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Commentary

The joys of fragile X: Understanding the strengths of fragile X and delivering a diagnosis in a helpful, holistic way

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Abstract

The purpose of this article is to provide a positive framework for a doctor, geneticist, genetic counsellor or other professional to deliver a fragile X syndrome diagnosis and to offer support thereafter. Our aim is not to glamourise the condition nor downplay its impacts but rather to suggest that a more accurate and holistic definition of fragile X syndrome would cover all the aspects of the condition, including its variability and positive aspects. Fragile X syndrome is commonly described in terms of the 'problems' associated with it and stigmatic language is used. Instead, we believe that giving diagnosis and support in a holistic and family-centred way is imperative. In delivering a fragile X syndrome diagnosis, the strengths of those living with fragile X syndrome should be the starting point, encouraging development that builds on those strengths while supporting the areas of need. This article will set out these more positive aspects of fragile X syndrome to assist those providing a diagnosis of fragile X syndrome.

Lay abstract

The purpose of this article is to provide a positive framework for a doctor, geneticist, genetic counsellor or other professional to deliver a fragile X syndrome (FXS) diagnosis and offer support. Our aim is to give a more accurate and holistic definition of FXS. This would cover all the aspects of the condition, including its variability and positive aspects. FXS is commonly described in terms of the 'problems' associated with it and stigmatic language is used. Instead, we believe that giving diagnosis and support in a holistic and family-centred way is important. In delivering a FXS diagnosis, the strengths of those living with FXS should be the starting point. These strengths should be built on at the same time as supporting areas of need.

Keywords

fragile X syndrome, fragile X premutation-associated conditions, holistic care, person-centred care, newborn screening, intellectual disability

Introduction

When people receive a diagnosis of a condition, they will want to know what it 'is'. 'I have condition Z: so what?' If they do not gain a full description of the condition from their doctor, they will seek it online, sometimes with detrimental effects and anxiety, if the information available is imprecise or inaccurate. The delivery of the diagnosis from the doctor and the material online introduces the individual, their family and friends to the new condition (Gratton et al., 2016). This is likely to set the agenda for how it is viewed and the allegedly correct response to it. The way the condition is described to people is, therefore,

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fundamental. It will impact how they understand themselves and how others perceive them. It is, therefore, crucial that when professionals deliver a diagnosis, they convey the positive aspects of a condition, alongside the areas where support might be beneficial.

In this article, we will look at how fragile X syndrome (FXS) is described in the medical literature, how this affects people's perception of the condition and how a diagnosis can be best conveyed. The lessons learned from this analysis can be important in the context of many other conditions and influence how genetic diagnosis is generally delivered. In the case of FXS, the average age of diagnosis is 36 months for boys (Bailey et al., 2009, please see a later detailed section on diagnosis for girls), but there is a high degree of variability in this experience, both in terms of who delivers the diagnosis, when and how (Bailey et al., 2000, 2003; Goodwin et al., 2015). Negative experiences at diagnosis have an impact on parents and their later likelihood of disclosing diagnosis to others (Raspa et al., 2016), including their children (Metcalfe et al., 2008). Therefore, the cascading effects of poorly delivered diagnoses have multiple important ramifications that we wish to redress. We will highlight three points in particular.

The first is that FXS is typically described in 'negative' terms in academic and public medical literature. The emphasis is on how a person with FXS may differ from 'a normal person' with the implicit assumption that all these atypicalities are inherently 'wrong' or 'a deficit' (e.g. Hagerman & Hagerman, 2021; Skomorowsky, 2022). The strengths observed in those with FXS as highlighted by parents and caregivers (Raising Children, 2021), as well as an emerging scientific literature (e.g. Ellis et al., 2023; Hoffmann, 2022) go unmentioned. We argue that the literature therefore fails to provide a holistic picture of those with FXS. We will go on to outline the positive attributes of FXS: our aim is not to glamourise the condition nor downplay its impacts but rather to suggest that a more accurate definition would cover all the aspects of the condition, including the strengths and the variability, and not just outline the 'negatives'.

The second is that the language used to describe FXS, and other conditions, is often stigmatic. It is striking how some medical professionals do not question offensive terminology which they would never use outside a medical context and, one suspects, they would never wish to be used about themselves.

Third, the issues raised in this article have significance around disability and echo many of the critiques of the concept made by disability scholars. The focus on the so-called 'negative' aspects of a condition is based on a norm of a perfect body and departures from it are seen as undesirable. This emphasises the way people with disabilities are different (they are 'othered'), rather that highlighting common humanity.

