TELEGENETICS SUMMARY DOCUMENT

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Purpose of This Report

Telemedicine is a type of clinical service delivery that may be used to provide various kinds of health care from a distance. Addressing inequities of access to genetics care is a major focus of Clinical Diagnosis & Management Cluster of the Region 4 Genetics Collaborative. This summary describes and assesses telemedicine and discusses its implications for the provision of genetics services. Experience with telegenetics will be reviewed in order to assess its potential as a regional activity that would increase access to quality genetics services and decrease inequities of such access. The emphases of this report include: 1) the relevance of telemedicine to access to genetics care; 2) regional relevance of telegenetics; and 3) special challenges for telegenetics.

What is Telemedicine?

Telemedicine is the "use of electronic communication and information technologies to provide or support clinical care at a distance" (U. S. Department of Commerce, 1997). It is an alternative model of medical care that does not require the health care provider to be physically in the same room as the patient. Telemedicine is one aspect of "telehealth," a term that that has been used to include all uses of electronic technologies and telecommunications to support "patient and professional health-related education, public health, and health administration" (U.S. Dept. Health and Human Services, 2001), such as, for example, medical information provided on the internet. Although education and health management may be ways in which telehealth might be used in clinical genetics, this report will focus specifically on the application of telemedicine to provide direct clinical genetic patient services.

Telemedicine has been used for several decades for certain limited purposes, such as its use by the United States Military Operations to provide radiology consultations to remote sites, and the development of a Mobile Medical Station by NASA in the 1970's. The first documented use of telehealth was half a century ago when the University of Nebraska in 1955 created long distance psychiatry services (Caputo, 2005). However, the feasibility of broader application of telemedicine was severely limited by expense and unsophisticated technology. As technology has progressed, the use of telemedicine as an alternative model of service delivery, especially in rural areas with shortages of medical specialists, has become more common.

The U. S. federal government has shown an increasing interest in telehealth as a tool for health care over the last decade. In 1997 the Department of Commerce submitted its Report to Congress on Telemedicine, in which telemedicine was touted as an emerging model of health care delivery (U. S. Department of Commerce, 1997). Many new telemedicine programs in the United States have been started with the help of federal grants intended to improve access to medical care, especially in areas that meet the definition of a Health Professional Shortage Area (HPSA). In 2005, the Office for the Advancement of Telehealth (OAT) awarded 92 telehealth/telemedicine projects a total of over $34.9 million in funding. In addition to providing seed money for telehealth activities, federal agencies, including OAT and the Health Resources and Services Administration (HRSA) of the U.S. Department of Health, have examined the current trends in telemedicine and identified key issues that still need to be addressed, namely:

lack of reimbursement; legal issues; safety and standards; privacy, security and confidentiality; and telecommunications infrastructure (U.S. Dept. Health and Human Services, 2001).

Telemedicine is one way to increase access to health care limited by geographic, medical, legal, or other factors. A number of initiatives have addressed locally relevant limits to access. For example, Maine created state-wide telemedicine services through collaborating Teleheath Networks to meet clinical health care needs of individuals living in rural areas of the state. For people on small islands off the coast of Maine, a mobile telemedicine boat was available (Edwards and Patel, 2003).

In addition to addressing inequities based on geography, telemedicine can be used to provide medical care to individuals who do not have the ability, liberty or means to travel to a primary care physician or medical specialist, such as prison inmates, soldiers and the home bound. Telemedicine can also be school-based to ensure appropriate health care access for children.

Several medical specialties have used telemedicine as a means to provide clinical services – mental health, cardiology, endocrinology/diabetes, asthma, ophthalmology, otolaryngology, radiology, neurology, dermatology, orthopedics, home health care, and trauma & critical care. Telemedicine is more easily adapted for some medical specialties than for others. For example, telecardiology for the assessment of children with heart murmurs has been available for decades, since phonocardiograms, echocardiographs and other studies can be transmitted long-distance (McConnell et al., 1999). Teleophthalmology services have been provided by analyzing digital retinal images sent via the internet (Gomez-Ulla et al., 2002). Other specialty care, such as for diabetes or asthma management, require little medical equipment and sometimes little in the way of physical assessment, and focus more on progress updates and patient education (Gelfand et al., 2003; Romano et al., 2001).

Telemedicine can be non-synchronous (medical findings are sent electronically and are reviewed at a later time) or synchronous (patient and health care provider interact in real-time). The equipment needed for a telemedicine clinic depends on the type of services being provided. Non-synchronous telemedicine typically requires store & forward technology. This could include using fax, email, electronic patient records, medical images, telephone, the internet, and/or compressed video (Antoniotti, 2005). Interactive telemedicine visits require more sophisticated equipment, such as videoconferencing units (Caputo, 2005). Document cameras can be useful in showing visual aids for patient education. Equipment for physical examinations can include a digital camera, a document camera, digital electronic stethoscope, fiberoptic otoscope, fiberoptic opththalmoscope, intra-oral camera, laser caries detector, and so forth (Antoniotti, 2005).

