"It was a lot tougher than I thought it would be"

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ORIGINAL RESEARCH

"It was a lot Tougher than I Thought It would be". A Qualitative Study on the Changing Nature of Being a Hemophilia Carrier

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Abstract Studies on carriers of genetic disorders mainly focus on the process of genetic testing and reproductive choices, and less on how psychosocial aspects of being a carrier change over time. Our study sought to understand more about the psychosocial aspects of hemophilia carrier status, and thereby improve counseling aiming to advance carriers' quality of life and well-being. We analyzed 16 in-depth interviews from women who were carriers of hemophilia and had a son with hemophilia. Three themes emerged: Guilt and sorrow across generations; the choices and future consequences of genetic testing; and preparing to have a child with hemophilia. Experience with being a hemophilia carrier is a process that changes over time while feelings of guilt and sorrow run across generations. The carrier status may create "mothers-in-waiting" living at risk of having a sick child or not. The women think they are prepared to have a son with hemophilia, but experience more sadness than they expect when a son is diagnosed. Our findings suggest that health professionals, especially clinical geneticists and genetic counselors, carriers, families and patient organizations need to be aware that women's experiences of being a carrier of hemophilia changes during the biographical life course. The women may benefit from several rounds of genetic counseling at different stages of life.

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Introduction

Hemophilia A and B are X-linked disorders characterized by deficiency in factor VIII (hemophilia A) or factor IX (hemophilia B) clotting activity resulting in prolonged bleeding. The disorders are chronic with a medical and psychosocial burden, not only on the patient, but also the family (Barlow et al. 2007; Goldstein and Kenet 2002; Torres-Ortuno et al. 2014; von der Lippe et al. 2017). Women who are carriers of hemophilia are usually without bleeding symptoms. Carriers with bleeding symptoms may experience inappropriate care, feel mistrusted and not taken seriously by the healthcare system (Renault et al. 2011), and have impaired health-related quality of life (Olsson et al. 2015).

Due to the increased risk of bleeding, a woman's carrier status has implications for child birth. Knowledge of a mothers' carrier status may also facilitate the diagnosis of hemophilia in a son (Chi et al. 2008; Street et al. 2008). Most hemophilia A patients and their at-risk female relatives find it important to know their (own) carrier status (Sorenson et al. 2003), and many parents want to know the carrier status of their young daughter before the daughter is old enough to consent (Dunn et al. 2008). Reasons for why parents want to know the carrier status in their child may be that parents find it comforting to know, and because the parents want to prepare the child to make reproductive decisions later in life (Vears et al. 2016). Even though patients, at-risk females and parents think it is important to know one's hemophilia carrier status, a positive family history for hemophilia does not mean that female relatives always know their carrier status (MacLean et al. 2004). A reason for non-disclosure may be that parents find it

emotionally painful and difficult to communicate about genetic risk with their children (Metcalfe et al. 2011). Feelings of guilt may also be a cause for non-disclosure. A review of carriers of different genetic diseases showed that carriers who already have an affected child are more likely to experience guilt and self-blame, and change their reproductive plans compared to carriers without affected children (Lewis et al. 2011). For hemophilia, it has also been documented that learning about being a carrier may lead to feelings of guilt for giving birth to a son with hemophilia (Ross 2000; Thomas et al. 2007).

Several studies on carriers have been investigating reproductive behavior and choices and attitudes toward prenatal diagnostics (PD). The fear of passing on the mutation to a child means that many women make a conscious decision not to have children (Kadir et al. 2000; Martensson et al. 2014). PD are used both to decide termination of an affected fetus (Varekamp et al. 1990), or as psychological preparation for having a child with hemophilia (Martensson et al. 2014). In a Finnish study of 167 first-degree female relatives of hemophilia patients, only 16% would terminate the pregnancy if the fetus was affected (Ranta et al. 1994). Almost all women in this group belonged to families with severe hemophilia. Experiences with complications of hemophilia or its treatment seem to make carriers to be more in favor of PD than carriers without such experiences (Tedgard et al. 1999b). The long-term psychological effects of carrier testing and PD of hemophilia do not seem to be negative (Tedgard et al. 1999a).

