



# Evaluation of Satisfaction of Parents With the Use of Videoconferencing for a Pediatric Genetic Consultation

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Telegenetics is a new development in the service delivery of Genetic Services in Australia. This project was designed to establish if it was an acceptable alternative to a face-to-face consultation in the genetic assessment of intellectual disability, including morphological assessment, of the patient. Ten children from two outreach clinics in rural NSW who were referred by their pediatrician were assessed by a single geneticist via telehealth and then seen again face-to-face as a 'gold standard'. Satisfaction surveys were then sent to both the parents and the referring pediatricians. After the face-to-face appointment, the clinical geneticist reviewed the recordings of both the transmitted footage and the high definition footage that was sent separately. There were very few morphological findings missed by the telegenetic assessments. The discrepancies that were noted could decrease in frequency as staff become more familiar with the methods. The parents of the patients reported no problem with the cameras and telehealth. They would have preferred face-to-face appointment but would be happy to have the telehealth appointment if it meant being seen earlier. This pilot study suggests that clinical genetic diagnostic assessment could be performed by telemedicine.

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The clinical application of telemedicine as a tool for genetic counseling is rapidly increasing (Zilliagus et al., 2009). The use of telemedicine for genetic counseling of clients with cancer has been evaluated in Australia and found to be generally accepted by clients and clinicians (Zilliagus et al., 2009, Gattas et al., 2001). Telemedicine has also been used for diagnostic purposes in areas such as pediatric genetics (Lea et al., 2005, Stalker et al., 2006), teledermatology (Warsaw et al., 2010), telepsychiatry (Boydell et al., 2010) and cardiology (Dowie et al., 2008); however, there are limited studies investigating telemedicine as a diagnostic tool for clinical genetics.

Genetic counselors in rural New South Wales are employed by local health services and coordinate clinics with a clinical geneticist who travels to the area from a genetic service based in a city. The demand for rural genetic clinics has increased significantly since their inception in the late 1980s; however, there has not been a concomitant increase in the number of clinical geneticists in that time.

A physical examination by a clinical geneticist is an essential diagnostic tool for children and adults with developmental delay. Pediatric patients usually attend a face-to-face appointment with a clinical geneticist and local genetic counselor. For rural families that can mean many hours traveling when the patient has to visit a metropolitan doctor, or a significant wait until a clinical geneticist can visit the patient's area. Waiting lists for patients to see a clinical geneticist in rural NSW are increasing, and genetic services are exploring other ways to reduce waiting lists.

Telegenetic services were first introduced to the Hunter New England area in 2004, for referred clients who did not require a physical examination by the clinical geneticist.

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The introduction of this service enabled clients referred for cancer genetic counseling to be seen very promptly by the clinical geneticist (Zilliacus et al., 2009), however there was little impact on the waiting period for a general genetics (face-to-face) appointment with the clinical geneticist.

From 1 July 2011 the Australian Federal government will introduce Medicare items for telemedicine consultations (Department of Health and Ageing, 2010). Prior to this Telemedicine consultations in Australia used ISDN connections of 384 kps, costing approximately A\$115 per hour. Internet protocol (IP) teleconsultations have enabled speeds up to 756 kps. This faster speed brings greater clarity of image, and has no direct cost to the service.

A pilot telegenetics project in the Hunter New England Area aimed to determine the feasibility of telemedicine as an assessment tool for the clinical and morphological assessment of children with developmental delay. Real-time high-definition images of the children were transmitted by videoconference and the results were compared to those of a face-to-face appointment with a clinical geneticist.

After the appointment, carers of children participating in the project were surveyed regarding their opinion about whether they thought a telegenetics consultation was acceptable alternative and whether they were satisfied with the consultations.

## Methodology

The genetic counselor at Taree NSW, the genetic counselor at Tamworth NSW, and a clinical geneticist at Hunter Genetics in Newcastle NSW participated in the project. The genetic counselors were chosen because they were experienced sole practitioners of an outreach service and were familiar with the types of examination and measurement performed during clinical genetic assessments. The outreach services were located within the same area health service as Hunter Genetics.

The Taree genetic counselor was provided with training on how to use the video equipment by the telemedicine coordinator of his region, and a representative of the company marketing the video equipment. The genetic counselor at Taree then trained the genetic counselor at Tamworth how to set up and use the camera and DVD with the telemedicine machine.

All participants were referred to the genetics service by their pediatrician, and a prior pediatric assessment had been conducted. Carers of the referred child received information about the study and signed a consent form that was discussed with them by the genetic counselor prior to the appointment. The study was approved by the Area Health service ethics committee.

A high-definition video camera (Sony HVR-A1p HDV 1080i camcorder) was used to transmit and record high-definition video content, as well as take still images

simultaneously. The camera delivered a live feed of the session to the geneticist via the telehealth equipment. The consultation was recorded on a high definition DVD, and still photos were included. The DVDs were sent to the clinical geneticist, and a copy went to the parents of the pediatric patients.

