Cynulliad Cenedlaethol Cymru Bil Awtistiaeth (Cymru) drafft Llythyr Ymgynghori DAB45	National Assembly for Wales Draft Autism (Wales) Bill Consultation Letter DAB45
Ymateb gan | Evidence from: Fragile X Society	

Please refer to questions in the Consultation Letter.

Introduction

I am pleased to have the opportunity to respond to the consultation on the draft Autism (Wales) Bill. I am a Board Director of the Fragile X Society and the parent of a child diagnosed with Fragile X Syndrome. For clarity, this consultation response has been drafted using the perspectives and experience gained in both roles.

Background

About the Fragile X Society

The Fragile X Society was formed in 1990 by families whose children had just been diagnosed with Fragile X Syndrome. At that time there were no facilities to support and inform families about any aspect of Fragile X. The charity has now grown to consist of a team of dedicated employees and volunteers, supporting thousands of individuals and families.

- Our vision is a world where people living with Fragile X are:
- Valued, included and have their individual needs met - and that Fragile X is recognised and understood by professionals and the public.
- Not alone - that they have access to an active community of people who understand.
- Empowered - through evidence-based knowledge about Fragile X.

Due to services' and society's attitudes, a lack of awareness, and features caused by the conditions themselves, people living with Fragile X and their families face wide-ranging challenges. Families regularly tell us that generic supports and services did not understand their needs adequately, or that they felt lost in broader organisations relating to autism or learning disability. We want to change this.

About Fragile X Conditions and Autism

Fragile X Syndrome

Fragile X Syndrome (FXS) is a condition with a known genetic cause, where the FMR1 gene has expanded (or lengthened) considerably. This switches off production of an important protein (FMRP), which has functions in the brain and throughout the body. Approximately 1 in 4000 men/boys and 1 in 6000 women/girls have FXS: the condition falls within the definition for a "rare disease" in relevant EU/UK policy.

FXS is associated with a number of psychological and physical characteristics. FXS can be identified with a blood test. By contrast, autism is diagnosed by evaluating patterns of behaviour. The exact causes of autism are still being researched, but a complex range of genetic and environmental factors are likely to be involved.

Many of the behaviours associated with FXS are “autistic-like”, such as avoidance of eye contact, social withdrawal, communication difficulties and repetitive behaviours. Approximately 30-50% of people with FXS have enough of these behavioural characteristics to meet the criteria for autism diagnosis. This proportion of people with a primary diagnosis of FXS who have an autism diagnosis accounts for approximately 2-6% of all cases of autism, making FXS the most common, known single-gene cause of autism.

Though much of the behaviour in FXS is “autistic-like”, there are differences in what drives the observed behaviour. In FXS, inattention, hyperactivity and anxiety interfere with learning from or participating in social interaction, resulting in autistic-like behaviour. Autistic-like characteristics also become more prominent in people with severe learning difficulties. As a result, it is important to recognise that not everyone with FXS is considered autistic, even if behaviour looks superficially similar.

Even when autism is diagnosed with FXS, observed behaviour may be subtly different from autism more generally. For example, people diagnosed with FXS and autism tend to be more socially responsive but show more repetitive behaviours, when compared to those without an FXS diagnosis. Therefore, when an individual is diagnosed with autism as well as having FXS as a primary diagnosis, it is very important to understand their genetic condition and how this affects them.

It is important that people who have FXS get the correct diagnosis. This allows better understanding of individual needs and the best way to support them. For example, FXS is associated with particular learning styles, and understanding them can help when teaching new skills. In addition, FXS is an inherited condition and immediate and wider family members would benefit from genetic counselling if their relative is diagnosed.

Fragile X Carriers

Some people have small alterations to the FMR1 gene and are described as being carriers (or having a “premutation”). Approximately 1 in 250 women/girls and 1 in 800 men/boys are carriers.

Fragile X Carriers may not necessarily be aware that they have this status, since they are not normally offered genetic counselling until someone in their family has been diagnosed with FXS.

There is a growing realisation that for Fragile X Carriers, although the small alteration to the FMR1 gene is not enough to "switch off" the gene (as in FXS), these smaller changes are important.

Firstly, the small alteration to the FMR1 gene makes it 'unstable'. This means that it might expand further in size when it is passed to the next generation. Therefore, this can lead to future generations having FXS.

