Telemedicine for genetic and neurologic evaluation in the Neonatal Intensive Care Unit

Tara L. Wenger, MD, PhD¹,², Jeffrey Gerdes, MD, MBA³, Katherine Taub, MD⁴, Daniel T. Swarr, MD¹,³, Matthew A. Deardorff, MD, PhD¹, and Nicholas S. Abend, MD⁴

¹Division of Genetics, The Children’s Hospital of Philadelphia, Philadelphia PA
²Section of Clinical Informatics, Division of General Pediatrics, The Children’s Hospital of Philadelphia, Philadelphia PA
³Division of Neonatology, The Children’s Hospital of Philadelphia, Philadelphia PA
⁴Division of Neurology, The Children’s Hospital of Philadelphia, Philadelphia PA

Abstract

Objectives—Evaluate whether telemedicine can be used to perform dysmorphology and neurologic examinations in the neonatal intensive care unit (NICU) by determining the examination accuracy, limitations, and optimized procedures.

Study design—Prospective evaluation of NICU patients referred for subspecialty consultation for dysmorphic features (n=10) or encephalopathy (n=10). A physician at bedside (bedside clinician) performed an in-person examination which was viewed in real-time by a remote physician (remote consultant). Standardized examinations were recorded and compared. Subsequently, a qualitative approach established technique adjustments and optimization procedures necessary to improve visualization.

Results—Telemedicine examinations identified 81 of 87 (93%) dysmorphology examination abnormalities and 37 of 39 (92%) neurologic examination abnormalities. Optimization of remote consultant visualization required an active bedside clinician assisting in camera and patient adjustments.

Conclusions—Telemedicine can be used to accurately perform many components of the dysmorphology or neurologic examinations in NICU patients, but physicians must be mindful of specific limitations.

Introduction

Telemedicine is defined by the World Health Organization as, “The delivery of health care services, where distance is a critical factor, by all health care professionals using information...
and communication technologies for the exchange of valid information for diagnosis, treatment and prevention of disease and injuries, research and evaluation, and for the continuing education of health care providers, all in the interests of advancing the health of individuals and their communities.”

Telemedicine may play an important role improving access to subspecialty expertise for patients in locations without access to specialists or for patients requiring time-sensitive care. For example, the use of telemedicine in adult stroke care has been shown to permit regional hospitals to retain patients and reduce costs while also improving thrombolysis decisions. These data have led to recommendations regarding telemedicine implementation to increase stroke care access. Many regional hospital neonatal intensive care units also provide care without immediate access to subspecialty consultation, forcing neonatologists to decide between forgoing subspecialty involvement and transferring critically ill patients to sometimes distant hospitals. Previous research has demonstrated that clinical agreement between neonatologists is good to excellent with telemedicine, and that clinicians had a positive impression of telemedicine use. However, studies of telemedicine to perform detailed dermatologic evaluations suggest difficulty with some features, cautioning that the use of telemedicine may need to be evaluated for each subspecialty indication.

Studies have not evaluated the use of telemedicine for neurologic or dysmorphology evaluations in the neonatal intensive care unit. We performed a prospective study comparing in-person and telemedicine dysmorphology and neurologic examinations of subjects in the neonatal intensive care unit. We aimed to establish the accuracy of telemedicine examinations in this setting and to identify measures to optimize telemedicine examinations.

**Subjects and Methods**

This was a prospective study performed in a quaternary care children’s hospital. The study was approved by the institutional review board and informed consent was obtained from each subject’s parent(s). All subjects received standard medical care by the appropriate clinical services.

Patients in the Neonatal Intensive Care Unit were eligible for participation if they had received: 1) consultation by the Genetics service for dysmorphic features or; 2) consultation by the Neurology service for encephalopathy. Data regarding demographics and their acute medical conditions were obtained. Each subject underwent telemedicine examination using an InTouch Health RP-Lite Remote Presence System (Chelmsford, MA) and a AMD-2500 General Examination Camera with a 50x Zoom Lens (Chelmsford, MA). Figure 1 presents a procedure schematic.

**Phase 1: Accuracy of Telemedicine Examination**

A remote consultant physician (geneticist or neurologist) in a remote location evaluated each subject with the assistance of a bedside clinician (geneticist or neurologist). The bedside clinician followed the instructions of the remote consultant physician (e.g. positioning of the infant, demonstrating reflexes) to optimize visualization. The examination components did not need to be performed in any particular order, and order was often determined by initial state and cooperation of the infant with the examination. The bedside clinician noted any
discrepancies between what they saw in person and what was displayed on the screen, but the bedside clinician did not communicate any of these observations to the remote consultant physician. Both the bedside clinician and remote consultant physician recorded their examinations using standardized forms.