We then address the importance of how a diagnosis of FXS is delivered (Goodwin et al., 2015). Multiple lessons from how to respectfully and responsibly disclose the diagnoses of other conditions, such as Down syndrome (DS), should be drawn into good practice for FXS, as negative biases by disclosing clinicians have long-lasting negative effects (Meredith et al., 2024; Skotko, Kishnani et al., 2009). Sadly, parents continue to report that even for a very well-known condition such as DS, clinicians disclosing diagnoses continue to focus on negatives alone (Artal et al., 2024). Diagnosis can be a positive experience, bringing understanding and enlightenment of symptoms that have been apparent to the person with FXS and their family perhaps for a long time. However, if a FXS diagnosis is only conveyed in negative terms, it can be a harmful and inaccurate process. It does not give a full picture of life with FXS and ignores the strengths and positive contributions that those living with FXS bring to those around them and to wider society.

Descriptions of FXS

The descriptions of FXS tend to focus on symptoms and needs, rather than on presenting a full picture of the variability of the condition, including the strengths people with FXS may exhibit. In a recent book about fragile X mental retardation 1 (FMR1) premutation carriers (Skomorowsky, 2022), the author gives an overview of FXS, which includes the following:

> Physically boys with full mutation Fragile X Syndrome have poor muscle tone, loose joints, and anomalies of their connective tissues. They tend to have large protruding ears, crowded teeth, and long narrow faces…They are prone to ear infections, gastric reflux and loose stools…. Nearly all fragile X men have large testes…. Fragile X Syndrome is the most common single-gene form of autism and intellectual disability. Intellectually, most boys with Fragile X Syndrome have moderate intellectual disability…. About one-third of girls with fragile X have mild to moderate intellectual deficits.

The description includes a detailed discussion of many negative impacts of the condition. This is all the more surprising, given that the author writes in her book warmly of those children with FXS that she has met.

Another example of a description of FXS that fails to refer to any positive aspects of the condition is provided by the Cambridge Community Services National Health Service (NHS) Trust:

> Fragile X is a genetic condition that may cause mild or moderate degrees of learning or intellectual difficulties. Children may have a longer shaped face with features

> such as slightly larger ears or more flexible fingers from low muscle tone. Children with Fragile X are sometimes more likely to have seizures, autistic features or hyperactivity.

A widely acknowledged paediatrician working in the field of FXS for decades explains it as follows:

> What is fragile X syndrome and what causes it? Fragile X syndrome (FXS) is an X-linked neurodevelopmental disorder and the leading heritable form of intellectual disability. Although the defining clinical feature is mild to severe cognitive impairment beginning in infancy, syndromic features include: social avoidance and anxiety; aggression and outburst behaviours, particularly in males; autism; and physical features of large/prominent ears, macroorchidism, high-arched palate, and tissue laxity. (Hagerman & Hagerman, 2021)

These descriptions, written by renowned experts in the field, focus on symptoms and needs, without a single positive characteristic mentioned. The absence of including strengths and variability is remarkable. Contrast this description: Children with Fragile X syndrome often have a strong sense of humour and enjoy having fun, which can help them overcome their social anxieties. And they're often really good at things like:

- learning visually using pictures or computers
- remembering things like songs, movies or sports events
- undertaking practical, relevant tasks
- doing essential daily tasks
- being compassionate, helpful and friendly (Raising Children, 2021).

Or this from a mother with a child with FXS:

> I realised that Michael has brought me an enormous amount of happiness and taught me many things. That thought is opposite of the normal way of thinking in society about people with special needs. Society tends to refer to them as a burden. And for sure, sometimes it is not easy, but a burden? My son has so many special talents; you only have to look to see them. And as society, we can really benefit from people like Michael, and a lot more than everyone thinks. (The Fragile X Society, 2018)

Why are medical experts describing FXS narrowly, without including the strengths and variability of this condition over the lifespan? Of course, it is important that individuals and families are informed of the challenges that those with FXS face and the interventions that may be appropriate. However, a balanced view is needed, whereby both positive aspects and areas where families may need support are set out together (Johnson et al., 2024).

Why are the positive aspects omitted?

Few parents of a child with FXS would describe the condition solely in the way medical professionals do above. So, why are the positive aspects of FXS omitted?