State Laws Regarding Telemedicine

The practice of medicine is regulated by each state through medical boards and state statutes. The Telemedicine Licensure Report (Center for Telemedicine Law, 2003) gives a state-by-state overview of existing laws regarding the practice of telemedicine. All states require a license to practice medicine but many do not specifically address telemedicine. Twenty-nine States have laws specifically addressing the licensure requirements in regards to the practice of telemedicine,

including in Region 4, Illinois, Indiana, Ohio, and Minnesota. Illinois and Indiana require a full medical license to practice telemedicine. However, Indiana state laws stipulate that the practice of medicine requiring a license applies to services that are provided "on a regular, routine, and non-episodic basis or under an oral or written agreement to regularly provide medical services." Therefore, providing sporadic or occasional telemedicine services to Indiana residents by physicians licensed in another state may be permissible. Minnesota has a special purpose license for telemedicine, but physicians practicing telemedicine across state borders into Minnesota do not need this if they provide services less than once a month or see fewer than ten patients annually. Ohio requires a telemedicine certificate and has no explicit provisions for infrequent or sporadic telemedicine services. The other states in the region – Wisconsin, Michigan, and Kentucky – have no laws that explicitly address licensure needs for telemedicine, although the Kentucky Board of Medical Licensure adopted a Telemedicine Policy Statement in 1997 stating that "Physicians living outside Kentucky but actively practicing medicine upon patients within Kentucky should be required to meet the same statutory qualifications and should be held to the same standards of acceptable and prevailing medical practice within the Commonwealth as are resident physicians practicing within the state," implying the need for licensure in order to provide any telemedicine services.

Most states include a "consultation exception" in their statutes; an out-of-state physician may be asked to consult by a physician licensed in the state who is responsible for the patient's care and may legally do so without that consultant being licensed in that state. In Region 4, all states except Michigan exempt out-of-state physicians from the licensure requirement when providing consultation services to an in-state physician (although many states note that this consultation should not occur on a frequent basis without licensure). Michigan does not explicitly allow a consultation exception, but does allow for the practice of medicine by individuals not licensed in the state in an "exceptional circumstance" when called in by a Michigan physician, or when "attending meetings or conducting lectures, seminars, or demonstrations under the auspices of professional associations or training institutions" in Michigan.

Most physicians providing regular and recurrent interstate telemedicine services apply for licensure in the other state(s) where patients make telemedicine contact as well as holding a license in their home state. Applying for multiple state licenses is time-consuming and expensive, and so is a significant barrier to the development of telemedicine across state lines. This issue is being explored by the Joint Working Group on Telemedicine (JWGT) sponsored by the U.S. Department of Health and Human Services, which is working with states to assess feasibility of creating common licensure application forms. Additional legislation that has been proposed includes an abbreviated telemedicine licensure process based on the Federation of State Medical Board's model act, licensure reciprocity similar to the interstate compact of the National Council of State Boards of Nursing, and/or exceptions to licensure for non-routine telemedicine consultations.

Reimbursement for Telemedicine

Just as laws and regulations regarding licensure for telemedicine vary by state, so do the statutes regarding Medicaid reimbursement for telemedicine services. These standards are important not only because they determine payment for those individuals covered by Medicaid, but also

because insurance companies often use Medicaid standards as the basis of their own reimbursement policies.

According to the Telemedicine Reimbursement Report (Center for Telemedicine Law, 2003), 15 states have statutes regarding Medicaid reimbursement for telemedicine; 2 states have statutes only regarding private insurance reimbursement; 5 states have statutes for both Medicaid and private insurance; and 28 states (and the District of Columbia) have neither. The number of states which reimburse telemedicine services though Medicaid had increased to 24 by July 2005, as shown report of that date from TeleHealth Connections for Children and Youth. Four additional states are planning to implement reimbursement laws.

Within Region 4, Illinois and Minnesota have statutes regarding Medicaid reimbursement for telemedicine. The Michigan Department of Community Health mandated Medicaid coverage of telemedicine services effective May 1, 2006 (MSA 06-22). Kentucky has laws regarding both Medicaid and private insurance. All of these states reimburse only for physician consultations using interactive video teleconferencing and require that the consulting physician be licensed within the state. These states also reimburse for non-physician health care providers associated with the telemedicine consultation (typically limited to RNs and other allied health professionals), reimburse providers on both ends of the telemedicine visit, use a fee-for-service payment method, and have specific telemedicine CPT codes and/or use a telemedicine modifier. Kentucky and Michigan explicitly do not reimburse for store-and-forward consultations. Indiana, Ohio, and Wisconsin have no state statutes regarding the reimbursement of telemedicine services. (Center for Telemedicine Law, 2003; TeleHealth Connections, 2005).

