such as the Kyphoscoliosis and Arthrochalasia types can engross severe difficulties that are potential threats to the life of the sufferer. Additionally, the fact that
the EDS of Kyphoscoliosis type may cause breathing problems and difficulties due to its significant contribution to a serious progressive curving of the spine (Genetics Home Reference, 2015). Dermatosparaxis type, shows a skin that sags and there is the presence of wrinkles on the skin surface (Ensor, 2013). If one is affected when still young, the chances are great that their skin will fold as they get older. There are many other forms of EDS causing various conditions of suffering to the patients. In this study, individual were found to be affected in the joints and the skin, and the effects vary with the individuals and the type of EDS. Affected persons have the propensity to bruise easily to any exposure to hazards or impact to the body.

II. Comments on Interviews

Two practitioner Dermatologists were selected and interviewed (see Appendix A). Dr. Maha Sulaiman is a Dermatologist in the Saudi Arabian Airline’s Medical Center, in Jeddah Saudi Arabia. Dr. Talib Al-Abbadi is also a Dermatologist in Saudi Arabian Airline’s Medical Center, in Jeddah Saudi Arabia. They were interviewed and were posed questions relating to: Ehlers-Danlos Syndrome (EDS). Different questions were posed to each one of them, but both practitioners used a similar set of criteria and symptoms in their approach to diagnoses. The overall outcome provided important insights on the medical perspectives on EDS.

Dr. Talib Al-Abbadi emphasized the importance of knowledge regarding the genetics of the various types of EDS. In particular, his genetic knowledge level was extensive and he clearly understood that mutations in different genes could cause the disorder to manifest in many different ways. He recognized that genetic transmission
could involve autosomal dominant, autosomal recessive, and X-linked forms, and that the latter form is extremely rare. He noted that EDS is primarily considered as an autosomal dominant disorder in most cases, but carriers of the recessive forms do exist. In the case of carriers, these individuals have the greatest possibility of passing the characteristics to their offspring. When EDS transmitted through the X-linked recessive gene, it usually affects only males due to their single X-chromosome. In females, the same version of the gene must be present on both X chromosomes in order for an individual to be affected. The signs of the EDS vary considerably with the form one is suffering. A constellation of similar symptoms are typically seen in individuals, which correlate and appear in most types of Ehlers-Danlos Syndrome. Finally, Dr. Maha Sulaiman re-stated how important it is to be on familiar terms with the clinical, scientific, and medical aspects of the Ehlers-Danlos Syndrome.

**III. Summary of Knowledge on EDS**

A. **Causes of the Ehlers-Danlos Syndrome**

Research reports at the EDS Network (2015) noted that most types of the syndrome are as a result of changes in one gene among several others. The manner in which the gene is oriented from one person to the other determines how the syndrome is inherited. The major three paths of transmission involve autosomal dominant and autosomal recessive in addition to the inheritance through the X-linked chromosomes, in an extremely rare case (EDS Network, 2015).

Mutation of the genes can cause the disorder to occur in many different ways. In autosomal dominant, there is a change of only one copy of a given gene required for a
person to be affected. In the recessive autosomal versions of the syndrome, both copies of the gene must be changed or mutated to a defective version, for one to be affected. By changing only one of the recessive alleles, a person becomes a carrier and shows no signs of the syndrome. Carriers, then have the possibility of passing the characteristics to one-half their offspring who will then be carriers or affected depending on the gene version from the other parent. EDS transmitted through an X-linked recessive version of the gene affects males, who only have a single X-chromosome. In females, the X-linked gene must be present on both X-chromosomes to cause an effect (EDS Network, 2015).

The EDS Network (2015) research also noted that though an effort to gather as much information as possible about the gene mutations that leads to the EDS, difficulties were found in establishing the real source of the problems that causes the whole syndrome. In all the primary sources examined the exact changes in the genes for all the types of EDS could not be determined (EDS Network, 2015).

B. What are the signs and symptoms of Ehlers-Danlos Syndrome?

The signs of the EDS vary considerably with the form one is suffering from (EDS Network, 2015). However, combinations of symptoms seem to correlate and appear in most types of EDS. The primary signs center around the defect or lack of sufficient levels of collagen in the body. It will affect the joints, the blood vessels, and the skin (EDS Network, 2015). The joints are hyperflexible, and are so unstable that they are easily vulnerable to joint dislocation and prone to sprain. The patient may be affected by the chronic diseases of the joints characterized by progressive deterioration and loss of function. In other cases of the disorder, there are serious signs that affect the spine. For
instance, one type of EDS, Kyphoscoliosis EDS is characterized by the deformation of the spine.