Medical literature, practice and diagnosis are dominated by the medical model (Farre & Rapley, 2017), a process of classifying a clinical problem (usually an illness or disease), in order to inform decisions around care to 'manage' or 'cure' the patient's problem based on explanatory models derived from research of the mechanisms that explain the symptom (Huda, 2019). Arguably, the medical model is well suited to diagnoses of disease and/or illness that are 'temporary' and treatable, as well as those viewed as being 'independent of the patient' (Seidlein & Salloch, 2019). It is understandable and useful that such conditions are categorised as a problem to be removed by medical intervention or treatment, so that the person 'recovers'. However, the use of a medical model framework pertains that a diagnosis of FXS will focus only on descriptions of the 'problems' associated with the condition. As discussed in the next section, we argue that this is problematic for a condition such as FXS, which is not only a lifelong diagnosis with no 'cure' but also a diagnosis that is closely intertwined with a person's sense of self and identity (Plesa-Skwerer et al., 2004). A likely unintended but crucial consequence of presentation of those with FXS in simply negative terms/with no mentions of the positive aspects is not only a limited and unidimensional representation of those with FXS, but one that has been reported by other parents of people with genetic syndromes such as DS to be unbalanced, hurtful and overwhelming (Artal et al., 2024; Skotko, Capone et al., 2009).

Of course, FXS is complex and associated with some symptoms, for example, epilepsy (Berry-Kravis, 2002), which do need identification and medical treatment. However, most research in FXS likely follows the medical model (whether intentional or not), thereby focusing on describing those areas of challenge in order to develop solutions to ameliorate them. Medical diagnoses and descriptions should be based on current research evidence base (Balogh et al., 2015), so if the medical literature focuses solely on challenges, the assumption is that this will filter down into descriptions and delivery of a FXS diagnosis to individuals and families. Therefore, what is needed now is more creative research on recognising the real benefits of building on the strengths evidenced by people living with FXS, so that the complexity of this community is accurately reflected during the delivery of a diagnosis.

The impact of the focus on negative aspects of FXS

As previously mentioned, for conditions which are permanent and have a significant lifelong impact on the person such as FXS, the condition becomes closely connected

with their identity. Evidence from autism suggests that the way in which a diagnosis is disclosed to parents has a crucial impact on them (Reed & Osborne, 2012), and by reflection, on their children's experience of diagnosis (Crane et al., 2019, 2021). Evidence from other groups, such as those with Williams and Prader-Willi syndromes, indicates that a person's genetic syndrome often forms part of their self-concept that is shaped not by their own perceptions of their condition but crucially, also from the reactions of other people towards them (Plesa-Skwerer et al., 2004). This may also be true for other genetic syndromes, such as those with FXS.

As such, unlike conditions such as cancer, often perceived as an insidious disease (Robb et al., 2014) that one must battle to survive (Smit et al., 2019), FXS may be seen as part of who they are. This, again, makes the lack of balance between the strengths and areas of challenge particularly hurtful because the line between describing a person and describing a condition disappears. Stigmatic language about the condition becomes stigmatic language about the person. Negative descriptions of the condition become negative descriptions of the person, which may inadvertently shape how an individual views themselves (Plesa-Skwerer et al., 2004).

In order to help a patient flourish, the natural approach of many medical professionals is to identify and address 'problems'. However, there is an unexplored opportunity to build on the strengths and develop the potential of individuals with FXS. Our responsibility as a society is to offer the same chance to each individual. If we want to build an inclusive environment, we should start considering what individuals with FXS bring to society instead of limiting them to what they cannot achieve.

The focus on negative aspects also carries the danger that it overlooks the wide spectrum of FXS. Many of the descriptions of FXS mentioned above are featured in some but not all people with FXS. It is crucial that people with FXS are recognised as unique individuals with their own particular personality. As advised by Bottema-Beutel et al. (2021), we avoid the use of medicalised language and suggest the use of 'increased likelihood' instead of, for example, 'increased risk'. Although these recommendations are for autism researchers, we argue that it is also appropriate to apply it when discussing those with FXS, as both diagnoses and their associated characteristics are likely to an important component of an individual's sense of self (Jaarsma & Welin, 2012; Plesa-Skwerer et al., 2004). The descriptions of people with FXS should be careful to specify that as a group, they have an increased likelihood of showing the associated characteristics, but this does not mean that every person with FXS will definitely show the same strengths and challenges. Identifying common characteristics associated is a helpful starting point to guide assessment and support for people with FXS. However, this common starting point should not be used to discount the need for individualised, person-centred approaches.