Although some of the above mentioned studies address the psychosocial impact of being a carrier of hemophilia, there has been limited focus on the emotional and psychosocial aspects experienced by women throughout their life (Cassis et al. 2012). It seems also that genetic counseling has a focus on medical issues more than psychosocial issues. A review of communication in genetic consultations suggested that clinicians are spending most time explaining information to clients, and not addressing psychosocial issues that may exist (Paul et al. 2015). Thus, in order to understand more about the psychosocial aspects over time of hemophilia carrier status, and thereby improve the counseling aiming to advance carriers' quality of life and well-being, we, in this paper ask: How do women experience being a hemophilia carrier over time? This study is part of a larger study investigating experiences of being a carrier of an X-linked disorder.

Methods

Design

We used an explorative qualitative research approach, specifically semi-structured in-depth interviews to gather insights into the women's experiences of being a hemophilia carrier.

Sample and Recruitment

A nurse at the national competence center for hemophilia, Centre for Rare Disorders, Oslo University Hospital, Norway, invited women known to be carriers of hemophilia A or B to participate. To be included, the women had to be carriers, have a child with hemophilia, and understand and speak Norwegian or English. We aimed for recruiting ten participants. To achieve variation in the sample, the nurse selected 11 women of different ages and from different parts of Norway. She approached four mothers during their sons' annual follow-up and phoned seven other mothers. Five additional women contacted the principal investigator after they had learned about the project from the Norwegian Haemophilia Society. All women received written information about the study and agreed to participate. Thus, the sample comprised a total of 16 women with an average age of 42 years (Range: 27–72). All participants gave written consent and decided when and where they wanted to be interviewed. For clinical and demographic characteristics of the sample, see Table 1.

Data Collection

The principal investigator conducted all 16 semi-structured interviews between February 2013 and March 2014 on the premises of a university hospital or at participants' homes. The interviewer, a clinical geneticist, was explicit about her role in the project as a researcher. The digitally recorded interviews lasted on average 70 min (Range: 40–100), and were transcribed verbatim. The interview guide included open-ended questions such as: Tell me about how, and when, you learned you were a carrier, and what you feel about being a carrier? How do you feel about having an affected child? How do you communicate about having a genetic disorder in your family? How does your son's disorder affect you and your family's daily life? What are your experiences with the treatment and the healthcare system? Even though all the participants expressed gratitude that a researcher was "finally asking about them", it was not easy to get the women to talk about themselves. As soon as they had the chance, they started talking about their son with hemophilia, about how he was diagnosed, his treatment and their experiences with the healthcare system. They had difficulty focusing on their personal feelings. As for example in the interview with Mette:

> Interviewer: *Can you tell me about when you first learned you were a carrier?*
> Mette: *It was not discovered until the youngest boy was 6 months old. He fell off the changing table, and got a lump back in his neck. I went to the doctor and he thought it was a collection of blood, and reffered him to the hospital…[goes on with what happened to her son].*

Table 1 Participants' clinical and demographic characteristics

Participant	Age	Type of hemophilia (A or B)	Number of affected sons	Carrier status known prior to having a son with hemophilia
Kari	35	Hem. A, severe	1	Yes
Lisa	32	Hem. A, severe	1	Yes
Anna	41	Hem. A, severe	1	Yes
Heidi	27	Hem. A, severe	1	No
Lisbeth	39	Hem. A, severe	2	No
Marianne	35	Hem. A, severe	1	Yes
Ingrid	27	Hem. B, moderate	1	Yes
Thea	30	Hem. A, severe	(1, early termination)	Yes
Lena	40	Hem. B, moderate	2	No
Tanja	65	Hem. A, severe	3	No
Wilma	55	Hem. A, severe	1	Yes
Mette	72	Hem. A, severe	2	No
Victoria	34	Hem. B, moderate	2	No
Jane	56	Hem. A, severe	1	Yes
Therese	28	Hem. A, severe	1	Yes
Astrid	49	Hem. A, severe	1	Yes

The interviewer thus guided the mothers to talk more about themselves and their experiences of being a carrier.