In most cases, the skin is affected by the disorder manifesting itself through fragility seen by the easy tearing skin, easy bruising of the skin, and extreme folding of the skin as the patient grows older (EDS Network, 2015). The Vascular type is detected by rupture of blood vessels and the uterus in females. It poses a threat to a possible progressive life threatening degree of valve prolapsed with the dilation of the ascending aorta that eventually ruptures if the patient is not put under treatment. Other complications of the EDS are the collapsing of the lungs after a severe damage, disorders of the nerves, lack of sensitivity to anesthetics while the platelets do not support blood clotting or clump together properly (EDS Network, 2015). All the types of EDS are characterized by increase chronic pain, weak and abnormally stretchable skin as well as brain disorders in the later stages that are usually life threatening.

C. People Living with Ehlers-Danlos Syndrome

Ehlers-Danlos Syndrome in most forms is commonly characterized by fatigue. The patients who have been diagnosed with the EDS are advised to ensure they conserve their energy and establish a competitive speed in their activities (NHS Choices 2014). The patients are restricted from being involved in activities that would tire them quickly. Most patients avoid heavy lifting, or staying in one position for long which could lead to joint problems. According to one patient at the NHS Choice website (2014) who had survived with the condition for over fifteen years now, it only takes simple measures to protect oneself and the joints from physical pain. Moderated exercise will help strengthen
the muscles and help minimize any cases of dislocation of the joints (NHS Choices 2014). Bandaging of the joints is important in the lower legs and the elbow when involved in activities to reduce skin damage. Parents that are taking care of children with the conditions should take precaution to protect them from harm of any kind.

However, it is considered important that the parents are not too protective to the extent of denying the affected children some freedom. They are instead, advised to allow the children who suffer from the EDS to have as normal a life as possible. Doctors recommend that those individuals with Vascular EDS avoid sports with a lot of physical contact and other activities that require any sort of strain, such as a sudden increase in speed or weight training. On enquiry, a patient explained that they have medical alert bracelets that help them check on their activities and notify them whenever there is a weird situation or a state that signifies any form of danger (NHS Choices 2014).

In the research reported by NHS Choice (2014), successful interviews were conducted with the EDS patients’ support panels all over the globe through the internet and electronic mail. In addition, physical interviews with other staff associated with the support of patients living with the deadly disorder were conducted. The research team was also able to gather some information from a series of interviews with the support group. NHS Choice (2014) advised that if one has problems with pain and movement, or they have relatives, children or friends suffering the symptoms, they are supposed to request their general practitioner to refer them to medical personnel. Most preferred of all medical professionals who specializes in the treatment of diseases, injuries or body deformities through physical methods (NHS Choices 2014). For example,
physiotherapists use methods like massage and exercise rather than drugs and surgery in the treatment of their patients.

The person suffering problems to do with pain and movement should be referred to physiotherapists who have a good understanding of the joint hyper mobility. The general practitioner can refer the suspected patient to an occupational therapist who will help them manage their day to day activities while giving them the advice they require on the equipment they will use to help them in their activities. Use of cognitive behavioral therapy can assist the patient suffering from EDS to muddle through with the pain that could have affected them for a long time. Cognitive behavioral therapy is accompanied by a lot of counseling to ensure it is a success. The general practitioner should be able to give advice regarding the neighborhood counseling services (NHS Choices 2014).

The interview drew the wrapping up that the support groups like the EDS support group of the United Kingdom are more than willing to give support to the sufferers and make the awareness of the syndrome accessible to all the people across the globe. They advise that more information should be sought from the genetic counseling and local genetics services. Here they will talk about the causes of the condition and in the process; (NHS Choices 2014) genetic counseling, will discuss the chances of one passing the syndrome on to the future children teaching on the ways to control the inheritance of the condition (NHS Choices 2014). The support group’s websites offer advice about the control and ways of overcoming sexual difficulties that are associated with the pain of the syndrome.
D. Diagnosis of Ehlers-Danlos Syndrome

Any symptoms that suggest extremity in the looseness of the joints and any abnormalities in the qualities of the skin of an individual along the family line can cause the step of diagnosis to be taken. Tests are done to the skin to determine the state of the health of the individual’s skin. The skin biopsy is a skin test that is done for several types of Ehlers-Danlos syndrome (EDS Network, 2015). The types diagnosed by this test include the Vascular and the Dermatosparaxis types of EDS. It absorbs the practice of removing a skin part as a sample that is used to check up the microscopic details of the skin structure. Kyphoscoliosis is diagnosed using a urine test. If parents took responsibility for having their child diagnosed, chances of having another child patient of the disorder will depend on the type of EDS the child has been diagnosed (EDS Network, 2015).