Studies on Effectiveness and Cost-Utility of Telemedicine

Telemedicine clinics are often created to meet patient health care needs in remote areas. While telemedicine technology has the capability of reducing geographic barriers to care for some patients, it does not come without cost. Is the investment of telemedicine equipment, extra health care provider time, and start-up costs for a telemedicine clinic justifiable? Furthermore, are the outcomes of a medical evaluation via telemedicine (diagnosis, recommendations, etc.) similar to (i.e. of equal validity) outcomes of face-to-face evaluations? The latter is critical if telemedicine is being considered an alternative means of health care delivery.

Numerous studies have been published about telemedicine in general and its potential benefits, including savings of time and travel costs, and the effectiveness of non-face-to-face encounters. Hailey et al. (2002) reviewed and classified 66 “scientifically credible” studies of telemedicine that included comparison with a traditional model of care. The studies were ranked by level of scientific evidence from highest (I) to lowest (IX) depending on study design (large or small sample size, randomized or non-randomized, retrospective or prospective, and whether they were cohort, case-controlled, descriptive, or case reports) and scientific rigor. Most of the studies presented data that suggested the effectiveness of telemedicine, but few had scientifically rigorous design and few examined the long-term use of telemedicine. The limitations of these studies were mainly attributable to constraints of circumstance, such as small sample sizes, use of retrospective data, and lack of follow-up. Other problems in many studies included inadequate or missing details about the design and/or data from the study; estimates of cost and/or

effectiveness based on the most optimistic alternatives; and conclusions that did not appear supported by the data. Whitten et al. (2002) systematically reviewed 612 articles about telemedicine and identified 55 studies that presented cost benefit data, which on the whole were "marked by poor design and inadequate technical quality". The authors conclude that "most of the studies analyzed were small scale, short term, pragmatic evaluations that added little to our knowledge of the costs and benefits that would be expected to result from the introduction of telemedicine services into routine clinical practice." Obviously, further studies of larger scale and improved design are need to provide adequate evidence that telemedicine is an appropriate means of health care delivery.

Barriers to Telemedicine

Several obstacles to the successful implementation of telemedicine have been identified:

- the cost of creating and maintaining telemedicine services;
- technological limitations;
- problems in reimbursement;
- licensure and liability issues;
- lack of willing and knowledgeable providers.

Cost. The start-up costs of installing transmission lines and buying videoconferencing and other equipment can be a serious investment of time and money. For many states or centers, this can be the determinative barrier to instituting telemedicine services. The cost is dramatically greater if transmission lines need to be installed or expanded, but even if there is a pre-existing transmission network, the cost of videoconferencing and other telemedicine equipment is significant. In addition, there is a need to have trained health care professionals on both ends of the telemedicine visit. Training is an added cost, and getting reimbursement for the individuals providing and facilitating the telemedicine services is often a problem.

States and centers have found different ways of creating and equipping telemedicine networks. Most telemedicine projects have been funded with the help of state or federal grants, such as those from the Office for the Advancement for Telehealth (OAT) (e.g. Georgia), or by state contracts with Medicaid (e.g. Arkansas). A few are private ventures (e.g. Marshfield Clinic in Wisconsin). Some have taken advantage other types of opportunities, such as Nebraska's use of a telemedicine network developed among state hospitals as part of an anti-bioterrorism federal grant. In Nebraska and in several other states, multiple medical specialties share the telemedicine network and equipment to make its use more efficient and cost-effective. Other programs purchased their own equipment but made use of pre-existing ISDN phone lines (Lea et al., 2005).

Technology. In the past, the quality of telemedicine services was in part determined by the quality of transmission of information. If the transmission speed is too slow, there is a time lag between the patient and the provider. If the transmission quality is poor, then an evaluation via telemedicine is less effective. Improvements of resolution of telemedicine equipment and increased transmission speed have helped improve the quality of telemedicine consultations. Still, equipment malfunctions can occur (Babineau and Ludman, 2004), and installation of new

transmission lines and equipment is typically associated with a period of trouble-shooting (Stalker et al 2006).

Another consideration relates to transmitting images that are both secure and of high quality. Transmission speed is usually measured in kilobits per second. Transmission via POTS (plain old telephone system) tends to be too slow (around 20-50 Kbps) for high quality interactive video images (Antoniotti, 2005). Most telemedicine clinics use multiple high-speed ISDN lines (around 128 or 256 Kbps), which are expensive to install but considered reasonably fast and fairly secure, security being effected by only a portion of the transmission being sent on each line. Some telemedicine clinics use T1 lines (1544 Kbps), which transmit information faster but can have potential security issues since images are transferred only on one line. The transmission of telemedicine images and information via the internet is a tantalizing option since any site with internet access could be used as a telemedicine clinic; however, the security of the transmission is a large and currently unsolved issue. Software is being developed to provide for secure transmission of medical images and videoconferencing (such as SSH tunnel technology available on Macintosh computers and a program being adapted for PC computers [personal communication, Heather Stalker, Sylvia Au]). If a truly secure method of transmission on the internet of telemedicine consultations and information is developed, it could greatly reduce costs and simplify initiation of telemedicine services.