Data Analysis

We adopted an inductive thematic analytical approach for analyzing the data (Braun and Clarke 2006). The principal investigator listened to the interviews, read the transcripts several times and summarized in writing the key aspects of each woman's experiences. The second and the fourth authors read transcripts, while the third author participated in the subsequent analysis and discussions. The first author coded the entire material line by line by using the software program HyperRESERCH (Hesse-Biber et al. 1991). All authors discussed the coding and grouped codes by main themes. We reviewed the themes and data extracts by going back to the transcripts and to the research questions, and selected quotes representative for the themes. The Norwegian authors translated the quotes and gave the participants pseudonyms.

During the data analysis we remained cognizant of mothers potentially downplaying the individual burden of being a carrier. This was kept in mind because of the fact that the mothers would spend much time in the interviews talking about the son's disease and treatment, and a certain phrase from one of the mothers stating: *There is so much treatment that every day just passes by very quickly so you give no space to your own feelings or thoughts. You displace your own feelings or experiences and focus on the son.* (Marianne)

This paper will focus on results concerning women's experiences over time with being a carrier. In the last stage of the data analyses in which this topic was singled out we especially found three themes to run distinctly through the interviews. These themes were: guilt and sorrow running through generations; the choice and future consequences of testing; and preparing for having a child with hemophilia.

Results

Guilt and Sorrow across Generations

Several of the women expressed feelings of guilt and self-blame for having passed on the mutation to a son. One woman even thought this was something everybody felt, and did not believe those who said they did not have feelings of guilt:

> *I would like to know that carrier who does not have bad conscience about it, I do not believe it. … I think everyone who is the cause of someone's illness will have bad conscience.* (Lena)

Feelings of guilt and self-blame were not restricted to the individual women, but ran across generations of a family. The women shared stories about sad feelings expressed by

their mothers who were carriers, fathers who had hemophilia, and even grandmothers:

My mother blames herself, so she feels she is the cause of me [being a carrier] and says "Oh no, should we pass this on?" and "let's just cross our fingers and hope we do not pass it on". (Marianne)
My dad thinks it is very, very sad [that his grandson has hemophilia]. Because I think he feels guilty because it comes from him. He has also said so. He has apologized. (Ingrid)

Heidi, even felt sorry for her partner:

I went through a phase where I had such a guilty conscience because my boyfriend ended up with me who gave him a son with an illness. It was my fault.

However, not everyone shared feelings of guilt. When told by the interviewer that others had expressed feelings of guilt for passing on the mutation a few responded that it was just the way it was [that they had a son with hemophilia] and that there was nothing they could do about it. Wilma elaborated on these thoughts and stated:

I have never had much problem with my son, never very difficult times. Maybe if there had been a lot of that [problems with hemophilia], maybe then one would have feelings like that [feelings of guilt]. But I have never had that.

The Choices and Future Consequences of Genetic Testing

Many of the women had a father with hemophilia, and they had been aware of their carrier status since their childhood or teenage years. It had been explained to them that they had inherited their fathers' X-chromosome and thereby were obligate carriers. Participants who knew their mother was a carrier were in a different situation. They had to make a choice whether or not they wanted to undergo genetic testing to find out if they had inherited the family mutation. Some had decided to get tested around the age of 16 years, which is the age from when you legally can be carrier tested in Norway. Most of the women expressed that they did not really pay much attention to the result of the genetic testing when they were young, because the consequences felt far in the future. Lisa had grown up knowing her male cousin had hemophilia, and knew her mother was a carrier. Lisa was tested when she was 16 years old, and found out she was a carrier. In response to a question about how she felt when she found out she was a carrier, she replied:

I didn't feel that much. I almost took it for granted, I know I wasn't sad. It just confirmed it. I felt I was like my mother and my aunts. I was maybe naive to what the future would bring. It was not a failure. It was not. I was more like the others. I grew up with the fact that women were carriers, and it was OK. ……. I did not feel it [the carrier diagnosis] changed my identity because, the consequence was so distant. When you are 16 you do not think about having children. So in a way I did not feel it changed me. It was nothing I reflected upon.