In addition, some carriers experience one of the medical conditions which are associated with carrier status. Fragile X Tremor Associated Ataxia (FXTAS) is a neurological condition which onsets later in life and some women may experience premature ovarian failure (FXPOI).

Finally, some carriers may experience cognitive and social/emotional effects. These effects may also be gender-dependent. For example, some women and girls are at higher risk for social anxiety and depression. Recent research has also suggested that some men and boys may be more likely to behave in ways consistent with the “broad autism spectrum phenotype”.

General comments on the need for legislation specifically targeting people with an autism diagnosis

That proposals for autism-specific legislation exist suggests that the current systems for education, health and social care in Wales do not consistently support people with an autism diagnosis and their families appropriately, which as discussed also includes some people who also have a primary diagnosis of FXS. We agree that this is wrong and must be addressed.

It is, however, not clear to us why autism-specific legislation is required as the mechanism to address this. If the main issues identified for autistic children, adults and their families relate to poor experiences of service provision of all kinds (e.g. assessment, accessing relevant support, support not available, etc.), we think that the Autism (Wales) Bill (“the Bill”) as drafted is unlikely to resolve these issues.

Arguably, existing systems also do not consistently support people with other neurodevelopmental conditions, learning disabilities or other medically recognised conditions appropriately at present either. Other groups may have needs that require support, though they may fail to meet the criteria to access services. Notable among these are children and adults without a specific diagnosis, those with “rare diseases” and those with conditions so rare that they are unnamed at present (“syndromes without a name” or SWAN).

It is estimated that 1 in 17 people will be diagnosed with a rare disease, so while individually the numbers of people diagnosed with one particular rare condition like FXS may be small, the number of people diagnosed with rare diseases in total is significant.

If autism-specific legislation is introduced, we are concerned that these different groups of people will need to secure their own legislation too, to ensure access to adequate services. This includes people living with Fragile X related conditions including FXS without a comorbid diagnosis of autism or ADHD, and Fragile X Carriers with unrecognised educational, health and social care needs.

It is important to also recognise that Fragile X Carriers will generally not be aware that they may have this genetic status, unless someone in their family has been diagnosed with FXS and they have chosen to have testing after genetic counselling.

We think that if existing systems of support and the legislation underpinning them worked effectively in practice for all children and adults, including those with neurodevelopmental conditions and/or learning disabilities, there would be little demand for separate diagnosis-specific legislation.

We would suggest that rather than introducing separate legislation for one particular group sharing a common diagnosis such as autism, more attention should be given to providing robust scrutiny of existing legislation and practice governing service provision. Recent examples include the introduction of new legislation for additional learning needs, new legislation governing social care and reforming mental health provision in Wales.

We would like to see robust systems to monitor service provision for all people with neurodevelopmental conditions and/or learning disabilities, including amending legislation where necessary to provide more accountability for service performance and reviewing levels of funding available for service provision more generally.

Question	Answer
01	**Definition of Autism**
02	We think that defining "autism spectrum disorder" in legislation is likely to be very difficult to achieve, for the following reasons: – The reference manuals used for behavioural diagnoses and the diagnostic criteria used are not static and will change over time (e.g. ICD 10 will soon be superseded by ICD 11, DSM 5 now available, etc.). – Behavioural diagnoses are underpinned by best practice guidance and assessment methods, which will also change over time (e.g. NICE guidelines, ADOS, ADI-R, etc.). – An emerging trend in research literature suggests that "autism" may not be best viewed as a discrete condition; rather, individuals with an autism diagnosis share similar behavioural characteristics though may also have very different etiologies, trajectories of development and support needs. This heterogeneity suggests that a "one size fits all" approach to autism service provision is not pragmatic. – Neurodevelopmental conditions are complex and with greater understanding, it is likely that definitions of conditions and their

