**“I’m Healthy, It’s Not Going To Be Me”: Exploring Experiences of Carriers
Identified Through a Population Reproductive Genetic Carrier Screening Panel
in Australia**

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Advancing genetic testing technologies mean that population-based carrier screening for multiple inherited conditions is now available. As the number of genetic conditions being screened increases, there is a need for research into how people experience these screening programs. This research aimed to explore how women experience simultaneous carrier screening for three inherited conditions: cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome (FXS). A qualitative approach was adopted using in-depth semi-structured interviews to explore the experiences of ten female participants: five SMA carriers, three CF carriers and two FXS premutation carriers. Eight participants were pregnant when

offered screening by their general practitioner or obstetrician and the decision to have screening was described as straightforward. Participants reported experiencing emotional responses such as anxiety and stress whilst waiting for either their partner's carrier screen result (CF or SMA carriers) or the pregnancy's CVS result (FXS carriers) and sought additional information about the relevant condition during this time. Most participants were in favor of population carrier screening for these conditions, preferably prior to conception. Genetic counselors played an essential role in supporting couples after they received a carrier result given the variable consent processes undertaken when screening was offered. Further research should focus on the development of reliable online information tailored to people receiving carrier results and strategies for raising awareness of the availability of population carrier screening within the community.

Key words: population carrier screening; fragile X syndrome; cystic fibrosis; spinal muscular atrophy; community genetics; qualitative research

INTRODUCTION

Advancing genetic knowledge and technologies have led to the identification of deleterious gene mutations for many inherited conditions, and have provided reliable and efficient ways to test individuals to ascertain carrier status of certain genetic conditions. Population carrier screening programs offer genetic screening to individuals who are not considered to be at an increased risk of being a carrier of a particular genetic condition(s), as they do not have a known family history. Ideally conditions screened for at a population level should be relatively common, with a high carrier frequency, have cost-effective and accurate tests available with good detection rates, and be serious conditions with a significant impact on the affected individual and their family (Edwards et al., 2015; Godard et al., 2003; Sanderson et

al., 2005). Furthermore, population carrier screening programs should be voluntary, with sufficient information about the test being provided before screening, and test results (particularly carrier results) should be followed by genetic counseling (Godard et al., 2003; Sanderson et al., 2005). Carrier screening enables informed reproductive choice for couples identified as being at increased risk of having a child with a particular genetic condition, where reproductive options include prenatal diagnosis, pre-implantation genetic diagnosis (PGD), adoption, and in vitro fertilization (IVF) using a donor egg, sperm or embryo, or using the information to assist in preparing for the possibility of a child with a genetic condition.

Recent developments in genetic testing technologies enable a move from screening for one genetic condition to screening for multiple conditions at one time point (Bell et al., 2011) providing the opportunity for cost effective and time efficient carrier screening. Carrier screening panels are currently available in many countries already, including the USA and UK, through companies like 'Counsyl' and 'this is my:' (Counsyl Inc., 2015; this is my:, 2015).

In Victoria, Australia, population-based carrier screening for CF has been available (for a fee) for approximately 10 years through Victorian Clinical Genetics Services (VCGS). The program was implemented in 2006, after three governing American organizations publically endorsed CF screening (Archibald et al., 2014; Ioannou et al., 2010). Evaluation of the program determined that people appreciated the offer of testing, those identified as carriers used the information during their reproductive decision making and experienced similar anxiety levels to those who received a low risk result. In late 2012, this program expanded to include carrier screening for SMA and FXS and was named the Reproductive Genetic Carrier Screening (RGCS) program. These conditions were added to the program because

they were considered severe, there were reliable tests available with good detection rates and research supports offering screening for these conditions (Archibald et al., 2009; Prior et al., 2010). As laboratories are now able to offer panel screening with multi-condition reproductive screening programs becoming available, the need for continued evaluation of these programs, particularly the impact on individuals who undergo screening through these programs, is paramount (Edwards et al., 2015).

Here we present the findings of a study exploring the experiences of women identified as carriers of CF, FXS and SMA through the VCGS RGCS program. As this study is the first step in evaluating a newly expanded population carrier screening program, we took an in-depth exploratory approach to learning how women who were identified as a carrier for one of the three conditions experienced the entire carrier screening process.