The in-person bedside clinician examination was considered the accurate gold-standard to which the remote bedside physician’s examinations were compared. Examination accuracy was determined by calculating the percent agreement between abnormal physical examination findings noted by the bedside clinician examination (“gold standard”) and the remote consultant physician examinations. Examination components identified accurately >90% of the time were considered “easy to identify”. Examination components deemed as “not achievable” were categorized as those unable to be appreciated without an in-person examination or those that were unidentifiable with the clinical resolution of the tested equipment.

Phase 2: Qualitative Limitations and Optimization

After completing the telemedicine assessment, the remote consultant physician came to the bedside for the second phase of the study. The bedside clinician and the remote consultant physician discussed any discrepancies between the telemedicine and in-person examinations and attempted technique adjustments to achieve optimal visualization of those examination components. These limitations and optimization techniques were recorded as qualitative observations. Supplementary figures 1–2 shows the bedside clinician and remote consultant views.

Results

Subjects

Ten subjects with dysmorphic features were evaluated by geneticists and ten subjects with encephalopathy were evaluated by neurologists. Subject characteristics are provided in Table 1.

Phase 1: Accuracy of Examination Using Telemedicine

Abnormal examination findings and accuracy of identification via telemedicine are indicated in Table 2 for the dysmorphology examination and Table 3 for the neurologic examination. Most abnormal examination components were identified with an accuracy of greater than 90%. For the dysmorphology examination, 81 of 87 (93%) dysmorphic features were correctly identified using telemedicine on the initial examination. Optimization of lighting and positioning resulted in visualization of two additional abnormalities (83 of 87, 95%). For the neurologic examination, 36 of 39 (92%) abnormal features were identified using telemedicine on the initial examination. Optimization of lighting and positioning resulted in visualization of one additional abnormality (37 or 39, 95%). All calculations were made comparing the assessments of the remote consultant physician’s initial telemedicine examination and their own subsequent in-person examination. Inter-rater agreement with the bedside clinician was not ascertained.
Phase 2: Qualitative Limitations and Optimization

Following the standardized telemedicine examination, the bedside clinician and remote consultant physician worked together to optimize visualization of the examination of features that could not be assessed well remotely. In many cases the viewing angle, lighting, or examination technique could be adjusted to optimize the evaluation. Considerations for examination optimization are provided in Table 4. Several common trends emerged.

1. Bedside Clinician Role: The bedside clinician plays a critical and active role in the evaluation, and must report their observations of the subject and their impressions regarding the camera’s view. The bedside clinician must evaluate and adjust the equipment’s angle and distance from the subject, camera’s position and zoom, lighting conditions, and patient positioning. For effective evaluation, the camera and/or subject must be repositioned during the evaluation, and often these adjustments must occur in a fluid manner that is responsive to the examination needs and activity of the subject. For example, identifying dysmorphologic abnormalities which are often small and subtle requires optimal positioning, and movement, tone, and reflex examination must occur when the subject is in the relaxed state.

2. Image Display: Although the telemedicine equipment permits different displays for the bedside clinician and remote consultant physician, having the same image displayed allowed the bedside clinician to more readily make changes to enhance the view of the remote consultant. Suboptimal lighting, camera angles or camera focal points sometimes resulted in an inability of the remote consultant to appreciate abnormal skull shape, pigmented differences (especially eye color), respiratory effort, cleft palate evaluation, eye movement abnormalities, muscle tone abnormalities, and deep tendon reflexes. In all cases, the bedside clinician knew the camera images were suboptimal and did not allow them to fully appreciate the examination finding they could see in-person.

3. Focal Point: Prior to the examination, it was useful to identify the minimal focal distance for the equipment’s cameras by zooming in on an object in the environment. Subsequently, placing the equipment so the camera was at or minimally further away allowed maximal zoom while retaining the ability to focus on features. For the equipment tested, this was approximately one meter from the patient.

4. Lighting: Lighting conditions varied based on overhead lighting of different rooms and with different camera positions. Overhead lighting adjustments were often needed to provide lighting that was sufficiently bright enough for the examination without creating bright spots on the subject that exceeded the camera’s ability to achieve good contrast balance. In addition, low angle lighting was at times useful to distinguish skin surface characteristics. Having identical images displayed on the bedside and telemedicine physicians’ screens allowed the bedside physician to best optimize the lighting.