Reimbursement. Costs can be a major barrier in creating and maintaining telemedicine clinics, and so reimbursement of services is a major issue for telemedicine. As noted, many states have laws regarding reimbursement for telemedicine services. In those states that have laws favoring telemedicine reimbursement, such as Texas, Georgia, and Hawaii, revenue is typically sufficient to sustain telemedicine services. Reimbursement for telemedicine services in states without Medicaid laws is typically poor, even from third party payers, and creates a significant obstacle to paying for telemedicine services. Often telemedicine clinics are started with the help of state or federal funding, but if the funding ends, there needs to be an alternative reimbursement strategy to maintain the program (personal communication, 9/29/06, Dale Lea). Some centers have found success by implementing contracts with remote sites by fee-for-service, regardless of the patient's insurance coverage (personal communication, 9/12/06 Heather Stalker).

Licensure and Liability. Licensure issues have been discussed. The need for multiple licenses is a significant hindrance to interstate telemedicine. Even in Canada, in which there is a national system for health care, licensure across province lines and reimbursement issues was noted to be a limitation (Babineau and Ludman, 2004). Some health care providers have been working to simplify licensure. The Federation of State Medical Boards (FSMB) has developed a model act for abbreviated licensure process for outside doctors providing telemedicine services (1996). So far, eight states have approved acts based on the FSMB model and require a "special purpose," but not full, license to practice telemedicine. The National Council of State Boards of Nursing (NCSBN) has developed a mutual recognition model for licensed nurses, and currently 20 states are using this interstate compact, including Wisconsin.

Even for telemedicine practice within a state, credentialing and liability issues with telemedicine sites can be problematic (Lea et al., 2005). The Joint Commission on the Accreditation of Healthcare Organizations (JCAHO) has standards for telehealth credentialing and privileging of

providers, which states in MS.5.16 that "Practitioners who diagnose or treat patients via telemedicine link are subject to the credentialing and privileging processes of the organization that receives the telemedicine service (JCAHO, 2003). That means that a provider needs to apply for and receive privileges not only in the home institution but for every remote site – a process that can be as complex and onerous as multiple state licensing.

Little has been written regarding liability issues for telemedicine. Coverage likely would need to explicitly address cross-state provision of care.

Willing and knowledgeable providers. Although most telemedicine studies have found that patients tend to be satisfied with their telemedicine encounters, health care providers have been resistant to embracing telemedicine (Gray et al., 2000; Babineau and Ludman, 2004; Lea et al., 2005). Part of this resistance may be due to unfamiliarity with telemedicine equipment and use. Several centers have found that demonstrations and presentations given to health care providers to educate about telemedicine services have been helpful (personal communication, 8/29/06, Steve Anderson), and Maine providers tended to rate telemedicine services higher after they had personally provided telemedicine services (Lea et al., 2005). Education and training for telemedicine facilitators/providers can also be provided by centers specializing in telemedicine, such as the Telemedicine Learning Center at the University of California, Davis (personal communication, 9/21/06, Sylvia Au).

Telegenetics

Telegenetics is the use of telemedicine for the provision of clinical genetics services. Clinical genetics is a medical specialty with several sub-specialty areas, such as prenatal genetics, biochemical genetics, cancer genetics, and pediatric genetics. Many genetic providers are even further specialized, with expertise in a class of genetic syndromes or even one particular genetic disorder. Clinical genetics has few providers, and genetic clinics tend to be located at major medical centers in cities, creating geographic barriers for individuals in rural areas. Access to a geneticist with true expertise in a specific genetic disorder is more dependent on chance than need. These issues are not improving. Indeed, fewer and fewer genetic practitioners are being trained (American Board of Medical Genetics website, 2006). As one alternative, several genetics centers have created telegenetics outreach clinics to meet patient needs.

Experience with Telegenetics Nationally and Internationally

Several early reports about telegenetics originated outside of the United States. One of the first pilot studies of telegenetics, conducted in 1998 in Wales, provided cancer genetics services (Gray et al., 2000). Four genetics nurse specialists and two genetics consultants participated in providing cancer genetic counseling via videoconferencing from a center in Northern Wales to patients located in Southern Wales. Another pilot telegenetics service originated from the Queensland Clinical Genetics Service in Australia, which offered genetic consultations via videoconference when a physical examination was not required, mostly for cancer genetics counseling (Gattas et al., 2001). A third was a program provided by the Maritime Medical Genetics Service (MMGS) based in Halifax, Nova Scotia with 8 outlying rural locations (Babineau and Ludman, 2004). The MMGS program began by piloting distance cancer genetics

services, and then expanded to other types of genetic cases for which a physical examination was not required, including prenatal genetic counseling, follow-up of metabolic disorders, result sessions for Huntington Disease predictive testing, and group counseling for hereditary hemochromatosis.