MATERIALS AND METHODS

Ethics Approval

Approval to recruit participants for this study was granted by the Human Research Ethics Committee (HREC) of the Royal Children's Hospital (Melbourne, Victoria).

Study Setting

The RGCS is currently offered through private obstetricians (OBs) and general practitioners (GPs) at a cost of \$385AUD to the patient. The program involves a two-step approach to screening, whereby the female undergoes screening first, and if she is identified as a carrier of CF or SMA, her partner is then offered screening for the relevant condition (see Figure 1). If she is identified as a carrier of a fragile X premutation, due to the condition's X-linked inheritance, there is no need for her partner to undergo screening; however if pregnant, the couple is offered prenatal diagnosis. All identified carriers and their partners are offered genetic counseling.

Study Design

This qualitative study adopted a narrative approach. Narrative inquiry allows the participant to tell the story of their subjective experience of a phenomenon, which in turn enables them to reinterpret and make further meaning of their experience (Gilbert, 2002; Liamputtong, 2009). This approach allowed an in-depth exploration of how women experienced the carrier screening process from their own point of view (Riessman, 2008). This framework particularly suited the study as little is known about the in-depth subjective experiences of women who undergo reproductive carrier screening for multiple genetic conditions.

Recruitment of Participants

Participants were women 18 years of age or older who speak and read English, all had received a carrier result through the VCGS RGCS program and had completed genetic counseling where any partner or pregnancy testing had been finalized at least two weeks prior to recruitment to the study. As the VCGS RGCS program had only been offered for a short period at the time of this study, no carrier couples had been identified. Purposive sampling was used to recruit participants who had received differing test results and had varying test experiences. Recruitment continued until carriers of all three conditions had been recruited and until data analysis revealed no new themes were emerging, suggesting data had reached saturation.

Data Collection

Semi-structured interviews were conducted, as this enabled an in-depth exploration of participants' experiences and interpretations of the carrier screening process. An interview guide was used which included questions exploring some pre-determined topics, including how they experienced each phase of the screening program and

their feelings towards population carrier screening programs, however the interviews were flexible so they could be guided by the participants' responses and accounts of their experience. Telephone interviews were conducted for consistency and convenience for the participants. Interviews were between 17 and 64 minutes in length, were digitally recorded, transcribed verbatim, de-identified, and participants were assigned a pseudonym.

Data analysis

Interview transcripts were analyzed using narrative analysis in order to allow the subjective experience of each participant to emerge. This process involved immersion in the data by reading and re-reading the transcripts (Clandinin et al., 2000). The transcripts were coded noting particular events or interactions, characters and their role within the experience, turning points and major impacts (Gilbert, 2002). To gain a broad overview of each experience, the transcripts and codes were arranged into a timeline (Gilbert, 2002). Each timeline was then further coded and key elements of the participants' experiences were identified by finding similar codes across the participants' stories. The transcripts and timelines were then co-coded by another researcher independently to ensure the data analysis was rigorous. Data analysis in this study was facilitated by QSR NVivo 10 software (QSR International Pty Ltd, Melbourne, Victoria, Australia).

RESULTS

Ten out of 30 women eligible to participate consented to take part in an interview: five SMA carriers, three CF carriers and two carriers of FXS premutations. All partners of the women who were carriers of CF and SMA received a low risk result. One of the two women who were carriers of a FXS premutation was pregnant at the time of screening, and she subsequently underwent a CVS procedure revealing an

unaffected pregnancy. Eight of the ten participants were pregnant at the time of screening (one IVF pregnancy), and all but one were initially offered screening by their OB, the other by their GP. Participants' ages ranged from 28 to 37 years and all were tertiary educated. All interviews took place by telephone between April 2013 and February 2015 and occurred between one and three months after the participant had completed the genetic counseling process.

Six main steps or elements of the participants' experiences were observed: the offer of the test, the decision making process, waiting for partner's test results or prenatal diagnostic test results, seeking further information, communicating with family members and feelings toward population screening.