The consultation was directed by the geneticist talking to the patient and carer for the telegenetics appointment. The genetic counselor took physical measurements (and still images) and used a hand-held camera to transmit images to the geneticist.

A face-to-face appointment was arranged for participants with the same clinical geneticist and genetic counselor 3 weeks after the telemedicine appointment. The clinical geneticist wrote to the referring doctor and carer documenting the outcome of the appointments. Surveys were sent to carers of the patients who participated in the project and their referring pediatrician 3 months after the appointments. Carers were also interviewed by the research coordinator (BH) over the telephone (due to poor response rate to postal questionnaire) 1 year after the initial appointment, after all the genetic test results became available.

## Results

Ten children and their carers participated in the project; seven girls and three boys between the ages of 8 and 14 years. Measurements taken by the genetic counselors and clinical geneticist varied slightly but not sufficiently to affect the assessment and alter a diagnosis. Numbers of participants were too small for statistical analysis.

A genetic diagnosis was not evident for all of the participants at the telemedicine consultation, nor the follow-up face-to-face appointment with the clinical geneticist. Clinically indicated tests were requested for several patients. Following both appointments, comparative genomic hybridization by microarray analysis confirmed a 17q microdeletion in one patient.

Four of the ten carers who participated in the project returned the 3-month questionnaire. All of the respondents reported that they were satisfied with the telegenetic concept for clinics. All of the respondents agreed that they had enough opportunity to ask questions at the telemedicine and face-to-face appointment. Half of the respondents agreed that telemedicine was an effective medium to see a geneticist for the purpose of trying to ascertain a genetic diagnosis for their child. One carer felt that a face-to-face appointment was important for a first appointment with the clinical geneticist 'because cameras don't show everything'. Another carer speculated that the effectiveness of telemedicine 'depends on how a child deals with TV intrusion', and a follow-up appointment with the geneticist in person was important.

Eight carers were contacted by telephone for a 12-month follow-up survey identical to the first survey, and

two were lost to the study. This was after all results were back from relevant tests and results returned to participants. All respondents were either satisfied or very satisfied with the telemedicine experience. Respondents were asked their opinion about improving the provision of telegenetics; however, no suggestions were made. All carers were given enough time to ask questions at both appointments and were happy with the information given at the telegenetic clinic. One parent commented that they 'did not know what kind of questions to ask' in the telemedicine appointment. The majority of participants (88%) agreed that telegenetics was an effective way of seeing a geneticist for assessment.

There was a 50% response rate from the referring pediatricians to the 3-month post survey. Three pediatricians had not seen the patient since referred to the genetic service, and declined to comment. One responded to the letter that was sent to them after the session stating that they had some reservations with the technology as 'several features were missed by telemedicine according to the letter I received'. These features were: double whorl, linear sebaceous nevus on forearm, dermal hypoplasia left calf and 5th toe clinodactyly. The patient and referring pediatrician had not mentioned these features.

## Discussion

Overall satisfaction with the technology was high, from the respondents, pediatricians and genetic clinicians. Some of the children needed to be coaxed to cooperate with the filming process. This behavior was likely to be a reflection of the intrusiveness of the large camera. This component could be overcome by new and emerging technologies that require a much smaller camera, which would be less confronting to the patient and family and not as bulky for the counselor to handle. A satisfaction survey of the participating children was not performed.

For the technology to realize its fullest potential both the clinical geneticist and the genetic counselor need to be familiar with the technology and know how to solve problems that arise during the consultation. The clinical geneticist and genetic counselor need to be assured of each other's technical and clinical skills. This has been recognized previously as an important factor for the effectiveness of telegenetics (Zilliaccus et al., 2009). Genetic counselors need to be able to take diagnostic measurements, and will need additional training to standardize procedures and achieve uniformity across various centres.

Not all clinicians would necessarily be confident enough to participate in this technology as it makes the contact seem more remote and can potentially turn the clinical geneticist to more of a 'visiting specialist' (Zilliaccus, 2009).

There should be an opportunity for the genetic counselors and clinical geneticist to review the recordings of the telemedicine consultations. With extensive practice

and debriefing after the review of the recorded consultations on DVDs, the clinicians would be more confident and efficient in identifying specific features. In retrospect, with more experience of telegenetics shared between the geneticist and the genetic counselor, the double whorl, linear sebaceous nevus on the forearm, dermal hypoplasia of the left calf, and 5th toe clinodactyly would have been identifiable via the camera.

This technology could be used in the future in home visits by the genetic counselor, or when the genetic counselor travels to remote locations and then links the geneticist by telemedicine. It could also be used for remote communities with less medical facilities. Other applications include the assessment of fetal morphology by ultrasound (Al-Kadi et al., 2009; Chang et al., 2001). Although the resources permitted study of only a small group of patients, this pilot project has demonstrated that telegenetic appointments for pediatric patients are generally accepted by clinicians and patients, and should encourage further research into the use of telemedicine for pediatric patients.

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