Fundamentals of language Acquisition

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Week 12

Lecture 058

Lec 58: other neurodevelopmental disorders

Hello and welcome back. We are in the 12th module now, which is the 12th week, the last week of the course. So, in this module, we have been looking at the various kinds of language-related disorders. Language disorders may sometimes co-occur with other cognitive disorders; sometimes they might occur on their own. So, we have looked at and discussed specific language impairment, and we have also discussed Williams syndrome, where there is no language-related disorder as such, but there is a cognitive disorder. Now these two disorders were discussed in terms of whether language and other cognitive mechanisms can be separate to some extent, if they are separate. And so today we will look at some other disorders, some other neurodevelopmental disorders where there seems to be a relationship between language and cognitive disorders; they are not separate. So while they have certain kinds of cognitive disorders and certain kinds of physiological issues, they also have language-related delays or disorders. So that is the second part of the understanding of how far language and other cognitive mechanisms are related, as seen through disorders. So this is lecture 3; here we will talk about the second group of disorders. Now, within the neurodevelopmental disorders of various kinds, we will not be able to go through all of them, but the few that we will discuss also share this same issue, which is the issue of inbuilt heterogeneity. Now, what do we mean by in-built heterogeneity? It is that even though this category groups diverse disorders together, there is often a significant amount of overlap. Now, the overlap can be in terms of the language-related problems; they can also be in terms of other cognitive disorders. So, it is not very easy to parse together certain features which are, let us say, part of ADHD that

can also be seen in the case of ASD and so on. So, similarly each of these also show a high level of heterogeneity.

So, for example, in the case of ASD (autism spectrum disorder), not all children suffering from ASD will have the exact same kind of clinical repercussions. So, they might have different levels of linguistic delay, or they might also have different levels of social behavioral output. So, as a result, these treatments or clinical interventions require the expertise of a diverse range of professionals, like child psychiatrists, psychologists, pediatricians, speech and language therapists, occupational therapists, and so on. And then, of course, this also needs the help and support of various sectors, like health and education, together, because when we are talking about special children, the special education needs of these children, of course, require the combination of both the education and health departments. So that is what makes NDDs a very specific kind of case.

So, heterogeneity in terms of symptoms, outcomes, and treatment responses across all neurodevelopmental and psychiatric disorders is a very common hallmark of these disorders that we will discuss together. So, we will start with developmental dyslexia; it is quite commonly understood and very well studied. In fact, they are also part of popular imagination because of films and so on. So, this is an area that has been studied more than the other neurodevelopmental disorders of similar nature. Now, what is dyslexia? This is a very prevalent neurodevelopmental disorder. Now, this typical hallmark of this disorder is difficulties with accurate or fluent word recognition and spelling. These are the most important and most visible problems with respect to developmental dyslexia. These people, they generally do not have much cognitive disorders or cognitive disabilities. They have normal sensory abilities; they will have comparable or average intelligence matched with their age and mental age, as well as with chronological age controls, and so on. So, on the surface, they are like anybody else.

However, there is a severe issue with recognizing and understanding words, as well as with the recognition of spelling. So, this has been, as I just said, studied for a long time. One of the oldest references that we have goes back to 1878, where he talked about word blindness because the recognition they do not understand means they do not recognize the words as they are. So, it was initially called word blindness to describe patients who had lost the ability to read but had their oral and nonverbal intelligence intact, oral as in verbal intelligence, verbal and nonverbal IQ that was intact. So, these were the patients he reported had lost their ability. They were not born with that disability; they had lost it as a result of a stroke or some other such issues. So, in this case, he reported that his ability to read was gone. However, all other abilities were intact. The name dyslexia

came a little later. This name was given in 1884 by Rudolf Berlin. Another important name in the early studies on dyslexia is that of Pringle Morgan. He described his famous case of congenital word-blindness. So, the term that was used to denote this kind of people and this kind of disorder was word blindness. So, word blindness in the case of the first one he reported on was a patient who had become word blind. But in the case of others that they reported, most importantly Morgan, he reported a child who was not able to read; his name was Percy.