“It was up to us whether we did it”: experiencing the offer of carrier screening

When being offered the screen, participants recalled receiving written information such as a flyer or pamphlet, or had a conversation with their OB/GP:

“[the obstetrician discussed] the pros and cons of doing it and what the test results would mean...I got a lot of written information to go home and read...”
[Sally, carrier of CF]

When asked about the level of information they were provided with prior to testing, participants felt comfortable with the information they received as they didn't anticipate an adverse outcome through screening:

“I guess it gave me everything that I needed...we didn't really feel the need to get down in to a level of detail about each of the conditions because we couldn't really relate and didn't really think there was going to be...a large chance of us having to deal with that...”
[Georgia, carrier of FXS]

Participants recalled being offered the test in a fairly casual manner and it was common for participants to recollect that testing was presented as an option or a choice:

“[The obstetrician wasn't] forceful either direction, [they]...said it was an option and it was up to us whether we did it.”

[Tina, carrier of SMA]

Participants were asked how they felt about being offered a screen for three different genetic conditions, with responses varying from not giving it much thought, to feeling the screen was more 'worthwhile':

"Firstly I was surprised that that was out on the market, but secondly I didn't expect that one blood test would be able to test for three different...genetic issues...I was happy about that, I thought, well that's good, that's almost like killing three birds with one stone (laughs)."

[Karen, carrier of SMA]

"Yep, let's do it": reflecting on a straightforward decision-making process

When reflecting on the decision-making process and discussing their reasons for choosing to have screening, participants valued the opportunity to learn their carrier status and described the decision as 'quick', 'easy' and straightforward:

"...we both like to know what's ahead for us and be informed so I think for both of us it was just a 'yep let's do it'..."

[Courtney, carrier of CF]

For some participants, the genetic carrier screen was performed in conjunction with other blood tests, including 1st trimester combined screening. This was perceived to be a convenient process:

"...when we were looking at the test that we needed to do for our 12 week scan...[the doctor] just talked through the fact that I could have [the carrier] test done at the same time...so wouldn't require two rounds of blood tests, so based on that we decided that it wasn't going to be a huge...inconvenience..."

[Chloe, carrier of CF]

In addition to convenience and valuing the opportunity to be informed, prior experience with compromised health was a factor that played a role in motivating some participants to consider screening:

"I was happy to participate...also the fact that my partner's cousin has a mental disability [so] we wanted to make sure that we weren't carrying anything..."

[Maria, carrier of FXS]

"It was just an anxious time": experiencing emotional difficulty whilst waiting

for their partner's result or prenatal diagnostic test results

Receiving a carrier result came as a surprise to all participants. Some participants reflected that because they had no family history they were not expecting a carrier result:

"...neither of us had any sort of family history or anything that would have indicated there could have been a problem...I guess I was a bit shocked actually, at the time [of receiving the result]..."

[Julia, carrier of SMA]

All participants had contact with a genetic counselor regarding their carrier result; in seven cases the carrier result was disclosed by a genetic counselor, whereas the remaining women were given the result by their OB and spoke with a genetic counselor afterwards. All participants reflected on their interaction with the genetic counselor as positive:

"When I got the call from [the genetic counselor], she was extremely informative and I got a really good grasp of what it actually is and what my chances were of, or what the baby's chances were of actually having a full mutation of the condition...we probably sat on the phone for 40 to 45 minutes or so as she explained to me what it actually was, so that was extremely helpful..."

[Georgia, carrier of FXS]

Although initially perceived as an 'easy' decision accompanied by a low sense of urgency, after receiving their test result participants recalled feeling a sense of urgency around arranging their partner's testing or the prenatal diagnostic testing. Participants were very appreciative of having this test arranged as soon as possible:

"I'm a do-er so I was really happy that [the genetic counselor] organized the test the next morning, 'cos I think it would have been a long night had [they] said I can't test your husband until next week..."

