THE ROLE OF ARTIFICIAL INTELLIGENCE IN GENETIC COUNSELING: A SYSTEMATIC REVIEW OF CHALLENGES, OPPORTUNITIES, AND PATIENT-CENTERED CARE

by

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With the increasing prevalence of artificial intelligence (AI) in society, it is no wonder that AI applications are starting to seep into healthcare. This thesis aims to synthesize existing literature on the patient experience with AI in various healthcare domains to assess how it may be implemented in the field of genetic counseling. The initial intention was a meta-analysis of AI in genetic counseling. However, there is a paucity of published studies in this narrow area of inquiry.Thus, a systematic search was conducted using PubMed and Web of Science for clinical studies published between 2014 and 2024 on AI and patient experience with the aim of better understanding how AI is being studied in patient-centered care domains to guide the rational application in genetic counseling. Based upon the predetermined search criteria, resulting studies that focused on the patient experience towards AI in their healthcare experience were included in this review. Given the plausibility of AI in genetic counseling, the intended goal of this systematic review is to determine common themes and challenges described in other AI enabled healthcare applications. Results showed that AI was particularly useful in diminishing clinician workload to allow for more thoughtful meetings with patients, but concerns arose about how this machine learning system would be programed and operated. While patient reticence was a common theme in earlier studies, more contemporary studies found that AI trust was less of a

concern highlighting the pace at which this technology is being deployed without study. Finally, while AI has immense promise, understanding key patient concerns and needs must be the focus of upcoming studies to make AI implementation meet the demand for complicated medical communication with the goal of providing space for enhanced patient – provider experience. Thus, utilizing the capabilities of AI in the field of genetic counseling can help to optimize patient understanding of complex health issues leading them to the ability to make better informed choices.

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Purpose

This thesis aims to explore how artificial intelligence (AI) impacts the patient experience in healthcare, with a focus on its potential applications in genetic counseling. It seeks to understand how AI can enhance accessibility, decision-making, and patient autonomy while addressing challenges like clinician workload and patient education.

Main Research Questions

1. How does AI influence the patient experience across various healthcare domains?
2. What are the benefits and limitations of using AI in patient-centered care?
3. How can AI tools be effectively integrated into genetic counseling to improve accessibility and patient outcomes?

Literature Review

Introduction

Human curiosity has driven monumental advancements in all aspects of humanity. From the discovery of fire to the introduction of artificial intelligence (AI), curiosity has been the driving factor in pushing society into the future. Over the last few decades, curiosity has turned inwards in the hopes of uncovering what makes humans, humans. Leading the way towards a deeper understanding of ourselves was the international project of the 90's, the Human Genome Project (HGP).

The HGP began on October 1, 1990, and set out to decode an entire human genome. According to the executive summary published at the time, "The Human Genome Initiative is a worldwide research effort with the goal of analyzing the structure of human DNA and determining the location of the estimated 100,000 human genes"(1990). This project took over a decade to complete and its fundamental goal was to change the way genetic information could be utilized in daily life. The hope from the project was that one complete human genome could be used as a roadmap for future healthcare applications. The idea at the time was to be able to understand and identify the genetic underpinnings of health and disease. With this knowledge, medicine could be tailored to an individual based upon sequence analysis enabling precision medicine.

While not a central goal of the project, the scientists, as expressed in their executive summary from the initiation of the project, understood that there would be specific legal and ethical implications of their work. Already by that time, Francis Collins (one of the key organizers of the HGP) had been among the first to successfully identify genetic mutation leading to disease at the molecular level with his group's publishing of the cystic fibrosis

mutation gene (Riordan, Rommens et al. 1989). Concerns began to emerge regarding the ability to determine disease risk based upon identifications at the molecular level and what types of potential would exist for social discrimination (1990).

By the completion of the HGP in 2003, as predicted, the technology evolved substantially to allow for more rapid sequencing of a human genome. Today, a human genome can be sequenced in the matter of hours for a cost under $300 (Illumina 2023). This is of course in contrast to the 13 years and billions of dollars involved in the HGP (Roberts, Davenport et al. 2001). It is this technological advance that has allowed what was once the domain of international governmental consortiums to be transitioned to the individual consumer level. With this access to advanced in-depth consequential information, the details of how an individual is to approach such data is the crucial next question that genetic medicine has been struggling with over the past two decades. Among the most ambitious goals of the HGP was to develop novel therapies to treat diseases based upon genetic understanding which has led to the modern therapies for cystic fibrosis such as Trikafta (FDA 2019). The benefits of the HGP are to this day still unfolding including the recent report of an apparently successful gene editing therapy to treat an orphan metabolic disease, severe carbamoyl phosphate synthetase 1 (CPS_1) deficiency (Musunuru, Grandinette et al. 2025).

Current Understanding of Genetics and Health

Inheritability of physical characteristics have been understood as a practical measure since 1866. Classic experiments demonstrating the inheritability of such characteristics can be traced back to Gregor Mendel's published work on pea plants, where he showed that traits such as flower color and seed shape followed predictable inheritance patterns (Mendel 1869). These experiments laid the groundwork for modern genetics.

Provided with the necessary molecular framework, progress was made on identifying the details by which DNA could confer inheritability leading to the creation of the central dogma of molecular biology: DNA is transcribed into RNA, which is then translated into proteins (DNA -> RNA -> protein) (Crick 1970). Thus, the details of DNA existing within ourselves is the core set of instructions that presents the most intimate details of our assembly.

Nature cleverly has encoded our information within a combination of four unique molecules, the nucleotide bases: Adenine (A), Thymine (T), Cytosine (C), and Guanine (G). Each nucleotide is composed of a deoxyribose, sugar molecule, attached to a phosphate group and one of the four nitrogenous bases (Brody 2025). The sugar-phosphate backbone is held together by phosphodiester bonds which provides the structure length of DNA (Nelson 2013). As DNA is a double stranded structure, it is stabilized by two or three hydrogen bonds that occur between complementary nucleotide pairs on each strand. Through this arrangement, Adenine (A) is complemented by Thymine (T) connected by two hydrogen bonds, and Cytosine (C) is complemented by Guanine (G) connected by three hydrogen bonds. The specific sequence of nucleotides determines the order of amino acids that make up proteins which then accomplish functions within ourselves (Crick 1970).

Furthermore, the portion of DNA that encodes proteins (genes) make up approximately 2% of the total genome. These protein coding regions of the DNA strands include “exons” and ‘introns’ (the non-coding regions within a gene) (Sakharkar, Chow et al. 2004). Exons themselves only account for one percent of the human genome (Venter, Adams et al. 2001). Much of the remaining 98% of the genome plays a vital role in how DNA is organized, accessed, regulated and expressed. For instance, there are areas of the non-coding regions that control how a specific gene is expressed (e.g., promoter) (Venter, Adams et al. 2001). Thus, to understand

how DNA functions in totality, we would need to understand the sequence of all the DNA, not just the one percent that ultimately produces proteins.

Types of Genetic Variation

Human genomes vary between individuals. First, the most common genetic differences in the human genome are single base pair differences. These are called single nucleotide polymorphisms (SNPs) and frequently occur about once every 1,000 base pairs (Shastry 2009). SNPs may affect promoter activity (gene expression), messenger RNA (mRNA) conformation (stability), subcellular localization of mRNAs and/or proteins, protein synthesis or chromatin structure which may lead to disease formation (Shastry 2009). It is possible to utilize SNPs to detect genetic patterns that may signify indicators of a disease. With a large enough study population, population with particular disease (i.e. diabetes) and nonaffected population, it is possible to use SNPs as markers to see if there are similar areas of change within the genome of the affected population to signal a potential genetic cause for further investigation (Gunter 2025).

Second, the next most common genetic variation are INDELs. INDELs are small insertions and deletions of nucleotide base pairs that can range from one to about ten thousand base pairs (Mullaney, Mills et al. 2010). They contribute to genetic variation across the human species as well as influence phenotypic changes. It has been estimated that INDELs are likely to make up 16-25 percent of all sequence polymorphisms in human (Mills, Luttig et al. 2006) As many INDELs function at important sites within human genes, they can have a heavy influence on genetic traits and diseases (Mullaney, Mills et al. 2010). However, INDEL variations have not been as widely explored as SNPs, thus further exploration is necessary to gain a deeper understanding of how variations influence the human genome.

Third, the genome contains repetitive regions within DNA. About 50% of the human genome consists of repeats but not all of these repeat sequences will be detrimental to the human (Bustos, Billingsley et al. 2023). Certain repetitive regions are important for the maintenance of genomic structures such as telomeres or centromeres but they can alter gene expression which may be harmful (Lower, Dion-Côté et al. 2019). Thus, sequence information including the degree to which certain sequences are repeated within a certain gene, can be predictive of a specific disorder. For instance, the number of CAG nucleotide repeats within the Huntington gene is predictive of the development of the disease (Moncke-Buchner, Reich et al. 2002). While there is still much to uncover about how repetitive sequences influence our genome, it is known that these repetitive DNA sequences do play an indispensable role in the physiological activities of organism (Liao, Zhu et al. 2023).

