

# Prediction and prognosis for SLPs in the age of human genome mapping

Helen Smith and Donna Dancer

## KEYWORDS

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Helen Smith  
(top) and Donna  
Dancer

Genetic testing is now available for as little as US\$100 online. The ability to map the human genome has allowed scientists and health care professionals to identify an increasing number of genetic mutations that may influence the health outcomes of both individuals and families. More specifically for SLPs, a number of genes, and genetic variations, have been linked to some forms of communication, swallowing, and hearing disorders. The importance of the environment and an increasing number of interventions on modulating the clinical expression of genetic traits should not be underestimated. This article reflects on some ethical considerations for SLPs when discussing prediction and prognosis with clients, particularly those who may wish to pursue or have undergone human genome testing.

**W**hy should DNA testing concern speech-language pathologists (SLP) when they are dealing with the ethical considerations in prediction and prognosis? In this “Ethical conversation” we consider clients’ “diagnostic Odyssey” (Parens, 2015, p. 18) in light of recent advances in human genome testing. Through reflecting on a parent’s story of seeking diagnosis and prognosis for their child, we consider the benefits and the harms that advancing technology in the field of human genomics may bring our clients.

## What is genetics?

Genetics is the science of biological inheritance (American Speech-Language-Hearing Association [ASHA], 2015). Genomics is the study of how genes, more specifically DNA sequences, interact within an organism and the environment. The complete human genome was first mapped in 2003. This has led to successful diagnoses of some rare diseases and rare forms of other diseases (Parens, 2015). One can now have a full genome map completed, on line for as little as US\$100–200. Genetic tests are, however, not as simple as a pregnancy test to interpret (Matloff, 2015). The answers provided by DNA mapping, even for known genetically linked disorders, are

often indicators of “lifetime risk” or predisposition to a disease rather than a determination that the disorder will occur (McInerney, 2014). Similarly, the nature of the clinical expression of the disorder such as age of onset, severity, and response to treatments may all vary (Matloff, 2015). Further, environmental factors and interventions can significantly influence the functional expression of a disorder (ASHA, 2015).

Accurate diagnosis is essential to predicting the natural course of a condition. Thus, genetic testing may influence a SLPs ability to provide a prognosis and guide interventions which may in turn influence medical, behavioural and psychological outcomes (McInerney, 2014).

## Genetic testing relevant to SLPs

Medical conditions encountered by SLPs in both paediatric and adult practice may have a genetic link. Examples include:

- cancers, for example BRCA 1 and BRCA 2 genes are linked to breast, ovarian, and prostate cancer (Matloff, 2015);
- neurodegenerative disorders, for example chromosome 4p in Huntington’s disease, familial Parkinson’s Disease, some forms of Alzheimer’s dementia, and Duchene’s muscular dystrophy (Fischbeck, 2014);
- autoimmune disorders, for example JAK-STAT in lupus erythematosus (Phillips, 2015);
- intellectual disability, for example trisomy 21 in Down syndrome, fragile X (Down Syndrome Australia, 2015).

More specifically, genetic mutations have been shown to be implicated in forms of: autism (Brooks, 2015), hearing loss (ASHA, 2015; Arnos, 2001), stuttering (Chen, et al., 2015; ASHA, 2015; Han, et al., 2014; Newbury & Monaco, 2010), cleft lip and palate (ASHA, 2015), dyslexia (Chen, et al., 2015), verbal dyspraxia (Kang & Drayna, 2011; MacDermot, et al., 2005), speech sound disorders (Lewis et al., 2006; Newbury & Monaco, 2010), and specific language impairments (Newbury & Monaco, 2010; Stromswold, 2008). ASHA has recognised the importance of genetics in the practice of speech language pathology and audiology by providing a free on-line education package (ASHA, 2015).

## Receiving a diagnosis and prognosis: a family’s story

The following is a discussion had with a parent before the age of genetic testing. We explore the issues around

diagnosis and prognosis. Finally, the parent reflects on whether they would have pursued genetic testing had it been available. Amy and her parents provided the information shared in this column.

Interviewer: As a parent when were you first aware/concerned that all was not right with Amy's communication?

Parent: I was concerned right from birth as she did not cry. The perfect baby!!!! She did not demand feeds, I would walk in to check her and she would be lying awake, waiting for me to feed her. She had no voice or idea how to make a sound – I actually thought she was deaf in my ignorance of how deaf babies present.

Our child was constantly sick and on antibiotics for ear, nose and throat issues. In fact, she had her tonsils and adenoids out at 2 years because they were septic. Her ENT health issues were always attributed to her global delay by our GP. At 18 months of age, she was still not crawling (she commando crept on her first birthday), grunting and pointing to make herself understood. She was very keen to communicate and interact with the family. I finally persuaded the doctor that I did not have post-natal depression and that in fact my child had issues. The paediatrician agreed with our concerns and referred us to an early intervention centre.