[Karen, carrier of SMA]

The most challenging aspect of this experience appeared to be the time between receiving their carrier result and waiting for the result of their partner's carrier test or the prenatal diagnostic test results. Participants who were pregnant at

the time of testing discussed varying emotional responses and spoke of feelings of anxiety and stress during this time:

"I guess it was just an anxious time 'cos...I was through almost the fourth month of my pregnancy...there's not much I can do about it...so it was like a whole week waiting (laughs), then I couldn't sleep the whole week."
[Sandra, carrier of SMA]

Participants described their thought processes during this stressful time, mentioning thinking ahead and trying to mentally prepare themselves for the worst case scenario. One participant in particular, who had undergone a termination of pregnancy the previous year due to a fetal abnormality, reflected on how challenging this period was for her:

"I tried to not even think that I was pregnant for that week 'cos I tried to get myself mentally prepared for going through what I went through last year...just to make it easier on myself...I was trying to...not even think about the baby as much and not talk about it as much in that week..."
[Grace, carrier of SMA]

Some participants reflected that the carrier result had been particularly challenging in that it had compelled them to contemplate the prospect of termination of pregnancy depending on the outcome of further testing:

"...at this stage, I was viewing this pregnancy as temporary, like a temporarily maybe kind of thing, I didn't want to commit to the pregnancy...like I didn't want to look [at] any baby clothes, I didn't want to start planning for the future, or like planning too seriously for this baby because I didn't know whether or not we were going to have it..."
[Georgia, carrier of FXS]

"It was scary to get a [carrier] result because then we had to...have the discussion of well what happens if my husband came up positive and we would then decide what to do, that was obviously really scary to...have to consider abortion.....I was well and truly attached and so the thought of having to abort was really devastating."
[Sally, carrier of CF]

Reflecting on their supports during this time, participants articulated that they felt they could call the genetic counselor back at any time, however none did. All participants who felt they needed support received it from existing supports such as

their partner, family, friends or work colleagues. Almost all participants discussed the role and approach of their partner as 'level-headed', highly supportive and calming:

"I mean he was worried and anxious but he was trying to be positive...which sort of calmed me down and like not to worry about it too much.....my partner was...just saying that he's not a carrier, he'll save the day (laughs)."
[Sandra, carrier of SMA]

Participants appreciated having access to genetic counseling as part of the screening program, with most commenting on the genetic counselor's expertise in their ability to answer questions, be reassuring and empathic:

"...[they] help[ed] make a difficult process as easy as it can be...incredibly empathetic and understanding...and...reassuring as well...[they were] vital in me not completely losing it..."
[Courtney, carrier of CF]

"Googling": seeking information online

Most participants commented that they appreciated the level of detailed information provided by the genetic counselor when they received their result:

"I really appreciated the amount of information that was given with the results, so it wasn't just you know this was the result and you need to come in and have a chat...it was probably sort of a twenty to half an hour conversation of ...the sort of different complexities of fragile X and how you can receive it and what the likelihood is of children with the condition and stuff like that..."
[Maria, carrier of FXS]

However, the majority of participants also talked about researching online or 'Googling' for further information about the condition and its prognosis after receiving their test result:

"So I ended up going home and jumping on YouTube...and watched a few video clips of what it's actually like to live with a child who has the fully mutated fragile X gene...and seeing that, I like...seeing that I kind of thought oh goodness this isn't just a small disability, this is severe..."
[Georgia, carrier of FXS]

One SMA carrier discussed being given basic information about SMA and struggling to find reliable sources on the Internet:

"We were told the basics and I suppose people just go and Google but...it might be an idea for future if there...are any good websites that we could be recommended?..."

[Grace, carrier of SMA]

When asked how the information they found online made them feel, some participants spoke of feeling worse or more concerned:

"Probably worse...I guess sometimes no knowledge isn't good, but too much knowledge isn't good either..."

[Tina, carrier of SMA]

"The potential impact for family members": communicating with family

When asked about speaking with family, the sense of urgency felt previously when organising their partner to undergo testing had dissipated for most:

"It certainly made me think about the potential impact for family members and sort of discussing it with them...it was something that I had to kind of think about."

[Julia, carrier of SMA]

"I told my Mum...as soon as I heard, it wasn't a blame thing but it was like 'well I got it off you or Dad' (laughs).....I didn't want to tell my family anything because...I wanted to make sure our baby was ok first...so once I found out that everything was ok...we told everyone in our family..."

[Grace, carrier of SMA]

Some participants indicated they had thought deeply about the best way to approach cascade testing:

"I've spoken with my parents... about...either of them being tested...probably my Dad because I do have two cousins on his side ...around my age...so I have spoken to Dad about him being tested, if he comes back negative then it's...come down my Mum's side and it stops kind of ..."

