How can we address inequalities in access to genetic services?

Clinical Genetics is a relatively small specialty when compared to most other areas of medicine. But that is not to say, that it is unimportant. Clinical Genetic Services see and help a wide variety of people, from parents looking for genetic testing on their unborn baby to patients wishing to know their risk of cancer due to family history. They can be involved in the specialist care of those with some of the rarest conditions and help towards early detection and prevention of more common ones. In the field of medicine, there has been increasing awareness surrounding health inequalities. Access to affordable health services forms part of the social determinants of health (Figure 1) and it is therefore important to ensure everyone has equal access to healthcare (1). Health inequalities have been recognised as an important topic and have consequently been included as part of the NHS Long Term Plan as something to be addressed (2). Inequalities in access to Clinical Genetic Services results in inequitable opportunity for early detection of genetic diseases (including cancers) specialist treatment, improved survivability, and psychological support via genetic counselling. The focus of this essay will be how we can address inequalities in access to Clinical Genetics Services in the United Kingdom only, although the same principles could be applied elsewhere.

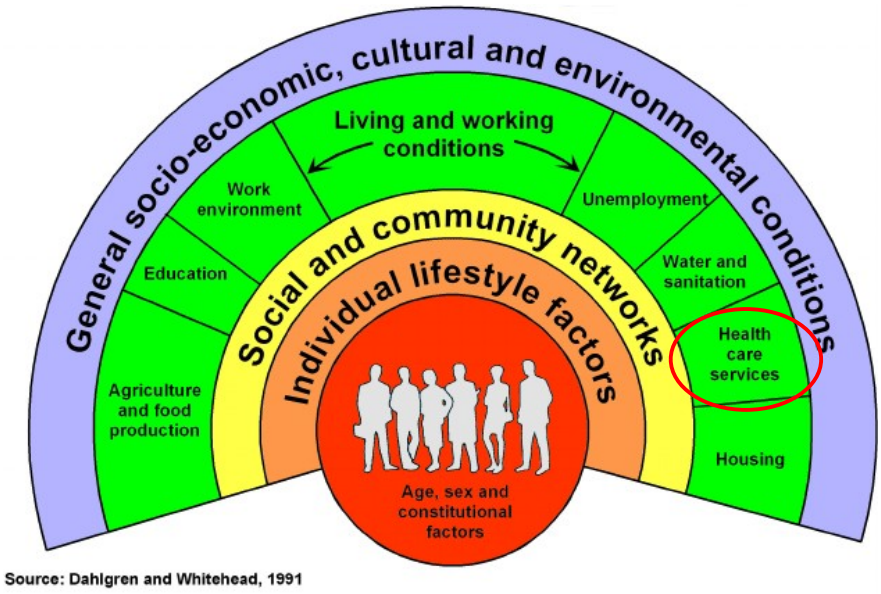

Figure 1: Social Determinants of Health, adapted to highlight healthcare services as one of its factors. Adapted from Kennedy, 2011 (3).

In the UK, there are currently 23 Clinical Genetic Services (4). The location of these services can be seen in Figure 2. Unlike larger NHS specialties such as cardiology which have services in main city hospitals as well as smaller district hospitals, Clinical Genetic Services tend only to be in main city hospitals. This can create issues with geographic accessibility as people are unable to make the long

journey, either financially or due to other responsibilities. This can be a particular issue in this field as users of the service could be severely disabled. To tackle this, these Clinical Genetic Services could provide outreach clinics is neighbouring districts or rural areas. This is an approach which Nottingham Clinical Genetic Services has taken. As well as running at their main site at Nottingham City Hospital, they hold clinics in Boston, Derby, Grantham, Lincoln, Mansfield, Skegness, and Spalding (5). Running clinics in these areas greatly decreases the distance required to travel and therefore the cost. For other Clinical Genetic departments where this might not be feasible, another option could be to subsidise transport costs to improve affordability.

Figure 2: A map demonstrating the 23 Clinical Genetic Services located across the UK. Made using Google Maps.

The usual journey a patient takes to being seen in Clinical Genetics is via a referral from their General Practitioner (GP) or a Secondary Care Physician. Less often, a Clinical Geneticist may be called to an inpatient ward to assess a patient with phenotypic abnormalities; this is usually on the neonatal intensive care unit. In recent years, there has been a surge in health promotion, which the World Health Organisation defines as “the process of enabling people to increase control over, and to improve, their health (6). Consequently, the market for Direct-to-Consumer Genomic Testing (DTCGT) has emerged. Companies such as 23andMe offer the opportunity to “see how your DNA can

affect your chances of developing certain health conditions" with their Health + Ancestry Service (7). Thus, a new patient journey is created. Some people, on receiving their DTCGT results, will consult their GP, in the hopes of a referral to Clinical Genetics to discuss their risks or receive more definitive diagnostic genetic testing. On the surface these services can seem appealing: why not be proactive and take control over your own health by finding out what you are at risk of? However, these services create a further gap in access equality. At $199 (roughly £150) this is a price that many people in the UK would be unable to afford. This access inequality contributes to a "two-tier" healthcare system whereby those of lower income must use public healthcare services and are subject to longer waits whereas those with higher income use private healthcare (8). However, there has been concern about the efficacy of these tests, as there are reports indicating high numbers of false positive results (9). The Royal College of General Practitioners have therefore recommended in their position statement that GPs do not refer patients to Clinical Genetic Services based on DTCGT alone, but rather refer them as they would anyone else: based on history, examination, family history and risk factors (10). This has helped reduce the access inequality created by these DTCGTs.