	diagnostic categorisation will change. Experience demonstrates that both of these can change quite markedly e.g. Rett Syndrome has been removed altogether from autism diagnosis in DSM 5. As a result, even if someone is diagnosed with autism at some point in time, the diagnosis: may have been given erroneously or superseded by a differential diagnosis (e.g. genetic, learning disability, developmental language disorders, mental health diagnosis etc.); the criteria may change over time, or; an individual may otherwise no longer meet the criteria. If the needs of the person have not materially changed but their diagnosis has changed, they may no longer benefit from the additional rights conferred by this Bill. This again does not seem to be a pragmatic basis for service provision. **Power to include other neurodevelopmental disorders in this legislation** In the consultation document it states that: "[…] if Welsh Ministers believe the provisions of this Bill should be applied to people with other neurodevelopmental disorders, they would have the power to do so." We would suggest that this is not an effective way to plan services. We think that services should be made available based on need, not based on having a particular diagnosis. Accessing services based on need is already challenging for the Fragile X Society's member families, for example because of children with FXS being referred to either a "learning disability" or "ASD" pathway for provision. We think that further "gatekeeping" in services based on diagnosis should not be encouraged. Assuming that this measure to include other conditions is introduced, we observe that the Bill would no longer be autism-specific. We would suggest that "Neurodevelopmental Conditions and Learning Disabilities Bill" is perhaps a more appropriate description in these circumstances. We would also see a great danger that the unique characteristics of conditions other than those falling within an autism diagnosis, particularly those of "rare disease" conditions like FXS, would not be considered appropriately. For example, we think that children and adults with FXS would suffer "diagnostic overshadowing": services designed for people with a primary diagnosis of non-syndromic autism may not be adapted sufficiently to support the specific profiles of strengths and needs found amongst people with a primary diagnosis of FXS. A substantial number of children and adults may have "traits" of

	autism or other conditions, though perhaps not sufficient to meet diagnostic criteria for autism or any other firm clinical diagnosis. In response to a question I asked recently at the Cross-Party Autism Group, it appears that services provided by the "Integrated Autism Service" have not been funded to provide support to children and adults in this group, or to those with complex needs including autistic traits. This trend in policy is worrying, for a number of reasons. These children and adults deserve support and should not be excluded from services that may potentially help them, though they will also potentially also not be considered in this legislation. We think that they should not be "left behind" as services potentially improve for other groups named by diagnosis in this legislation. We think that "gatekeeping" in services based on diagnosis should not be reinforced via this Bill. Although we recognise the good intentions behind this, we therefore would not support this definition, even if it did include Fragile X conditions as part of the appendix. For example, a significant proportion of children may not be diagnosed with FXS until later in childhood: they will still struggle to access appropriate support until their diagnosis is confirmed, regardless of FXS being a named diagnosis in the appendix to the legislation. No doubt similar arguments can be made for children/young people and adults who are diagnosed with autism in later life, rather than in early childhood. To summarise, we think that early recognition of each individual person's needs, timely assessments and good access to adequate support throughout their life should be based on need, rather than based on a diagnosis made at a single point in time.
03	As previously described, we do not agree with the original premise that autism-specific legislation is required or desirable. If we did, however, it would make sense to include a power of direction. We would prefer to see such powers used in relation to education, health and social provision more generally for children and adults with recognised needs for support e.g. neurodevelopmental conditions, genetic conditions and learning disabilities.
04	Section 4 of the draft Bill states that relevant bodies must have regard to the autism strategy and guidance when exercising their functions. Our view, given the earlier discussion on the heterogeneous nature of autism, differential diagnoses and the widespread prevalence of "autistic traits"/"autistic-like features" in children and adults without a formal autism diagnosis suggests that this is an approach that

	potentially excludes many children and adults requiring support. Assuming that the goal is to allow any person with needs relating to neurodevelopmental traits and/or learning disability to access the services they require in a timely manner, we would suggest an alternative approach is required: – Identifying individual needs in a timely manner. – Identifying provision to meet those needs. – If the provision does not exist to meet needs, to undertake to commission it or provide a direct payment system to the individual to secure it. – Review the above periodically to make sure that needs are assessed and adequate provision is available. Similar frameworks already exist in several different bodies of legislation (e.g. SEN/ALN for education). We propose that legislative scrutiny and effort should be focused on ensuring that service funding, guidance, codes of practice, staff training etc. allow the delivery of adequate support for all children and adults who require it. We see the creation of separate autism legislation as a potential distraction from addressing current difficulties on the ground with service provision.
05 06 07	As previously described, we do not agree with the original premise that autism-specific legislation is required or desirable. If we did, however, it would make sense to include specific timescales such as these in the legislation to ensure that action is taken by the relevant public bodies in a timely manner.
08 09	We think that diagnosis should already be happening in accordance with NICE guidelines and separate legislation should not be required to implement this. If, as appears to be the case, these guidelines are not consistently followed, further investigation is required to find out why this is happening and intervention is required. For example, diagnostic services may be under resourced and unable to meet the standards. In this instance, we would suggest that putting additional targets based on NICE guidelines into place may not necessarily result in the desired changes in practice. We also observe that an assessment of education, health and social support needs does not and should not need to wait until a diagnosis is confirmed. For example, a request for assessment of Special Educational Needs can be made to a local authority by parents independent of diagnosis: it is based on the child's needs, not on a