[Chloe, carrier of CF]

"Ultimately it comes down to the individual": feelings toward carrier screening

When asked about their thoughts on genetic screening being offered to the entire population, the majority of participants felt that genetic screening should be offered to everyone, rather than a select few and that it should be offered as a choice:

"It's a woman or a couple's right to find out this information if it's available to them...I think it's fine as long as everyone realizes it's a choice and no one's

judged for the choice that they make.....but at the end of the day I think that people should be informed and have the option..."

[Courtney, carrier of CF]

A few participants discussed their concerns with the cost of the test resulting in inequitable access to screening:

"I guess my concerns might be that...it's costly so that sort of cuts down the availability to younger people and people that don't have stable jobs and those sort of things, so accessibility would be something that I would be like, oh you know, but again maybe if it's offered more widely the government might subsidize or something..."

[Maria, carrier of FXS]

All participants were asked when they felt was the best time for women and couples to be offered this test. The vast majority preferred that the test be offered prior to conception:

"It definitely made it more stressful for us, because we were already pregnant...I think...had we had the test done...a year ago.....I think we still probably would have gone through similar emotions but not with the same gravity..."

[Chloe, carrier of CF]

DISCUSSION

This study is one of the first to take an in-depth approach to explore attitudes of carriers identified through a reproductive genetic carrier screening panel, and is an important step in evaluating the potential impacts of these panel screening programs as they become more common globally. The findings demonstrate that women appreciated the convenience of being able to undergo screening for three inherited conditions simultaneously. Furthermore, population-based carrier screening was widely supported by participants provided screening is presented as a choice, and, similar to other studies, the best time to offer such screening to women and couples was considered to be in the preconception period as opposed to prenatally (Archibald et al., 2013; Ioannou et al., 2010). The difficulty in obtaining truly informed consent prior to testing was highlighted by the participants' shock in receiving a

carrier result especially in the absence of a family history, and the observed need to search for further information online. The women in this study valued the opportunity for genetic counseling and it was evident that more direction and availability to reliable information after receiving a carrier result was required.

Ensuring informed consent is achieved is an important process for any genetic test, and includes: that the test is offered as a choice, adequate information is given, residual risk is outlined, and potential implications for family members are disclosed (Edwards et al., 2015; McQueen, 2002). When it comes to screening for multiple conditions simultaneously compared to a single condition, the knowledge levels of those undergoing screening has been shown to decrease (Ioannou, Massie, Lewis, et al., 2010). All of the study participants were offered screening through a non-genetics health professional, and when offered during pregnancy, the test seemed to be presented at the same time as other prenatal testing options. These factors coupled together indicate that optimal informed consent is difficult to achieve in this setting. This poses a challenge for the development and implementation of population genetic carrier screening programs where resources are limited. Additionally, genetic screening and testing is becoming increasingly accessible by being offered in mainstream settings (Martin et al., 2009). This raises questions about what level of informed consent is sufficient prior to population-based genetic carrier testing.

Whilst most participants appreciated the level of information they received from their genetic counselor, the internet was the primary resource used by women and their partners to obtain further information about their result. Given the increased use of the Internet for health related inquiries, there is potential for women/couples to become misinformed or experience psychological harm if accessing unreliable or

obstructive web pages (Case et al., 2004; Gerber et al., 2001). To ensure accurate information is sought and therefore assist with coping (Shiloh et al., 1997), it is important that within genetic carrier screening programs women/couples are either provided with or guided to trustworthy sources of information to meet their needs. This is an important consideration for the introduction of programs offering carrier screening panels to the population.

Anxiety was experienced whilst either waiting for partner's carrier screening result or the pregnancy's diagnostic test result. The main concern for these participants was the prospect of having to consider a termination of pregnancy, which for some prompted them to distance themselves emotionally from the pregnancy as a way to cope during this period. Based on the participants' recollection of their experience, there did not appear to be a difference between the anxiety and subsequent dissociation from their pregnancy experienced by women who awaited their partner's SMA or CF test results compared with the woman who underwent a CVS for FXS. This was an interesting observation, as it seemed the women who were identified as a carrier for an autosomal recessive condition mentally prepared themselves for their worst case scenario of having an affected pregnancy. The potential for difference in psychological impacts on women and couples who receive carrier results for conditions with varied inheritance patterns is an important consideration as reproductive carrier screening panels expand. As with other studies, most of the women in this study felt their anxiety subsided after receiving test results determining the pregnancy was considered low risk for the condition (Levenkron et al., 1997; Mennie et al., 1993). For this reason, all but one participant felt this screening test was best offered prior to pregnancy, which is in line with other studies (Archibald et al., 2013; Ioannou et al., 2010; Metcalfe et al., 2008).