5. Viewing Angle: Features perpendicular to the plane of view were seen most accurately. Thus, for most components of the examination the optimal camera
angle was directly above the isolette and angled down at the prone or supine subject since this approach maximized the amount of the subject perpendicular to the camera. When examining particular body parts, positioning them perpendicular to the camera through repositioning of the equipment and the patient was essential. For dysmorphology examination, this was particularly important when evaluating hand/finger characteristics or limb proportions. For neurologic examination, this was particularly important when evaluating reflexes or spontaneous movement.

6. Multiple Views: While it may be difficult to maneuver large equipment around an isolette, the bedside clinician must be an active participant and can turn the subject as needed to optimize examination. For the dysmorphology examination, viewing the subject from angles rotated 180 degrees, so that the feet extend away from the camera permits better visualization of skull shape, hand shape, and dorsum of foot evaluation (syndactyly, overfolded toes). Turning the subject to the side permitted a lateral view which also aided in assessment of head shape. For the neurologic examination, repositioning of the subject’s face perpendicular to the camera aided in evaluation of facial movement, suck, rooting, and eye movement evaluation. Placing the subject so that the angle of the joints was perpendicular to the camera aided in assessment of spontaneous movements and reflexes.

7. Special considerations for the hands: Because neonates often have their hands clenched at rest, the hand must be held open and flat by the bedside clinician for proper evaluation. The dorsum of the hand is best evaluated with the subject’s head pointed towards the camera. Nails were easily visualized. Hand proportions can be distorted when the hand is not placed perpendicular to the camera viewing angle. When viewing the palmar aspect of the hand, creases could be easily visualized. However, clinodactyly might not be identified since holding the fingers open made the fingers appear to curve inwards. See Supplementary Figure 3.

Discussion

We describe a prospective study to determine the accuracy of dysmorphology and neurologic examinations in subjects in the NICU, and to identify optimal approaches and limitations of telemedicine examinations in this setting. For both dysmorphology and neurologic examinations, telemedicine could be used to perform generally accurate examinations, but certain limitations were identified. Importantly, we found that the bedside clinician played an essential role in optimizing the examination. The bedside clinician was able to indicate if the displayed image did not accurately reflect the in-person view, which could prompt adjustments to the camera, viewing angle or patient positioning.

These results suggest telemedicine has the ability to improve access to pediatric subspecialty expertise for patients in locations without in-person subspecialty access and may reduce the need to transfer of critically ill patients. Currently, patients may be transferred to a tertiary care center for evaluation by subspecialists who are not available at the referring hospital. While some transfers are necessary so that the subspecialty service can provide ongoing management, some infants are transferred for a single evaluation by a geneticist or neurologist. Significant health care costs, medical risks to the critically ill patient, and stress
to the patient and family could be prevented if a subspecialist could reliably assess the infant from a remote setting using telemedicine. By providing for greater subspecialty access, telemedicine could play a role in making feasible recent calls for enhanced neonatal neurocritical care. However, there are no published studies addressing telemedicine for subspecialty consultations in a NICU setting.

While telemedicine has been used to facilitate access for patients with more chronic conditions such as epilepsy, its most widespread use has been to manage patients with neurologic conditions requiring rapid intervention such as stroke. Studies have documented the reliability of a remotely administered stroke scale, shown that telemedicine is associated with a high retention rate at the local level and reduced patient costs, and demonstrated that thrombolysis decisions were more often correct using telemedicine than phone consultation. Therefore, guidelines for stroke management have recommended implementing telemedicine to increase access to acute stroke care. In a more general neurocritical care setting, telemedicine has been shown to improve physician responsiveness to unstable patients and reduce costs. A recent report from the American Academy of Neurology Telemedicine work group concluded that tele-neurology would be of greatest benefit to patients with restricted access to neurologic care, would improve neurologists’ ability to meet the demand for specialty care in underserved areas, and overall could promote rapid evaluation of patients in remote locations. While most prior work has focused on adults requiring stroke or neurocritical care so the guideline could not address pediatric care specifically, occur data indicate that telemedicine may also be of benefit to neonates and infants requiring neurologic care.

