Genetics and Telemedicine: Extending Our Reach

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Clinical Genetics Attending
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Invited Presentation

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- No financial disclosures

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Objectives

- Provide general background on telemedicine and potential benefits
- Illustrate how we are incorporating telemedicine into our clinics using three case examples
- Discuss barriers, limitations, and future directions

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Telemedicine:

Improving the genetics and metabolic medical home

- CN-RDI is one of the largest clinical genetics groups in the US with over 8,500 visits per year
- Telemedicine experiences providing virtual clinics and consults in Mississippi, DC, Maryland, Virginia, Idaho, and Thailand
- **"Genetic Deserts"** = states or regions with no geneticists or a single geneticist (usually with extended wait times and inpatient demand)

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Telemedicine:

Improving the genetics and metabolic medical home

ACCESS

Decreases wait times

Allows simple issues to be addressed quickly

Saves clinic slots for patients that need to be seen in person

BENEFITS OF A VIRTUAL HOME VISIT

Patients with autism can be seen in home setting

Dieticians can view inside the home refrigerator

Enables increased frequency of visits for high risk patients

REMOVING BARRIERS TO CARE

Transportation

Child care

Fear of exposure to illness for at risk patients in hospital setting

Missed work

Combine with in-person visits when deemed appropriate

MULTIPLE SPECIALTIES

Easy for multiple providers/ locations to share screen and coordinate telemedicine visits

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Telemedicine:

Improving the genetics and metabolic medical home

STREAMLINED CARE

- Call center offers new and follow up visits
- Emergency room offering follow-up visits to decrease bounce backs

COMMUNITY AND NATIONAL OUTREACH

- Support for genetics deserts
- Distance support for metabolic patients and centers

EPICENTER AND SUPPORT

- International consultation capability
- Incorporating new tools such as facial recognition to reduce diagnostic odyssey

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- Illustrate pilot virtual clinic program and three case examples of use
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Virtual Clinic Goals

- Shorten referral times
- Maximize efficiency
- Partner with primary care doctors to diagnose rare disease more quickly
- Incorporate innovate virtual genetics materials into these visits (Bear Genes)
- Shorten the diagnostic odyssey for rare disease

https://childrensnational.org/departments/rare-disease-institute/beargenes

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Virtual Pilot Clinic: How Does it Work

- New patients and follow-up patients seen
- Scheduled through call center
- Virtual Visit Coordinator e-mails family an app and instructions for download
- HIPPA compliant video tool used
- Carefirst grant for all uninsured and Medicaid patients in MD, DC, and VA
- Several other insurances cover direct to patient

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Case 1: More than Autism?

- 16 year old who is the size of a primary school child and had history of autism/ behaviors resistant to therapies
- Major aggression with abnormal movements
- Eating: only chicken nuggets, ketchup, chips, fruit snacks, juice
- OCD behaviors: obsessive behaviors with scratching self, staring, seems to be OCD about his arm. Will have repetitive arm movements
- Medications: Ativan, Zoloft at night

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Family History

History of FTT in NICU

Autism and Non-verbal

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Diagnosis and Outcome: Case 1

Atypical 22q11 deletion syndrome

- Deletions of chromosome region 22q11.2 are associated with a broad spectrum of clinical phenotypes including conotruncal heart defect, often associated with facial dysmorphisms, cleft palate, thymus hypoplasia, and learning disability. One of critical genes is *TBX1*.
- Most deletions (84–90%) encompass ~3 Mb, known as the typically deleted region. Smaller deletions, spanning 1.5 Mb, are found in about 7–14% of the cases .
- Likely autosomal dominant with various expression in this family
- Able to get 17-year-old into telemedicine neurology; reach out to psychiatry; get children into neurodevelopment and help with counseling
- Other two children also seen for care, counseled, monitored using guidelines

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Case 1: Lessons Learned

- Telemedicine may be an option for some children with severe autism/ children who could not otherwise come to care
- Several other patients seen with similar issues
- This has resulted in initiative of planned **genetics autism clinic** with combination of **telemedicine** and **in-person visits**, tailored to child

Case 2: Telemedicine Follow-Up

2 year old with a history of cardiofaciocutaneous syndrome, constipation, developmental delay, feeding problems (gastrostomy in place), reflux.