Fourth, there can be structural variance within the packaging of the DNA that can be important to a variety of human conditions. Pre-dating our understanding of DNA sequencing, was our understanding of chromosome abnormalities. Humans have 23 pairs of chromosomes, 22 autosomes and one pair of sex chromosomes. There is an abundance of human genetic disorders or conditions that are the result of chromosome abnormalities, in which there is a net gain or loss of genetic material (Theisen and Shaffer 2010). Down syndrome is a chromosomal abnormality whereby there is a third 21st chromosome (Patterson 2009). There are a number of other chromosomal abnormalities that are only detectable by visualizing a person's chromosomes. These include errors in organization such as breaks in the chromosome and miss attachment to alternative sites such as translocations (Bell 2025). Chromosome abnormalities can impact phenotypes because they can cause an imbalance of one or more dosage-sensitive genes (Theisen and Shaffer 2010).

Lastly, there can be alterations to the structures around the genetic material that alter the interactions with genes. There are mechanisms that can explain how environmental factors and lifestyle choices influence gene expression without directly changing the genome (e.g., epigenetic) (Dupont, Armant et al. 2009). For example, there are molecules involved in the organization of DNA strands called histones. Chemical modification of histones (and other types of epigenetic modifications) can alter DNA structure making some genes less accessible and others more accessible leading to altered patterns of gene expression (Baccarelli and Bollati 2009). These epigenetic changes can have lasting impacts on cellular function (Baccarelli and Bollati 2009). For instance, there is an entire body of literature devoted to better understanding the role that stress and stress hormones place on gene functions leading to disease (Yaribeygi, Panahi et al. 2017). Some of these epigenetic changes are thought to have transgenerational impacts helping us to better understand how environmental impacts may create lasting health concerns for entire populations (Baccarelli and Bollati 2009).

Types of Genetic Diseases and Conditions

Genetic disorders are health conditions that are caused by changes to one's genome and can be categorized based upon how they were inherited. The genetic combination of an individual is known as the genotype and the manifestation of the genotype that is in observable characteristics is called the phenotype. The molecular mechanisms that underlie a difference in sequence and functional output are numerous. There are a few important essential mechanisms to understand.

To begin, the concept of parental variation leading to offspring variation was first delineated by Gregor Mendel and is frequently referred to as Mendelian inheritance. (Strome, Bhalla et al. 2024). Single gene or mendelian inheritance follows predictable pattens and each

gene contains two alleles, one inherited from each parent. There are five basic modes of inheritance for single-gene diseases: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, and mitochondrial (Genetic Alliance and District of Columbia. Department of Health 2010). Autosomal dominant inheritance means that an individual receives a single mutant copy of the disease associated gene from either parent (Chial 2008). An example of an autosomal dominant disease is Huntington's disease. Autosomal recessive inheritance only occurs when the individual receives two mutant copies of the allele from both parents (Chial 2008). For example, cystic fibrosis is an autosomal recessive disease. X-linked dominant inheritance occurs no matter the sex. This means that the individual who inherits the mutant X-linked disease will express the symptoms associated with the inherited disease (Chial 2008). Hypophosphatemic rickets, a bone disorder, is an example of an X-linked dominant disease. X-linked recessive inheritance relies on how many X chromosomes that individual inherits (Chial 2008). If a typical female sex, XX, individual receives two mutant alleles then symptoms will be express. However, if only one mutant allele is inherited then that individual will not express symptoms but instead be a carrier. A male sex, XY, individual inheriting a mutant allele will express the disease since they only have one copy of the X-chromosome. An example of a X-linked recessive disease is hemophilia A, a blood clotting disorder. Finally, mitochondrial inheritance can affect both female and male sexes but typically gets passed along by females (mothers) (Genetic Alliance and District of Columbia. Department of Health 2010). For example, Leber's hereditary optic neuropathy is a mitochondrial inherited disease.

Next, there are three types of chromosomal abnormalities that lead to genetic disorders; numerical, structural, and sex chromosomes. Numeric abnormalities are the result from the gain or loss of an entire chromosome (Theisen and Shaffer 2010). This type of chromosomal

abnormality is most common and results from improper segregation of the chromosome pairs during meiosis. An example of numeric abnormality is down syndrome or trisomy 21 which has the presence of three chromosomes copies rather than the typical two copies. Structural abnormalities are the result from the breakage and then reunion of chromosome arms (Theisen and Shaffer 2010). These structural abnormalities often involve deletions or duplications of a segment of a chromosome segment leading to a gain or loss of genetic material. Additionally, there are structural abnormalities that do not result in the gain or loss of any genetic material. Such rearrangements include inversions, which are caused by a two-break event and the end-to-end reversal of the intervening chromosomal segment; translocations, which result from the exchange of chromosome segments between two or more chromosomes; and insertions, which occur when a segment of one chromosome is translocated and inserted into a new region of the same chromosome (Theisen and Shaffer 2010). Finally, sex chromosome abnormalities are the gain or loss of the entire or parts of the sex chromosomes (Berglund, Stochholm et al. 2020). Typically, female sex has XX chromosomes and male sex has XY. However, it is possible for an individual to have one sex chromosome, a broken sex chromosome pair, or even additional sex chromosomes. An example of a sex chromosome abnormality is Turner syndrome, where that individual is partially or completely missing an X chromosome (Gravholt, Andersen et al. 2017). This abnormality can lead to neurocognitive deficits, social challenges, and other health related problems.

Furthermore, there can be patterns of inheritance that simply do not follow mendelian inheritance and are referred to as non-mendelian inheritance (Strome, Bhalla et al. 2024). For instance, polygenic traits, such as human height or eye color, are controlled by two or more genes (Hindorff 2025). Many polygenic traits are also influenced by the environment.

Public Understanding of Genetics

To learn and have a sufficient understanding of genetics can require a lengthy period of time to master. In the United States, high school biology curriculum typically includes fundamentals of genetic understanding (Dougherty, Pleasants et al. 2011). Survey data evaluating US adults with a secondary education found that comprehensive genetic literacy can be as low as 1.2% on direct testing (Chapman, Likhanov et al. 2019). In an alternative evaluation of US population, only 57% were even aware of genetic testing highlighting a large gap in the public's understanding of these important concepts (Krakow, Ratcliff et al. 2017). While molecular genetics is clearly an area of advanced study, the basics can be made accessible to most people. In reality, the need for genetic interpretation and understanding is already widespread given that direct to consumer genetic testing includes specific health information. The obvious mismatch between baseline genetic understanding and the desire for that information to influence health decisions is already evident. This is where new technologies may facilitate a more rapid deployment of genetic understanding to enrich individual use of genetic testing to their healthcare as the evolution of technology unfolds to effectively realize the aim of the human genome project.

Types of Genetic Tests and Their Uses

Genetic testing can uncover information about an individual that can have a life-changing impact on them as well as their family. In 1999, the task force on genetic testing defined a genetic test as:

> *the analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes, or karyotypes for clinical purposes. Such purposes include predicting risk of disease, identifying carriers, establishing prenatal and clinical diagnosis or prognosis. Prenatal, newborn, and carrier screening, as well as testing in high risk families, are included* (McPherson 2006).

Note that paternity testing is not included as a genetic test as it does not signal an inheritable disease. Given the potential gravity of the results, detailed understanding of the test and its pitfalls is important in making an informed decision. A comprehensive review of all genetic tests is beyond the scope of this systematic review; however, a functional understanding can be had by following our basic understanding of genetic organization from sequence to chromosomes. Since genetic tests are categorized by their purpose, there are two main categories of tests; diagnostic and predictive.

First, diagnostic is the most recognizable example of genetic testing. Diagnostic tests are DNA-based tests used to confirm or eliminate a suspected genetic disorder in an individual showing symptoms to confirm or rule out a specific genetic disorder (McPherson 2006). For example, if an infant is brought to a physician with symptoms that match cystic fibrosis, a diagnostic genetic test may be performed in combination with other testing to confirm the diagnosis (Riordan, Rommens et al. 1989). The result would instruct the healthcare professionals on how to move forward with treatment; cystic fibrosis has robust treatment options for individuals with the most common genetic mutations (Riordan, Rommens et al. 1989). As cystic fibrosis is a genetically inherited Mendelian disease, family members of this individual may want to participate in carrier screening in order to assess for further familial risk. A carrier screening genetic test is used to identify individuals who have a gene mutation on one allele (autosomal recessive) for a Mendelian disorder (Pagon, Hanson et al. 2001). Typically, carriers do not show

symptoms of the disease itself but the implication of being a carrier can leave a lasting effect on reproductive choices.

Beyond reproductive choices, individuals who know of a genetic disease in their family may be more interested in predictive genetic testing which has two distinct categories: presymptomatic and predisposition. In presymptomatic genetic testing, a healthy person is tested for a condition with delayed onset (McPherson 2006). A positive result indicates that the individual will develop the condition, but it does not indicate when it will occur (McPherson 2006). Thus, presymptomatic testing is offered to adults who are at risk for a specific genetic disorder (Bove, Fry et al. 1997). However, presymptomatic genetic testing should be used with caution and can come with significant ethical considerations (Bove, Fry et al. 1997). If proven treatment options or a preventative measure can be taken to prolong a life, early testing may be beneficial (e.g. familial adenomatous polyposis) (Swartwood, Morales et al. 2024). Other diseases that do not have treatment options such as Huntington's disease, presymptomatic testing may have terrible psychological effects (Group 2004). Therefore, genetic counseling for presymptomatic testing requires great skill, sensitivity, and a nondirective approach (Bove, Fry et al. 1997).