This study focused on the physical exam component of dysmorphology exam, as previous literature has documented patient and provider satisfaction with remote genetic counseling and outpatient visits. However, these studies did not include an in-person evaluation, and were therefore not able to assess the accuracy of physical examination findings. Most relevant to the current data was a study addressing the accuracy of telemedicine for dermatology examination, although this study used transmission of static images as opposed to our study utilizing real-time audio-video assessments. They found some skin lesions were readily diagnosed (e.g. eczema, acne, psoriasis, and skin infections) while others were more difficult to accurately assess (e.g. pigmented lesions, actinic keratosis and basal cell carcinoma). This study is the first to provide data about the limitations of the equipment for dysmorphology evaluations, and recommend a systematic approach to an examination using telemedicine. Similarly, while components of the neurologic examination have been validated using telemedicine in the adult (e.g. for stroke evaluation), this is the first study to assess the accuracy of thorough neurologic examination of neonates and infants.

Several studies have addressed telemedicine accuracy and impact when used by neonatologists. A recent study comparing bedside and telemedicine neonatologist evaluations of 46 neonates found excellent or intermediate-to-good agreements for almost all components of the evaluation. However, poor agreement was identified for several physical examination features including auditory components (breath, cardiac, bowel sounds) and capillary refill time. In a second study, 5 of 19 neonates who underwent tele-consultation avoided transport, and as a result use of telemedicine was associated with
substantial cost savings.\textsuperscript{20} Overall, clinicians report having a positive impression of telemedicine for neonatal consultation.\textsuperscript{8} There have been more limited studies of telemedicine use for subspecialty evaluation of neonates, and they have generally focused on image interpretation and not performance of a full subspecialty physical examination. An early study with a low cost video-teleconferencing system for surgical consultation reported that in six neonates, an accurate diagnosis was established in all, leading to earlier care implementation for some neonates.\textsuperscript{21} A study of 63 neonates with suspected congenital heart disease reported that a diagnosis could be made in 97\% using transmitted echocardiographic images of neonates with suspected congenital heart disease. Congenital heart disease was excluded in 19, diagnosed but did not require transfer in 28, and required transfer in 14.\textsuperscript{22} A telemedicine program focused on retinopathy of prematurity screening performed 582 examinations on 137 infants. Thirteen infants were transferred and nine required laser treatment, and there were no poor outcomes.\textsuperscript{23} In addition to being useful for image transmission, our data indicate telemedicine may allow performance of a more complex examination. One similar study has been reported in 15 older children being evaluated in an emergency department as if they required transport by in-person and telemedicine examinations. The sensitivity of the telemedicine physician to identify abnormal findings was 88\% and the specificity of the telemedicine physician to identify normal findings was 93\%.\textsuperscript{24}

There are several limitations to this work. First, since we aimed to define the limitations of the equipment with optimal use, subspecialists were involved in the patient evaluations at bedside and remote positions. Additional steps are needed to generalize these data since with clinically implementation the bedside clinician would not be a subspecialist. Although the bedside clinician was instructed to follow directions of the remote consultant and not provide feedback about their own observations during the first phase of the study, it is possible that having a bedside physician with less familiarity with these specialized examinations could negatively affect accurate demonstration of abnormal findings. Second, this study focused on the ability to identify examination abnormalities and did not assess whether management decisions would differ based on in-person or telemedicine examinations. Further study is needed to determine the impact of examination omissions or inaccuracies on clinical diagnosis or management. Additionally, it is unclear whether these management changes would exceed the medical risk and financial cost of transferring these sometimes critically ill patients to a center for a specialty consultation. Third, our study did not select patients based on their hypothesized suitability for “avoidance of transfer.” Fourth, we utilized a single in-person remote consultant examination as the gold standard. However, even with in-person examination there will be inter-examiner variability. This study was designed to mimic the real-life scenario of a remote physician evaluating a neonate or infant via telemedicine and then repeating their examination if the patient was transferred to their facility. A more robust strategy to determine the sensitivity of the telemedicine evaluation would employ multiple expert evaluators of the same patient. Finally, we used one model of telemedicine equipment used and the sensitivity may vary depending upon technical specifications of the cameras and data transfer. The telemedicine device used in this study was similar to that employed in prior evaluations of infants in a
NICU setting so it is possible that studies employing different technology could have different findings.