A further difficulty in focussing on the 'problems' of FXS is that it can misrepresent the difficulties as static in time, overlooking the possibilities of adaption. Much longitudinal work involving children with FXS and their families has shown that even areas that are at higher likelihood of presenting early challenges (e.g. focusing attention) on average improve over time (Scerif et al., 2012; Tonnsen et al., 2015).

Time can be significant in another way too. A characteristic that can be seen as negative if present for a short term can be seen as neutral or positive if long term or permanent. Indeed, many personality traits are neutral, it just depends on the context and they each have pros and cons. For example, being orderly has its advantages and disadvantages! Autistic features such as a strong will or need to have things in a routine order might be considered problematic at first; however, in the course of life, proper scheduling or an orderly environment can indeed be highly advantageous.

The inadvertent focus away from adaptation perhaps comes from the different models and traditions with which professionals approach disability. Following a medical model, the disability is located within the body of the individual and seen as a problem which needs to be overcome. While under the social model the challenges different people's bodies face is a result of soceity's failure to provide accommodations and wider social structures (Hogan, 2019). So, for example, under a medical model, we might see that the need to use a wheelchair is negative and the person's mobility ability needs to be 'fixed' or 'improved' by a provision of equipment. But if we are to take the social model of disability, we may see the wheelchair as a positive way of changing someone's immediate environment, that is, they are now less disabled by their environment (Beaudry, 2020; Hogan, 2019; Robinson & Herring, 2024). Indeed, the person may discover all kinds of new activities they can only do with a wheelchair. Furthermore, the social model emphasises the need for the environment surrounding the wheelchair user to be adapted to facilitate access and thereby remove barriers to achieving the same goals as non-wheelchair users. Therefore, the harm in certain conditions might mostly lie in the eye of the beholder or in the period of adaptation. For lifelong conditions, adaptation is a positive as it is accepting of who one is, with all social justice rights and aims to be happy and fully recognised. This kind of approach can apply not only to physical matters but also issues such as neurodivergence (Woods, 2017). These all highlight the importance of social and familiar support as key to the experience of a person with a condition such as FXS. Their life experience will depend very much on this wider social and environmental context.

Another side to this is the adaptation of family members. Parents might see some traits as negative initially and then,

after adaptation, come to see them as neutral or positive. This might be so partially because they change their sense of identity as parents or partially because they change their minds on their perception of certain traits.

The point here is that the aspects of FXS cannot be entirely divided up into 'negative' or 'positive' aspects because they can be perceived differently, often neutrally, and are variable across people and throughout the life course. Whether attributes are lifelong or not, or adapted to or not, these characteristics can affect a medical professional's assessment. It is important, therefore, that when clinicians explain the aspects of FXS they acknowledge that these can be viewed in a negative and positive way and attitudes towards them can change over time. What might at first be an irritation can become a charming characteristic. What at first might be seen as challenging, can become part of routine life.

The positive aspects of fragile X

Earlier, we gave some mainstream descriptions of FXS, in terms of its 'negative' aspects. In this section, we set out the positive attributes derived by an emerging research literature, as well as coauthors (KJ, EW and JR) experiences of enabling services to families via their roles in national and international support organisations. In areas where quantitative research has lagged behind, we provide verbatim quotations from lived experience to evidence qualitative themes, as it is the best practice in rigorous qualitative research (Braun & Clarke, 2006). We do not engage in formal thematic analysis here, but hope that the theme of strengths below will inspire future quantitative and qualitative research. Some of these strengths are straightforward goods making people with FXS enormously enjoyable company. Others can be brought out if society was able to provide them with the right tools and inclusive environment (Nasen, 2022).

Long-term memory. Research has shown that many people with FXS have good strengths in long-term memory, including visual (Bennetto & Pennington, 1996) and verbal memory. They often have a wide vocabulary that exceeds expectations beyond their general cognitive ability (Hoffmann, 2022) and are readily able to learn by heart everything concerning things they feel passionate about. This can give them valuable skills in activities and jobs that require good memorisation; such as acting and journalism (especially in sport), foreign languages, and jobs requiring facial recognition skills, for example, in museums, cinemas, restaurants and schools. Of note, functional academic skills such as reading and writing are a prerequisite for these occupations, and they can, in turn, benefit from strong long-term memory as a strategic support for learning how to read and write. However, reading and writing vary greatly across adults with FXS, particularly among females, with some achieving high functional competence in reading and writing and others struggling (Raspa et al., 2018). In turn, this variation, and its impact on career prospects and independent living, emphasises the need to leverage strengths (e.g., long-term memory) as well as focusing on weaknesses (e.g. phonological awareness), to support functional literacy (Adlof et al., 2018), as it has been argued for DS (Fletcher & Buckley, 2002).