In contrast, predisposition genetic testing is used to determine the potential risk for a specific disease to manifest in an individual (Bove, Fry et al. 1997). Unfortunately, it is not clear whether a particular disease will manifest at all and if it does how severe it will be. Predisposition testing is used to provide results for proactive action in one's healthcare. A common example is BRCA mutation testing. These individuals with mutations are at significantly elevated risk for both breast and ovarian cancer, among others. Knowledge of this mutation may help to make lifestyle (e.g. reducing controllable risk factors such as smoking) or

healthcare decisions to limit the risk of these types of cancers (Lawrence, Peshkin et al. 2001). This type of predictive genetic testing often applies to cancer predisposition testing in which a positive result indicates a need for increased surveillance, while a negative result implies a risk similar to the general population but is not negligible (McPherson 2006). While the field of genetic testing is exploding with new subspecialties, at the core of each is to provide informative information to help facilitate the person(s) in making the most beneficial decision for themselves.

Genetic Counseling

The application of genetic principles to human health in America began to emerge in the late 19th century predating the HGP and was advocated by the Progressive movement of the time. By the early part of the 20th century public health steps were being taken (e.g. force sterilization laws in over 30 states (NIH 2022)). Patient centered genetic counseling, in its earliest form, first emerged in the 1940s, with its origins partly linked to the eugenics movement of the 1930s. Eugenics is the scientifically inaccurate theory that humans can be improved through selective breeding of populations (NIH 2022). The fever pitch of the eugenics movement are undoubtably the criminal application by the Nazi's which subsequently discredited eugenic ideas in America. As the science of molecular genetics advanced, the notion that we could pursue purposeful, ethical, individual genetic engineering emerged by the 1970's. Hence an expanding need for personalized understanding of genetic conditions thus creating a new specialty in healthcare. However, it was not a formally recognized medical specialty until the 1970's following the creation of first graduate degree program in genetic counseling at Sarah Lawrence College as well as the National Society of Genetic Counselors (NSGC) (Counselors). The NSGC defines genetic counseling as the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease (Uhlmann, Hoskovec

et al. 2020). Additionally, at this time the field of genetic counseling defined core values such as non-directiveness counseling and the autonomy of the counselee as a way for the historical responsibility of Nazi ideology and other eugenic practices to not fall on geneticists (Zuckerman 2021). Thus, the purpose of genetic counseling is to provide personalized, empathetic, and science-based support for informed healthcare decisions. Therefore, genetic testing allows for one to increase their own autonomy through uncovering more about oneself on a molecular level. In contrast, the eugenics movement utilized genetics as a form of discrimination. Genetics was used as a tool in a misguided attempt to form a "healthy" society based upon ideology not on individual wishes. Hence, a tool that was supposed to empower autonomy within the population was used to strip away autonomy instead.

Following the completion of the HGP and a high-profile op-ed piece published in the *New York Times* by Angelina Jolie regarding her BRCA1 diagnosis in 2013, societal interest in genetic counseling increased. Her piece created what is now known as the *Angelina Jolie Effect* and has generated an increase in referrals for breast and ovarian cancer screening as well as BRCA tests (Troiano, Nante et al. 2017). Increasing the awareness of the benefit of genetic testing in turn grew the field of genetic counseling as more and more people went seeking their own genetic answers.

The technology of course expanded rapidly beyond simple screening for mutations with a known pathology to include identification of variants with unknown or unclear significance (Kelly, Mahon et al. 2023). While these variants may yet yield important prognostic information, the meaning is yet to be determined. It is important to note that the presence of sequence variation does not necessarily mean this variation will have health implications. As the number of genomes sequenced and analysis increases, the number of variants that are then

identified subsequently increases (Kelly, Mahon et al. 2023). Importantly, many of these variants will not negatively impact health. Such a nuanced finding is often times difficult for many individuals, even amongst those who are highly educated in other fields, to comprehend which leads to increased demand for specialized counseling to avoid undue anxiety and misunderstanding (Jez, Martin et al. 2015).

A genetic counselor can be a source of information regarding genetic conditions, available genetic tests, and other genetic related health questions. Since its creation in 1993, the American Board of Genetic Counseling (ABGC) has certified genetic counselors which requires recertification every five years to ensure ongoing competency in a rapidly changing field. Since they are trained to present complex and difficult-to-comprehend information about genetic risks, testing, and diagnosis to families and patients, they are of upmost importance when it comes to patient care applications of genetics (Genetic, The New York-Mid-Atlantic Consortium for et al. 2009). Their responsibilities include walking individuals and families through the entire genetic testing process and related procedures, some of which can be invasive. For instance, a family deciding on prenatal genetic testing of the fetus may consider an invasive test such as an amniocentesis which directly tests for genetic variation within a fetus but also carries certain medical risks to the pregnancy (Salomon, Sotiriadis et al. 2019). There are many areas of health that often benefit from genetic counseling including prenatal testing, the genetic contribution to cancer care, rare disease identification and management, and optimization of drug treatment based upon genetic variations (e.g., pharmacogenomics).

Upon the first visit, genetic counselors will take a detailed personal and family history to help assess whether this person(s) needs to undergo genetic testing and if so, which test is most appropriate. These conversations are often complex given the evolving nature of testing

availability. For instance, a family may come with a specific concern that focuses on a narrow subset of mutations. In those situations, focus testing is often more impactful than broad screening. At other times, when the underlying cause of the condition is uncertain, broad screening for genetic variance through whole genome sequencing may offer the best hope at achieving a molecular answer.

Helping patients understand why they may choose a particular route of testing and consequently understanding the results requires both strong scientific knowledge and communication skills. Each genetic counselor needs to assess each patient's baseline understanding of genetic inheritance. Invariably, this will involve a degree of education of the science of genetics to bring a family's baseline understanding to a sufficient level to make appropriate risk-benefit decisions regarding testing. It is this baseline education that may require at times biologic explanations to families that may not have fully understood the material presented at the high school level. Beyond testing and results interpretation, genetic counselors are often times a bridge to specialized physicians or support groups.

Direct-to-Consumer (DTC) Genetic Testing versus Genetic Counseling

For good reason, clinical lab tests upon which healthcare decisions are based upon are not accessible to the public without a providers involvement (Wilkinson and Pontius 2003). This is to ensure that mean critical components are part of the healthcare experience: high-quality laboratory testing and high-quality result interpretation (Power, Fell et al. 2013). DTC genetic tests are a form of pseudo-diagnostics in which *consumers* are directly marketed for their participation rather than being instructed by a healthcare professional (FDA 2019). In a unique circumstance, DTC genetic tests fall outside of regular laboratory surveillance supervision. These tests passed simple FDA clearance and given the legal framework of non-discrimination

against individuals with genetic testing results, privacy concerns are left to the individuals involved (i.e. the consumer) (Su, Borry et al. 2013). Further empowering the commercialization of this medical testing process is a robust set of disclaimers that these results do not substitute for "official" clinical testing (Eissenberg 2017). Hence, DTC genetic testing companies (e.g., 23andMe) have emerged through the convergence of several forces: consumers' interest in taking a proactive role in staying healthy, the public's fascination with genetics and the promise of personalized medicine, the rise of the internet, and concern for protecting the privacy of genetic information (Myers and Bernhardt 2012). If companies wanted to reach a wide audience of willing participants, they needed to make taking genetic samples easy. To solve this dilemma, companies turned to a "spit-to-screen" method in which a participant spits into a tube and sends it back to the company for testing and analysis. From the moment one sends their spit for testing and analysis, it can take between 4-6 weeks to see results. Users of DTC tests may not initially be after health-related results (more intrigued by ancestry), but may be persuaded to pursue that avenue as well. However, those results can have an impact on one's life and consumers must use their judgement when ordering and interpreting their results from these tests (FDA 2019). How DTC tests are marketed can persuade the consumer that they oversee their own healthcare, but this is a slippery slope as many DTC companies, such as 23andMe, do not provide conclusive results on whether the consumer will develop a disease of not (Oh 2019). Consumers are cautioned in the fine print to understand these results simply as for entertainment and that for true medical decision making, robust clinical test with appropriate counseling should be pursued (Eissenberg 2017). And yet, these testing platforms do market health results as part of their advertising strategy. Clearly taking advantage of a regulatory loophole to, in fact, practice medicine (Saukko 2013). For example, 23andMe screens for variants that can be associated with

late onset Alzheimer's disease. The results do come with an abundance of information attempting to place the risk in context and in fact encourages consumers to seek genetic counseling and provide a link to NSGC. While DTC platforms are presented in a way that seems educational, the results can be hard to interpret if one stumbles across something that was unexpected. In what could be interpreted as an effort to limit risk liability, these companies do offer access to an interpretive service for incidental findings (Saukko 2013). However, as outlined above, pretest counseling is crucial in deciding which test to run and why. Whereas, if genetic testing was sought through a genetic counselor, one would be led through a more thorough process to ensure that the genetic test they choose will be the most beneficial for what conclusion they are after. Further, your results would be discussed in depth, and you could in that session come up with a plan that you feel supported in, instead of having to deal with upsetting news while also being proactive about how to help yourself. The public's increasing access to genetic testing along with a demonstrable gap in basic genetic understanding is likely to create increased demand for genetic counseling.