Cardiofaciocutaneous (CFC) syndrome: **cardiac** abnormalities (pulmonic stenosis and other valve dysplasias, septal defects, hypertrophic cardiomyopathy, rhythm disturbances), distinctive craniofacial appearance, and **cutaneous** abnormalities (including xerosis, hyperkeratosis, ichthyosis, keratosis pilaris, ulerythema ophryogenes, eczema, pigmented moles, hemangiomas, and palmoplantar hyperkeratosis).

Diagnosis is based on clinical findings and four genes associated: ***BRAF* (~75%), *MAP2K1* and *MAP2K2* (~25%), and *KRAS* (<2%).**

In this child, *MAP2K1* gene variant (c.389A>G).

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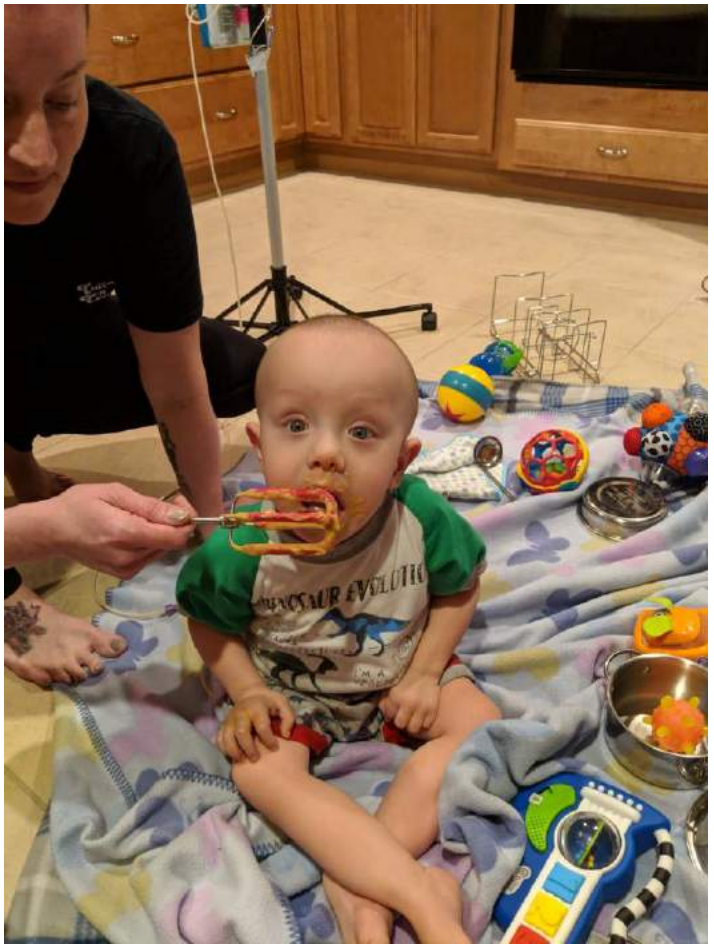
Multidisciplinary Care

- Specialty visits include ophthalmology, neurology, development, cardiology, and genetics
- Mom lives 2.5 hours away from CNMC
- Works as air-traffic controller



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What is your day like?

Mom and family provide following feeding schedule:

Pediasure Peptide 1.0 and 1.5 (1.25 calories) 51 ml per hour, 11 hours per day. He gets breaks (6 am to 10 am) and break (1:30-6:30) **(10p,-2 am).** When on breaks, G boluses.

The mom does homemade blends and uses fitness app to track.

1000 calorie per day: 300 calories per blends J: 700 calories.