The parents’ status of the form the child was diagnosed of will also determine the fate of the children to come in future. X chromosome linked recessive autosome causes EDS with a more complex prototype of inheritance. If the father passes his X chromosome to the children, boy children will be safe from the infection of the syndrome while the female children will be carriers of EDS. If the mother is a carrier of the disorder, the sons can have a fifty percent chance of being either positive or negative of the EDS group of symptoms that consistently occur together in a characteristic combination of behavior (EDS Network, 2015). The female children of a carrier mother can be unaffected by the syndrome or half a chance, they can inherit the carrier ability from their mother depending on the other chromosome that their father contributed.
There are many more other types of diagnosis done for particular forms of EDS. Prenatal diagnosis is a test only possible in types of the disorder where the defect, primary to the cause of EDS is found in separate member of the family.

E. Management of Ehlers-Danlos Syndrome

Management will mainly involve genetic counseling. This will help the individuals affected together with their respective families to understand how the syndrome impacts on their family history hence the family future members (EDS Network, 2015). There is no cure yet for the disorder. Carefully keeping an eye on the cardiovascular system and occupational therapy can be helpful in the management process. Instruments for the correction of deformities of bones and muscles are crucial for the prevention of more damage to the joints. They could help especially in long distances though doctors discourage individuals from becoming dependent on the orthopedic instruments unless the options for mobility are limited. A physiotherapist may prescribe casting to ensure the joints are stable and consult an occupational therapist to help the patient strengthen the muscles (EDS Network, 2015).

They teach people how to use joints correctly and preserve them healthy in the long run. Aquatic therapy is used to promote the development of muscles and muscular coordination. Manual therapy ensures the joints are gently mobilized to the maximum range of motion possible manipulating them to work to the advantage of the patient. Precautions out of the ordinary standards have been taken all through the times by doctors and medical care workers to control the complications that seem to arise in the patients suffering from EDS. Before the pregnancy period, patients are advised to get
genetic counseling (EDS Network, 2015). The patients suffering should be provided with a lot of information on the disorder for them to understand the reasons for some restrictions against normal life activities like participation in contact sports. From the point of view, the support groups are immeasurably helpful in guiding the patients on the best lifestyles to live and advising them on how to deal with significant changes in lifestyle and poor health conditions.

The people close to the patients should be taught and provided with information about EDS so that they can assist the people suffering as necessary. The conservative therapy has failed to control and prevent the instability caused by the disorder in the longer term. However, it is the most recommended way of dealing with the disorder compared to other surgical methods that only apply when therapy totally proves to be impossible to help the patient (EDS Network, 2015). The surgical intervention comes in when the instability of the joints causes extreme pain and dislocation out of the joint. From previous research was done in the earlier years, that after surgical procedures there is a degree of stability in patients, the pain reduces and the patients experience an improved degree of satisfaction.

However, the surgical procedures are discouraged by many occupational therapists. One claimed that surgical treatment has strings attached. He further explains that the surgery reduces the strength of the tissues, and the fragile blood vessels are a significant hindrance to operation hence could be so problematic (EDS Network, 2015). Another problem associated with surgery is the poor healing of the wound in the period after surgery, often delayed or at times even does not complete. The study has it that
anesthetics cause higher risk in the solid swelling of clotted blood within the tissue when used for surgery in patients suffering from the disorder. Surgery, if necessary with these patients, will highly require careful handling of the tissues and less mobility from that point forward (EDS Network, 2015). Special anesthetics should be recommended for the EDS patients to improve personal patient wellbeing features.
In conclusion, Ehlers-Danlos Syndrome (EDS) is a very serious medical condition that requires a lot of attention and careful treatment. The medical condition under study has been established to have no cure. Medical therapies depend on the management symptoms, and the primary aim is to prevent any further complications. The point of view in appearance for a person with the defect depends on the type of the syndrome they are diagnosed. Symptoms will always vary depending on the level with which the condition is severe. Some individuals have symptoms that are easily negligible. It is a lifelong state. A person affected may face obstacles in their social life on daily basis. Some people are not diagnosed until they are adults. Although persons with the defect go through different challenges in their lives, it is very significant to note that each has their uniqueness, and they are distinguished in qualities and potential.