Interviewer: When you first consulted with a speech pathologist how did they discuss Amy's diagnosis and prognosis? How did you feel they broke the news? Can you remember how you felt?

Parent: I remember feeling great relief that someone finally believed me that my child had communication difficulties. I don't recall discussing prognosis with the first speech pathologist. I know alarm bells started to ring with us as the other children at the early intervention clinic had various degrees of disabilities, so the reality struck that our child may have a disability, but I just assumed we would be there for a short time – this was the miracle cure and we would go on with our lives. The speech pathologist introduced us to Makaton signs almost immediately to facilitate communication and Amy took to it immediately. I remember this time as being very positive as we were finally getting some help!!! When Amy was 2 and a half we shifted from Queensland to Perth. She was referred to a disability organisation to receive her therapy services so I guess although we were never given a diagnosis/prognosis, common sense made us realise it was going to be a long haul. At this stage Amy's dad was more realistic than I was. He understood that her issues were ongoing, but as her mother I was still of the belief that all this therapy would fix her.

Interviewer: What did you think Amy's communication difficulties would mean with regards to her starting childcare/kindergarten/school?

This worried me greatly. Arriving in a new city far from family support, I wanted her to be in an environment where her communication and other issues were not seen as a barrier to her interacting with other children and being part of the program. I researched nearby day cares and kindergartens that had vacancies and willing to accept a child with communication and coordination difficulties. A day care with a kindergarten next door accepted Amy and the bonus was the

kindergarten had a speech pathologist on site attached to the kindy program. The speech pathologist worked with the children individually as well as having a great deal of input into the program.

It was this speech pathologist who first made me aware of SPEECH (WA) Inc. My husband and I joined this organisation which then started our education into understanding language disorders and being the parents of a dyspraxic child. It changed all our lives in that it gave us hope that Amy would be provided with an appropriate education. Both my husband and I became heavily involved in SPEECH (WA) Inc., being officer bearers in all capacities over a number of years.

Amy started pre-school at a Language Development Centre. Amy was supported at the LDC for the next 7 years prior to us moving to England to live. This wonderful school still caters for children with various language disorders in a small classroom setting. To my knowledge no similar school existed in Australia in the early 1990s. We feel so lucky that these professionals came into Amy's life. The foundation and development of her language and literacy that she received at this school has been one of the greatest gifts she has been given.

Upon returning to Australia, high school became an issue as there is no school in Australia like the LDC for teenage children but we all survived!!!

Interviewer: What did you think this diagnosis of dyspraxia would mean with regards to her communication with you/extended family/friends/community?

Parent: Amy has always been surrounded by a large accepting family. When we shifted to Perth she was very young and we moved into a very large accepting community, so her inability to speak clearly did not seem to inhibit her interaction with others. What it has done has inhibited friendships and the forming of relationships outside the inner accepting circle of people.

Amy is 28 years of age and still has severe expressive language difficulties. She has always been an over communicator in so far as she will not stop until you understand her. As a child she would speak, play charades to act it out, sign it, draw it and even prior to literacy, write it down. Now she speaks and if you have not understood her she writes it down or finger spells for clarity. Social media is her greatest tool. She is able to communicate with others via the internet which negates the necessity to speak.

Interviewer: As she made progress and became older, how did her therapists/yourself/and she personally change or modify your hope and expectations regarding her prognosis with regards to formal education/social options/employment options/life choices?

Parent: We don't recall ever getting a prognosis as such and we just became pragmatic about her development. In the early stages, it just became apparent that her education needed to be about becoming as self-sufficient and independent as she possibly could with work and life skills. She was keen to get a job and wanted to firstly go to open employment. We went down that path but she came

to the decision that she did not like it as much as in supported work where she had done work experience. Supported employment provides her with social interaction, a friendship base and social activities. She has attended TAFE since the age of 16 years and continues to do so. Amy now wants to leave home and live on her own which we see as a giant step in her development. With the introduction of NDIS, we're optimistic that Amy will be able to move into her own unit and have the support she needs to facilitate her independent life.

Interviewer: Were there any key milestones or points when you needed more assistance from your SLP with predicting the impact of her communication disorder on her life? If these milestones could have been managed better or differently by the SLP, how would that have looked?

Parent: I don't believe we could have done anything differently for Amy. Once the hurdle of convincing medical staff that she had a problem and getting the appropriate referrals, our speech pathology journey could not have been any different I believe. The early intervention and her signing skills were a godsend to open up communication. Her education which was language/literacy based gave her the gift of literacy. Expressive language, although not clear, continues to develop.