Honesty. Many families have reported to us that most individuals living with FXS are unable to lie (Johnson et al., 2024). They are so honest that if they do something wrong, they will readily confess. They require time to understand and integrate rules, but once they have, they will neither forget, nor surpass them. They appreciate clear guidelines and are eager to please.

Humour and imitation skills. All the coauthors can attest that those living with FXS are known for possessing a great sense of humour, a characteristic that has been commented on by other experts (Yankowitz, 2022) and parents. One mother says her child's spontaneity creates funny situations where they can surprise their audience by making simple observations that highlight the irony of a moment or a hidden meaning, visible only to those who know how to enjoy every moment of life (Weight, 2021). She goes on to say that there is no hidden agenda in their reactions; they are not trying to please or get their own way. She feels that such honesty and spontaneity serve as a reminder to those living without FXS about the essentials in life that that are often invisible to neurotypicality. These spontaneous and in the moment reactions may build on strengths in observing and imitating others. Indeed, research suggests that many toddlers with FXS have good imitation skills (Rogers et al., 2003). Parents see this in day-to-day life due to their children's fabulous ability to mimic others; often borrowing expressions from things they have heard in the past.

Sociability. Families report that their loved ones with FXS are friendly, sociable and empathetic (Fragile X Association of Australia, 2019.). However, many medical summaries of sociability in people with FXS (e.g. Centers for Disease Control and Prevention, n.d.) often paint a picture of low levels of social interest by focusing on their avoidance of eye contact and high levels of social anxiety. These summaries likely reflect an overreliance by the research literature on standardized assessments and measures of sociability that lack precision and fail to capture the complexity of both the strengths and challenges experienced by people with FXS (Ellis et al., 2023). More fine-grained and in-depth methods have better illustrated the many social strengths shown by people with FXS (Ellis et al., 2023). For example, research that uses in-depth behavioural observations of males with FXS

across a variety of social situations has shown that they display similar levels of social motivation to people with other conditions who are characterised by high levels of sociability, such as DS (Crawford et al., 2019). This is in keeping with descriptions highlighting that people living with FXS show behaviours indicative of their willingness or desire to interact with others despite experiencing high levels social anxiety (Crawford et al., 2020). The 'fragile X handshake' describes how individuals living with FXS display a wish to initiate social interaction by approaching a social partner and offering a handshake while simultaneously avoiding eye (Cornish et al., 2008), perhaps demonstrating the push and pull between both the social anxiety and social motivation experienced by people with FXS. Our opinion is that this reflects the extraordinary amount of bravery that people living with FXS show to not just overcome their anxiety to get along with others but to also take part in research that often involves meeting new people and going to new places, which in turn has helped to capture these social profiles in the first place. The willingness to be so brave thus reveals their enthusiasm to be sociable with others. With a little patience, time and space with new people, research has shown that people with FXS will 'warm-up', demonstrated by an increase in social approach behaviours (Roberts et al., 2019) and eye contact (Hall et al., 2009) over an interaction.

Kindness and helpfulness. Despite experiencing social anxiety, people living with FXS nevertheless exhibit a desire to be helpful – a characteristics both reported by parents, experts (Yankowitz, 2022) and corroborated by research. For example, Ellis et al. (2023) demonstrated that boys with FXS do very well in helping tasks: they understood when adults were struggling to complete a task and pitched in. On a day-to-day basis, we note that they see people as they are and are not bound by any traditional classification of society. For example, individuals living with FXS often happily interact with older people and disabled people, never considering them to be of less value than younger or able bodied people (Weight, 2021).

Sensitivity to others. Individuals with FXS are sincerely interested in others. Weight (2021) reported that her son can seems to be able to read his family members' minds and feel their emotions. Eye tracking studies have demonstrated that people with FXS are sensitive to the emotions and social cues of other people. For example, males living with FXS exhibit similar implicit discrimination of facial expressions of emotion as neurotypical children and adults. This supports other research showing that boys and adult males living with FXS use visual and auditory stimuli to accurately identify emotional expressions (Simon & Finucane, 1996). In addition, despite many people with FXS showing gaze avoidance (Wall et al., 2022), eye tracking has shown that, unlike children with autism, boys with FXS follow another person's attention direction cues to an object of interest just as frequently as neurotypical children (Ellis et al., 2024).