For those who first learned about their carrier status after a son was diagnosed, and where other relatives did not have the mutation, the psychosocial consequences were different from those who knew their carrier status since their childhood or teenage years. Heidi describes how this can lead to conflicting emotions:

I remember about half a year ago when we found out that my mother was not a carrier, I became upset, because then it was just me. Then it was not something the family had in common… I can't tell him [her son with hemophilia] grandmother has it – it is just stupid me... then again it is something that binds us [she and her son] very tight together. It is not like I love one of my children more, but this is a "thing" with him and me.

Once a woman in a partnership knows she is a carrier she also has to make a choice about when to tell her partner. Most of the participants in our study had chosen to tell their partner about their carrier status early in the relationship, as they believed their partner should know. For example, Wilma shared the following:

I think I told him I was a carrier on our second date. Right away, I told him. I thought I just needed to have his reaction on this because there was nothing to build a relationship on if he was negative about it [me being a carrier].

Almost all had experienced affirmative reactions about their carrier status and support from their partner. However, not everyone experienced this support. In one family nobody had talked about the risk of being a carrier, except the grandmother mentioning hemophilia as "something distant in the family". The mother had never given it a thought that she could be a carrier – not until her first son was diagnosed with

hemophilia at six months of age. She had never felt she had practical or emotional support from the children's father or his family, and she expressed her feelings about the lack of support:

It was very difficult. I felt I was never good enough. I probably wasn't [good enough] in his family, to his parents. My children were in a way never accepted.

Preparing to Have a Child with Hemophilia

As mentioned previously, the majority of the participants were familiar with hemophilia from their adolescence, either because a maternal grandfather, brother, male cousin or father had hemophilia. They knew about bleeding episodes, pain, hospital treatment, home treatment, and risk of developing inhibitors to the factor treatment. Many had witnessed the improvement in medical treatment that has taken place in recent years and they believed these experiences made them well prepared to have a son with hemophilia. The women, who knew they were carriers either before the first child was born, or after they had a son with hemophilia, talked about deciding to have children or not, or trying to have a child without hemophilia. However, even though they thought themselves well prepared for having a son with hemophilia many shared stories of profound sadness when their son was diagnosed with hemophilia, and as Marianne said:

But, It is Obvious; You cannot Prepare yourself for that Anyway

The women were surprised by these feelings of sadness because when looking back to when they learned they were a carrier they never thought they would have such feelings, as articulated by Lisa:

It was a lot tougher than I thought it would be. My common sense disappeared and my emotions took over. Even if I was very, very prepared that he [my son] could have hemophilia, it [the son getting the diagnosis] was a "sentence" that was much harder than I had thought. So I had a lot of sad feelings and reflected a lot about it for a period. At the same time, I constantly told myself this would be OK, things would be OK, but it was much harder than I could imagine.

Preparedness was not just something the women expected of themselves but something they felt others expected of them. When Ingrid's son was diagnosed with hemophilia her friends thought she should not be sad and feel sorry, because she had been "prepared", knowing she was a carrier. Ingrid also felt she had to be the strong one in the relationship with her partner because the mutation "came from her".

Even though some felt it was easier to learn about one's carrier status and risk of having a son with hemophilia before a son was born, this knowledge could create fear; fear of the son bleeding while in utero and fear of doctors not providing the right treatment during birth. Thea knew from childhood there was a risk she could be a carrier but she did not have a genetic test done until she was pregnant. Undergoing genetic testing had been a difficult decision and she stated:

It is not being a carrier that's difficult, what's difficult is when you find yourself in a situation where it has a consequence.

She explained that she herself did not face any problems with being a carrier, but she thought it was very difficult to know she could have a child with hemophilia. She had experienced hemophilia in several generations in her family, and was well aware that treatment was available. Nevertheless, she thought it was difficult to make the choice of having a child with hemophilia. She believed that having a son with hemophilia would make a great impact on daily life in the family, and the dynamics in the family such as the relationship with her partner and any healthy siblings of a boy with hemophilia.

Victoria, who first learned about her carrier status after a son had been diagnosed, reacted with surprise because she had to make choices she could not have imagined making previously:

I was a bit surprised [when I learned I was a carrier], because you read about these things in the paper, people who have to make these kind of choices [to have children with a disorder or not]. I never pictured I would be one of them.