Interviewer: Finally, if with the magic of a time machine you could return to the beginning and have your daughter's genome sequenced would you have this done? If Amy's genome map identified a genetic link to verbal dyspraxia, and/or a specific language disorder, would this change any of your decisions/actions / expectations (positively or negatively)? Would Amy want to have genetic testing done now if she were thinking of having children?

Parent: The reality is that we would not have made decisions any differently than we have. I didn't even know what verbal dyspraxia was when I fell pregnant and a language disorder would not have changed our minds. Had genome testing been available in the 1990s we would have undertaken it with Amy around 5 years of age in an endeavour to predict the clinical course of Amy's verbal dyspraxia and general speech disorders. We think it is unlikely that this testing would have caused any change to the intervention program for Amy.

Amy will not be having any children so there will not be any reason for her to undertake this form of testing. We would not be considering undertaking this testing for Amy at this time to assist or guide any clinical intervention by a speech pathologist.

## Ethical considerations in genetics testing relevant to SLPs

With the increasing availability of pre- and post-natal and adult genetic testing, many ethical considerations arise.

### *Informed consent*

As the cost decreases and availability of genetic testing increases, the use of an appropriate consent process before testing becomes ever more important. What is known can never be unknown and this may have significant implications for the person him/herself, parents, and

families (and extended families) once genetic information is disclosed. Human genome mapping means that while a person may want a genetic test for a particular disease, the lab results may reveal a disease not even imagined (Bush & Rothenberg, 2011). When considering prenatal testing, informed consent should involve consideration of how the results of the testing may be used. People will have differing ideas about what a "good life" means for themselves and their children (Parens, 2015). Fully informed consent requires that the person seeking testing is made aware of the potential psychological burden of the information as well as the potential broader implications – for example, for employment and insurance (Arnos, 2001). With regards to prenatal testing or adult testing, before any clinical signs occur and before a test is conducted, careful consideration of the risks, benefits and limitations are essential. Amy's parents reflecting on the possibility of human genome testing would not have considered or even known to consider the implications of testing prenatally.

### *Beneficence*

Genetic testing may be viewed as beneficent in some disorders, allowing early confirmation of a diagnosis and greater predictability about the clinical course of the disorder. This may in turn relieve or reduce anxiety (Arnos, 2001; Parens, 2015). It may also facilitate timely environmental or medical intervention (Lewis et al., 2008; Matloff, 2015) and aid accurate prognosis and planning. Amy's parents reflect that at age 5, if genomic testing had provided some answers, it might have been useful in fully understanding the clinical course of Amy's communication disorder.

### *Do no harm*

Genetic tests can be poorly interpreted and incidental findings may be inadequately communicated (Matloff, 2015). The process of completing an ethical genetic test should include a skilled and knowledgeable genetic counsellor who is available for both pre- and post-testing support. A trained genetic counsellor is also able to take a detailed family history, conduct a risk assessment, provide both medical and genetic education, and access to appropriate primary prevention where appropriate (ASHA, 2015). In prenatal testing, many parents assume they are accessing health care to ensure the birth of a healthy baby. Often, however, prenatal genetic testing is to help prepare parents for a baby with a disability or the risk of a future disability (Parens, 2015). This knowledge needs to be made clear to parents as part of the consent process. Similarly, the potential impact of results not only on the individual requesting genetic testing but their wider family may need to be discussed. A plan may need to be made with the genetic counsellor and the individual being tested with regards to how results of their testing may be shared with others in an ethical manner to ensure other family members, who had not sought testing, are not overwhelmed with information they may not wish to have known. As they have not sought testing, Amy and her family have not had to deal with this consideration.

### *Conclusion*

While genomics is still in its infancy, genetic mutations have been linked to communication, swallowing, and hearing disorders. The case discussion highlights some ethical considerations around human genome testing across the life-span that are relevant to SLPs. Some SLP roles may

require SLPs to develop their basic knowledge in the area of genetics. SLPs may also need to increase their knowledge of local resources such as genetic counsellors who may assist their clients when making decisions around the choice to undergo genetic testing. Prediction and prognosis for SLPs in the age of human genome mapping may bring a brave new world of ethical challenges for their clinical consideration and reflection.

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Helen Smith is a senior member of the Speech Pathology Australia Ethics Board (member since 2008) and is manager, speech pathology, Central Adelaide Local Health Network, South Australia. Donna Dancer is member of the Speech Pathology Australia Ethics Board (Community representative) and parent to an adult daughter with dyspraxia.

Correspondence to:  
Helen Smith  
Manager, Speech Pathology  
The Queen Elizabeth Hospital and St Margaret's Rehabilitation Hospital  
Central Adelaide Local Health Network  
28 Woodville Road, Woodville South, SA 5011  
email: [helen.smith4@health.sa.gov.au](mailto:helen.smith4@health.sa.gov.au)