Eye tracking research has supported observations that carers have reported to national and international support organisations of their child's social interest in other people. For example, allocation of visual attention across naturalistic social scenes was similar between males with FXS and neurotypical children (Bouras et al., 2003), and after an initial brief period avoiding looking to people in naturalistic scenes, boys and girls with FXS returned to looking to those social elements within scene as neurotypical children and young people (Guy et al., 2020). These studies do not conform to the oversimplified representation of people with FXS being 'socially impaired' as is so often described in the medical literature (Crawford et al., 2017).

'Human metronome'. Parents have mentioned to authors that those living with FXS are not able to follow the often-hectic rhythm of modern society's pace of life, and so keep a slower, more humane pace. They cannot be rushed – their inability to go as fast as society requires us to go saves them from rushing and wasting their life, always running faster to catch impossible superficial dreams of consumerism. Trying to live life at their rhythm, enjoying every moment, taking time to appreciate things, allowing ourselves to rest and get bored, sometimes is precious for human ecology. This point is slightly different to the others mentioned above because (unlike, e.g. humour) it is not necessarily a positive under dominant values. Rather, it challenges the dominant values which focus on speed and efficiency. This aspect requires further research to establish its connection with FXS.

Stigmatic terminology

The second theme we wish to explore is the use of stigmatic terminology (Boysen et al., 2023). Until 2022, the name of the gene causing FXS was offensively called *FMR1*. Fortunately, the name was changed in 2022 to *Fragile X messenger ribonucleoprotein 1* (HGNC, 2024; OMIM, 2022; UniProt, 2024) with the welcome removal of offensive and derogatory terminology, which referred to only a single aspect of FXS (Herring et al., 2022). This gene and protein name change, which will take a while to permeate through the research ecosystem, have already had a profound impact on how FXS is perceived.

Despite this, references to retardation and the now archaic terminology persist globally following the welcome change of the *FMR1* gene name in 2022 by the HUGO Gene Nomenclature Committee and other protein databases (HGNC, 2024). Hopefully, as the new name becomes more widely published, the offensive terminology will be abandoned (Mousavi et al., 2020). As Lenti et al. (2020) explained,

> Stigmatisation can be defined as the identification of negative attributes that distinguish a person as different and worthy of separation from the group, often leading to the person losing social status and facing discrimination. Social stigma is a complex and continually evolving phenomenon. It includes one's own perception, the internalisation of negative behaviours held by other subjects, and the implementation of behaviours by other subjects.

When the *FMR1* gene name was first introduced, it was not widely regarded as offensive, although now the term retardation is widely accepted as stigmatic (Herring et al., 2022). Nick Marcelino, objecting to the use of the word 'retarded' in relation to his sister who had DS, said: 'What you call people is how you treat them. If we change the words, maybe it will be the start of a new attitude towards people with disabilities' (whitehouse.gov, 2010).

In a study in 2007, identifying the worse words to describe those with disabilities, the word 'retard' was seen as the top one (BBC n.d.). An international campaign has been operating for many years now to stop the use of the words 'retard' and 'retarded' (Spread the Word, 2022), a campaign supported by the Special Olympics. In 2013, The *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition* changed the terminology 'mental retardation' to 'intellectual disability' (Regier et al., 2013). Indeed, in the USA, 'Rosa's Law' means that as a matter of federal law, 'mental retardation' should be referred to as 'intellectual disability' for all legal purposes (Public Law 111–25, 2010). Many medical journals have declared they will no longer use the terms retardation or retard (Schalock et al., 2007). The Oxford University Press Online Dictionary says the use of the term 'retarded' to refer to someone with intellectual impairment was old fashioned and offensive. It is now rare for the term to be found outside a medical context (Norri, 2019).

In spite of all these welcome improvements, offensive terminology is still used in many countries, particularly in relation to FXS. It is not that medical professionals are unaware of the offensiveness of this terminology. A study of American doctors found that 'the majority of parents indicated that they would be upset if a physician used the term mental retardation. Some professionals reported being criticized for using the term' (Nash et al., 2012). For example, articles in 2023 appearing in *European Journal of Paediatric Neurology* (Elmazny et al., 2023), *Molecular Biology of the Cell* (Ledoux et al., 2023), *Death and Disease* (Yao et al., 2023) and *Neurochemical Journal* (Zeng & Yang, 2023) all used the offensive terminology. No doubt the authors are not intending and/or are unaware of the nature of the terminology, but international journals that are publishing these articles need to begin to reflect and consult more openly on terminology. While accepted terminology may not be consistent across contexts and countries, it is important to develop sensitivity for voices within the communities that research focuses on.

