R.C.P.U. NEWSLETTER

Editor: Heather J. Stalker, M.Sc.
Director: Roberto T. Zori, M.D.

R.C. Philips Research and Education Unit

Vol. XXIX No. 2 **A statewide commitment to the problems of mental retardation** **June 2018**

R.C. Philips Unit ♦ Division of Pediatric Genetics, Box 100296 ♦ Gainesville, FL 32610 ♦ (352)294-5050
E Mail: stalkhj@peds.ufl.edu; zorirt@peds.ufl.edu
Website: http://www.peds.ufl.edu/divisions/genetics/newsletters.htm

Metabolic Clinic Updates

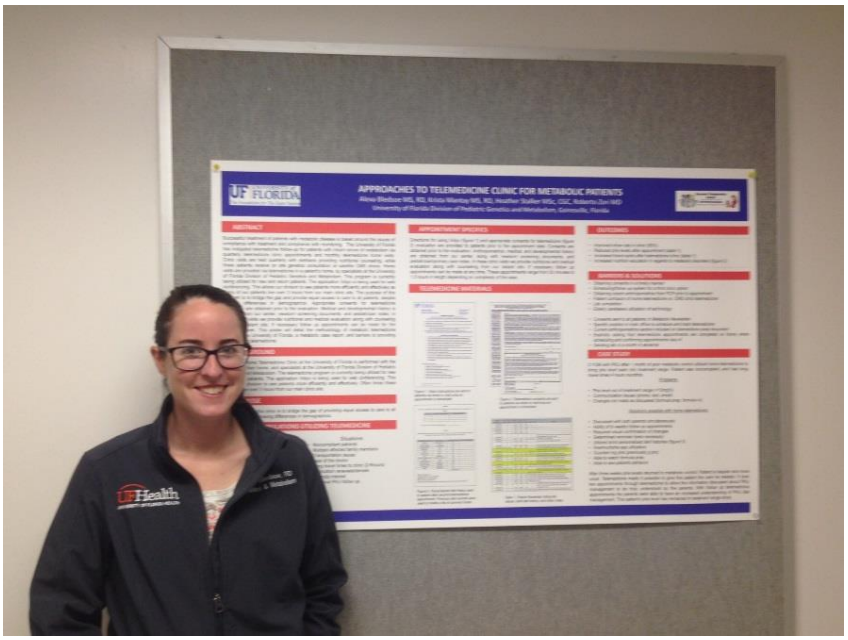
Krista Mantay & Alexa Bledsoe, MS, RD
Division of Pediatric Genetics & Metabolism
University of Florida

Home Telemedicine Clinics are live!

We are now offering home telemedicine clinics monthly! The purpose of this clinic is to bridge the gap of providing equal access to care to all patients despite growing differences in demographics. Our hope is that these clinics will greatly reduce the stress of the many factors that go into attending a clinic appointment. We currently have telemedicine appointments open and available through December 2018. At this time, telemedicine clinics are at various times on Tuesdays and are open to almost all diagnoses. The application Vidyo is currently being used for web conferencing. This allows our division to see patients more efficiently and effectively as many of our patients live over 3 hours from our main clinic site. The purpose of this program is to bridge the gap and provide equal access to care to all patients, despite growing differences in demographics.

We decided to start utilizing telemedicine with patients who currently have a poor show rate to scheduled appointments as well as generally non-compliant patients as a way to intensively help them be successful with their treatment. Many of the situations patients present as a barrier to arriving to our Gainesville clinics include but are not limited to multiple members of the family with the same disorder, transportation issues, fear of doctor's offices, and long travel times to Gainesville (2-6 hours), necessity of frequent appointments for high-risk patients.

Our initial Telemedicine outcomes have been published on a poster and will be presented at this years' Southeastern Regional Genetics Group conference (SERGG).



This poster not only explains our process for home telemedicine but describes our recent experience with home telemedicine, highlighting a particular case.

2 YOM with PKU after 1 month of poor metabolic control utilized home telemedicine to bring phe level back into treatment range. Patient was noncompliant, and had long travel times 4 hours roundtrip.

Problems:

- Phe level out of treatment range (>10mg%)
- Communication issues (phone, text, email)
- Changes not made as discussed (formula prep, formula rx)

Solutions possible with home telemedicine:

- Discussed with both parents simultaneously
- Ability of bi weekly follow up appointments
- Required visual confirmation of changes
- Determined reminder texts necessary
- Utilized strict personalized diet histories (figure 3)
- Howmuchphe app utilization
- Counted mg phe (previously g pro)
- Able to watch formula prep
- Able to see patients behavior
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After three weeks phe levels returned to metabolic control. Patient is happier and more vocal. Telemedicine made it possible to give this patient the care he needed. It took two appointments through telemedicine to allow the information discussed about PKU management to be truly understood by the parents. With follow up telemedicine appointments, the parents were able to have an increased understanding of PKU diet management. This patient's phe level has remained in treatment range since

New Medication for PKU Patients: Palynziq

Biomarin recently received standard approval for Palynziq (pegvaliase-pqpz) Injection for treatment of Adults with Phenylketonuria (PKU), who have uncontrolled blood phe concentrations on existing management. Palynziq is a PEGylated recombinant phenylalanine ammonia lyase enzyme substitution therapy to target the underlying cause of PKU by helping the body breakdown Phenylalanine. The following includes excerpts from the 2018 Palynziq press release.