She reflected further on this, and thought it was not a problem to have one son with hemophilia, and things were easy with the treatment of her son after he had an intravenous access device and they could treat him at home. However, thinking about having more children with hemophilia made her sad and emotional. With tears in her eyes she expressed the moral dilemma this question raised for her:

It is not really a problem, but what I have been thinking is if it is OK, now that I know I am a carrier, to have another child without making sure the child does not get

the disorder. Is it morally right to get a child with a serious disease, now that we could avoid it?

Interestingly, Anna, who thought treatment was going well, expressed it was not a big problem to have a child with hemophilia:

It is a very little burden it is just part of the morning ritual. (Anna)

A few expressed it was not an option for them to choose prenatal diagnostics and one expressed her surprise it was even an option to choose termination of an affected fetus:

Why do they talk about that [termination]? Is that an issue? (Lisa)

Others described negative feelings, when they had been informed about prenatal diagnostics, and Victoria said with a cracking voice:

My impression when I had the genetic counseling was that they discouraged me not to have any more children ´the normal way`.

Some expressed that it would even been easier if their second child also had hemophilia so that they could treat all their children the same way.

Discussion

The aim in this article was to better understand how women experience being a carrier of hemophilia over time. This study shows that the women's experiences change across their life span. Feelings of guilt and self-blame for passing on the mutation to their sons were common for most of the participants. These results are consistent with earlier published literature on carriers of genetic disorders (James et al. 2006; Lewis et al. 2011; McConkie-Rosell et al. 2000; von der Lippe et al. 2016). Experiences of not feeling good enough for ones' partner - or his family - which were present in our findings may enhance feelings of guilt. A few women, however, did not have feelings of guilt. These women had sons who did not have severe symptoms of the disease, and did not have challenges with the treatment. Thus these women's statements suggest therefore that there is a link between the physical experience of the medical manifestations in the son with hemophilia and feelings of guilt.

Knowledge about carriers' tendency to feel guilty about passing the mutation on to a child has been known for many years. It is therefore important to note that so many of the participants in this study still had feelings of guilt, despite having had genetic counseling. One reason could be that this issue is not addressed properly in a genetic counseling session (Paul et al. 2015), or that it is addressed at a time in life when the women do not reflect upon it, or because it is not relevant to them at that point in life. Another reason could be that feelings of guilt have a strong foundation in generational knowledge of hemophilia, as the women in the present study also talked about how their own feelings echoed their mothers', fathers' and grandmothers' feelings of guilt and shame. This pattern supports earlier research that shows that the psychosocial aspect of genetic conditions goes beyond the personal level as it is also a family matter (Featherstone et al. 2006; Lehmann et al. 2011; Sobel and Cowan 2000). Issues on personal feelings of guilt, and how guilt runs through generations should be addressed in genetic counseling. Even though carriers believe they are prepared for having a son with hemophilia, feelings of sadness, shock and guilt when the son is diagnosed is common. Health professionals and people close to the mothers should be aware of these reactions and be prepared to provide support. If a mother expresses sadness or guilt, either verbally or non-verbally, one should not confirm the emotion by telling her that you understand very well why she experiences these feelings. Such a strategy may only enhance feelings of sorrow and guilt (Frich et al. 2007). Alternatively one may use one's authority to normalize such feelings by telling the patient that you know that it can be difficult sometimes and that in your experiences other mothers have felt the same.

Carrying the mutation may not just create feelings of guilt, but may create a feeling of internal family connection, that is "being one of us" (Duncan et al. 2008), as described by several in our study. For those who were the only carrier in their family, not being able to "share the blame" created feelings of loneliness and overwhelming responsibility for being a carrier and passing on the mutation to a son.

From another study about reproductive decision making in women known to be carriers of X-linked conditions, it is shown that the women feel responsible to inform partners of the reproductive implications of being a carrier early in the relationship, and to carefully consider reproductive decisions (Kay and Kingston 2002). In our study women also described the importance of informing their partners about the carrier status, and most of the women had experienced support from their partner in this regard. Positive support from a partner, as most of the participants in our study had, seems to be an important factor in mitigating negative feelings about carrier status (Kaneko et al. 2015).