It is puzzling why medical professionals are so persistent in their use of terminology which they know to be offensive. It is hard to believe they would use such language when talking to friends in a personal capacity. In part, this may simply be unthinking use of terminology they have been taught in years gone by and/or which has been made established in textbooks. Or it may be that doctors are imagining that patients will not access the technical medical information that appears in journals and so will not be offended. That is not convincing in this age of the internet where there is ready access to academic material. In any event, the terminology appears on websites, which are clearly written for members of the public.

> The opening line from one prominent website discussing FXS stated: Fragile X syndrome…is the most common cause of inherited mental retardation… (Jewell, 2022)

During the writing of this article that terminology has been updated, but it is remarkable it existed until very recently. This may be the first message that parents might find online after receiving a diagnosis. It may be that children being told of their condition by doctors will find it carries a word they will only have heard in the context of bullying in the playground. This is not just an English-language problem. For example, in German, the word 'retardiert' translates to 'zurückgeblieben', which is again very offensive; yet, 'retardiert' is widely used in the medical sector. Sadly, even today, families and friends searching about FXS online are likely to come across the terminology of retardation.

Ableism and describing FXS

We have seen so far two striking features of much of the medical literature on FXS. First, that the condition is very often described in entirely negative terms, with no reference to the abilities of those living with FXS. Second, that stigmatic and offensive terminology is used to describe people with FXS in the medical literature, which would never be used outside that context. Indeed, it is the kind of terminology that if a teacher used in relation to a pupil would be likely to lead to serious disciplinary proceedings.

To those familiar with the critical disability studies literature, this is typical of wider problems. First, there has been much work within disability studies on the concept of ableism (Goodley, 2014). While disability studies have traditionally focused on the status of people with disabilities and discussed disadvantages suffered as a result of the differences in their bodies or minds, ableism shines the spotlight on 'the able'. As Dan Goodley (2014) writes, a

danger with disability studies is that the focus is on those with disabilities, and there is no critiquing of the concept of what it is to be able:

> When disabled people become solely objects of study, are reduced to fetishized products of professional or academic knowledge, are fixed as untroubled entities, are conceptualised only as social actors caught up in processes of oppression, then we risk limiting not only the lives of individuals fixed in this gaze but also the possibilities of the study of dis/ability.

It highlights how we assume the 'able' is the ideal. Departures from that ideal are automatically assumed to be disadvantages. Ableism opens up the initially shocking possibility that having a disability might be an advantage or might have advantages. In Andrew Solomon (2012)'s remarkable book, *Far From the Tree*, the author looks at families raising children with disabilities and describes the mixed pictures of disability. In candid discussions people with disabilities explain the challenges, as well as the joys, that disability can bring. For example, on deafness, he writes (p. 62):

> Most hearing people assume that to be deaf is to lack hearing. Many deaf people experience deafness not as an absence, but as a presence. Deafness is a culture and a life, a language and an aesthetic, a physicality and an intimacy different from all others. This culture inhabits a narrower mind-body split than the one that constrains the rest of us, because language is enmeshed with the major muscle groups not just the limited architecture of the tongue and larynx.

As he goes on to note, while initially he was considering whether through sign language a deaf person could communicate as well as a hearing person, he suddenly realised that, in fact, it may be that sign language is a more effective mode of communication than talking.

The focus in discussions on the negative aspects of FXS (and many other conditions) is a fine example of the point ableism is making. The 'norm of the able' is taken for granted and FXS as described in terms of the (inevitably negative) departures from that norm. We have already seen this in the way FXS is described in medical literature, and how diagnoses are conveyed. Another good practical example of ableism can be found in the debates around including FXS in newborn screening (Tassone, 2014). In the UK, protocols for newborn blood spot population screening require an evidence base (GOV.UK, 2018) and adherence to stringent protocols, with the most recent review deciding not to include FXS in antenatal or newborn screening programmes (GOV.UK, 2019). The newborn blood spot debate tends to be framed over whether the screening can lead to interventions, which can avoid the harms associated with the condition (GOV.UK, 2022). Little attention is paid to exploring whether early screening can lead to interventions which help bring out benefits (Landa, 2018) and build on the strengths associated with a condition, allowing a person to achieve their full potential; for example, by enabling stronger language skills with early speech therapy (Fielding-Gebhardt et al., 2020). It seems to be assumed that conditions of the kind that would be screened for only come with negative aspects (Bailey, 2021; Boardman, 2021).