However, there is little guidance for clinicians on who should actually receive these referrals. A 2009 study from the University of Birmingham found that 50% of UK GPs felt they would not be able to recognise when referring a patient to Clinical Genetic Services would be appropriate (11). This means that there could still be disparity in the patients that are referred to Clinical Genetic Services, as someone that one doctor may refer, may not be referred by another. At secondary care level there are resources to help Clinical Geneticists find relevant information and make decisions. The online interactive book and search tool GeneReviews® gives excellent, peer reviewed articles about many genetic conditions (12). Additionally, the National Genomic Test Directory gives doctors the clinical indications for genetic testing and what specific gene panels may be requested (13). Yet there is little in the way of guidance for clinicians who are not geneticists on when or how to refer. Providing guidance, could mean that doctors have clearer understanding on when is appropriate to refer, and patients receive more equitable access to Clinical Genetics. An example of useful referral guidance that exists currently is the National Institute for Health and Care Excellence (NICE) guidance on familial breast cancer referrals (14). Furthermore, other studies show that GPs recognise their need for education on identifying patients with inheritable disease risk, taking a detailed family history, and interpreting it and recognising patterns of inheritance amongst other things such as the clinical management of genetic conditions (15). Addressing these educational needs through training days/ workshops or online training can also help bridge the gap in inequality of access as well as improve the overall quality of referrals to Clinical Genetics.

On the other hand, you could argue that another way to address inequalities in access to these services is to focus efforts into making Direct-to-Consumer Genomic Testing more affordable (ideally free!) for all. This is something that has been discussed in the media for several years and is cause of constant debate. There have also been suggestions of whole exome sequencing as a nationwide form of screening for rare conditions in newborns (16)! If there was national roll out of whole genome sequencing kits, delivered straight to patients' doors, would this not be the ultimate accessible service? However, despite these advantages, there are several potential disadvantages associated with DTCGT. For example, there is concern that people will overestimate their risk of a condition causing unnecessary anxiety, search of unnecessary treatment or changes to their health-related behaviours. On the contrary, others upon receiving their results may underestimate their risks and be falsely reassured causing them not to seek professional medical help when required. There are also more practical considerations. Who/ what will deliver the results, particularly in cases of people receiving bad news? Will the large sum of genetic data collected be kept private or be used for national research purposes to aid in the detection of more disease-causing mutations? Will this not create a huge burden on the NHS? (7) Due to this level of uncertainty and controversy, this is something that is unlikely to occur successfully for many years, if at all.

Finally, I would like to consider ethnicity and equitable access to Clinical Genetic Services. There is evidence from multiple studies that minority ethnic groups are underrepresented in specifically cancer related genetic services (17) (18) (19) . A study from 2003 of five regional Clinical Genetic Services in England found only 2.5% of users were non-white despite ~14% of the population being non-white in the same time period (20). Studies analysing the reason for this found a few potential explanations. Within South Asian and African populations, there was stigma and fear around cancer diagnosis or being at risk of cancer which prevented attendance. Additionally, some people had fatalistic viewpoints of familial cancers as they thought nothing could be done and therefore avoided testing (21). This issue could be addressed with increased awareness of genetic conditions and what can be done to help patients at Clinical Genetic Services. This could be done with posters or leaflets in the GP practices where there is high prevalence of minority ethnic patients, in a range of languages (which is discussed further later). A more innovative method could be to host webinars with local community leaders such as Imams or hold clinics in culturally significant locations to populations such as places of worship. This is something that has been done successfully in Leicester when there was found to be low uptake of the COVID-19 vaccine in minority ethnic populations (22). Another barrier discovered was language. Explaining to patients the complexities of DNA and Mendelian inheritance is difficult enough, without additional difficulty of a language barrier. Use of interpreters helps solve this, but some patients fear that interpreters choose which information to

present them and therefore influence their decision making (21). This is a difficult issue to try and solve but an idea could be to source interpreters who not only speak the same language as the patient but are of the same ethnicity or religious/ cultural background so that the patient can feel more represented, and more trust is gained. Similarly, there are visual aids Clinical Geneticists can use to aid in their explanations of genetics such as Illumina's Patient Genetic Counselling Guide which is a book with images to help explain inheritance (23). However, resources such as this have very low representation of minority ethnic patients. Representation matters, as without it people will not feel welcomed or like they belong. Adapting some of these resources, including the advertising posters and leaflets, to show more minority ethnic patients can help people feel encouraged to attend Clinical Genetic Services. There is no evidence suggesting that there are lower rates of cancer in minority ethnic populations, yet this disproportionate accessibility exists. More needs to be done to enable minority ethnic groups equitable access to Clinical Genetic Services.

To summarise, although initially the task may appear insurmountable, there are several methods to address inequalities in access to genetic services. A reasonable first step would be to collect more data on the demographics of people who attend Clinical Genetic Services. It was difficult to find any *recent* studies showing such data, but this sort of information is incredibly important to decide where changes need to be prioritised. Addressing access inequalities is something that escapes no medical speciality and so the work should not stop there. The methods we've discussed in this essay include: outreach genetic clinics; subsidised transport; DTCGT; clear guidelines for clinicians regarding referral and education for non-geneticists. Particular attention needs to be paid towards increasing accessibility for minority ethnic populations which could be done by increasing awareness of Clinical Genetic Services with advertisements, hosting events with local community leaders or clinics at community hubs, using interpreters of the same ethnicity and having greater representation.

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