Challenges Faced by Genetic Counselors

There are many barriers to accessing genetic counseling. Firstly, genetic counselors are not in abundance. The ABGC reports that as of April 1st, 2025, there are 7,514 certified genetic counselors in the United States (Counseling 2025). With a current population of over 300 million people in the United States, if everyone sought out a certified genetic counselor, each genetic counselor would be buried in work. Access to genetic counseling can vary significantly depending upon the urgency of the consultation. However, some provider groups within Oregon state can arrange for an urgent appointment within 48 hours (Wellness 2020). Anecdotal

reporting though the CDC indicates that some clinics have a 3 to 4 month wait list (Garrison 2020).

Second, genetic test results can vary in complexity and consequently the time needed to interpret the findings can vary significantly. Additional time is needed to educate and counsel the patient. Some results are simple to interpret (e.g., Hemophilia A) and can be explained from a single DNA sequence mutation, as a binary "yes or no" answer. However, other results may not be so discrete. A common finding on broad screening tests, such as whole exome sequencing, are variants of uncertain significance (VUS). VUS encompasses DNA differences that are statistically distinct from the control population, but there is not enough information to determine if it is connected to a health condition (Burke, Parens et al. 2022). Since a VUS finding may be worrisome and confusing or cause other negative responses, including disappointment, sadness, frustration and decision regret it is an important part of a genetic counselors training to assess one's psychological standing to move forward with genetic testing (Burke, Parens et al. 2022). Additionally, as more research is done regarding a genome mutation, a genetic counselor, who has kept track of patient's VUS's, can relay a more in-depth response as new information has been made available. Thus, highlighting another important role in genetic counseling; ongoing follow up. As it stands, genetic counselors in the outpatient setting spend on average 2.3 hours on patient related tasks for one hour time of direct patient encounter, thus highlighting the complexity of the workload (Osborne, Bland et al. 2025).

Third, explaining the results from a genetic test requires time and empathy. Genetic counselors must be able to be skilled in dealing with individuals or families that are experiencing life altering news. The potential for accumulating emotional and psychological stress on the provider is an ongoing factor (Bernhardt, Rushton et al. 2009). Since patients come to the

counseling session with experimental, emotional, religious, and situational concerns that will influence not only their perception and interpretation of risk but also the manner in which they receive the information presented to them, it is up to the genetic counselors to accurately assess the needs of each patient (Andrews, Fullarton et al. 1994). Often, multiple visits are needed to address each concern and to make sure that the patient fully comprehends the intended use of genetic testing (Osborne, Bland et al. 2025).

Fourth, this resource comes at a financial cost. Comprehensive genetic testing involves both pre-and post-counseling visits along with the cost of testing directly. While genetic testing costs have improved substantially, specialized testing can be particularly expensive. For instance, whole exome sequencing has an average cost of $2,439.00 (Pathology 2016). The cost of genetic counseling is of course separate and varies with the number of complexities of the visits. Available online estimates for initial consultation range from $175-$250 (Genetics 2025). Thus, financial considerations are a known barrier to genetic counseling (Lawrence, Peshkin et al. 2001). While insurance may cover a portion of the cost, there may also be out-of-pocket expenses for certain tests. Given the costs associated with genetic testing, counselors will often be faced with barriers to providing comprehensive available testing to patients who simply cannot afford it, or whose insurance companies refuse to cover these costs (Andrews 1994). Often times, these decisions are being made based upon the understanding that these tests are being done at the cutting edge, and there is a reasonable argument that the tests may yield information that is inconclusive and borderline experimental.

Finally, as discussed above, genetic literacy in the general population is poor. Thus, genetic counseling often times involves baseline education in order to ensure a good faith effort towards informed decision making. Genetics is a topic that is only included in the standard K-12

curriculum to a limited extent and often avoids the use of technical language (Dougherty, Pleasants et al. 2011). The avoidances of technical language have been identified as a prominent source of student misconception (Elmesky 2013). Basic biology which is introduced in high school gives a brief overview of the inner workings of a cell which includes DNA and basic concepts of inheritance (Dougherty, Pleasants et al. 2011). However, due to genetic complexity, often patients need a form of an accelerated biology class to comprehend their options to make a truly informed decision. That is why comprehensive genetic counseling includes education, pre- and post-test counseling including interpretation of test results, discussion of the implications of that information, answering all question (in a language and manner understandable to the person being counseled), providing supportive counseling, and offering information about community support groups and other follow-up resources (Andrews, Fullarton et al. 1994).

Potential for Artificial Intelligence Use in Genetic Counseling

With technological advancements happening right before our eyes, a new tool has been introduced to society, artificial intelligence (AI). AI refers to computer systems capable of performing tasks that typically require human intelligence, such as data analysis, pattern recognition, and decision-making. Each AI platform needs to be customized for that particular use or else it will not produce the desired output (Russell 2021). Therefore, done properly, AI can process data and come to its own conclusions about what that data means, and it can then carry out a set of actions based on what it has learned (Ronald M. Razmi 2024). With the right customization, AI can be a tool that greatly enhances the medical field. It can be applied in predictive analytics, diagnostic tools, administration, and clinical decision support systems (Ronald M. Razmi 2024). With any innovation there are pros and cons to applying this to the healthcare field. For one, AI can effectively handle large datasets and repetitive tasks which

improves efficiency. Additionally, it can also enhance the efficiency of administrative tasks which allows healthcare providers to focus more intently on their patients (Bajwa, Munir et al. 2021).

However, there are legitimate concerns about this rapidly evolving technology especially in the use of sensitive areas including healthcare. Specifically, AI can raise ethical concerns about bias, privacy, and autonomy (Guidance 2021). Based on how AI is programmed, which is on predetermined data sets, AI is typically bias towards the majority of the population which can place those in the minority at a disadvantage (Guidance 2021). Further, privacy concerns regarding personal health data are another factor that needs to be considered when implementing AI into healthcare. Since the software to run AI is based on predetermined data sets, questions arise about sensitive data security (Guidance 2021). The lack of privacy may either harm an individual (such as future discrimination on the basis of one's health status) or cause a wrong, (such as affecting a person's dignity if sensitive health data were to be shared or broadcasted to others) (Guidance 2021). Respect for autonomy has a part to play in the protection of privacy but moreover it has to do with ensuring humans remain in full control of the healthcare systems and medical decisions. AI may not fully be able to compensate for autonomy as a human trait (Prunkl 2022). Thus, AI should be designed to assist humans, whether they be medical providers or patients to ensure total autonomy (Guidance 2021). Since it is not a human, it will not be able to provide human empathy in patient interactions which can lead to the patient feeling that they are not as important as someone else or that their condition which may feel life-altering is not being taken seriously (Prunkl 2022).

Given the potential for reward, genetic counseling assisted by AI may offer an unrealized opportunity. In this particular instance, we have a situation where there is frequently a large gap

in baseline knowledge that needs to be acquired in order to adequately protect patient autonomy through informed decision making. Furthermore, adult learners acquire and retain information more rapidly when presented in an interactive medium (Palis and Quiros 2014). What is not yet clear is if AI offers significant advantages over more primitive interactive teaching such as digital media or games (He, Chen et al. 2024). Since AI is obviously suitable for repetitive tasks to reduce human workload, the baseline genetic education component of genetic counseling may be perfectly suited to AI. The hypothetical advantage is that the teacher (AI in this case) can learn from the student (the patient) about their specific gaps in understanding the way that an in person skilled genetic counselor would behave. The situation is of course in contrast to more agnostic digital media currently available. There are other repetitive tasks that could theoretically be offloaded to AI including large dataset interpretation or more mundane follow up assignments. However, with any computer driven application the parameters as decided by the users (humans) will drive effectiveness.

Thus, looking to apply AI into genetic counseling needs to be done with human judgement to implement it where it is and is not appropriate. If AI were to be implemented into genetic counseling it could be used as a tool to assist with genomic data interpretation, patient education as well as virtual assistants to handle frequently asked questions or follow-up tasks. As genetic counselors are contextual experts who take genetic test results and analyze it through a personalized lens, AI systems could help to augment part of the process but not replace it. Most importantly, any healthcare interaction values empathy as a core component which as of yet cannot be adequately offloaded to a virtual entity. That is, there's no replacement for a caring human.

