The challenge of understanding autism in genetic syndromes

Imagine that two people walk into a meeting. As you observe these people you note that both walk with their eyes averted from others. You might note that they appear very similar in their mannerisms. However, if anyone asked you to say why the people were averting their eyes you might give a number of different answers. Perhaps they are shy, perhaps they are focused on an object, they could be confident but disinterested in others, or they may be motivated to interact with others but do not realise that they need to make good eye contact to initiate this. From this one observation alone it would be difficult to tell what was driving a particular behaviour and you could not be certain that both individuals were averting their eyes for the same reason.

When a person is assessed for Autism a similar issue arises for clinicians. Autism assessments usually come about because a person has some difficulties when interacting or communicating with others. In addition, the person may show repetitive behaviours. Autism is assessed by looking at the behaviours a person shows as well as some of the environmental factors that might impact on a person. Usually, if the person shows a certain number of behaviours associated with autism they fall above a cut-off for a diagnosis. However it starts to get very difficult to diagnose autism when the person has a severe intellectual disability. This is because many of the behaviours associated with autism are present in people with severe intellectual disabilities who do not have autism (i.e. difficulties communicating, or unusual social interaction skills). This means teasing this apart can be a challenge. One question that is sometimes asked during an autism assessment is 'does the person ask socially inappropriate questions or make socially inappropriate statements?' However, a high number of people with moderate to severe intellectual disabilities without autism will make social faux pars simply because they have difficulty remembering social conventions.

Knowing about someone's genetic syndrome may be important when the person is undergoing an autism assessment or their needs are being assessed. In some genetic syndromes behaviours have been observed that would be a marker for autism on a diagnostic assessment, for example, poor eye contact or a reluctance to join in social games. However, the reasons underpinning these behaviours may be different to the reasons in autism, and the pattern of behaviours may be subtly different. For example, in Cornelia de Lange syndrome and fragile X syndrome, research has shown that reluctance to join in with others and averting eye gaze is more likely to be underpinned by social anxiety than autism. Another example is children with Angelman syndrome who are more likely to show high levels of social motivation alongside high levels of stereotyped behaviour and a greater degree of disability. It may be that stereotyped behaviour and behaviour associated with a greater degree of disability could trigger a diagnosis of autism, even though that person may have a very different profile of social interactive abilities. This is not to say that people with these syndromes should not receive a diagnosis of autism as a diagnosis can be helpful for a number of reasons, and some children with these syndromes will have autism. However, it is important to be mindful of current

research when working with someone who has a genetic syndrome because the underlying reasons for why the person shows a behaviour may be different. This is important because it ultimately impacts on the type of support a person may need.