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Dyslexia: Is it genetic and what does this mean for social inclusion?

What does it mean to be told you are dyslexic?

When children or adults are given a diagnosis of dyslexia, it can be a relief because it seems to explain the difficulties they have experienced, for example in learning to read. School experiences can be painful, as children can come to feel they are not as good as others because of these difficulties. This may in turn lead to anxiety or depression, either directly or when children slip behind in school and do not achieve their potential, or are disadvantaged in relation to their chosen field of employment. A diagnosis of dyslexia early on may lead to support being put in place to prevent children slipping behind, or to help them work out how best to achieve their goals in light of their individual strengths and weaknesses.

Is dyslexia genetic and what does it mean if it is?

Sometimes people are told dyslexia is a genetic condition, and that therefore it is not their fault. Yet the assumption of a genetic cause may not reduce the feeling of blame. It has been shown that believing a severe mental health condition to be genetic can increase stigma (Angermeyer, Holzinger, Carta, and Schomerus, 2011). So perhaps educators and dyslexia testers should not be trying too hard to push a genetic explanation of dyslexia.

That, of course, ignores the very important question of what evidence there is for genetic causes. If dyslexia is clearly caused by our genes, then it would make sense to tell people this. If any condition can clearly be shown to have a genetic cause - it is often argued - there may one day be a medical treatment for it. This is the suggestion in one of the papers I review in this article. This paper (Gialluisi et al., 2016) reports a study in which the researchers looked at people's genetic material to see if there was a link between their genes and identified dyslexia-related difficulties.

Can a child's environment cause dyslexia and what does it mean if it can?

The debate about the causes of dyslexia, as with mental health difficulties, often comes down to 'nature versus nurture' – that is, either our genetic inheritance or our environment was the

main cause. This is an over-simplification. There is always interaction between the genes we are born with and the environment into which we are born. It is just the same for a plant seed that is sown in rich soil or poor soil. Even a seed with the best genes is unlikely to germinate without water.

The second paper I review here (Fuller-Thomson and Hooper, 2015) examined whether there is a link between dyslexia and childhood physical abuse. If a strong link were to be found, then perhaps there should be a check on whether children identified as dyslexic have been physically abused. However, it would be wrong to assume that children identified as dyslexic had been abused by their parents. Abuse can happen at the hands of those other than parents. Furthermore, physical abuse may only be one of several causes of dyslexia. So even if we find a strong link between dyslexia and physical abuse, that does not mean that every child with dyslexia has been physically abused. It just means it is a possibility.

What sort of genetic changes can cause problems?

Bits of repeated genetic code: You may not be very familiar with genetic research. I am not a genetic scientist and I have looked up explanations of some of the words and phrases used in articles that report genetic studies. I will explain two important phrases before going on to describe the studies and their findings. One type of genetic difference (as in difference from normal genes) is when a sequence of DNA – the material in our genes that represents our blue-print or genetic code – is repeated. This repeated sequence is called a copy number variant (CNV). The number of copies differs between people. Repeats of short sequences of DNA are in fact common and most are harmless. But not so common is when there is a very large number of repeats of the same small sequence. This happens in the DNA of people with Huntington’s disease. In this neurological disease, Macdonald et al. (1993, p. 971) found that a short sequence of genetic code was repeated many times more “than the normal range” on a particular chromosome in “all 75 disease families examined”.

Tiny changes within a gene: Another difference that can be found between the same genes in different people is a tiny change in one base pair. A base pair is a pair of molecules that are linked and form a 'rung' on the twisted 'ladder' that we are used to seeing in visual representations of DNA. A large number of base pairs forms a gene. One of a single pair can be a different molecule than normal, and this is called a single nucleotide polymorphism (SNP). These are also common and there can be many of them in any person's genes without causing any problems.

However, one of these single variations has been found to underlie sickle cell disease, in which the red blood cells are malformed. This can cause fatigue and episodes of severe pain. The SNP responsible is found in the gene that codes for haemoglobin, the oxygen-carrying molecule in red blood cells. If two copies of the faulty gene are inherited, one from each parent, sickle cell disease is the result (US Department of Health and Human Services, 2016).

Huntington's and sickle cell are two clear examples of inherited diseases. But when it comes to disorders that are what we call 'functional' – like dyslexia and mental health difficulties such as anxiety, depression and psychotic disorders – the picture is much less clear. There is no neat link as there is in these particular physical diseases. However, these kinds of genetic changes are what researchers look for. Exploring genes has become more possible due to technological advances.

Difficulty finding a genetic difference behind reading and language difficulties

The mystery of the missing genes

One study that reported fairly high heritability for dyslexia (Willcutt, Pennington, Olson and DeFries, 2007) looked at twins where one of the twins had a high level of reading difficulty compared to the rest of the population. They compared the difficulties experienced by the twin's sibling when the twins were identical and when they were non-identical (born at the same time but with a different set of genes). If dyslexia is highly heritable, one would expect high level of reading difficulty in the identical twins but not in the non-identical twins. In fact, what was found was statistically

lower but still relatively high levels of reading difficulty in the non-identical twins. These were twins reared in the same household, and so the possibility of a strong effect of their environment cannot be ruled out as contributing to reading difficulties in all cases. Willcutt et al. (2007) hardly mentioned the potential influence of the children's environment.