Using AI to Address Genetic Counseling Challenges

While there are several challenges a genetic counselor faces, there are two challenges in particular where AI could help alleviate the growing demand for access to them. Firstly, to address the high patient volume, virtual AI assistants can be implemented for pre-counseling education and intake. Here patients can be assisted virtually to input personal and family history into an AI tool that will sort and highlight important aspects for the genetic counselor. Similarly, AI can generate a preliminary risk assessment and education materials based upon that to support the patient. This use of AI will save the genetic counselors time and improve accessibility. However, it may reduce personal connection and would require robust data security.

Second, AI can be used to interpret complex data. From customized machine learning algorithms, it would be able to comb through the genetic test results for variant interpretation. The algorithm would be programmed to flag all clinically significant genetic variants and suggest next steps. Using AI in this manner would reduce the workload for genetic counselors while enhancing precision. Unfortunately as an AI algorithm is produced by an iterative process that uses examples and experiences as opposed to predefined rules, this can lead to bias which may misclassify results (Ronald M. Razmi 2024). As AI is incapable of detecting hidden bias, it can inadvertently provide skewed, wrong, or unfair recommendations, thus leading to problematic results (Ronald M. Razmi 2024). Furthermore, if the AI is only looking for clinically significant variants, this would require human oversight since it may miss a huge portion of results that fall in the category of VUS.

With all the promise of AI in genetic counseling, it would be useful to know how AI has already been studied in the other healthcare settings. Taking what we can learn from the implementation of AI in the patient experience can help us to tailor the implementation of AI and

genetic counseling in particular to appropriately design studies to answer meaningful questions as determined by AI's evolution in other healthcare domains.

Methods

The central aim for this systematic review was to evaluate how AI incorporation into healthcare has influenced patient experience. To do so, I first established criteria to lead my literature search to gather the appropriate studies to be included in the review. Second, I developed subcategories to sort the selected articles from my search criteria which included results that had to do with *patient experience* (Caldwell and Bennett 2020).

Criteria for Literature Search

Search Strategy and Terms

To perform this systematic review, I used both PubMed and Web of Science, with the final search occurring on January 10, 2025. To ensure relevancy, I constrained the search results to only include articles that had been published within the last 10 years, 2014-2024.

Types of Studies Included

The following types of studies were included: Clinical Trial, Clinical Study, Multicenter Study, Observational Study, published between 2014-2024.

Exclusion Criteria

Review articles and case reports were excluded from this systematic review. Furthermore, articles that were not directly related to AI, Machine Learning, or Deep Learning, such as robotics, were also excluded.

The keywords that were used in the search were the following terms:

artificial intelligence	AND	patient experience
machine learning	AND	patient experience
deep learning	AND	patient experience
artificial intelligence	AND	patient counseling
machine learning	AND	patient counseling
deep learning	AND	patient counseling

Table 1: "keywords" that were entered in combination into PubMed and Web of Science to generate a collection of articles to then further categorize into themes under patient care

In total, the search identified 458 articles across both PubMed (n=144) and Web of Science (n=315). To pull out the articles that had to do with the patient experience, I sorted the 458 articles into categories by looking at their titles and abstracts. Categories include *patient experience, clinician training/education, clinician workload, clinician experience, clinical decision-making, diagnostics, patient management* and *miscategorized*.

n=41	**Patient Experience** the focus of the research was to uncover patient's attitudes, experience, or feelings towards an AI driven mechanism
n=22	**Clinician Training/Education** the focus of the article was to help clinician's advance their skill and knowledge
n=13	**Clinician Workload** AI impact on healthcare time, resource, effort including the term "workload"
n=17	**Clinician Experience** clinician's attitudes in respect to an AI-driven tool
n=8	**Clinical Decision-Making** AI and the term "decision making" as a study aim
n=108	**Diagnostics** AI and diagnosis as a study aim
n=75	**Patient Management** AI and "patient management" as a study aim
n=174	**Miscategorized** unrelated to artificial intelligence, such as a robot led investigation

Table 2: Categories used to find the articles that had results that incorporated "patient experience".

Only articles categorized as related to *patient experience* (N = 41) were chosen for further analysis. While the remaining categories include articles with relevance to AI use in healthcare, they are not specifically related to the patient experience. Figure 1 below depicts the process of identifying the selected articles for review.

Of these 458 articles, 41 were initially categorized as *patient experience*. *Patient experience* was defined as those studies whose results were derived from patient obtained data including satisfaction surveys, patient outcome metrics, or provider perception of patient

engagement. These articles were reexamined, this time looking at the abstract and the results. Upon this closer analysis, some of the articles were replicants or were not actually related to patient experience such as how clinicians were perceiving the patient experience with AI. These articles were removed. Following this second review, the list of *patient experience* articles decreased from 41 to 22.

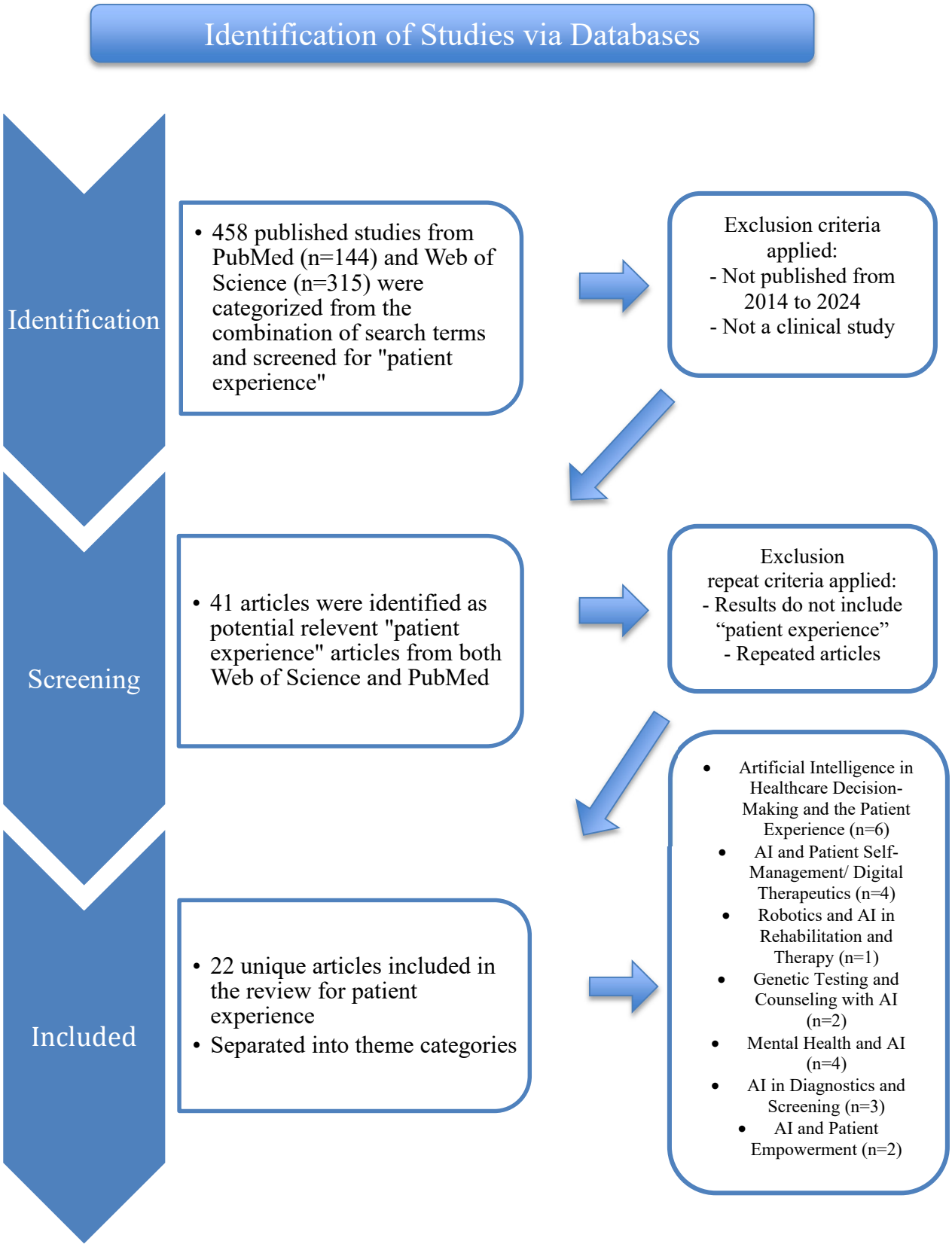
Synthesis and Analysis

Each of the 22 articles were then categorized into one theme based upon the *main* result. The seven themes include *artificial intelligence in healthcare decision-making and the patient experience, AI and patient self-management/digital therapeutics, robotics and AI in rehabilitation and therapy, genetic testing and counseling with AI, mental health and AI, AI in diagnostics and screening,* and *AI and patient empowerment*.

n= 6	**Artificial Intelligence in Healthcare Decision-Making and the Patient Experience** the focus of the results was for AI to assist in decision making and how it impacted the patient experience
n=4	**AI and Patient Self-Management/ Digital Therapeutics** the results of the articles in this theme were focused on how AI can be utilized in the management of patient of their own health conditions as well as tools to support treatment
n=1	**Robotics and AI in Rehabilitation and Therapy** results included robots and AI to facilitate rehabilitation and therapy treatment
n=2	**Genetic Testing and Counseling with AI** results included patient experience with AI in the field of genetic testing and counseling
n=4	**Mental Health and AI** the results showed how AI can be employed to support mental health
n=3	**AI in Diagnostics and Screening** the focus of the results in this theme were to see how AI can assist in diagnostics and screening to increase the patient experience
n=2	**AI and Patient Empowerment** articles were placed into this theme if their results had to do with how AI influences patient empowerment

Table 3: Themes found based upon the main result of "AI and the patient experience" articles

Figure 1. The Search and Selection Process of Retrieved and Selected Articles



Results

Performing a systematic review that focused on what is known about AI and the patient experience allowed me to make an informed conclusion on how it could strongly benefit the field of genetic counseling.