Most of the early pilot studies of telegenetics studies were created to address geographic barriers to access to care. At the time of the report, the MMGS in Nova Scotia delivered care to approximately 2 million people and was the source of genetic services to people as far as 900 km away (Babineau and Ludman, 2004). The authors report that the telemedicine program was developed out of necessity, as some patients were not willing to travel that far and phone discussions were not felt to provide optimal services. This was similar to the experience with the Queensland Clinical Genetics Service in Australia, which was the only genetics service provider in the state with a population of approximately 3.5 million people (Gattas et al., 2001).

A common theme of the telegenetics reports from other countries was that consultations requiring a physical examination were not provided via telemedicine. This was both because of the cost of high resolution cameras and equipment, as well as the special training of the support staff that would be needed (Babineau and Ludman, 2004).

Although telegenetics pilot studies were first reported in centers abroad, a few pilot studies of telegenetics services have been developed in the United States. Georgia was one of the first states to implement statewide telemedicine beginning in the 1980s, funded by a Title V health care program, Children's Medical Services (CMS). An early project called the Georgia Statewide Telemedicine Program (GSTP) published a study on pediatric telemedicine consultations from 1995-1997 (Karp et al., 2000). Overall, 333 children with special health care needs were evaluated via telemedicine by various pediatric specialists, and 52 of those consultations (16%) were specifically for genetics. The consultation duration times ranged from 5-45 minutes, and included diagnoses such as Cornelia de Lange syndrome, Williams syndrome, chromosomal translocation, and Sanfillipo disease. The article reports that 8 diagnoses were made via telemedicine involving dysmorphic features and/or multiple congenital anomalies, although it was unclear how many children already had a diagnosis before the telemedicine consultation. Recent telephone follow-up with the Medical College of Georgia found that telegenetics clinics to Waycross, GA are still taking place once a month, although they serve as an adjunct to traditional outreach clinics and are used primarily for follow-up and routine consultations (personal communication, 9/21/06, David Flannery).

The first U. S. experience with a program exclusively of telegenetics was a 3-year pilot project in rural Maine funded by the March of Dimes (Lea et al., 2005). In addition to direct telegenetics services, the project included a strong educational component, with numerous genetics and educational presentations made across the state. The project made use of pre-existing ISDN phone lines to 24 rural sites and utilized Maine public health nurses to assist with telegenetics consultations. During the three year period, 125 patients were evaluated and/or counseled via telemedicine. These consultations were primarily for pediatrics genetics and/or neurologic genetic issues (64%), although the consultations also included ones for cancer genetics, other adult genetics, and reproductive genetics (prenatal and preconception). While diagnostic effectiveness was not specifically measured in this project, it was felt that telemedicine was

effective as a dysmorphology screening tool, in that patients could be referred for a further evaluation in person if a dysmorphologic genetic syndrome was suspected (personal communication, 9/28/06, Dale Lea).

A recent telegenetics project in northwestern Florida explored the effectiveness of telemedicine for genetic diagnosis (Stalker et al 2006). Fifty patients were evaluated via telemedicine over a two year period, with a genetic counselor taking a family history and a pediatrician performing a physical examination to screen for potential genetic syndromes. The telemedicine clinics alternated every other month with traditional face-to-face outreach clinics. After two years the average waiting time for a new patient consultation at outreach sites decreased from 16.9 months to 3.0 months. The first 8 patients evaluated via telemedicine were also assessed by a clinical geneticist by a face-to-face consultation. Subsequently only patients with a complex or possible dysmorphologic genetic diagnosis, as noted by telemedicine contact, were seen for an in-person evaluation. The authors reported that no diagnoses were missed by the telemedicine evaluations that were detected by the face-to-face consultation or deemed to be inaccurate. Although the sample size was small, the authors found that "the telemedicine approach was successful in establishing the diagnosis of several dysmorphologic syndromes and in identifying individuals in need of further 'hands on' dysmorphologic review." The telegentics/outreach clinic in Pensacola is still currently operating, and telegenetics services were expanded to the NICU at a Pensacola hospital for genetic consultations with neonatologists (personal communication, 9/12/06, Heather Stalker).

A novel and highly successful application of telemedicine is the Antenatal and Neonatal Guidelines, Education and Learning System (ANGELS) in Arkansas (Britt et al., 2006). With support from Medcaid, this program provides maternal-fetal medicine (MFM) consulting services to physicians at numerous sites across Arkansas. Two MFM specialists at the University of Arkansas for Medical Sciences have real-time ultrasound and video access to 21 remote sites (another 21 sites have videoconferencing but not real-time ultrasound equipment), resulting in almost 100 consultations per month. A genetic counselor is present for most telemedicine consultations. Additional components of this program include training and support for physicians and sonographers, distribution of evidence-based guidelines, and research (personal communication, 9/8/06, Diana Jackson Moore).