Willcutt et al. (2007) also point out that "60% of children with reading disability (RD) meet criteria for at least one additional diagnosis" of a mental disorder. One could speculate that genetic differences causing reading difficulties also cause mental disorders, or alternatively that some children have a poor childhood environment that leads to high anxiety and stress, and these in turn interfere with their ability to learn to read and are labelled as mental health difficulties or behaviour disorders. One such difficulty often identified in children with reading difficulties is attention-deficit hyperactivity disorder (ADHD).

Willcutt et al. (2007) did not study actual genetic material, and they concluded that there was a need for researchers to do so. They suspected that there are genes that "confer risk for more than one disorder". Studies like that of Willcutt et al. (2007) have indeed led researchers to look at children's and adults' genetic material. However, many studies have not found any clear genetic differences, or been able to find the same ones as other studies. This has led to an assumption that there are genes to be found that will explain "the missing heritability" (Gialluisi et al., 2016, p. 2). However, it is possible that heritability has been overestimated, and that further studies could make this problem disappear.

Existing studies do not agree with each other

Gialluisi et al. (2016) start their paper by discussing previous efforts to find genes linked to dyslexia. Different studies have suggested that different genes might be important. However, it seems rare for more than one study to produce the same genes as possibly linked to reading and language difficulties, except in very severe cases that are more easily identified. These severe cases might be different in nature from what we usually think of as dyslexia. Some of the studies looked at

the whole genome – that is, all of the genes in children identified as dyslexic, and children without dyslexia. One problem with this is that when so many statistical tests are carried out on so many thousands of places in the genome, it is possible to find that a difference seems to be linked to dyslexia in a study sample, but another study does not find the same, because it was just a chance finding. Many correlations – what seem like links – can be found when large numbers of statistical tests are done, where in fact there is no real link between two things of interest.

Links reported between specific genetic variations and dyslexia are weak

Most of the genetic differences that studies suggest might be causes of dyslexia are of the type where there is a single change – the SNP (see earlier explanation in “What sort of genetic changes can cause problems?”). However, many such changes are harmless, so it is important to establish how strongly linked any particular one is with dyslexia. One thing the studies show is that none of these suggested genetic differences is strongly linked to dyslexia. They are all quite weakly linked to it, according to Gialluisi et al. (2016).

This lack of evidence for SNP-type genetic difference led Gialluisi et al. (2016) to wonder whether they should look at the other main type of genetic difference – the CNV (see earlier explanation). They tell us that not many previous studies looking at CNVs have done so in relation to dyslexia. None, they report, found a perfect relationship between a genetic variant and dyslexia. There is clearly no single gene for dyslexia. If genetic differences are the cause, it is complex.

Could it be that dyslexia is due to a large number of small genetic differences?

Gialluisi et al. (2016) decided to look at the total amount of variation (CNV) in the genes of children identified as having reading and language difficulties and compared them with those in their unaffected siblings. If there is a strong genetic link between these genetic variations and reading and language difficulties, they would find a difference in the number of genetic variants in the children with difficulties compared to their siblings. Gialluisi et al. (2016) were thorough in that they also checked whether there was a correlation between the amount of genetic variation and the level of

reading and language skills in all the children. This is a more fine-grained analysis that can pick things up that the previous one might miss. There were over 600 children in the total sample. The analyses were done both with reading and language ability, and these abilities were corrected for the child's IQ, in case some children's difficulties might be due to their IQ level.

Previously suggested gene variants were not confirmed

Gialluisi et al. (2016) found no link between overall amount of gene variation and reading and language scores. When they looked at especially large areas of CNV there was one family in which two children had reading and language difficulties and one did not, and the two with difficulties had this large difference that affected several of their genes. But this was just one family out of over 600, and therefore cannot be a general explanation for dyslexia. Another, different genetic variant was found in one other family to be only present in the siblings with difficulties. However, Gialluisi et al. (2016) found no evidence that genetic variants suggested by previous research were linked to dyslexia.

Despite their various different sorts of analyses, none of these links was upheld. There appear to be a few children who may have a specific and relatively large amount of genetic difference, where these variations contribute to reading and language difficulties. However, the majority of children with these difficulties did not have any evidence of CNV-type genetic variants, and more importantly, did not have those suggested by previous research. Because Gialluisi et al. (2016) were thorough and did many different kinds of analysis, they gave themselves a good chance of finding evidence if it was there. Previous studies may not have been as careful. It is easy to find associations that are just due to chance.

Could a child's environment cause dyslexia?