	particular diagnosis being made. A more interesting question is why it appears that people need to have a diagnosis of autism to be confirmed in order to access the support they need. We think that this should not be happening and may perhaps be an indication of service failures and/or lack of resources to provide adequate services.
10	Again, it is not clear to us why this is required. If practice as set out in NICE guidelines etc. is not being followed routinely within service provision, then perhaps this is an indication of wider service failures requiring further action.
11	See previous answer.
12	See previous discussion in relation to service provision above.
13	Yes. It should also make clear provision to safeguard against identifying individuals, particularly those having rare conditions like FXS as a primary diagnosis.
14	There appears to be ample scope for individuals to be identified from such detailed information and we question whether this should be passing through multiple organisations. Apart from the risks of data loss and loss of privacy, with each step in the chain further from where the data are collected, the risk of error in documentation and interpretation increases. Individuals would also need to give consent each time their data are processed. We think it is not acceptable to assume that data collated for one purpose may be passed to a different organisation or processed for another purpose. In terms of the suggested types of data collected, we are not sure why the Welsh Ministers need such detailed information. We think that the Welsh Ministers need assurance that relevant good practice guidelines are being followed e.g. NICE guidelines. It is not clear why these data are required by the Welsh Ministers for that purpose. If there are concerns that, for instance, diagnosis is not being made in a timely manner or guidelines are not being followed appropriately, perhaps consideration should be given to providing a statutory code of practice to accompany the Bill.
15	We think that any data types should be specified on the face of the Bill and that this needs to make reference to individual rights to view any data collected about them. We also think that individuals should

	have a right to determine how their personal data are processed. This should include the right to withdraw consent for their data to be collected for this purpose. Any proposals made should comply with the incoming General Data Protection Regulation.
16	We are not sure why Welsh Ministers require this data. The reasons for this need to be explained further.
17	We observe that there are many "third sector" organisations "raising awareness" about ASD already. While raising awareness can be a worthy aim in itself, we would like to see activities focused on improving the lives of autistic people and their families. We would observe that the "awareness" such campaigns engender is of variable quality and does not always appear to include the whole "autism spectrum". For example, we observe that very little awareness is raised about individuals with autism and/or learning difficulties who perhaps require the greatest degree of support. This group includes the most vulnerable people who are perhaps least likely to be able to advocate for themselves. Recent "grassroots" campaigns have proved effective for this group e.g. the "7 Days of Action" campaign was launched in 2016 to raise awareness of the experiences of young people and adults diagnosed with autism and/or learning disability within assessment and treatment units (see for example https://theatuscandal.wordpress.com/, last accessed 15 April 2018). There is also very little awareness of conditions like FXS, which give rise to "autistic like" behaviour, which is subtly different even where a co-morbid diagnosis of autism is made. Perhaps what would be more useful is to focus scarce resources on better training for staff involved in all aspects of service provision, from commissioning to the front line, including real participation of autistic people and their families in training and service design. Where autistic people and families provide their input to such processes, we also think that they should receive appropriate recognition of their status as "experts by experience" and be appropriately paid for their input (to ensure that a more representative cross-section of people are able to participate). We would of course recommend expanding this approach to include all neurodevelopmental conditions, genetic conditions and learning disabilities.
18	As previously described, we do not agree with the original premise that autism-specific legislation is required or desirable. We think we

	have already addressed these questions previously in our responses. In addition, we think there may be a significant increase in children and adults seeking an autism diagnosis in order to access services they need, should this legislation be introduced. This may be an unintended and not necessarily desirable consequence of this approach to policy.
19	To summarise, while this Bill has the very best of intentions, we think that it also sets a worrying precedent. We think that services should be accessed on the basis of what each person needs, rather than on the basis of a diagnosis made at a particular point in time. A particular diagnosis should not be required to access a service and provision should be made as required for each individual to meet their needs: one size does not fit all.

Selected references and further reading on Fragile X conditions and Autism
Selected internet sources

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