A number of centers have begun to offer genetic services via telemedicine that are not documented in literature. In the GeneTests clinic directory, funded by the National Institutes of Health, genetics centers in 23 states are listed as providing telemedicine services. Telephone follow-up found that many of these genetics centers in reality did not currently offer clinical telegenetics services and were listed for various reasons: videoconferencing was available but not used for direct patient care; telemedicine services were offered in the past but discontinued; telemedicine capabilities were being explored and expected to be offered in the future; the clinic was simply listed in error. Of 33 clinics listed by the GeneTests clinic directory as offering telemedicine services (not including outreach sites), 18 (55%) are currently offering telegenetics for direct patient care. There were 3 additional centers identified in the United States that offer telegenetic services not specified in the GeneTests clinic directory (in Hawaii, Oregon, and Texas).

There are an unknown number of additional telegenetics programs not identified either in the literature or through GeneTests. One example is the Cook Children's Medical Center in Fort Worth, Texas, which has a HRSA grant for a pilot project of telemedicine for subspecialties including genetics to a site several hours away (personal communication, 8/29/06, Steve Anderson). Hawaii offers intrastate telemedicine for genetics services with neighbor islands 2-3 times a year, primarily for follow-up visits (personal communication, 9/21/06, Sylvia Au). Often, telegenetics services serve as "gap fillers" to supplement outreach services (in Oregon, Texas, Colorado, and Missouri). In several large states with smaller populations and fewer health care providers (such as Montana and Nebraska) telemedicine is used to meet the basic clinical genetics needs. Some centers provide telemedicine for cancer risk assessment counseling only including the Allegheny General Hospital in Pennsylvania, University of Kansas Medical Center, and the Memorial Sloan-Kettering Cancer Center in New York.

All of the pilot studies regarding telegenetics services have reported high patient satisfaction as ascertained with questionnaires (Gray et al., 2000; Babineau and Ludman, 2004; Lea et al., 2005; Stalker et al., 2006; Coelho et al., 2005). Surveys of genetics providers were not always as positive. Antecedent training may be critical in acceptance by care providers. For example, after extensive educational efforts of care providers, a survey of those involved with the Maine telegenetics project found that they felt confident in evaluating and managing patients using telemedicine (Lea et al., 2005). However, training may not allay all provider concerns. In several studies, the genetics providers expressed concerns about establishing rapport (Gray et al., 2000; Babineau and Ludman, 2004).

Another common theme across telemedicine clinics in several states was that telemedicine works best as a complement to face-to-face service delivery, not as a replacement of traditional clinics (Karp et al., 2000). Even those studies reporting success with genetic diagnosis via telemedicine recommended in person follow-up and physical examination for individuals with a newly suspected genetic disorder (Lea et al., 2005; Stalker et al., 2006). Telemedicine can be an effective screening tool for individuals with possible genetic disorders as well as being useful for follow-up of cases to promote efficiency of the clinics and reduce wait time (Stalker et al., 2006; personal correspondence, 9/21/06, Sylvia Au). Many providers offering telegenetics services alternate the telemedicine clinics with face-to-face outreach clinics at the distant site, which they think best meet the needs of the patients (Karp et al., 2000; Stalker et al., 2006; personal correspondence, 9/21/06, Sylvia Au). Whether telegenetics can provide stand-alone, comprehensive genetic services long term has not been assessed.

Regional Telegenetics

Although licensure requirements make the delivery of medical care across state lines more difficult, there are some examples of interstate telegenetics programs. In particular, the Western States Genetic Services Collaborative (Oregon, Washington, California, Alaska, Hawaii, and Guam) is incorporating telemedicine services in their goal to "refine, pilot, and evaluate a regional practice model that improves access to specialty genetic services, comprehensive primary care, and care coordination for children with heritable conditions living far away from comprehensive genetics and metabolic centers" (Western States Genetic Services Collaborative website, 2006). This model will recommend the appropriate number of urban genetic sites, the

number of outreach sites, the number of providers, and extent of telemedicine consultation with the aim being to provide the most efficient and effective care for the target population (personal communication, 9/21/06, Sylvia Au). Currently, the Idaho Department of Health has contracted to have telegenetics services from an Oregon provider. Also, Washington will begin providing telegenetics services to Alaska this year, and telemedicine will be used from Hawaii for genetic consultations to Guam. Outside of the Western States region, Nebraska also offers interstate telegenetics services; in addition to numerous sites within its state, the University of Nebraska Medical Center also has 3 telemedicine sites in South Dakota and 8 in North Dakota that are used periodically for genetics consultation.

Telegenetics in Region 4

Currently, the only center in Region 4 offering telegenetics services is the Marshfield Clinic in Wisconsin. These services are just starting, with the first patient scheduled for November 2006 (personal communication, 10/10/06, Kristen Meddaugh), and therefore no data has been collected. Reimbursement for telegenetics services has been a concern (personal correspondence, 5/16/06, Philip Giampietro), especially since Wisconsin does not have any state laws regarding Medicaid reimbursement of telemedicine services. The Children's Hospital of Wisconsin is exploring the use of telegenetics and hopes to have 1-2 telegenetics clinics started by the end of the year (personal correspondence, 8/29/06, Cristin Griffis). No other centers in Region 4 are known to be in the process of arranging telegenetics clinics.