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Impression/ Plan: How Can We Make Life Easier?

1. Modifying his feeding schedule so that the mom or family member no longer has to get up at 2 am in the morning
2. Reminded that his poor growth is related to CFC rather than lack of nutrition
3. Recommended adding prune juice for constipation (had worked in past)
4. Recommended following up with audiology
5. Reviewed gene reviews guidelines: https://www.ncbi.nlm.nih.gov/books/NBK1186/
6. Wrote medical necessity letter for stander that PT had recommended

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Case 2: Lessons Learned

- By taking focus away from travel/ the waiting room, more time on coordinating care
- Seeing a child in home setting is like a virtual home visit: it is very different and different insights/information gained

  (social determinants of health)
- Impact of visit can sometimes have reverse correlation with time spent

(KISS model: Keep it simple and salient)

Case 3: Palynziq Clinic

- Palynziq: used to lower blood levels of Phe (phenylalanine) in adults with PKU (phenylketonuria)
- Uncontrolled blood Phe levels above 600 micromol/L (10 mg/dL) on their current treatment

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Risks to Palynziq

- **Anaphylaxis requiring pre-medication and epinephrine pen**
- Skin reactions
- Joint pain
- Anxiety (?)

Telemedicine and Palynziq Clinic

1. Telemed visit initiated to review medication, benefits versus risks
2. Patients interested then reviewed safety video about epi pen on-line, signed clinic contract
3. Insurance approval submitted
4. In-person visit when drug improved and training reviewed

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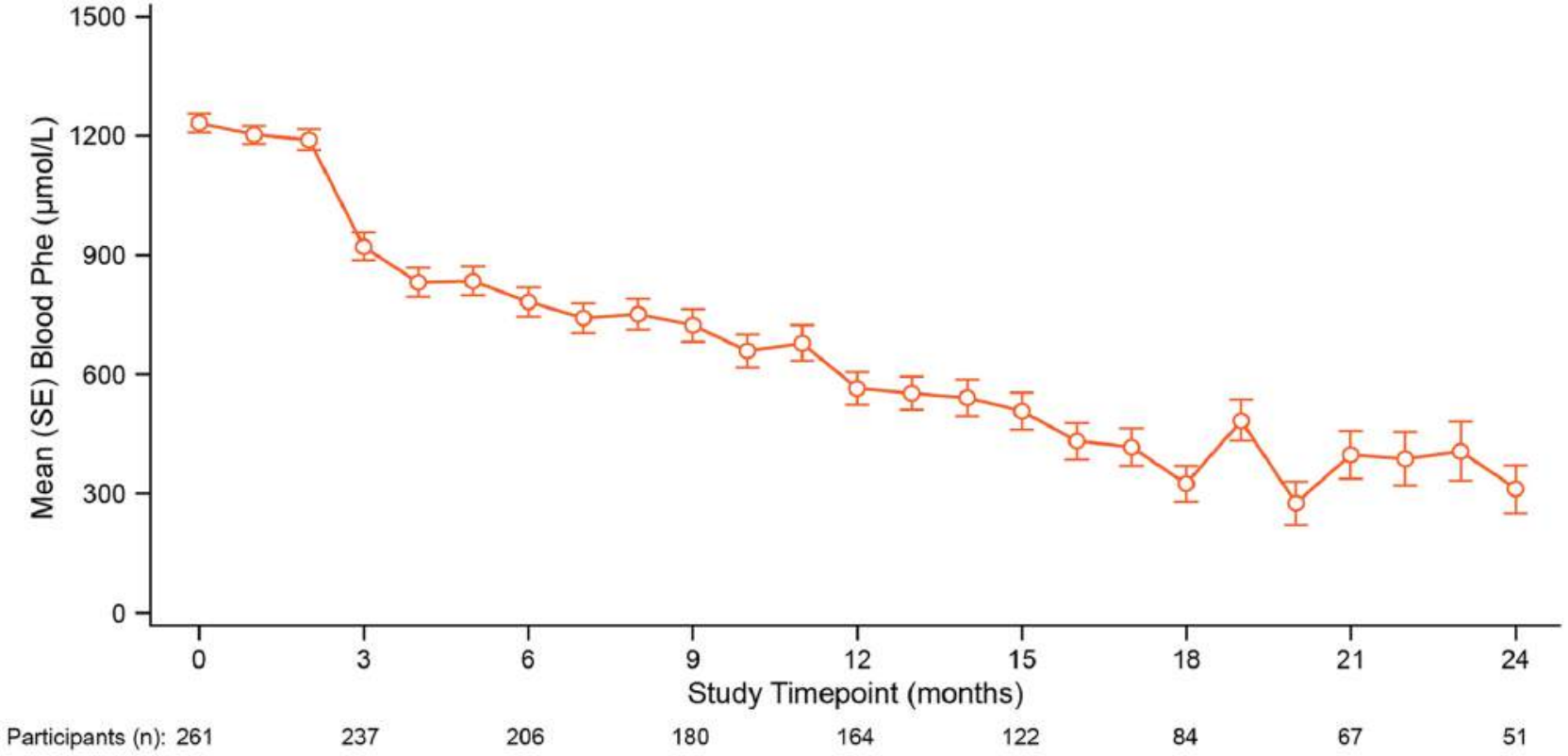
Mean Blood Phe Concentration