Testing prior to conception allows more reproductive options as well as time to make choices. However, there are difficulties with attempting to offer population carrier screening prior to conception, including that women who are not planning a pregnancy are less interested in carrier screening and that not all pregnancies are planned (Miedzybrodzka et al., 1995; Poppelaars et al., 2003; Stark et al., 2013). Building awareness around carrier screening within the population could act to provide more autonomy for women and couples choosing to take up the offer of the test, as they have the choice to undergo testing at the time most suited to them. Furthermore, increasing awareness around carrier screening in the population can improve the general public's genetic knowledge and therefore the ability to obtain informed consent prior to undergoing screening.

Amidst the issues experienced by the women in the study, including receiving varying levels of information prior to screening and experiencing anxiety after receiving their carrier result, from their narratives it was clear that the women in this study valued the availability of genetic counseling. Given the varying informed consent processes and that many women undergo screening during pregnancy, it is crucial that genetic counselors are involved in expanded population carrier screening programs, to help provide support when an adverse result is received.

An outcome of carrier screening, other than providing reproductive choices, is that it can provide information for other family members who may also be at risk of being a carrier for an inherited condition. Once participants in this study were given a combined low risk for their pregnancy, the crisis and sense of urgency resolved, and although all participants discussed the need to speak to family members about the familial implications of their carrier status, the urgency had dissipated. Studies show that the uptake of cascade testing in at-risk family members after screening is low

(McClaren et al., 2010; Ormond et al., 2003), which can be due to passive non-disclosure (Hodgson et al., 2013). Given the increased chance of receiving a carrier result, population-based panel carrier screening programs need to consider the best approach to facilitating and encouraging the sharing of information regarding the carrier risk within the family, at an appropriate time in a woman/couple's life when they can shift their focus to this task. We suggest consideration of introducing an agreed intervention with the women/couple either after the pregnancy has been identified as low risk and/or in a year's time, when the woman/couple are no longer pregnant (Hodgson et al., 2014).

This exploratory qualitative study aimed to identify the experiences of a group of women who had received a carrier result through a novel reproductive genetic carrier screening program, and therefore results are not generalizable, nevertheless they establish a foundation for further evaluation of carrier screening experiences on a larger scale. It should also be noted that all participants in this study could afford the testing fee and had achieved a tertiary level education, which likely reflects a higher socio-economic group, however this is representative of the demographic of women currently undergoing screening through this particular screening program. Finally, half of the participants were employed in the healthcare sector, which might be a reflection of the types of people who choose to take up the offer to participate in research within the healthcare setting.

To help guide and develop expanded population-based carrier screening programs for multiple conditions, studies need to be conducted to determine knowledge retention, informed consent, community attitudes, potential harms, as well as the impact of receiving a carrier result for a condition for which screening may not have been routine within the general population previously. In addition, it will

be important to identify both the outcomes and in-depth experiences of couples who have both received a carrier result for the same condition. Further research is also needed to explore health professionals' knowledge and attitudes towards expanded population carrier screening programs, particularly the health professionals involved in offering screening to their clients.

As technology advances and the number of genetic conditions being screened increases, we need to ensure that public awareness of carrier screening is improved to enable couples the choice to undergo screening prior to pregnancy and to allow for more equitable access to screening. Due to the difficulty in obtaining informed consent prior to multi-condition population carrier screening, as well as the anxiety and distress women/couples can experience after receiving a carrier result in an emotionally heightened period of their lives, we have established that genetic counselors play a crucial role in these screening programs. As population-based carrier screening programs continue to expand, there is a need for continued evaluation in order to set a foundation and framework for optimal service delivery in the future.

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DECLARATION

The authors have no conflicts of interest to declare.

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Figure I - VCGS RGCS program process v2 - Copy .

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