Conclusions

Using telemedicine consultation, geneticists were able to accurately identify 93% of dysmorphic examination features and neurologists were able to identify 92% of abnormal neurologic examination features in encephalopathic patients. These data extend previous evaluations of telemedicine in children by indicating that beyond simple image transmittal, telemedicine may allow subspecialists to interpret more extended physical examinations accurately. However, our data also indicate that physicians must be mindful of specific telemedicine limitations, that the bedside clinician must play an active and educated role in the process, and that collaborative communication between the bedside clinician and remote consultants is essential. These findings indicate that while equipment improvements are certainly important, optimization of telemedicine consultation also requires attention to implementation. Our data indicate that telemedicine examinations are generally accurate which provides an impetus to study additional implementation issues. We did not involve parents in the study, so our study cannot provide any data regarding acceptance of telemedicine consultation by parents/families. Further study is certainly needed focused on implementation issues, such as family and physician opinions regarding speaking and providing consultation over telemedicine. Establishment of telemedicine programs in which physicians understand the limitations of this approach and continually work to improve upon its use will likely be of most benefit to patients. With further development of these types of programs, telemedicine may offer specialized care to patients in the NICU on a larger scale than could be accomplished by conventional in-person consultation models.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgements

The authors would like to thank Stephanie Tuttle, Newton Buchanan and Michael Gould. Funding for this project was provided by NIH T32 GM008638, NIH KO8 HD055488 (NICHD), and NIH K23 NS076550 (NINDS). The equipment used in this study was supplied in part by a grant from the Verizon Foundation.

References

1. Organization WH. A Health Telematics Policy in Support of WHO’S Health-For-All Strategy for Global Development: Report of the WHO Group Consultation on Health Telematics (11–16 December, Geneva, 1997). 1998.
2. Wechsler LR, Tsao JW, Levine SR, Swain-Eng RJ, Adams RJ, Demaerschalk BM, et al. Telenurology applications: Report of the Telemedicine Work Group of the American Academy of Neurology. Neurology. 2013; 80(7):670–676. [PubMed: 23400317]
3. Demaerschalk BM, Miley ML, Kiernan TE, Bobrow BJ, Corday DA, Wellik KE, et al. Stroke telemedicine. Mayo Clin Proc. 2009; 84(1):53–64. [PubMed: 19121244]
4. Harno K, Paavola T, Carlson C, Viikinkoski P. Patient referral by telemedicine: effectiveness and cost analysis of an Intranet system. Journal of telemedicine and telecare. 2000; 6(6):320–329. [PubMed: 11265100]
5. Demaerschalk BM, Raman R, Ernstrom K, Meyer BC. Efficacy of telemedicine for stroke: pooled analysis of the Stroke Team Remote Evaluation Using a Digital Observation Camera (STROkE DOC) and STROkE DOC Arizona telestroke trials. Telemedicine journal and e-health : the official journal of the American Telemedicine Association. 2012; 18(3):230–237. [PubMed: 22400970]

6. Schwamm LH, Pancioli A, Acker JE 3rd, Goldstein LB, Zorowitz RD, Shephard TJ, et al. Recommendations for the establishment of stroke systems of care: recommendations from the American Stroke Association's Task Force on the Development of Stroke Systems. Stroke. 2005; 36(3):690–703. [PubMed: 15689577]

7. Garingo A, Friedlich P, Tesoriero L, Patil S, Jackson P, Seri I. The use of mobile robotic telemedicine technology in the neonatal intensive care unit. J Perinatol. 2012; 32(1):55–63. [PubMed: 21617643]

8. Armfield NR, Donovan T, Smith AC. Clinicians' perceptions of telemedicine for remote neonatal consultation. Studies in health technology and informatics. 2010; 161:1–9. [PubMed: 21191153]

9. Third CK, Brooker I, Ormerod AD. Teledermatology: a tool for remote supervision of a general practitioner with special interest in dermatology. Clinical and experimental dermatology. 2011; 36(5):489–494. [PubMed: 21507041]

10. Bonifacio SL, Glass HC, Peloquin S, Ferriero DM. A new neurological focus in neonatal intensive care. Nat Rev Neurol. 2011; 7(9):485–494. [PubMed: 21802977]

11. Ahmed SN, Mann C, Sinclair DB, Heino A, Iskiw B, Quigley D, et al. Feasibility of epilepsy follow-up care through telemedicine: a pilot study on the patient's perspective. Epilepsia. 2008; 49(4):573–585. [PubMed: 18076644]