The timing of when the women in this study learned that they were a carrier differed. Some knew about hemophilia from a family member, and subsequently about their carrier status before they had a son with hemophilia, while others learned about their carrier status after a son was diagnosed with hemophilia. Our results show that learning one is a carrier as a child or youth does not make a big difference at that point, but this changes abruptly once the carrier is planning to have a child, or when she is pregnant. This should be kept in mind when some argue that parents should be allowed to test their children for carrier status before the child is old enough to decide (Vears et al. 2016). Carrier testing of children and young women for X-linked diseases may not influence the young women's lives (Jarvinen et al. 2000). Later in life however, when planning to start a family, and especially when planning to have children, being a carrier has a consequence mainly related to telling a partner and the risk of having a son with hemophilia. Carrier testing as a child or youth may prevent the women from seeking genetic counseling when needed. Obligate carriers and at-risk carriers should be offered genetic counseling for carrier testing, with a focus on the emotional consequences of being a carrier, when the timing is right for them.

As such our analysis indicates that learning about a carrier status does not seem to be "a telling moment" (Bamberg 2006). Being a carrier seems more about a process that develops over time, the same way any diagnosis may be seen as a process (Jutel and Nettleton 2011) based on an assemblage of cues and clues involving a particular context (Locock et al. 2016). Inheritance is part of ordinary talk, but there is often misconceptions and lack of knowledge in lay understanding of genetics (Chapple et al. 1995). Perceived stigma may influence reproductive decisions (Clarke 2016), and may lead to the moral question as expressed by one mother – is it right to get a child with a serious disease when one can avoid it? The fact that the women face these moral dilemmas is an important fact to be aware of when counseling the women.

Another important finding is that even though the women who knew they were carriers thought themselves prepared to have a son with hemophilia, the realities of their son's diagnosis triggered shock and sadness. Knowledge about being a carrier does not seem to prevent feelings of sadness, and seems be fully understood only when the consequences of being a hemophilia carrier become relevant (Gregory et al. 2007). In the genomic era that characterizes this century, newborn screening (Timmermans and Buchbinder 2010), and predictive gene testing (de Vries-Kragt 1998) may create patients-in-waiting, living between sickness and health, as for example young people testing positive for Huntington disease. Carrier testing of hemophilia, and perhaps other X-linked disorders, may create "mothers-in-waiting" contemplating the risk of having a sick child or not. It is important that information about how experiences of being a hemophilia carrier changes over time is highlighted in the communication process of genetic counseling.

Study Limitations and Strengths

There is no registry of carriers of hemophilia in Norway. We tried to randomly invite women of different ages and from different parts of Norway to participate. We also included five women who contacted the first author directly. The selection of participants may not be representative for all carrier mothers and another limitation is that only ethnic Norwegian mothers participated. However, we recruited a sample with varied age, and women who had different experiences of being a carrier of hemophilia. The researchers involved in this study have different backgrounds in healthcare and social science, which we consider a methodological strength.

Practice Implications

Health professionals - especially clinical geneticists and genetic counselors - carriers, families and patient organizations need to be aware that experiences of being a hemophilia carrier change with different stages of life, and carriers may need genetic counseling more than once during their lifetime. Feelings of guilt are common, and run through generations of the family. This needs to be addressed in genetic counseling. Even though carriers believe they are prepared for having a son with hemophilia, feelings of sadness, shock and guilt when the son is diagnosed are common. Health professionals and people close to the mothers should support those in need on this.

Research Recommendations

Research on how carriers of other disorders experience being a carrier over time, may provide novel insights into issues of gender associated with the diagnosis.

Conclusion

Carriers of hemophilia may need genetic counselling more than once during their lifespan. Even though women claim to understand the consequences of being a carrier at a young age, our findings showed that this understanding changes over time. The women may not be prepared for the shock of having a son with hemophilia and the feelings of guilt it may create. Health personnel, especially genetic counsellors and clinical geneticists, need to be aware of this and facilitate follow-ups.

Hem. A; hemophilia A, Hem. B; hemophilia B, PD; Prenatal diagnostics.

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Compliance with Ethical Standards

Conflict of Interest Charlotte von der Lippe, Jan C. Frich, Anna Harris and Kari Nyheim Solbrække declare that they have no conflicts of interest.

Human studies and informed consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

Animal studies No animal studies were carried out by the authors for this article.

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