So, that is why he called it congenital word blindness. So, in the initial stages of dyslexia, it was considered to be primarily a problem of visual perception, specifically a problem with understanding something related to visual analysis and visual acuity. The reason for the disability was basically pinned on a visual processing problem that selectively targeted written words, which the visual processing problem did not affect anything else was only affected in the case of written words. So, that is when the problem happened because they could not read. Now, another problem with dyslexia is that it is often confused with or even used interchangeably with poor reading capacity, and that becomes a very difficult issue to solve because dyslexia can also be severe, less severe, mild, moderate, and so on. So, where to draw the boundary between a poor reader and a person suffering from dyslexia is also a matter of discussion in this domain. In any case, the initial diagnosis was about a visual processing problem that would selectively affect only written words, and this was considered to be hereditary. A little later in the early 1900s, Samuel Orton focused on letter and word reversal, which is a very common error among dyslexics. So, they will read, for example, 'was' and 'saw' interchangeably, as you know. So, if it was reading 'was' for the word 'saw'.

So, this is where a reversal is happening. This is something that was studied by Samuel Orton. Based on his studies, he hypothesized that the visual problem arose because of a hemispheric dominance failure in which mirror images of visual stimuli were not inhibited. So, it is a visual processing problem, and that probably came out of a hemispheric dominance failure where the mirror image could not be inhibited. So, they were looking at the word, but they were actually processing the mirror image; that is how he proposed it. Later research followed, and then the idea was that around the 1950s, after generative syntax came into existence, Chomsky's influence, generative phonology was born out of it, and the entire understanding of dyslexia changed as a result of that; hence, the visual perception angle was kind of sidelined. It was not considered important anymore because now the understanding of dyslexia was from the perspective of phonology. So, though it is kind of coming back, some understanding of visual processing and visual acuity may have something to do with dyslexia, but on a larger scale, visual perception is not the culprit; that is where we are today. So, of late, the

scientific consensus for the last several decades is that dyslexia is a language-based disorder whose primary underlying deficit involves problems in phonological processing. So, this is where we are as a result of the phonological understanding of dyslexia, which in turn goes back to the understanding of generative phonology.

So, this is later because they have this understanding that there is this particular thing missing, which is phonological awareness. Remember, we talked about phonological awareness and the understanding of mapping between the sound and the orthography, as well as phoneme-grapheme mapping. So, that part is apparently missing in these patients. So, if it is missing, this will later cause problems in processing with other kinds of languages, namely written language. So, that is where there was a shift from the visual perception angle to the phonological perspective.

So, as a result, the phonological theory of dyslexia changed the way we look at it. So, according to this theory, dyslexic children fail to learn to read because they fail to acquire the skill of separating the sounds in a word to match them with their visual letter counterparts. So, the phoneme-grapheme mapping is what they are not able to do. This is the understanding. So, the establishment and automatization of letter-sound correspondence, which happens pretty early in life, is what dyslexics do not have. So, this is, of course, what we just referred to as phonemic awareness. Now, though the majority of the data and initial studies on dyslexia come from alphabetic languages like English. It was considered like a grapheme; there is a direct grapheme-phoneme connection. So, you have the letter corresponding to some kind of sound. So, that was the initial finding.

However, later studies have also taken into account other kinds of languages like Chinese, and a similar kind of problem was reported from there as well. Now the current view is that we remember we talked about the multi-deficit versus single-deficit understanding of various kinds of disorders. So based on that, we now know that the current view on dyslexia is that a single core phonological deficit is not sufficient to explain reading difficulties. So, like any other behavioral disorder, the etiology of dyslexia is also complex and multifactorial. So, today's understanding of dyslexia is that it is multifactorial and that there are various kinds of issues that have been pointed out.

So, for example, phonological awareness, rapid serial naming, verbal short-term memory, vocabulary, and other aspects of broader oral language skills and graphomotor processing speed. So, all of them have to have been found alongside just the reading difficulty. So, children with multiple cognitive deficits are at a much higher risk of having dyslexia, and those factors are these. So, it is not only one kind, unlike the SLI, which is a specific language impairment of any specific kind of problem; here we are seeing that dyslexia can also have a multifactorial origin.