12. Ahmed SN, Wiebe S, Mann C, Ohinmaa A. Telemedicine and epilepsy care - a Canada wide survey. Can J Neurol Sci. 2010; 37(6):814–818. [PubMed: 21059544]

13. Ganapathy K. Telemedicine and neurosciences. J Clin Neurosci. 2005; 12(8):851–862. [PubMed: 16326267]

14. Shaqat S, Kvedar JC, Guanci MM, Chang Y, Schwamm LH. Role for telemedicine in acute stroke. Feasibility and reliability of remote administration of the NIH stroke scale. Stroke. 1999; 30(10):2141–2145. [PubMed: 10512919]

15. Vespa PM, Miller C, Hu X, Nenov V, Buxey F, Martin NA. Intensive care unit robotic telepresence facilitates rapid physician response to unstable patients and decreased cost in neurointensive care. Surg Neurol. 2007; 67(4):331–337. [PubMed: 17350395]

16. Lea DH, Johnson JL, Ellingwood S, Allan W, Patel A, Smith R. Telegenetics in Maine: Successful clinical and educational service delivery model developed from a 3-year pilot project. Genetics in medicine : official journal of the American College of Medical Genetics. 2005; 7(1):21–27. [PubMed: 15654224]

17. Abrams DJ, Geier MR. A comparison of patient satisfaction with telehealth and on-site consultations: a pilot study for prenatal genetic counseling. Journal of genetic counseling. 2006; 15(3):199–205. [PubMed: 16779676]

18. Coelho JJ, Arnold A, Nayler J, Tischkowitz M, MacKay J. An assessment of the efficacy of cancer genetic counselling using real-time videoconferencing technology (telemedicine) compared to face-to-face consultations. Eur J Cancer. 2005; 41(15):2257–2261. [PubMed: 16176873]

19. Armfield NR, Bensink M, Donovan T, Wootton R. Preliminary evaluation of a system for neonatal teleconsultation. Journal of telemedicine and telecare. 2007; 13(S3):4–9. [PubMed: 17288651]

20. Armfield NR, Donovan T, Bensink ME, Smith AC. The costs and potential savings of telemedicine for acute care neonatal consultation: preliminary findings. Journal of telemedicine and telecare. 2012; 18(8):429–433. [PubMed: 23148301]

21. Robie DK, Nautly CM, Parry RL, Motta C, Darling B, Micheals M, et al. Early experience using telemedicine for neonatal surgical consultations. Journal of pediatric surgery. 1998; 33(7):1172–1176. discussion 1177. [PubMed: 9694117]

22. Mulholland HC, Casey F, Brown D, Corrigan N, Quinn M, McCord B, et al. Application of a low cost telemedicine link to the diagnosis of neonatal congenital heart defects by remote consultation. Heart. 1999; 82(2):217–221. [PubMed: 10409539]

23. Weaver DT, Murdock TJ. Telemedicine detection of type 1 ROP in a distant neonatal intensive care unit. Journal of AAP: the official publication of the American Association for Pediatric
24. Kofos D, Pitetti R, Orr R, Thompson A. Telemedicine in pediatric transport: a feasibility study. Pediatrics. 1998; 102(5):E58. [PubMed: 9794988]
Figure 1.
Schematic of study design.
Table 1