There is evidence of links between trauma and learning difficulties

Fuller-Thomson and Hooper (2015) introduce the subject of environmental influences on children by explaining that there is already evidence that “children with various disabilities are 3 to 4 times more likely to be physically abused than nondisabled peers” (p. 1584). In addition, poor progress at school has been found to occur for children who have been traumatised, Fuller-Thomson and Hooper (2015) tell us, and they back this up with a long list of studies. They also mention studies that have found abuse and trauma to lead to problems in brain development for the children concerned. These neurological effects of trauma and abuse include the sort of problems that can affect reading and language skills. However, there had not been an investigation to see whether dyslexia and childhood physical abuse were linked within a large sample of members of the public.

Dyslexia and abuse in Canadian members of the public

Fuller-Thomson and Hooper (2015) used data from a national community health survey carried out in Canada. They focused on people in two states – Manitoba and Saskatchewan, and there were 13,640 people in the sample, all of whom had been interviewed in 2005. However, because of some missing data, the final sample was 12,750. Within this sample, the number of people who reported having been physically abused in their childhood was 1,020, and the number reporting having been given a diagnosis of dyslexia was 77. Fuller-Thomson and Hooper (2015) say that the number of people reporting dyslexia is much lower than would be expected, which could be due to the sample being of all ages over 18. Dyslexia testing is much more common now than in previous years, so many of the older people in the sample may have had dyslexia that was not picked up. Indeed those reporting dyslexia were on average 11 years younger than those not reporting it.

How the researchers identified dyslexia and abuse

People had been asked at interview whether they had received a diagnosis of dyslexia from a health professional, as well as questions about other diagnoses. Fuller-Thomson and Hooper

(2015) point out that this could lead to further cases of dyslexia being missed in the survey, since dyslexia is not always diagnosed by a health professional. It could be an education professional who does the test. People had been asked whether they were ever physically abused by someone close to them either during childhood or teenage years before leaving home.

People with abuse history six to seven time more likely to have dyslexia

Fuller-Thomson and Hooper (2015) report that 7.2% of people with no abuse history had dyslexia, and 34.5% of people with abuse history had dyslexia. When they adjusted for age, sex and race, people with an abuse history were found to be seven times more likely to have received a diagnosis of dyslexia compared to people without experience of abuse. It is possible that both abuse and dyslexia are more common in people who had other childhood adversity such as their parents divorcing or being out of work or having addiction problems, all of which were also enquired about in the interviews. So Fuller-Thomson corrected the analysis for these things. After this correction those who had been physically abused were still six times more likely to have had a diagnosis of dyslexia than those who had not been abused. When Fuller-Thomson and Hooper (2015) looked at the age of people, they found that as they looked at increasingly older people, there was a lower and lower likelihood of reporting a diagnosis of dyslexia, and the link with abuse was also smaller. This could be due to dyslexia being missed in older people.

Fuller-Thomson and Hooper (2015) suggest that children born with genetic differences that make it harder for them to learn could become easily frustrated, as could their parents, and both of these could lead to conflict in the family and to children being physically abused. However, it is also possible that physical abuse can cause stress and psychological trauma, and Fuller-Thomson and Hooper (2015) cite evidence for links between post-traumatic stress disorder and problems in brain development and learning. It is possible that a combination of some – perhaps weak – genetic differences combined with abuse could be joint causes of dyslexia. Whatever the cause of the reading problems, Fuller-Thomson and Hooper (2015) suggest that “children and adults with a

history of dyslexia should be screened for abuse, and those with a history of abuse should be screened for dyslexia or associated learning problems” (p. 1588).

Fuller-Thomson and Hooper (2015) note some problems with their study, including the fact that they only had self-report data, and no actual test results for dyslexia. However, they point out, as has already been mentioned, that if anything the reported rate for dyslexia was probably lower than it should be. This would weaken the ability of their study to find any link between dyslexia and abuse, so the fact that they still found a link suggests it may be even stronger than their findings suggest. They also point to previous evidence that people under-report childhood abuse, and this would have a similar weakening effect on their findings.

Conclusions

This article is not a review of the available research on genetic and environmental causes of dyslexia. It is only a review of two recent papers. However, it demonstrates some of the difficulties and problems in research on dyslexia. Children and adults may be told, on being given a diagnosis of dyslexia that it is not their fault because it is genetic. Although probably well-intended, it may be both inaccurate and unhelpful to tell people this. If they have developed dyslexia because of negative experiences in childhood, then it is still not their fault.

There may, however, be complicated issues for the person diagnosing dyslexia to deal with. They may not see it as their role to enquire about childhood abuse or neglect. But perhaps there needs to be more attention given to training in how to do just this as a matter of routine, or to refer a child or their family to an appropriate expert for further investigation. This is never an easy area to discuss, since parents can easily be – or feel - blamed when they have not done anything wrong, or sometimes when they thought they were doing the right thing but others disagree. Corporal punishment is an example of this. There should never be an automatic assumption that parents have done something wrong. Equally, however, children need the best possible environment for their

development and to reach their full potential. Adults may find themselves trapped in difficult situations, but children are much more vulnerable, especially when young.

Whatever the causes of a child's reading and language difficulties, a genetic explanation may not increase their social inclusion, and it may reduce the possibility of protection from abuse.

Attention to children's individual learning needs and to any possible abuse experience could greatly enhance their current and future inclusion.

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