Analysis of Selected Articles

The 22 included studies were screened for common themes to better understand how AI was being understood in the patient experience in healthcare. The goal of this analysis was to better understand if there were common areas of inquiry across studies. What emerged from this analysis were the following seven themes:

n=6	Artificial Intelligence in Healthcare Decision-Making and the Patient Experience
n=4	AI and Patient Self-Management/ Digital Therapeutics
n=1	Robotics and AI in Rehabilitation and Therapy
n=2	Genetic Testing and Counseling with AI
n=4	Mental Health and AI
n=3	AI in Diagnostics and Screening
n=2	AI and Patient Empowerment

Table 4: Themes found from analysis of the articles categorized as "patient experience"

Artificial Intelligence in Healthcare Decision-Making and Patient Experience

Jayakumar et al. (2021) explored how patients perceive and respond to their own decision-making process when considering a total knee replacement for advanced osteoarthritis. The study utilized the CollaboRATE survey to gauge how patients felt about the decision-making process with an AI-enable decision aid in comparison to traditional educational

materials. Their study suggest that multifaceted decision aids integrating patient education, preference assessment, and AI-enabled analytics built with patient-reported outcome measurements data can provide a personalized, data-driven approach to shared decision-making for patients with advanced knee osteoarthritis considering total knee replacement (Jayakumar, Moore et al. 2021). The AI-enabled decision aid allowed the patients to feel that they had more involvement in the decision and thus had increased satisfaction with their care.

Knitza et al. (2022) sought to uncover the online patient experience with traditional search engines in comparison to the AI assisted ones in the realm of diagnostic decision support systems (DDSS). Two AI tools were utilized, the AI-based symptom checker (Ada) and the questionnaire-based self-referral tool (Rheport), to determine what patients preferred when researching symptoms in relation to musculoskeletal complaints. Overall, the study found positive results such as time efficiency, perceived helpfulness, ease of use/usability, as well as recommendation and acceptance. Some challenges did emerge such as lower usability with the elder population and limited familiarity with DDSS but with increasing digital literacy and implementing these new technologies into healthcare will push society to gain all the positives of AI-based diagnostic tools. Additionally, patients increasingly assess their symptoms independently online which signals that there is a growing need for more accurate symptom checkers to ensure that patients feel that they are getting clear and appropriate information while maintain their own autonomy (Knitza, Muehlensiepen et al. 2022).

Chung et al. (2024) assessed if ChatGPT could safely streamline the pre-vasectomy consultation process. Thus, increasing efficiency and patient satisfaction. The study concluded that pre-vasectomy counselling with ChatGPT was associated with a higher provider perception of patient understanding of the procedure and a decreased length of in-person consultation

(Chung, Sidhom et al. 2024). This study highlights how AI chatbots can be utilized in healthcare as a supplementary source that has positive outcomes.

Mahlknecht et al. (2023) aimed to evaluate patient and physician experience with the integrated AI-led symptom checkers in general practice. They found that patients had positive views of the AI-led tools and had a sense of autonomy in their healthcare as well as being more informed going into a conversation with the practitioner However, there were still downfalls that needed to be addressed. As AI-led online symptom checkers get more advanced, it should aim to empower patient self-management and relieve the burden on healthcare services (Mahlknecht, Engl et al. 2023). Thus, AI implementation into healthcare will supplement some of the leg work done by physicians and instead can entrust patients to explore their questions instantly online. Additionally, this study highlights that patients and providers use AI differently which could signal a conversational disconnect between them.

Simon et al. (2019) evaluated and discussed the creation of an AI-based decision-support tool for cancer patients. This study was done prior to public access to ChatGPT or other AI-based applications and thus speculated on the wonders of what AI could do when implemented into medicine. The researchers found that an AI-led tool could help patients navigate complex decisions, enhance their confidence, and foster a more productive discussion with clinicians. They noted that an absolute requirement of AI in medicine is to be transparent (Simon, DiNardo et al. 2019). With this, AI can be used to prolong a prospectus study as participants drop out and join over time. Therefore, in the current moment as AI becomes more advanced and spills into our everyday lives it is important to know what it is we are getting involved with and have the confidence in our decision-making skills to make the right one for ourselves.

Oconnell et al. (2024) explored the use of conversational AI as a tool for patients to access prior to their appointment with clinicians as well as the patient concern inventory to highlight their concerns to potentially streamline their appointment. The ability for patients to access such a tool no matter the time can offer a great amount of flexibility and reassurance. Additionally, conversational artificial intelligence (CAI) provides a tool to improve the quality and quantity of information flow between patients and professionals remote from the consultation (Oconnell, Gilmartin et al. 2024). This benefits both clinicians and patients and it does not require the clinician to discuss the basics of the medical issue and can allow patients to seek information outside of their scheduled time. Thus, allowing for patients to feel that they are learning about their condition on their own time, and this also allows clinicians to spend more time in person discussing the more complex matters.

AI and Patient Self-Management / Digital Therapeutics

The first evaluated study in this category, Marcuzzi et al. (2024) sought to uncover if patients would be responsive to an AI driven app as a tool to use for self-management of pain. They found that patients found the app to provide reassurance which in turn empowered them to take control of their condition. Unfortunately, the app's uptake was relatively low, but still both patients and health care practitioners had a positive opinion about adopting an app-based self-management intervention for low back and neck pain as an add-on to usual care (Marcuzzi, Klevanger et al. 2024). This is an example of how AI use can be a beneficial tool for patients who want to be more informed and involved in their healthcare decisions. This technology is still new, but it is showing great promise that an adoption of the advancing technology into healthcare shows tremendous benefit for every party involved.

Kamdar et al. (2024) aimed to explore the impact of an AI enabled digital therapeutic app (ePAL) on managing pain in patients with advanced cancer. They found that the app had improved pain management, increased engagement and monitoring, had educational value, reduced hospitalizations, and the AI algorithm effectively triaged symptoms. This work demonstrates the promise of digital therapeutics for improving patients' symptoms while reducing burdensome hospitalizations (Kamdar, Jethwani et al. 2024). As small computer devices walk around with every person, allowing healthcare to be as accessible as "Candy Crush" provides patients with a sense that they do not have to wait for care, and they can monitor their symptoms for a tool they use every day.

Svendsen et al. (2022) aimed to explore the patient experience with the AI enabled app selfBACK for self-management of low back pain. While they found a majority positive results, the researchers did find a few places where patients would like to see improvements, primarily with personalization. With the app feeling generic, patients felt that it may not be responsive to their specific needs and wants and thus less participation would occur. However, this is just a place for future improvement and the high prevalence of low back pain globally coupled with the advantages of providing help through an app offers opportunities to help countless people dealing with low back pain in regards to daily management of their pain (Svendsen, Nicholl et al. 2022). Thus, the app offers a promising avenue for patients to self-manage their pain without constant visits to physicians.

The final study in this theme, Kotov et al. (2024) aimed to explore how patients would perceive a neural conversational agent for weight loss counseling. They found that personalization, accessibility, and integration into healthcare would be a successful tool for weight loss patients. While this protocol is not implemented into society yet, this study found

that if proven effective, large language model- based counseling agents can become a cost-effective approach for addressing the obesity epidemic at a public health level (Kotov, Carcone et al. 2024). This is an example of how AI-based tools are in the trial phase for future healthcare implementation, and we have the ability to judge how patients are going to interact and react to such tools as to make adjustments to them to gain the most benefit from these tools as they hit the market.

Robotics and AI in Rehabilitation and Therapy

In Logan et al. (2019), the investigators aimed to analyze the implementation of social robot technology into a pediatric inpatient setting. They found the social robots to have a positive impact on those children who interacted with them. As they can be a tool to enhance the emotional well-being for hospitalized children, it may provide new ways to address their emotional needs, potentially increasing access to emotionally targeted interventions (Logan, Breazeal et al. 2019). However, it should be noted that further personalization, individual preference and scalability need to be taken into consideration when implementing these social robots in the future.

Genetic Counseling and Testing with AI

The first of the two studies with this theme explored the comparison between the use of an AI-programed chatbot versus traditional counseling with a genetic counselor in the hopes of determining what the patient experience is with each. Al-Hilli et al. (2023) found that both were comparable in satisfaction and education regarding genetic testing, the chatbot provided higher levels of accessibility and convenience, the patient concern of potential delay in treatment was not a factor, and that decision-making confidence was not impacted using a chatbot. Thus, the utilization of this technology can offer improved access to care and a much-needed alternative

for pre-testing counseling (Al-Hilli, Noss et al. 2023). Therefore, the use of a chatbot as a supplemental tool is widely beneficial if used in the right context.