| Subspecialty | Age (weeks) | Gender | Indications for Consultation |
|--------------|-------------|--------|-----------------------------|
| Genetics     | 12          | Male   | Congenital diaphragmatic hernia, bilateral lip/palate, lagophthalmos, failed hearing screen |
|              | 5           | Male   | Complex congenital heart disease, unusual facial features, hypoglycemia |
|              | 11          | Female | Mandibular hypoplasia, cleft palate, myopia, spondyloepiphyseal dysplasia |
|              | 16          | Female | Skeletal dysplasia, thoracic insufficiency syndrome, unusual facial features, failed hearing screen |
|              | 7           | Male   | Tetralogy of Fallot, cerebellar hypoplasia, unusual facial features, clubfoot, failed hearing screen |
|              | 6           | Male   | Congenital diaphragmatic hernia, cleft palate, patent ductus arteriosus, hypotonia, rib and vertebral abnormalities, failed hearing screen |
|              | 5           | Male   | Patent foramen ovale, small for gestational age, hypoglycemia, hypotonia, hypothyroidism |
|              | 3           | Male   | Pierre Robin sequence, cleft palate, syndactyly, unusual facies |
|              | 16          | Female | Complex congenital heart disease, giant omphalocele, cleft lip and palate, choanal atresia, atrial septal defect, duplicated digits, craniosynostosis, natal teeth, imperforate anus (status post repair) |
|              | 20          | Female | Intracranial growth restriction, pulmonary hypoplasia, pulmonary vein stenosis, atrial septal defect, inguinal hernia, widened cranial sutures. |
| Neurology    | 2           | Male   | Term, neonatal encephalopathy with presumed hypoxic ischemic encephalopathy managed with therapeutic hypothermia, brain MRI normal. |
|              | 2           | Female | Term, respiratory distress, encephalopathy, septum primum with ASD closure, brain MRI with mild periventricular leukomalacia. |
|              | 8           | Female | Term, micrognathia with mandibular distraction followed by CN VII palsy and encephalopathy. |
|              | 3           | Male   | Term, neonatal encephalopathy with presumed hypoxic ischemic encephalopathy, brain MRI with hypoxic ischemic brain injury. |
|              | 1           | Female | Term, neonatal encephalopathy with presumed hypoxic ischemic encephalopathy managed with therapeutic hypothermia, brain MRI with mild hypoxic ischemic brain injury. |
|              | 2           | Female | Term, focal seizures, encephalopathy, brain MRI with right middle cerebral artery infarct. |
|              | 3           | Male   | Term, neonatal encephalopathy with presumed hypoxic ischemic encephalopathy, seizures, brain MRI with hypoxic ischemic brain injury. |
|              | 8           | Male   | 34 weeks gestational age, trisomy 21, encephalopathy, hypotonia, abnormal movements of unclear etiology. |
|              | 7           | Male   | 33 weeks gestational age, encephalopathy, brain MRI with large porencephalic cyst. |
|              | 6           | Female | 32 weeks gestational age, encephalopathy, seizures, brain MRI with parietal infarct. |
Table 2
Accuracy of abnormal dysmorphology examination findings.

| Feature                      | Agreement | Comment                                                                 |
|------------------------------|-----------|-------------------------------------------------------------------------|
| Almond-shaped eyes           | 1/1 (100%)|                                                                         |
| Abnormal skull shape         | 4/4 (100%)| Degree of severity better appreciated in person                        |
| Cleft palate                 | 1/3 (33%) | Could not be visualized in two patients even with optimal positioning and lighting |
| Clinodactyly                 | 4/5 (80%) | Incorrectly identified in infant with clenched fists when fingers were held open |
| Deep creases                 | 2/2 (100%)|                                                                         |
| Clubfoot                     | 3/3 (100%)|                                                                         |
| Downturned mouth             | 1/1 (100%)|                                                                         |
| Duplicated digits            | 1/1 (100%)|                                                                         |
| Everted lower eyelid         | 1/1 (100%)|                                                                         |
| Frontal bossing              | 3/3 (100%)|                                                                         |
| Helical abnormalities        | 5/5 (100%)|                                                                         |
| Hemangioma                   | 3/3 (100%)|                                                                         |
| Hydrocele                    | 1/1 (100%)|                                                                         |
| Hyperpigmented lesion        | 2/2 (100%)|                                                                         |
| Hypertelorism                | 2/2 (100%)|                                                                         |
| Labial anomaly               | 1/1 (100%)|                                                                         |
| Lateral facial cleft         | 2/2 (100%)|                                                                         |
| Low-set, posteriorly rotated ears | 5/5 (100%)|                                                                     |
| Micrognathia                 | 3/3 (100%)| Degree of severity better appreciated in person                        |
| Nail abnormality             | 3/3 (100%)|                                                                         |
| Nasal anomaly                | 5/5 (100%)|                                                                         |
| Natal teeth                  | 1/1 (100%)|                                                                         |
| Nipple spacing abnormal      | 1/1 (100%)|                                                                         |
| Omphalocoele                 | 1/1 (100%)|                                                                         |
| Palpebral fissure slant      | 3/3 (100%)|                                                                         |
| Periorbital edema            | 2/2 (100%)|                                                                         |
| Pigmentary abnormalities     | 2/3 (67%) |                                                                         |
| Proptosis                    | 3/3 (100%)|                                                                         |
| Scar                         | 3/5 (60%) | Small healed scars difficult to appreciate via telemedicine             |
| Single or bridged palmar crease | 3/3 (100%)|                                                                     |
| Skin flaking                 | 1/1 (100%)|                                                                         |
| Small thorax                 | 1/1 (100%)|                                                                         |
| Sternal abnormalities        | 1/1 (100%)|                                                                         |
| Syndactyly                   | 2/2 (100%)|                                                                         |
| Synophrys                    | 1/1 (100%)|                                                                         |
| Thin hair                    | 3/3 (100%)|                                                                         |
| Y-shaped gluteal crease      | 1/1 (100%)|                                                                         |
| Feature | Agreement | Comment |
|---------|-----------|---------|
| Total   | 81/87 (93%) |         |
Table 3

