



Comprehensive Study on Pregnancy, Childbirth, and Childcare of Women with Ehlers-Danlos Syndromes

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**We have no COI
with regard to our presentation.**



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Ehlers-Danlos Syndrome (EDS)

The most frequent types are the three listed below.
Women with EDS have potential risks for pregnancy.

Types and prevalence	Gene	Heredity	Risks in pregnancy
Classical Type 1/20,000	COL5A1, COL5A2	Autosomal dominant	Premature birth, PROM Laceration of the perineum Uterine prolapse, Dislocation
Vascular Type 1/50,000~	COL3A1		Aortic dissection Uterine rupture Premature birth
Hypermobile Type 1/5,000	Unknown		Preterm labor PROM Serious low back pain

Levy HP. Ehlers-Danlos Syndrome, Hypermobility Type. 2004 Oct 22 [Updated 2016 Mar 31]. GeneReviews® [Internet]. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1279/>


古庄知己: エーラスダンロス症候群の臨床・疫学, 日本胸部臨床, 70, 2011より参照

Introduction

Medical-Record

Questionnaire

Analysis



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Background

At the Center for Medical Genetics at Shinshu University Hospital, we have provided diagnosis and genetic counseling to patients with various types of EDS nationwide

Pregnancy of women with EDS is an important life-event that could be associated with physical risks and affect the following generations, the condition has never been comprehensively investigated in Japan.

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Purpose

To clarify experiences of women with various types of EDS in Japan in their perinatal periods in view of establishing effective perinatal management,

We performed

1. Medical record-based investigation
2. Questionnaire-based Survey



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Medical record-based investigation

Subjects


Women with various types of EDS that have received genetic counseling and/or medical care in Shinshu Univ. Hospital.

Periods

April 1, 2000 to March 31, 2018.

Collected information

The ages at diagnosis, married or not, medical histories, courses of pregnancy and delivery, number of children including their physical status (affected or not), etc.



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Result 1) The results of all 60 adult women with EDS


60 adult women with EDS
mean age at diagnosis
: **29.4 ±12.0 (years)***

*Mean age of the first marriage in Japan : 29

Out of the 60 women, 21 gave birth,
and 18 births were before the diagnosis
Mean age at diagnosis : **34.3±12.0 (years)**
Mean age of birth age: **28.1 ±4.3 (years)**

EDS types	Women with EDS (n)	Married (n)	Multipara (n)	Delivery (n) [per woman]
Classical	8	5	3	7 [2.3]
Vascular	20	17	12	21 [1.8]
Hyper mobile	26	14	5	6 [1.2]
Musculocontractual	3	0	0	0
Arthrochalasia	1	1	1	3 [3.0]
unknown	2	0	0	0
Total	60	36	21	37 [1.5]

*国民衛生の動向2017/2018: 一般財団法人厚生労働統計協会,第2章人口動態, 2017; 64: 59-83参照



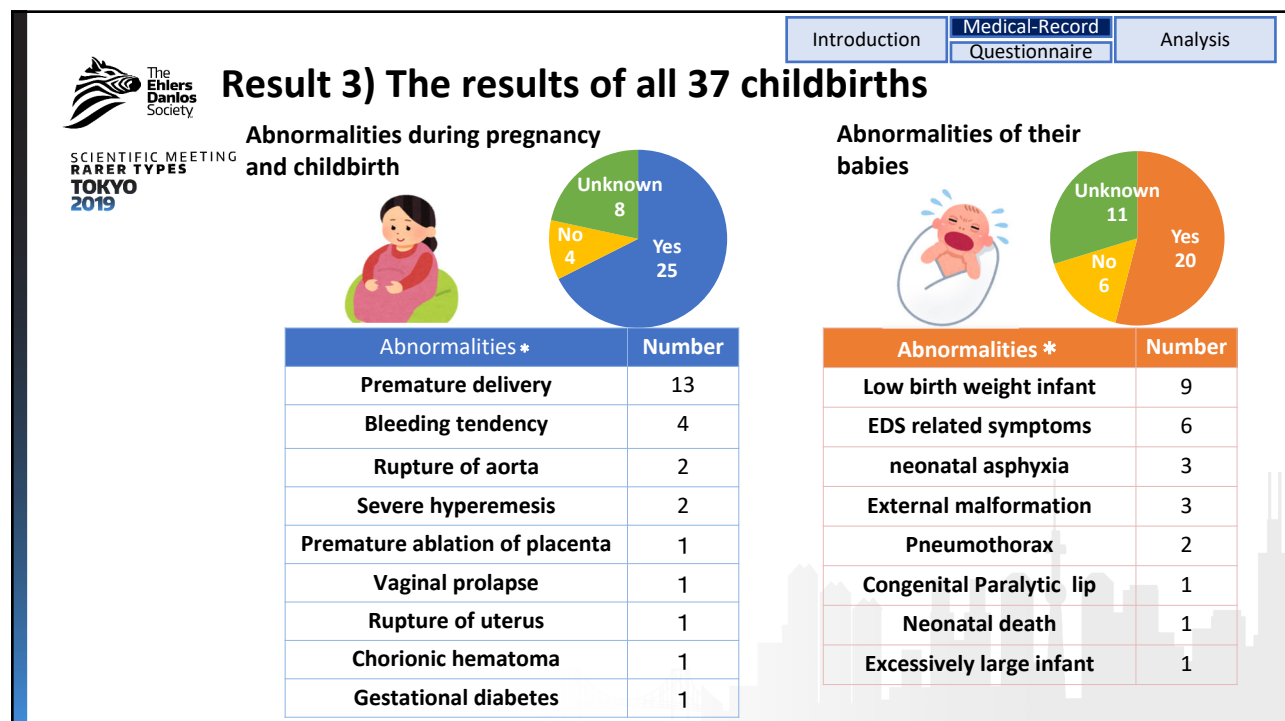
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Result 2) 6 women diagnosed as EDS throughout their childbirth