So, this is a more current view. So, starting with the very specific visual perception problem that affects only reading, from there to the phonological theory of dyslexia, where we are looking at only the grapheme and phoneme mapping problem, we now see that there is a multifactorial probable origin of dyslexia that affects many other domains. Of late, there is a renewed interest in visual deficits as well. So some recent studies have brought new understanding primarily about visual attentional deficit. So it is not about the processing deficits in the sense that it was seen before, but now it is looked at from the visual attentional deficit perspective. So in a recent study, they demonstrated that performance on a visual attention task in preschool children significantly predicted reading difficulty 2 years later.

So, similar patterns of results are reported across writing systems and with varying degrees of letter-sound relationship. So, for example, in the case of French, the mapping between sound and the letter is not that straightforward, let us say. So, comparing Italian and French, they have also found that this kind of pattern of results can hold for both types of languages. So, where the letter-sound relationship is consistent and where it is not consistent, and so on. So there seems to be a mapping between the visual attentional level and reading ability at a later stage. Of course, now we also have a lot of brain imaging studies conducted on dyslexic patients. So, we also have some understanding of the brain basis of dyslexia. Imaging studies have reported significant abnormalities in cerebral connectivity as well as in the cortical structure. So, it is not only the anatomical aspect, not only the cortical structure, but also the connectional architecture that has been found to be affected by this in the case of this kind of patients. There is quite a bit of study in a specific domain in the case of brain imaging studies; for example, the magnocellular development, which is the focus of many studies recently.

Magnocellular development is slightly different in the case of dyslexic patients as opposed to typically developing children. So, this is one. In dyslexics, the magnocellular responses are reduced in both the retina and the lateral geniculate nucleus, which is the LGN. So, in these two areas, the magnocellular structure is the easiest to explore, but it also exists in other areas of the brain. But these are the two areas where it is more clearly visible, and hence most of the data come from these two areas, and their underactivations have also been found in two posterior left hemisphere regions: one is the temporoparietal region, believed to be crucial for phonological processing and phoneme-grapheme conversion, which is expected, and then we also have the occipitotemporal region, including the so-called visual word form area.

So, these two areas have also shown underactivation, as in less than optimal activation; these two areas are very important for reading, visual word processing, and visual

language processing. So, you see under activation in this area also. So, activation in certain crucial brain regions has been found in the case of dyslexic patients, specifically the visual word form area, as well as the area that is thought to be responsible for grapheme-phoneme conversion. So, these are important findings regarding the brain basis of dyslexia. And then we also have the comorbidity that dyslexia can often co-occur with other cognitive disorders, most notably with ADHD, which is attention deficit hyperactivity disorder.

Comorbidity with ADHD and dyslexia is also present with two other language related disorders, for example, language impairment and speech sound disorders in terms of language development. So, yet another domain that is now being explored is what is called environmental genetics. This is yet another area where they basically look at the environmental impact and its relationship with the genetic structure. So, environmental genetics is another domain that is now being explored in terms of what the other factors might be for dyslexia patients, what else might be affected, and how it might be affected. So that is what we have now seen about dyslexia: though it is predominantly a language disorder, it can have comorbidity with other behavioral problems, for example, ADHD, as well as other language-related problems. Similarly, it is not just a simple problem of identifying the phonemes with respect to their sounds and visual representations; it also has a brain basis. Now, let us move on to autism spectrum disorder; this is yet another domain that has been studied quite a lot, and there is an adequate amount of literature available as well. So, ASD is characterized by social and behavioral impairments; primarily, this is a social and cognitive problem, including behavioral impairments such as verbal and nonverbal communication, social functioning, and patterns of restricted and repetitive behavior or interests. This is the definition given by the American Psychiatric Association about ASD (autism spectrum disorder).