The patients have a normal life and have families as well as the chance to be accomplished citizens while they overcome the difficulty of their challenges from the disorder. The difference in the presentations of the disorder is brought about by mutations in the Collagen gene, including the collagen alpha–1 (III) sequence of the collagen. Different transcripts have been determined for this gene resulting in the disorder. Note, collagen tissue is produced before tougher tissues are made by fibroblasts. It helps the
body tissues to fight deformation and is a very significant help in the provision of strength and resistance to pressure of the skin, muscles, ligaments, and blood passages.

The patient with EDS is time and again confused for depression and fatigue. The skin cannot hold stitches in cases where they are required due to skin problems. The syndrome is clinically diverse with the major variations occurring by virtue of the difference in the abnormality of the collagen lying beneath its occurrence. It is very important to be on familiar terms with the clinical, scientific, and medical aspects of the EDS one is dealing with or is suffering from.

This research was narrowed the ten major types, in 1997, down to six major types of the syndrome as classified in the previous research. The names of these types of EDS are: Classic type; Hypermobility EDS; Dermatosparaxis type, on the other hand, shows a skin that sags and there is the presence of wrinkles on the skin surface (Ensor, 2013); Arthrochalasia type; and the Kyphoscoliosis type of EDS which can engross severe difficulties that are potential threats to the life of the sufferer.

Additionally, EDS of the Kyphoscoliosis type may cause breathing problems and difficulties due to its significant contribution to a serious progressive curving of the spine. The most dangerous is the Vascular type. Hypermobility EDS is Type III and is inherited only through autosomal dominant or recessive genes. The Classical type of EDS includes both Types I and Type II, merging two of the previous ten types. Vascular EDS is an effect of a dominant autosome defect in the production of Type-III collagen, not to be confused with its particular classification as Type VI EDS. Dermatosparaxis EDS is a
recessive genetic form, known as Type VII-C, and is considered to be very rare such that almost no case has been reported for several years.

Most types of the syndrome are as a result of changes in one gene among several others. The manner in which the gene is oriented from one person to the other determines how the syndrome is inherited. The major three paths of transmission involve autosomal dominant and autosomal recessive in addition to the inheritance through the X–linked chromosomes which is an extremely rare case. Any symptoms that suggest extremity in the looseness of the joints and any abnormalities in the qualities of the skin of an individual along the family line can cause the step of diagnosis to be taken. Management will mainly involve genetic counseling. This will help the individuals affected together with their respective families to understand how the syndrome impacts on their family history hence the family future members.

Fighting the disorder is mainly based on the prevention and control aspects since the Ehlers-Danlos Syndrome has no cure. Any signs of joint problems, problems to do with blood vessels and abnormality in the conditions of the skin should be reported to the medical authorities as soon as possible. Patients should be diagnosed and proper medication sought as quickly as possible. The best fight however is the spread of awareness about the disorder. The results of this research are meant to support the awareness of the syndrome to ensure that all people are educated on the dangers. The contribution in the provision of information to save the entire world from deadly silent killers that would rather go unnoticed. Salvation begins with us.
REFERENCES CITED


Retrieved from URL: omim.org/entry/130000, updated 09/15/2015, accessed 10/28/2015


BIBLIOGRAPHY


Downloaded: 11/09/2015


http://www.mayoclinic.org/diseases-conditions/ehlers-danlos
syndrome/basics/definition/con-20033656

http://www.nhs.uk/conditions/ehlers-danlos-syndrome/Pages/Introduction.aspx

Retrieved from URL:omim.org/entry/130000, updated 09/15/2015, accessed
10/28/2015

version-Pediatrics. Merck Sharp Dohme Corp, Kenilworth, NJ. Retrieved from
https://www. Merckmanuals.com/professional/pediatrics/

(2013). Dermatosparaxis (Ehlers-Danlos type VIIC): prenatal diagnosis following
a previous pregnancy with unexpected skull fractures at delivery. *Medical


APPENDICES

APPENDIX A

Interview #1: Maha Sulaiman, M.D. 29
Interview #2: Talib Al-Abbadi, M.D. 30

APPENDIX B

Key Terms
Medical Terminology 32
Symptoms 32
Genetics 32
APPENDIX A

Two practitioner Dermatologists were selected and interviewed. I have had known one of them since I had an experience with, Dr. Maha Sulaiman, through treatments in the Saudi Arabian Airline’s Medical Center, in Jeddah Saudi Arabia. The other, Dr. Talib Al-Abbadi, my father knew, and arranged the interview with him.