Special Barriers to Telegenetics and Special Opportunities for Telegenetics

Little data regarding telegenetics barriers are available. Nonetheless, certain problems seem likely. Unlike many medical specialties, genetic services greatly vary and are not organ-specific. An ophthalmologist examines only the eyes; a cardiologist the heart, a pulmonolgist the airways and lungs. A geneticist may need to perform a complete and detailed physical and neurological examination not only of the initial patient (proband) but also of members of the proband's family. Presumably because of their complexity, currently few centers provide initial consultations requiring physical examination via telegenetics; it is used primarily to provide genetic counseling and routine follow-up care.

Another challenge specific to genetics is the shortage of trained professionals. As fewer clinical geneticists are being trained, those who are in practice are more stretched for time and less able to meet needs outside of their center. Telegenetics clinics may add an additional burden that may undermine efforts to provide equitable service.

Because clinical genetics is a relatively small field, there likely will be increased need for interstate service. A clinical geneticist may provide services in neighboring states, such as Nebraska offering telegenetics clinics in South Dakota and North Dakota (personal communication, 9/13/06, G. Bradley Shaefer). All of the issues of multistate practice may then need to be addressed.

As noted earlier, many genetic providers are even further specialized within genetics, and access to a genetics professional with expertise in a specific genetic disorder or class of disorders (a

"supersubspecialist") may be even more difficult. Telemedicine might provide a means for patients with rare genetic disorders to access supersubspecialists in different areas of the country. For an example, an individual in Ohio who has a diagnosis of Albinism may wish to consult with clinical geneticists at the International Albinism Center in Minnesota. However, these types of consultations by nature would not be routine or from one specified telegenetics clinic, and there would be significant barriers of licensure, credentialing, and reimbursement if telegenetics was used to provide non-regular access to supersubspecialists. Clinical geneticists would probably be less motivated to spend the time and money applying for licensure in other states if they were only going to consult with one or two patients in those states.

Lack of knowledge and increased training needs may be another barrier specific to telegenetics. A few studies on telegenetics commented on the need to have a knowledgeable health care provider or assistant trained in genetics at the telegenetics clinic to assist with measurements and observations during a physical examination (Stalker et al 2006; Lea et al., 2005). In addition, the lack of understanding of the role of genetic services on the part of physicians was also found to be an issue with telegenetics; if providers do not know who to refer for a genetics evaluation, patients will not obtain appropriate care even if telegenetics services are available (Lea et al., 2005)

Needs for Additional Telegenetics Research

Most of the reports that have been published to date regarding telegenetics have been pilot projects with small sample sizes. Because of the limited scope of telegenetic services at this point, large-sample, randomized controlled trials at multiple centers have not been possible. Two telegenetics studies were case-controlled studies, with patients assigned to either telemedicine or traditional face-to-face clinics (Gattas et al., 2001; Coelho et al., 2005). However, both of these studies had very small sample sizes. Another study in which patients were evaluated both by telemedicine and in person to examine the diagnostic effectiveness of telemedicine (Stalker et al 2006) also evaluated a rather small sample.

Additional studies on the costs and the effectiveness of telegenetics are needed. A recent editorial in the journal *Genetic in Medicine* calls for prospective, well designed studies of telegenetics consultations that would look at accuracy of diagnosis, diagnostic impact, and patient outcomes (Mitchell and Demiris, 2005). The authors write that the challenges lie in "extensively evaluating telegenetics technologies, properly adopting them, and making informed decisions about their appropriate use." Ideal studies not only would have adequate sample size but would be designed to study the effectiveness of the telemedicine encounter beyond measuring patient satisfaction, have random assignment to telemedicine or face-to-face consultation, and be evaluated by a neutral party to determine if quality of care was affected by the type of consultation. Multi-provider and multi-center studies would be preferable. Although studies could not, obviously, be blinded, large, controlled and carefully analyzed investigations are feasible if funding for their development is made available.

Is there a future for Telegenetics?

Although the studies on telegenetics are few in number and small in sample size, these early reports suggest that telemedicine may be appropriate as an additional genetics service delivery

tool to reduce inequalities in access to care. Patients receiving telegenetics services have been satisfied with their experience and expressed appreciation at having reduced wait times and/or shorter travel distances. Genetics providers have been more reluctant to use telemedicine technologies, but most have been accepting of the practice after appropriate education or experience. Most programs have been hesitant to use telemedicine for genetic physical examination and physical diagnosis. Telemedicine may be used most appropriately as a tool to screen for individuals with potential genetic diagnoses, to see patients for follow-up, and to provide routine counseling. Telemedicine may also be used for second opinions on genetic conditions and special consultations with supersubspecialists. However, until tested, its utility in replacing all traditional genetic clinic services is unknown.