	Type	Course	Delivery
1	Vascular type	Rupture of aorta at GA 32 weeks Neonatal asphyxia in the baby	Emergency caesarean (undiagnosed)
2	Vascular type	Rupture of uterus at GA 36 weeks	Emergency caesarean (undiagnosed)
3	Vascular type	Premature ablation of normally implanted placenta Death in the baby	Emergency caesarean (undiagnosed)
4	Vascular type	Rupture of left iliac artery at GA 34 weeks Neonatal asphyxia in the baby Rupture of right iliac artery in the postpartum period	Emergency caesarean (undiagnosed)
5	Vascular type	Disclosed that her father was diagnosed as vEDS at GA 10 weeks Followed by her own diagnosis as vEDS	Elective caesarean (diagnosis during pregnancy)
6	Hypermobility type	Suspected as having Marfan syndrome at pregnancy check-up.Followed by the final diagnosis as hEDS	Vaginal (diagnosis during pregnancy)

Undiagnosed women with vEDS have significant pregnancy-related risks!




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Discussion 1

- The average age of diagnosis as EDS women is the same as the average age of marriage for Japanese women.
- The pregnancy and childbirth for women with vascular EDS without being diagnosed carry high risk.



- Early diagnosis and perinatal genetic counseling are required for a safe childbirth.
- If a woman with EDS wishes to have a baby, a perinatal medical system with enhanced cooperation with other hospital departments is crucial.



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Questionnaire-based survey

Subjects

15 Women with various types of EDS, who experienced childbirth.

*Women with mental illness or those who couldn't have a baby were excluded, considering a psychological potential burden of this survey.

Contents of the questionnaire

- I) Whether their experiences of pregnancy, childbirth, and childcare were a positive or negative one for them, according to the scale of "1-5".
- II) Free description about their experiences of pregnancy, childbirth, and childcare.



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Result 1) Self assessment of their experiences


9 out of 15 women answered the questionnaires (response rate, 60%)

1. Negative experience 2. Slightly negative experience 3. Intermediate
4. Slightly positive experience 5. Positive experience

thought their
experiences as
positive
in each period

thought their
experiences as both
negative or positive

Woman	Types of EDS	Pregnancy	Childbirth	Childcare
A	Classical	5	5	5
B	Vascular	5	5	5
C	Vascular	5	5	5
D	Vascular	5	5	5
E	Vascular	4	5	5
F	Vascular	5	5	5
G	Hypermobile	3	3	4
H	Hypermobile	1	4	4
I	Hypermobile	4	2	1





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Result 2) Free description


Concern about heredity (by 8 women)

I'm most worried about inheriting EDS to my children.  (Vascular EDS)

 *I feel very sorry for my children inheriting EDS, just because I have EDS.* (Classical EDS)

 *I worry more than necessary when my children say they have pain somewhere.* (Vascular EDS)

NEGATIVE effects of EDS diagnosis





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Result 2) Free description


Acceptance of EDS (by 4 women)

I became depressed after I was diagnosed with EDS.  (Vascular EDS)

I didn't have the slightest idea that childbirth could have risk on my life, so I'm very grateful that I was diagnosed as EDS beforehand.  (Vascular EDS)

 *After I was diagnosed with EDS, I realized why I was not good . . .* (Hypermobile EDS)

NEGATIVE or POSITIVE effects of EDS diagnosis



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Result 2) Free description

No daily physical symptoms (by 2 women with vEDS)


I know that aortic-dissection will happen someday, and I'll pass away. So until then, I'm willing to give and share love for my child unstintingly.

Even though I was diagnosed with EDS, since I have had no subjective symptoms at all, the disease has never affected childcare.


NOT NECESSARILY NEGATIVE impacts of physical symptoms in vEDS



(Vascular EDS)



(Vascular EDS)



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Result 2) Free description

Physical burdens (by 2 women with hEDS)


I regret to this day that I could not carry my child and play with her because of chronic pain around my body.

Each time I scolded my children because of pain, I felt a strong sense of guilt.


NEGATIVE impacts of physical symptoms in hEDS



(Hypermobile EDS)



(Hypermobile EDS)




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Result 2) Free description

Supports reducing the burden (by 7 women with EDS)




I am always worried if my children inherited my EDS, but I receive support by my husband, older sister, and doctors.

(Vascular EDS)



I'm very grateful to my parents, husband, and mother in-laws helping with my housework while I was hospitalized.


(Classical EDS)



My illness has made my husband take care of my child. A woman with EDS should marry someone who likes childcare!!

(Vascular EDS)

POSITIVE impacts of external support




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Result 2) Free description


Lack of understanding of EDS (by 2 women with hEDS)

No matter how painful it was, no one understood EDS like now, and I carried my children while taking painkillers.



(Hypermobile EDS)

Even though I companied that I'll have childbirth any time, the nurses and my husband didn't take it seriously because it was my first child. I'm still disappointed about that day.



(Hypermobile EDS)

NEGATIVE impacts of external support



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Discussion 2

- Experiences of childbirth for women differed depending on the type of EDS, especially chronic pain made perinatal experiences difficult for women with hEDS.
- Impacts of diagnosis, physical symptoms, and support from others seemed important.



- In genetic counseling, empathy for women with EDS would be important in view of their physical and psychological burdens based on the type of EDS.



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Collaborators

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Acknowledgment

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