The second study in this theme, D'Amours et al. (2024), evaluated a potential protocol for implementing a platform that would collect medical and family histories, provide pre and post genetic counseling as well as help to facilitate the return of genetic testing results in both pediatrics and adults. Of the patient experience considerations taken in this study, the investigators assume that it would enhance accessibility, use a patient-centered approach, and would need to be integrated with traditional care. Since patients have found e-health tools to be acceptable, easy to use, trustworthy, and be able to facilitate personalized care in various clinical settings, their integration into genetic counseling is around the corner (D'Amours, Clausen et al. 2024). As this technology becomes widespread, it would be critical to learn how to facilitate this technology to prevent issues that could become problematic.

Mental Health and AI

In the first of four studies in this theme, Ronneberg et al. (2024) aimed to explore the option of a voice-based virtual coach, Lumen, for problem solving therapy (PST) to address critical gaps in the treatment of depression and anxiety. They found that the patient experience with this AI-led tool could be enhanced by the increased accessibility, reduced stigma, and the flexibility to engage with therapy as one sees fit. Their finding also provide evidence that voice-based virtual coaches have the ability to improve the reach and impact of psychotherapy, mitigating access, cost, and stigma barriers for people with depression and/or anxiety (Ronneberg, Lv et al. 2024). Thus, as technology becomes more advanced, this form of AI-led tool could serve adjunct with human therapists or even standalone all while improving mental healthcare access.

The second study, He et al. (2022), sought to evaluate a cognitive behavioral therapy based chatbot, XiaoE as a way for adults to seek treatment for depressive symptoms over the COVID-19 pandemic. They found that this chatbot had a positive impact on the tested population and that it is a feasible and engaging digital therapeutic approach that allows easy accessibility and self-guided mental health assistance for young adults with depressive symptoms (He, Yang et al. 2022). Therefore, future applications of a chatbot should be explored as a way to make healthcare access more widespread and allow for a place where people can divulge what they are feeling without the stigma they may have in their head.

In the third study, Southwick et al. (2024) explored patients' and mental health therapists' experience with a patient-generated digital data dashboard for psychotherapy. The research points towards the benefit of integrating patient-generated digital data as a source that can enhance patient engagement, self-awareness, and collaboration between patients and therapists. It was also found that patients reported that receiving their digital data helped them stay "accountable," while therapists indicated that the dashboard helped "tailor treatment plans" (Southwick, Sharma et al. 2024). While data privacy concerns are a potential challenge for this type of AI- based tool being implemented into the mental healthcare field, it shows great potential and with the right guidelines and structure could alleviate those concerns.

The final article in this theme, Juergensen et al. (2024) discussed the factors that are influencing the likelihood that individuals with depressive episodes are going to seek professional mental health care. With the use of machine learning models, the investigators found several key predictors such as self-stigma, previous treatment experience, and subjective symptom perception that impact attitudes, intentions, and actual help-seeking behavior. Additionally, the authors state that to effect changes in help-seeking behavior and improve the

mental health of populations, an approach that considers the interaction between individual influencing factors and structural elements is necessary (Juergensen, Peter et al. 2024). Thus, it is important to see patterns from individual experiences that may not be picked up outside of a machine learning analysis to ensure a well-rounded plan of action to knock down barriers and sigma that surround mental health.

AI in Diagnostics and Screening

In the first of three articles in this theme, Sadasivam et al. (2016) investigated the effects of personalized messages in relation to their quitting progress. Two types of online devices were utilized, computer-tailored health communication (CTHC) and a machine learning tool, Patient Experience Recommender System for Persuasive Communication Tailoring (PERSPeCT). This allowed the investigators to see which one had a stronger impact on their test population. They found that compared to standard CTHC with proven effectiveness, PERSPeCT outperformed in terms of influence ratings and resulted in similar cessation rates (Sadasivam, Borglund et al. 2016). Thus, as technology advances more people respond better to technology that seems to respond to them personally rather than a generic system.

In the second study, Sharma et al. (2023) focused on exploring the use of Amazon Alexa, a voice assisted AI-based system, as a way to screen for severe acute respiratory syndrome coronavirus 2 for caregivers or patients with heart failure. They found that Alexa performs comparably with a COVID-19 health care professional in administering a screening questionnaire (Sharma, Marques et al. 2023). This shows that voice-assisted AI options can perform just as accurately as a human and can decrease movement from home if an individual does have COVID-19. Therefore, other applications for voice-assisted screening show great potential in additional areas of healthcare.

In the third and final study in this theme, Deniz et al. (2024) investigated the potential of ChatGPT's introduction into healthcare. They found that ChatGPT remain accurate in some questions but answered other incorrectly or provided answers that seemed to be all over the place. Although ChatGPT exhibited improving potential as a clinical support tool in thyroid nodule management although it showed varied performance for binary and multiple-choice questions (Deniz and Guler 2024). This study highlights the concept of noninferiority but is subject to change as more AI studies are produced and this technology become further integrated into society. Furthermore, this study supports the concept of placing AI in healthcare, but it points out that at the current moment the technology is not advanced enough to have it be a useful supplementary tool.

AI and Patient Empowerment

In the first of the two studies in this theme, Perotti et al. (2024) set out to explore how older adults (>65 years of age) engage with an e-learning platform designed to help navigate electronic personal health records. While their results did not show exactly what the researchers were after, it did highlight areas that will need to be improved for increased engagement and usability. They also found a strong correlation between usability and the perception of how learning content was presented (Perotti, Stamm et al. 2024). Therefore, this study shows how elder adults interact with this shift to electronic health platform and how to help mitigate the confusion found with this population.

In the other study in this theme, Zhou et al. (2022) explored how the SMART Choice tool influences the decision-making of patients debating to undergo a total knee replacement. Since this tool is designed to be patient led and without clinician input, it allows patients to assess their own outcomes with the hopes that it can provide insights into the surgery, so they feel that they

have total control over the decision to undergo this intensive surgery. The investigators found that with the use of this tool, the delicate balance of surgical decision-making swings towards patients who can make more informed decisions about their own healthcare needs (Zhou, Weeden et al. 2022). Thus, allowing patients to perceive that they are in control of their healthcare decisions can feel empowering when it often feels daunting to put full trust into someone else for your own well-being.

Discussion

Genetics/ Healthcare and Artificial Intelligence

In modern medicine, technology is almost as important as the clinicians. Computers, monitors, and other automated processes are essential to the efficiency of healthcare. Over the past few years, patient records have been moved from large hardcopy paper files to easily accessible electronic health records (EHR). This move has allowed patients and providers to access medical records, order tests, refill prescriptions, etc. at the click of a button. In the United States, the increase in EHR adoption was stimulated by the 2009 stimulus plan's Meaningful Use initiative (Evans 2016). The Meaningful Use incentive program was run by the Centers for Medicare and Medicaid Services which provided a financial incentive to adopt EHR technology into practice (Anumula and Sanelli 2012). This is an example of how technology changed the way healthcare functions.

It is apparent to any casual observer that AI is being deployed into multiple domains of the human experience simultaneously. In fact, all of us are currently unwitting participants in a large prospective trial. As we move forward, it is going to be important for us to define how we are going to understand the impacts of this new technology in the human experience. Much has been identified of the potential safety concerns surrounding artificial intelligence. Another dimension highlighted in this work is that if we are thoughtful about our ongoing understanding of this technology, we can be more efficient in our integration of AI as we learn how humans best interact with artificial intelligence (De-Arteaga 2020). At first glance, we as expert smart phone application users, have multiple preconceived notions of how we will interact with artificial intelligence based on past experience with computers. However, we are likely to be surprised by how we *actually* interface with AI. Healthcare settings are fraught with bias both

on the patient and provider sides of the interaction. We anticipate that AI will not have those barriers, but such assumptions will need to be tested. We may learn more about our own fundamental psychology through the study of the patient experience and the human response to a virtual entity.

How does AI influence the patient experience across various healthcare domains?

The studies categorized under *Artificial Intelligence in Healthcare Decision-Making and Patient Experience*, suggest the use of an AI chatbot or online service does help to alleviate the workload of the clinician. Furthermore, the survey feedback from the Jayakumar et al. (2021) study found that there is an increase in patient perception of greater autonomy with an AI-enabled decision aid. These findings contributed to the identification of the second theme, *AI and Patient Empowerment*. The results of the studies categorized into this theme as well as aspects that appeared in some of the other studies, led to the conclusion that AI created the impression that patients felt empowered based upon potential increase in understanding of the questions being asked. These kinds of results are the anticipated benefit of AI and the individual advantage of providing tailored individual instruction. Thus, it should not be surprising that when patients have a real sense of a greater understanding of their situation that they would feel a sense of control and empowerment.