So, this prevalence is 18.5 per 1,000 people in the general population according to some recent estimates. So, it is quite prevalent. So, the characteristic feature of ASD is the lack of social and communication skills. So, it is not only the nonverbal behaviour but also the verbal behaviour that gets affected in the case of ASD, and that is why we are discussing this here. So, autism is argued to be the result of an impairment in the domain-specific mechanism of meta-representation dedicated solely to the processing of social stimuli. So, what does it mean basically? This means there is a deficit in the theory-of-mind module. Remember when we discussed the concept of theory of mind ; we discussed that this is one of the important markers for diagnosing atypical children in certain domains. So, ASD is one such domain because it has a domain-specific problem, a domain-specific

impairment that is responsible for theory of mind development. So, other than when non-social impairments are noted, they are explained in terms of secondary effects. So, non-social impairments are also present, but they are typically explained as a secondary effect rather than the primary problem. Now, in terms of language deficits in our ASD, social communication impairment is, of course, the most telltale sign. So, where they will not be able to take part in conversations. In a conversation, we have seen that there are certain rules of conversation in any kind of language scenario, be it any language or any community. So, you have to maintain the topic; you have greetings, and all those things are part of the conversation. In the case of autistic children, those things are missing.

So, they cannot take appropriate turns; turn taking is difficult for them, they cannot maintain the topic, and they do not have greetings as part of their vocabulary, and so on. Because these skills depend on the theory of mind and because theory of mind is severely affected, language output in those domains is also severely affected. Like all other cases we have seen before, there is some sort of asymmetry between expressive vocabulary and receptive vocabulary; expressive vocabulary is always less than receptive vocabulary, and the same thing can be seen here as well. Semantics and syntax also get affected. So, there is an overall language-related delay and disorder, as you see in the case of children with ASD. However, this is variable because it is a spectrum, as we said. So, the children who fall on the other end of the spectrum, where the severity rate is high, will have all areas of language function affected severely. So, syntax, semantics, and vocabulary will be very severely affected. But if you come to the moderate range, then their language abilities may not be that severe, and in fact, there are some children who are still within the spectrum but might have good, comparably good enough language skills; that is also possible. So, it is a variable in which you see a lot of variability within the same spectrum.

The subdomain of vocabulary production and comprehension of personal pronouns, mental state terms, and prepositions is generally found to be affected. Another important domain with respect to the ASD population is the domain of executive function. The executive function mechanism is one of the higher mental functions and higher cognitive functions in humans. So, this includes shifting attention, inhibition, working memory, planning, and so on. So, basically, this is something that is part of your flexible fluid intelligence, as we call it today. So, this is what decides how you allocate attention, how you selectively attend to the goal-oriented stimuli for that particular moment, and then similarly, at the same time, you have to be open to updating your attention in terms of the different kinds of incoming stimuli. And then, of course, working memory is another part of this entire EF network and planning, of course. So many studies find that children with ASD are deficient in all of these tasks, but not all, again because it is a spectrum. So, some children actually do quite well in terms of selective attention. In fact, many children

with ASD have been found to exhibit one of the typical markers of ASD: they are able to fixate on one thing for a prolonged period of time, and repetitive behavior is a hallmark of their disorder.

So, sometimes some of these functions are actually present. So, selective attention is present, but in many cases, it is not. Often these deficits are found when the relevant task has a language component to it. So these deficits, when you have a language component to them, tend to result in worse outcomes. Studies have also reported a connection between language and EF deficiencies in children with ASD. Probably this arises out of, remember we talked about it initially when we discussed theories of language evolution and theories of language acquisition; there was this constructivism that talks about language appearing at a stage when they have completed the sensory motor stage.

So, the schemata are already in place, and language gives them a label, and what follows is something called internal dialogue. They talk to themselves, and over a period of time, that speaking to oneself—egocentric speech—becomes what is called thought. This is something that is understood to be missing in the case of these children, and that is why it is said that the connection between language and ASD exists. The connection between language and EF in ASD children is found because they do not use language skills internally to facilitate goal-directed behavior, which is a common hallmark of children. Children speak to themselves over a period of time, and that self-talk becomes silent; this is what becomes thought. Probably, autistic children do not engage in that, and that is why we see the markers that we see. So now there are certain factors responsible for ASD. Now one is of course heritability. Both ASD and SLI are considered relatively highly heritable.