Evidence- and consensus-based recommendations for the use of **pegvaliase** in adults with phenylketonuria. Longo N, Dimmock D, Levy H, Viau K, Bausell H, Bilder DA, Burton B, Gross C, Northrup H, Rohr F, Sacharow S, Sanchez-Valle A, Stuy M, Thomas J, Vockley J, Zori R, Harding CO.Genet Med. 2018 14. doi: 10.1038/s41436-018-0403-z., PMID: 30546086

Outcomes Palynziq Clinic

- Approximately 8 patients started on Palynziq
- 1 patient dropped out due to reactions; several patients responded and now increasing protein in diet
- Program ongoing and one of several in country on forefront of treating PKU

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Case 3: Lessons Learned

- Model of care included education/ assessment/ and plan
- Telemedicine visit met criteria for insurance authorization
- Patients uninterested in treatment did not lose work time/ travel time
- Patient and provider choose between telemedicine and in-person visits

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Limitations to Telemedicine

- Insurance reimbursement barriers
  (site of service restrictions-clinic based telemed vs direct to patient)
- Reluctance to try a new care delivery new model
  (external communications)
- Technological challenges (migrating to new technology platforms)

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Summary

- Patients have been seen who would have been otherwise unable to come to hospital
- Reduced travel time, missed work, allowed in home assessments
- We are evaluating financial model and no-show rate effects (lower no-show rate with telemedicine appts)
- Over all, positive experience for both patient and provider
- Due to positive feedback, we have decided to expand our pilot program and offer telemedicine to more genetics patients in VA, MD, and DC
- Many types of patients but autism key area of interest
- We will continue to combine telemed and in-person visits

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Genetics Autism Clinic: Designing A Better Journey

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Interested Patient?

- Call to schedule an appointment through call center **1-888-884-BEAR**
- Virtual team sends an app to download

VirtualVisit@childrensnational.org

Dr. Shur contact info:

nshur2@childrensnational.org

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Thank You

- Telemedicine Division: **Clarence Williams**, Director of Telemedicine; **Tara McCarthy**, Telemedicine Coordinator; **Alejandro Lopez Magallon**, MD
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- Division Head: **Marshall Summar**, MD
- RDI technology committee: **Jamie Fraser**, MD, Chair technology committee
- NYMAC: **Alyssa Terry, Beth Vogal, Alissa Bovee**
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- **The wonderful patients whom we serve and families featured**

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