In the theme *AI and Patient Self-Management / Digital Therapeutics*, I found that AI is used to self-manage symptoms and help with accessibility. Unsurprisingly, these studies highlight that patients want to have personalized attention when it comes to healthcare and do not want to feel like they are a box on a clinician's checklist. This need is again suited to AI in a way that standard non-human approaches can't match. It is interesting to reflect upon the psychology that values the sensation of personalized attention, and I would not be surprised if

psychological analysis of human behavior is further understood based upon AI's ability to mimic this. The ability of the AI to provide feedback and personalization is a theme that ran through most of the patient reported experiences. While many of the studies focus on efficiency of patient education and workload impacts, improved patient engagement through enhanced autonomy seems to be a novel side effect and merits more direct study. This dimension, highlighting important aspects of human interactions, directly applies to genetic counseling. Patient with complex results need to be able to ask questions to clarify their own uncertainties. As these studies highlight, patients value personalized attention even in the form of AI. The ability of AI to address highly individualized queries is likely to be viewed positively based on the results above.

While still in its infancy, there is a study evaluating AI implementation of a "social robot" (Logan, Breazeal et al. 2019). The concept of the inclusion of a physical dimension to artificial intelligence using robots has been an idea since science-fiction first imagined such a possibility. Yet today, we can see from the studies reviewed that 'social robots' may serve to help patients that are looking for multiple dimensions to their AI interactions.

What are the benefits and limitations of using AI in patient-centered care?

There are a number of repetitive and mundane tasks associated with healthcare and AI may be better suited to more efficiently handle these tasks. Ultimately, using AI in this way would enable human practitioners to focus their limited time and effort on counseling patients face to face, facilitating the need for increased human to human interactions in healthcare. AI also offers many advantages in education (e.g., patient education in this context) including personalized learning, immediate feedback, the ability to create and supplement content through the inclusion of a greater degree of available resources, all of which can lead to a more rapid

educational experience (He, Chen et al. 2024). Through these ways, we may be able to improve both the practitioners' and patients' experiences. In order to best serve this end, understanding how humans best interact with AI will be crucial. This systematic review aims to begin to understand how humans respond to a virtual entity in an intimate healthcare setting.

It should be noted of course, that there are significant concerns with the use of AI in healthcare that still need to be addressed including privacy concerns, the potential for bias, and the concern about reduced human interactions (Yadav, Pandey et al. 2023). Since AI is programmed off of a large data set such as medical records, protecting one's privacy regarding their medical records comes into question before wide spread implementation. Further, those who participate in EHR predominately come from white or European countries lending itself to fall victim to racial bias just based on the data sets available. Additionally, since AI is a form of technology those who are not well versed in technology, older populations and those who do not have access, can be at a disadvantage when AI becomes more prominent in healthcare.

Furthermore, actual achievement data demonstrating these particular points of strengths of AI have yet to be demonstrated to any conclusive nature and thus we are still in the testing phase of the deployment of this new technology. In my review of AI and the patient experience, seven main themes (Table 4) became apparent that will help us when developing and evaluating integration of AI and genetic counseling.

How can AI tools be effectively integrated into Genetic Counseling to improve accessibility and patient outcomes?

In this time of advanced computing, we now are able to link up artificial intelligence and the interpretation and use of big data associated with individual human genetic analysis. The revolution that allowed us to understand our own coding from a species to an individual in the

past 50 years has enabled a detailed evaluation of each of us. With such large amounts of intimate knowledge, we need the benefit of expert interpretation. Prior to the advent of artificial intelligence, this meant a genetic counselor. However, the number of genetic counselors available today - a time when we are encountering an even greater need for accurate genetic counseling – has not kept pace with demand. The other reason genetic counseling may be particularly suitable to use of AI is the significant need for human education in a particularly narrow area (molecular genetics). As noted above, baseline understanding of genetic concepts is extraordinarily poor in the adult US population (Elmesky 2013). The educational needs vary significantly since the exact starting point for each patient/consumer is highly variable. Thus, an adaptive teaching model such as AI, may offer distinct advantages based upon what is known about effective methods of adult learning (Palis and Quiros 2014).

It would be an interesting study to compare the effectiveness of AI on baseline genetic counseling education versus a human genetic counselor. We assume that the human dimension of genetic counseling that provides empathy is the crucial element to patient satisfaction. Yet, we may be surprised to find that patient's adaptive interaction means more than a compassionate delivery. Thus, negating the importance of an in-person human genetic counselor for the educational understanding aspect of genetic counseling. Upcoming studies could randomly assign patients with similar genetic counseling needs (similar genetic findings such as abnormal prenatal screening results) and then determine if AI truly leads to patient empowerment as demonstrated by survey data. Obviously, it would be up to future investigators to determine if they wanted to quantify understanding through testing, but from a practical perspective it may not make much difference in terms of informed decision making since pre-testing patient comprehension it’s not a clinical standard.

Al-Hilli et al. (2023) and D'Amours et al. (2024), (described in *genetic counseling and AI* theme) were limited in scope but do show feasibility in workload reduction in a couple of areas: patient history taking and patient education. It is not clear if the outcomes are improved. The dimensions that need to be studied include patient satisfaction, patient engagement, quantifiable provider workload impacts, provider satisfaction, and critical error rates. Carefully observing for AI bias and error will be important when assessing the ability of AI to gather patient information. As seen in the mental health applications, AI can participate in a meaningful way to screen for patient symptoms through interactions, but caution is needed as this technology expands (Davenport and Kalakota 2019). Further, as a diagnostic tool, AI offers the ability to utilize a vast data set (virtually all of published medicine) but the ability to make decisions are susceptible to the bias inherent to the human programing. Planned quality control based upon expert human review should be a part of any future implementation in this arena.

While not included in the studies selected for review based upon the search criteria, it should be noted that AI is uniquely suited to working with large data sets such a genomic data. This power was demonstrated when AI (DeepMind) was able to accurately predict protein folding based solely upon peptide sequence and most recently with protein – DNA/RNA interactions (Service 2020, Abramson, Adler et al. 2024). In this way, AI may also contribute to genetic research itself by better linking VUS with disease phenotypes.

Study Limitations

Due to the emerging nature of this field of study, there are a few limitations of my review. Firstly, the studies used in this systematic review vary widely in subject, methodologies, tools, and datasets. The heterogeneity of my dataset made direct comparisons between studies

difficult since there is not enough data on a single area of interest. Second, most of the studies published regarding AI in medicine have to do with clinician experience or operational efficiency rather than patient focused outcomes. Third, while a serious attempt using broad search terms to identify all of AI and patient experience, there is the possibility of missing studies not evaluated. Finally, only one reader was reviewing and categorizing the chosen articles to be included.

Conclusion

This systematic review highlights the transformative potential of AI in healthcare through decision aids, increasing patient's perception of autonomy, and decreasing clinician's workload. With offering almost rapid data analysis and data-driven decisions, this technology is reshaping how individuals interact with the healthcare system.

From the discussion section above, the findings found in the articles categorized in *Artificial Intelligence in Healthcare Decision-Making and the Patient Experience*, *Mental Health and AI*, *AI in Diagnostic and Screening*, and *AI and Patient Empowerment*, reveal that AI tools not only assist in improving clinical efficiency but also empower patients by enhancing their sense of autonomy. In addition to improving patient autonomy, personalization is another key aspect that has been highlighted in this review. People respond highly to courses of action that feel personal to them. This can be in the way a clinician introduces themselves, takes the time to explain how something works, or just a connection that they feel. AI is unable to relay this type of connection to patients which is why it is important that AI features be used when appropriate. Furthermore, a main point that appears in all of the articles reviewed is that AI would improve clinician efficiency by fast-tracking patient intake, analyzing data faster, and completing repetitive tasks that take time away from patients. In this way, AI would allow clinicians to spend more time with the personalized aspects of healthcare. By refocusing clinician attention on their patients and away from the administrative work, AI will help alleviate some of the disconnect patients feel about the healthcare system.

As for genetic counseling, the introduction of AI has tremendous potential. As genetic counseling becomes more prevalent, AI-driven tools can assist in interpreting complex genetic data which in turn enables counselors to focus on the delivery of personalized guidance and

emotional support. Further, these tools would not only improve the efficiency of the genetic counselors but also improve turnaround time for delivery of results. This would allow patients to receive timely and accurate information regarding their genetics and would relieve the prolonged anticipation anxiety around their results. Finally, AI offers promising solutions to address workload pressures and increase access to services, but it should complement human expertise to maintain empathy and trust.

However, the introduction of AI in healthcare does present challenges that need to be addressed to fully harness its potential. For example, disparities in digital literacy pose a barrier to equitable adoption, particularly among marginalized or less tech-savvy groups. Additional barriers pop up when discussing genetic counseling. For one, ensuring that AI systems are comprehensible and culturally sensitive is critical for diverse populations to trust and benefit from these advancements. Ethical considerations around data privacy, bias in algorithms, and the balance between automation and human interaction are particularly important in genetic counseling, where personal and familial implications are profound.

While AI holds immense promise in fostering a patient-centered healthcare ecosystem, addressing existing barriers and embracing AI's potential, healthcare systems can become more responsive, efficient, and empowering for all involved.

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