Second, a constant complaint of people with disabilities is that they are seen as less than human. They are treated as invisible or as not having the same feelings or emotions as 'the abled'. Again, the use of the language 'retardation' is apparent: the entirely negative descriptions of a condition which is core to a person's identity (as described above) is hard to understand but for such negative feelings (Martinez et al., 2011). Other professionals avoid the use of retardation and if they do not they face serious professional repercussions (Howard, 2020; Mirror.co.uk, 2012).

Delivering a FXS diagnosis

The delivery of a FXS diagnosis requires care and skill. It should be remembered that, typically, the diagnostic odyssey can last years and can lead to parental ill-health with stress and anxiety. It is in that context that the news about the diagnosis is given. As already emphasised, if the condition is presented in a way which overly focuses on the challenges without emphasising the benefits, or by using stigmatic language, this can cause the person with FXS and their carer unnecessary pain.

The lived experience of many families, including several of the authors, is that the diagnosis is often given in a remote (by phone or letter), insensitive way with an undercurrent of doom and gloom. Broader research has highlighted similar experiences and better ways to deliver a diagnosis (Riggan et al., 2024). Diagnoses should preferably be delivered in person, by someone trained to support the family in processing the information. A FXS diagnosis should be holistic – it impacts the entire family (Johnson et al., 2024). For them, it is the start a lifelong journey with a person who will have unique characteristics, strengths and love them to bits.

That is why it is so important for professionals when delivering a diagnosis, to convey the positive alongside the common areas where support might be beneficial. This diagnosis should be accompanied with information on the strong network of patient associations (Fragile X International, n.d.). They have many resources to provide an accurate picture of FXS, which may include material prepared by people living with FXS. There may also be the opportunity to meet other families and people living with FXS to gain a better understanding of the spectrum

and variability of FXS across the life course. Above all, the family should be encouraged to enjoy their baby and cherish them.

The emphasis on the alleged negatives of FXS means it often goes undiagnosed. It is striking that many girls living with FXS go undiagnosed and unsupported throughout their entire lives, unless they are tested for FXS following a (typically male) diagnosis in their extended family. Girls/women are affected, some severely and others in less obvious ways (Lightbody et al., 2022). Movaghar et al. (2021) reviewed health data from 3.8 million people, showing the underdiagnosis and health inequalities evident in FXS: 'The estimated rate of underdiagnosis in women was considerably higher than in men (86.75% vs. 61.06% in Marshfield Clinic; 71.88% vs. 58.04% in UW Health)'. This failure to recognise FXS can reflect the fact that many girls/women display the positive aspects of FXS and so are not seen as 'having an issue' requiring medical attention. If FXS was understood as leading to both positive aspects and aspects which required more support, then the rates of diagnosis of girls may well increase.

Conclusion

One parent (Weight, 2021) has had this experience: 'Our specialist told us, make sure you believe in him otherwise he will feel your fears and doubts and will not try to develop himself. I have experienced since the truth of this valuable advice every day'.

Delivering a diagnosis is a key aspect of the FXS journey. It is important that it highlights the strengths of those living with FXS, as well as the areas in which extra support is required. It is also important to convey that the presentation of FXS is variable: every individual living with FXS has a unique journey, with mosaicism, gender, environmental factors and early support all being factors impacting on the experiences. The explanation given to newly diagnosed families of those living with FXS through the life course is vital: often there is an emphasis on deficits and problems, which need to be fixed through education and support treatments. The impression is given that the situation is dire and there will be one struggle after another. One author (KJ) was told there was no mainstream school in the county that would accept her daughter – a clearly untrue statement with a wholly negative perspective on a child who now flourishes, loves to cook and is studying art at university. Those with FXS can live life to the full: hold down jobs, enjoy social groups, have hobbies and travel independently.

Every person with FXS is unique, and there will always be challenges just as all of us will experience within our lives. What is missed in the medical literature, and therefore often failed to be conveyed during diagnosis, is the many joys and positive aspects associated with FXS. The distorted view that the person with FXS is simply deficient, as discussed above, leads to inequality and discrimination. A positive approach that is more inclusive and affirming would celebrate each person living with FXS along with their humour, talents, honesty and perspective right from the moment of diagnosis.

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