We have seen that an entire family had multiple members with SLI. ASD may not be that prevalent in the family, but there are some studies that have reported heritability of it as well. So, this is the reason an older diagnosed child or a first-degree relative with a similar disorder can lead researchers to predict the same or at least take the probability into consideration for a still undiagnosed child at risk. What this means is that if there is a child in a family, the eldest child, let us say, having ASD, then the researcher can predict that another close relative or a sibling might ultimately be at risk. They may not be showing the signs of ASD yet, but they might eventually do so. So, this is one outcome of this being possible only if it is heritable, right? So, there are certain cases where this has been found to be true. Genetic encoding of NDD involves many disorders like Down syndrome, Williams syndrome, and Fragile X syndrome, which have genetic causes and

can be diagnosed before birth as well. Today, of course, we have progressed so much in medical science that they can be diagnosed before and sometimes even interventions are prescribed. However, finding genetic links to SLI and ASD is a recent development, and studies are quickly catching up on this front as well. Prior to this, these two conditions have been exclusively diagnosed based on behavioral criteria. So, only when the child is exhibiting the signs of the problem is when they were diagnosed, but now we can diagnose them before birth as well. Genetic markers for ASD, like FOXP2, are associated with SLI. ASD susceptibility loci have also been found, and there were many loci that have been identified for ASD susceptibility as well. SLI and ASD have also been found to share some genetic vulnerability factors. Several genes that are regulated by FOXP2 have been associated with ASD and SLI, including a particular gene called CNT-CNAP2; this gene is associated with both language impairment and ASD. So, FOXP2 controls certain other genetic components; one of them is this, and this has been found to be connected to the development of autism spectrum disorder as well. A recently discovered copy number variant on chromosome 16p11 is associated with a range of phenotypes, including ASD and speech and language impairment associated with ASD.

So, there appear to be some genetic markers for autism spectrum disorder as well. As we have already seen, SLI does have a genetic marker, which is the FOXP2 gene. Similarly, for ASD, some findings have pointed towards the same. Neural markers also exist for ASD. Recent trends in research, particularly with brain imaging studies, have shown atypical lateralization of the language network, which is not the case for typically developing children. So, temporal cortices were found to have reduced activation compared to TD children when listening to bedtime stories. So, not only is the lateralization level difference of the network is present, but also an activation level difference has been found with respect to children with ASD. Also, they show higher activation in the right anterior superior temporal gyrus rather than in the left. So, basically, the activation pattern is not comparable to that of typically developing neurotypical children. So, the activation, as well as some kind of atypical lateralization, has been found in terms of these children.

Also decreased spontaneous inter-hemispheric synchronization in terms of the language-related areas. So, there is some amount of, when we speak; when we engage in language-related activities, there is a spontaneous synchronization between the hemispheres. So, that is also found to be decreased in the case of ASD children. Now, this decreased synchronization are directly connected to language as well as other markers of ASD. For example, this is positively correlated with language ability and negatively correlated with the severity of ASD symptoms. Also, this is not present in comparable SLI toddlers, so this kind of problem, this decreased synchronization, is not part of SLI toddlers, which

means this factor is more connected to social communication rather than language per se. This comparison is important because in the case of SLI, they do have a language deficit, but it is not a social problem. So, this is what they find using brain mapping as well: that in the case of SLI children, this decreased synchronization is not present, which means this synchronization is an important marker for social communication rather than language-related understanding or language-related underlying capacity. So, atypical lateralization was found in infants as young as 6 to 9 months, well before other symptoms emerged. So, because of this, now you can diagnose ASD much earlier, as the time for language to evolve has not yet come. So, between 6 to 9 months is before they start speaking in words as well. So, even at that time, atypical lateralization has been found. So, that means this is as early as it gets in terms of diagnosis. Comorbidity is also present; as we have seen for many other diseases, this ASD also has comorbidity. So, because of this, treatments and interventions focus on the predominant features, and they have their own benefits because, in the case of ASD, the primary problems are social communication, social theory of mind, social behavior, and interaction with other agents in society, and so on.

So, these are the predominant features. So, even though there are co-morbidities, treatment will typically address the predominant features. However, hierarchical methods may lead to a lack of diagnosis of certain disorders in individuals with multiple neurodevelopmental problems because there are hierarchical methods. So, if you are not focusing on all of them, then you might be missing out on something. So, today's intervention methods actually take into account a multiple multi-pronged approach, not just looking at the most dominant disorder, but also looking at all the connected disorders. So, that is where we are today. So, this is where we will end lecture 3. In lecture 4, we will take up another disorder as well as some intervention methods for today. Thank you.