Accuracy of abnormal neurologic examination findings.

| Feature                          | Agreement | Comment                                         |
|----------------------------------|-----------|-------------------------------------------------|
| Neurocutaneous                   | 1/1 (100%)| Café au lait lesion not appreciated via telemedicine but appreciated in-person. |
| Abnormal palmar reflex           | 2/3 (67%) |                                                 |
| Abnormal suck/root reflex        | 5/5 (100%)|                                                 |
| Facial weakness                  | 1/1 (100%)|                                                 |
| Hypotonia – upper extremities    | 3/3 (100%)| Degree of severity better appreciated in person |
| Hypotonia – lower extremities    | 4/4 (100%)| Degree of severity better appreciated in person |
| Hypotonia – truncal              | 6/6 (100%)| Degree of severity better appreciated in person |
| Hypotonia – face/neck            | 5/6 (83%) | Degree of severity better appreciated in person |
| Increased tone – lower extremities| 3/3 (100%)| Degree of severity better appreciated in person |
| Increased DTR – lower extremities| 4/4 (100%)|                                                 |
| Increased DTR – upper extremities| 1/2 (50%) | Better appreciated in person                    |
| No response to auditory stimulation| 1/1 (100%)|                                                 |
| **Total**                        | 36/39 (92%)|                                                 |
### Table 4

Considerations to improve examination.

| Ease of Telemedicine Assessment | Examination Component | Considerations |
|--------------------------------|-----------------------|----------------|
| **Easy to Assess**             | Eyes                  | Eye spacing can be obtained if infant opens eyes. Bedside clinician can hold measuring tape up to face to measure palpebral fissures. |
|                                | External ear          | Flexion/extension of head may cause normal ears to appear low-set. Zoom in to evaluate for ear pits. Bedside clinician should fold back helix to assess for creases. |
|                                | Nose                  | Accurate view of nasal bridge requires side view of face. |
|                                | Lips                  | Zoom in to evaluate for lip pits. |
|                                | Neck                  | Bedside clinician must demonstrate extranuchal skin exam. |
|                                | Chest                 | If inter-nipple distance or chest circumference are desired, the telemedicine physician can observe the technique of the bedside physician. |
|                                | Arms and Legs         | Must be extended to accurately assess proportions. Repositioning to have a perpendicular view of joints helps view deep tendon reflex movements, and evaluate for spontaneous movements. |
|                                | Hands                 | Creases, nails, syndactyly, cortical thumbing, palmar reflexes. |
|                                | Umbilical stump       | Zoom in, particularly if umbilical lines are in place. |
|                                | Genitalia             | Not difficult except for testicular exam, which is not achievable due to need for tactile exam. |
| **Possible with Optimization** | Hair                  | Hair whorls easier to appreciate in infants with darker hair. |
|                                | Skin                  | Pigmented lesions, rashes, skin flaking and scars were seen easily. Overall skin tone, faint capillary hemangiomas require optimal lighting. |
|                                | Spine                 | Repositioning required so spine is perpendicular to camera. |
|                                | Eyes                  | Iris color, colobomas, proptosis (better appreciated with eyes open and on lateral view), pupil size and reactivity, eye movements. |
|                                | Skull shape           | Multiple views of head must be obtained, but still difficult since head is not a planar structure. |
|                                | Chin                  | Micrognathia is better appreciated on lateral view of face. |
|                                | Hands                 | Clinodactyly, palm length:finger length. |
|                                | Feet                  | Soles easily visualized with feet pointed towards camera. Dorsum best visualized with neonate rotated so feet away from camera. |
|                                | Muscle Tone           | Bedside clinician must help ensure neonate is relaxed when assessing. Appendicular tone can be assessed when clearly increased or decreased since leads to position changes (ie. frog leg positioning) but not achievable with mild changes since requires tactile examination. Axial tone can be assessed by positioning neonate perpendicular to camera for vertical suspension and horizontal suspension. |