Dr. Maha Sulaiman and Dr. Talib Al-Abbadi, both are Dermatologists, MD. They were interviewed and were posed questions relating to: EHLERS-DANLOS SYNDROME (EDS). Different questions were posed to each one of them, but with some similarities of the symptoms. The overall outcome gave insights of the EDS.

Interview #1: Maha Sulaiman, M.D.

Question: How do the different types of EHLERS-DANLOS SYNDROME (EDS) affect victims?

Dr. Maha: They all involve a defect of the genetic arrangement of the collagen and the synthesis of the connective tissue as well as the connective structures.

Question: Which parts are affected most in an individual?

Dr. Maha: Ehlers-Danlos Syndrome tends to affect blood vessels, joints and the skin in most cases. People with the syndrome usually have excessively flexible joints and a fragile skin. This idea is also referenced in (Ensor 2013, p 2), (Mayo Clinic Staff 2012, p 1).

Question: How fragile can the skin be?
Dr. Maha: The skin cannot hold stitches in cases where they are required due to skin problems.

Question: What is the cause for the many types of the same infection?

Dr. Maha: The syndrome is clinically diverse with the major variations occurring by virtue of the difference in the abnormality of the collagen lying beneath its occurrence.

Question: What should be taken into consideration when dealing with a patient?

Dr. Maha: It is very important to be familiar with terms with the clinical, scientific, and medical aspects of the Ehlers-Danlos Syndrome one is dealing with or is suffering from. One may have concerns over what the cause of the Ehlers-Danlos syndrome is.

Interview #2: Talib Al-Abbadi, M.D.

Question: How is EDS transmitted?

Dr. Talib: There are three major paths of transmission that involve autosomal dominant and autosomal recessive in addition to the inheritance through the X – linked chromosomes which is an extremely rare case. This is notion is confirmed in Sanders 2009.

Question: Please explain how the mutations affect the disorder from one person to another.

Dr. Talib: Mutation of the genes can cause the disorder to occur in many different ways. In autosomal dominant, there is a change of only one copy of a given gene for a person to be affected.
**Question:** What is the mechanism of the recessive transmission?

**Dr. Talib:** The recessive version of the autosomal chromosomal syndrome requires that both copies of the gene change for one to be affected. Changing only one of the recessive, the person becomes a carrier showing no signs of the same.

**Question:** How does the carrier transmission occur?

**Dr. Talib:** Carriers have a great possibility of passing the characteristics to their offspring. EDS transmitted through the X-linked recessive gene affects only a single X–chromosome. In females, the similar gene in both X-linked chromosomes must be changed in order to cause an effect. This notion is also confirmed in Sanders 2009.

**Question:** What are the signs and symptoms of Ehlers-Danlos Syndrome?

**Dr. Talib:** The signs of the EDS vary considerably with the form one is suffering from. This notion is also confirmed in Sanders 2009. However, we had a combination of symptoms that seem to correlate and appear in most types of Ehlers-Danlos syndrome. The signs go around the defect or lack of sufficient levels of collagen in the body. It will affect the joints, the blood vessels, and the skin. This notion is also confirmed in Sanders 2009.

**Dr. Talib:** The joints are hyper flexible and so unstable that they are easily vulnerable to joint dislocation and prone to sprain. The patient may be affected by the chronic disease of the joints characterized by a progressive deterioration and loss of function. In other cases of the disorder, there are serious signs that affect the spine; for instance, one type of EDS, the kyphoscoliosis EDS is characterized by the deformation of the spine.
APPENDIX B

Key Terms
Associated with Ehlers-Danlos Syndrome, based on the interviews.

Medical Terminology
Dermatologists, treatments Ehlers-Danlos Syndrome (EDS), collagen, joint dislocation, prone to sprain, chronic disease, progressive, deterioration, spine.

Symptoms
Synthesis of the connective tissue, connective structures, blood vessels, joints, skin, syndrome, flexible joints, fragile skin, stitches, skin problems, Infection, abnormality, transmission.

Genetics
Autosomal dominant, defect of the genetic arrangement, autosomal recessive, X – linked chromosomes, mutations, genes, autosomal chromosomal syndrome, the carrier transmission, chromosome.