This approval comes after almost 10 years of research and clinical trials with one of the larger trial sites being here at the University of Florida. Palynziq significantly and substantially reduced blood Phe levels as demonstrated in the pivotal Phase 3 PRISM-2 study, which met the primary endpoint of change in blood Phe compared with placebo ($p<0.0001$). During the PRISM-2 double-blind, placebo-controlled, randomized withdrawal period trial (RWP), participants were randomized in a 2:1 ratio to either continue their maintenance Palynziq dosage (20 mg once daily or 40 mg once daily) or to receive matching placebo for a total of 8 weeks. Palynziq-treated patients maintained their blood Phe concentrations as compared to their randomized withdrawal baseline, whereas patients randomized to matching placebo returned to their pretreatment baseline blood Phe concentrations. In the Phase 3 program, 57% of patients were taking medical food at baseline and 16% were on a protein-restricted diet at baseline (defined as receiving greater than 75% of total protein intake from medical food).

Palynziq substitutes the deficient phenylalanine hydroxylase (PAH) enzyme in PKU with the PEGylated version of the enzyme phenylalanine ammonia lyase to break down Phe. Palynziq is administered using a dosing regimen designed to facilitate tolerability; Palynziq's safety profile consists primarily of immune-mediated responses, including anaphylaxis, for which robust risk management measures effective in clinical trials are in place.

The dosing and administration of Palynziq follows an induction, titration, and maintenance paradigm. Treatment is individualized to the lowest effective and tolerated dosage. Prescribers may consider increasing to a maximum of 40 mg once daily in patients who have not achieved a response with 20 mg once daily for at least 24 weeks. Prescribers are instructed to discontinue treatment in patients who have not responded after 16 weeks of continuous treatment with the maximum dosage of 40 mg once daily. Periodic blood Phe monitoring is recommended, and patients should be counseled on how to adjust their dietary intake, as needed, based on blood Phe concentrations.

Palynziq has the potential to be a game-changing therapy for adults in the PKU community who have continued to struggle throughout their lives to control their phe levels despite rigorous management.

Upcoming Events

Group clinics are coming back!

Be on the look out for invitations to group clinics to do some cooking, learning and meeting others with low protein food needs.

Patient Events

In the coming year, we will be trying to organize more patient events in the different cities we serve:

· Gainesville

· Jacksonville

· Tallahassee

· Panama City

· Pensacola

· Daytona.

Patient-run Networking

We would like to support the coordination of patient networking within our treatment areas. If you are interested in networking with others with PKU, please email us your info and fill out the attached form. This way we can safely distribute contact information.

Recipe Corner:

Hawaiian Shish Kabobs

Ingredients:

- 1/3 c. French's Worcestershire sauce
- 2 TBSP oil
- 2 TBSP pineapple juice
- 1 TBSP brown Sugar
- ½ tsp prepared mustard
- ½ tsp onion powder
- ½ tsp ginger
- Vegetables of your choice!

Directions:

1. Stir together all the marinade ingredients
2. Place vegetables on skewers
3. Grill for 15 minutes
4. Spread marinade on vegetables every 5 minutes while cooking
5. Enjoy!

Newborn Screening Program Update:

Beginning May 1, 2018 all newborns screened will be tested for X-linked Adrenoleukodystrophy (X-ALD). This addition to the panel of disorders screened in Florida was recommended in 2016 by the Florida Genetics and Newborn Screening Advisory Council and funding was authorized during the 2017 legislative season. Infants with presumptive positive results will be referred to the nearest Genetic Specialty Center in Gainesville, Tampa and Miami. If the diagnosis is confirmed, the baby will then be referred to Endocrinology and Neurology for additional management.

Follow-up staff from the Newborn Screening Program will direct these procedures and advise you of the actions needed. These procedures will only apply to X-ALD; no changes will be made to the protocols currently practiced for other conditions on the screening panel.

Florida HEALTH

About the RCPU

The Raymond C. Philips Research and Education Unit began in 1978 when the legislature established section 393.20, F.S., of what is now known as the "prevention" legislation. It is named after Raymond C. Philips, who was the Superintendent of Gainesville's Tacachale (formerly Sunland) Center for 38 years, and was an acknowledged state and national leader in services for mentally retarded persons. The Unit is located on the Tacachale campus and is funded through a contract with the Department of Children and Families and the Department of Health.

The purpose of the R.C.P.U. is to treat, prevent, and/or ameliorate mental retardation through medical evaluations, education and research. The unit provides direct evaluations and counseling to families and promotes service, education, and prevention projects.
Some of the conditions currently under study at the RCPU involve Angelman, Velo-Cardio-Facial, Prader-Willi, Fragile X, Williams and Smith-Lemli-Opitz syndromes.

The R.C. Philips Unit is a resource for all Floridians interested in the diagnosis, treatment and prevention of mental retardation. Staff members are available for consultation and for educational programs for health.

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