In order to create sustainable telegenetics clinics, reimbursement of telemedicine services (including providers on both ends) needs to be sufficient. The states who have reported the most success with reimbursement for telemedicine services have been those with state statutes requiring Medicaid reimbursement. Therefore, providers and other interested parties in states that currently have no such statutes (Indiana, Ohio, and Wisconsin in Region 4) may wish to advocate for this legislation. Some states, such as Hawaii, have telemedicine advocacy groups with the primary purpose of promoting telemedicine legislation (personal communication, 9/21/06, Sylvia Au). Even without state statutes regarding Medicaid reimbursement, centers interested in providing telemedicine services can approach local third party payers to lobby for reimbursement. Otherwise, centers may need to work on fee-for-service contracts with the remote sites or use federal and/or state contracts.

To further the successful growth and implementation of telegenetics, it would be helpful to develop specific standards and guidelines for telemedicine in the field of clinical genetics. Several organizations, such as the American Psychological Association and American Dermatology Association, have created specific telemedicine standards and guidelines for their fields of specialty. The ANGELS network in Arkansas has created a model program for the delivery of maternal-fetal medicine care, and the Western States Genetic Services Collaborative model of genetics service delivery will include recommendations for telemedicine based care. In addition, the Office for the Advancement of Telehealth is working with groups to expand clinical guidelines and promote safety and security. No similar initiative explicit to clinical genetics services has yet been initiated.

Should Telegenetics be Regionalized or Nationalized?

Telegenetics services have been successfully implemented as state-specific programs. The question remains, should telegenetics be regionalized or nationalized? Is there a valid reason or incentive for the creation of a regional or national network of telegenetics? The answer may depend on the characteristics of the region as well as the purpose of the telegenetics services – whether it is to provide general genetics care to geographically remote areas or, alternatively, to allow greater access to the expertise of genetics supersubspecialists.

Telegenetics within a multistate region (or nationally) would be essential for providing specialized expertise to small populations of individuals affected by relatively rare genetic conditions. Currently, access to genetic supersubspecialists is differentially available to individuals with rare genetic conditions who have the financial means to travel or who, by chance, are geographically close to the relevant subspecialist. Anecdote suggests that care through such a specialist is superior to that otherwise provided; however, no studies have been completed to test whether this assumption is true.

Regionalization for *general* genetics care may be appropriate only in circumstances of exceedingly sparsely populated regions. The most obvious examples are states with few or no clinical geneticists, such as Alaska, Idaho, and the Dakotas.

The more practical question may be: can telegenetics be regionalized or nationalized? Developing a regional plan for telegenetics poses challenges, related to constraints of licensure and the cost of a telehealth network. For a number of reasons (including licensure, reimbursement and economies of scale) the development of routine, long-term interstate telemedicine clinics with a few designated genetics providers may be more feasible than non-routine telemedicine consultations provided by an array of regional genetics specialists. However, this is counter to one of the justifications for telegenetics on a regional or national level – access to genetics supersubspecialists. This type of network would require involvement of a large number of genetics specialists, most of whom would participate in non-regular consultations at multiple telegenetics sites across the region or nationwide. Barriers are substantial, perhaps insurmountable. Benefits are probably substantial, too, but have never been measured or assessed.

Recommendations for Possible Future Activities of the Region 4 Collaborative

1. Needs assessment by professionals and patients

To determine if clinical genetics care needs are being met for populations in Region 4, there should be documentation of the geographic distribution of genetic providers, including distribution of supersubspecialists by area of expertise. (This has been partially completed by the database of Region 4 genetics providers including areas of expertise.)

2. Query interest in regionalization for possible telegenetics activities

Genetics providers can be surveyed to determine interest for regional telemedicine activities.

3. Demonstration/assessment of inadequacies from a patient perspective

Do patients feel that access to clinical geneticists with condition-specific expertise is necessary? For individuals in geographically remote areas, is travel to genetics centers a hardship or barrier to quality care?

4. Explicit identification of the structure and purpose of a regional program

Should a regional telegenetics project aim to provide general genetics care to geographically remote areas or to allow greater access to the expertise of genetics supersubspecialists? How many providers and centers in the region would be equipped for telemedicine consults? What will be the aim and scope of a regional telegenetics program?

5. Preliminary cost-consequence assessment

The expenses related to the creation and maintenance of telegenetics sites should be assessed, including costs of equipment, transmission lines, training, and personnel. Expected benefits/outcomes of a telegenetics program should be quantified.

6. Consideration of non-direct care components of telegenetics

Other aspects of telehealth that may increase knowledge and expertise of genetic providers in the region should be considered. These might include: teleconferences, regional case conferences, educational seminars, etc.

7. Develop testable hypotheses

If a telegenetics program is to be created as a part of a regional model, the program should be designed to measure effectiveness and cost-utility.

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