

# Mountain States Regional Genetics Collaborative Grant Update 7-13-07

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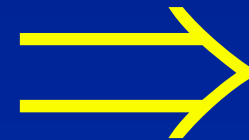
# Goals of Grant

1. Develop disease specific care plans for all metabolic diseases detected by newborn screening.
  - input from all regional care providers
  - “shared baselines”
  - long-term plans
  - not to be confused with the ACT sheets
2. Develop performance indicators and outcome measures for each disorder.
3. Review neuropsychological testing tools for long-term developmental outcome measures.
4. Implement care plans throughout the Mountain States region.
5. Utilize comprehensive database to track outcomes.

# Disease Specific Care Plans

- Special Considerations
- Diet Considerations
- Frequency of Visits
- Laboratory Studies
  - initial labs
  - monitoring labs
  - labs when ill
  - yearly labs

- Emergency Management
- Other Evaluations
- Neuropsychological Evaluations



# Performance Indicators and Outcome Measures

- **Performance indicators:**

Data to measure, track, and compare.

- **Outcome measures:**

End result of the intervention.

# Example: PKU

- Special Considerations
  - cofactor studies
  - tyrosine supplementation
  - hyperphe +/- diet
  - maternal PKU
- Diet Considerations
  - Phe-restricted diet
  - Breast-feeding mothers

# PKU

- Frequency of Visits (minimum)

Age	Clinic Visits
0-6 months	every 2 months
6-24 months	every 3 months
>2 years	every 6 months

- Emergency/Illness Management
  - None

# PKU

- Laboratory Studies
  - Initial (with consideration of DDx)
    - quantitative Phe/Tyr level
    - urinary pteridine profile
    - blood dihydropteridine reductase enzyme assay
  - Monitoring
    - quantitative Phe level
    - weight check with every Phe level up to 24 mos

# PKU

- Monitoring Labs (minimum)
  - quantitative Phe levels

Age	Frequency**
0-6 months	weekly
6-12 months	every 2 weeks
> 12 months	monthly

\* = if levels within desired treatment range

\* = minimum frequency

# PKU

- Yearly Labs
  - prealbumin, ferritin or transferrin
  - folate, vitamin B12, lipid profile > 12 years
  - consider zinc and/or selenium
  - consider other nutritional testing depending on formula
  - consider yearly quantitative plasma amino acids

# PKU

- Other Evaluations

- yearly developmental questions (parental questionnaires)
- developmental screening at 3 and 6 years
- neuropsych evaluation at 9 and 18 years
- consider psychiatric evaluation
- DEXA scans at 9 and 18 years

For other disorders this may include ophthalmology evaluations, cardiology evaluations, MRIs, speech evaluation, NCV, etc.

# PKU Performance Indicators

- Age of diet initiation
- Age of metabolic control
- Laboratory studies
  - Phe levels and frequency
  - other labs
- Growth
- Frequency of clinic visits
- Illness frequency
- Diet
  - frequency of dietitian visits
  - frequency of dietary analysis (3-day diet records)
  - tyrosine supplementation
- Neurosych assessments
- Neurologic examination findings
- Developmental services

For other disorders this may include frequency of hospitalizations, days of ICU care, need for surgeries, dialysis, transplantation, etc.

# PKU Outcome Measures

- Development
  - IQ
  - level of functioning
- Presence of psychiatric issues
  - anxiety, panic, depression
- Presence of ADHD and use of medication
- Growth (final adult parameters)

# Outcome Measures

- For other disorders may include:
  - presence of osteopenia, liver disease, pancreatitis, eye disease, renal disease, cardiac disease
  - presence of abnormal MRI findings
  - outcome of transplantation
  - mortality

# Collaboration

- Metabolic care providers representing AZ, CO, MT, NM, UT, and WY met in July, 2006 to begin the process of care plan development after preliminary plans were developed by IMD clinic.
- Second meeting occurred in December, 2006 which also include a representative from TX and from region 4.
- Third meeting just this week!

# Goals of Workshops

- Review drafts of care plans and establish minimal treatment and monitoring criteria.
- Determine performance indicators and outcome measures.
- Discuss data collection and database availability and feasibility.

Not to dictate treatment, but rather discuss current practices and come to a consensus about what management is minimally appropriate and necessary!!

# Secondary Benefit

Has allowed regional care providers to meet, talk, discuss, and collaborate!!

*Thanks Mountain States!!!*

# Progress to Date

- Completion of care plans for all metabolic disorders detected by ms-ms NBS.
- Investigation of neuropsychological tools.
- Early use of the care plans throughout the region.
  - pilot data for PKU entered
- Evaluation of possible databases including CHIRP, CHARM, the Oregon database, and Region 4's database.
  - reassessing CHIRP

# Progress to Date

- Beginning development of parent handouts for each disorder that follow care plan outlines.
- Collaboration with Drs. Botkin and Longo and their colleagues at the University of Utah for data collection tools and database functionality.

## Next 12 months....

- Ongoing review and revision of the disease specific care plans and outcome measures.
- Ongoing implementation of the care plans throughout the region and begin to collect data utilizing identified tools and database.
  - review performance indicators and outcome measures with the CHIRP database designers
  - each center to collect data on 10 patients as pilot study to determine utilization of care plans, database, time constraints, etc. (feeds back to first point above)

# Next 12 months...

- Integration of nutritional recommendations by GMDI (??).
- Ongoing development of parent handouts.
  - Word documents to be modified by each clinic
- Ongoing collaboration with regional care providers.
  - sharing of emergency medical letters
  - sharing of Spanish language resources
  - increased involvement of Texas care providers
  - hope for follow-up meeting in January, 2008.

(Joyce? Camille? John? 😊)

# Our Project Partners

- Colorado
  - Dr. Renata Gallagher
  - Dr. Johan VanHove
  - Dr. Celia Kaye
  - Laurie Bernstein, RD
  - Cindy Freehauf, RN, CGC
  - Joyce Hooker
- New Mexico
  - Dr. Claire Leonard
  - Dr. Susan Root
- Utah
  - Dr. Nicola Longo
  - Dr. Marzia Pasquali
  - Dr. Jeff Botkin
  - Sharon Ernst, RD
  - Rebecca Anderson
- Arizona
  - Dr. Kirk Aleck
  - Sarah Cox, CGC
- Texas
  - Dr. Jim Gibson