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SO, YOUR METABOLIC PATIENT IS AN ATHLETE:
Integrating Sports Nutrition Concepts for Patients with Amino Acidopathies

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DISCLOSURES: Karen Dolins

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- I am receiving an honorarium from Nutricia for this presentation
- I am a board member of the MSUD Family Support Group

- ***None pose any conflict of interest for this presentation***

The opinions reflected in this presentation are those of the speaker and independent of Nutricia North America

DISCLOSURES: Stephanie Hacker

- I am receiving an Honorarium from Nutricia for this presentation
- I have consulted with BioMarin on topics related to Wellness, Sports, and Fitness in patients with phenylketonuria
- I have served on Advisory Boards for BioMarin and Cambrooke

- ***None pose any conflict of interest for this presentation***

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LEARNING OBJECTIVES

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Participants will:

- Describe the physiological changes which occur with exercise training and their impact on substrate utilization.
- Understand nutrition strategies for sports performance.
- Modify and apply sports nutrition principles for active patients with amino acidopathies, identifying ways to optimize the patient's metabolic control.

EXERCISE PHYSIOLOGY 101

Physiological adaptations to exercise
Cardiorespiratory and muscular fitness
Substrate utilization

Sports nutrition is a blend of nutrition and exercise physiology
Requires some "art" to apply scientific principles to humans
Aim is to provide the nutrients needed to support specific level of exercise
Must understand exercise physiology to understand nutrient needs

PHYSIOLOGICAL ADAPTATIONS LEADING TO CARDIORESPIRATORY FITNESS/ENDURANCE

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- Increased plasma volume
- Heart ↑ in size and power for stronger contraction
- Increased VO_2 max (maximal oxygen consumption), RBCs and HgB
  - More O_2 consumed and delivered to cells
- Increased capacity for aerobic metabolism
  - Enhanced use of lipids/glycogen sparing

Plasma volume increases within 24 hours

Dilates L ventricular cavity like water in a balloon.

Drives increase in stroke volume along with increased heart size and power

Aids in thermoregulation

Increased stroke volume allows more blood to be pumped with each beat

Decreased heart rate for given sub-maximal exercise intensity (lower pulse rate)

Increased capacity for aerobic metabolism:

↑ capillary density

↑ mitochondria

↑ oxidative enzymes

↑ Type 1 (endurance) muscle fibers

MECHANISMS OF INCREASED MUSCULAR SIZE, STRENGTH, AND ENDURANCE

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- Muscle overload training stimulates:
  - Neural adaptations
    - Improved efficiency in neural recruitment
    - Increases strength independent of hypertrophy
  - Muscular adaptations
    - Muscle protein breakdown and synthesis (MPS) results in remodeling
    - Hypertrophy occurs when MPS is greater than breakdown
      - Slow process – max gain about 1/3 pound/week
    - Muscle stores of ATP, PCr, and glycogen increase

Motor neurons send signals to muscles, causing them to contract.

With resistance training, the nervous system adapts in ways which allow a more rapid and potent response.

Neural adaptations occur more rapidly than hypertrophy, which is a slow process.

Experience increase in strength without muscle hypertrophy.

SKELETAL MUSCLE ANABOLISM

- Food and exercise act as stimuli to induce gene expression for specific proteins
  - Activated by mammalian target of rapamycin (mTOR) pathway
    - Triggered by leucine and/or insulin and resistance/ endurance exercise
    - Note: elevated keto-acids in MSUD inhibit the mTOR pathway despite excess leucine availability
  - Muscle protein synthesis occurs during recovery from exercise

Must have both overload and appropriate nutrition.

FUEL FOR EXERCISE: SUBSTRATE UTILIZATION

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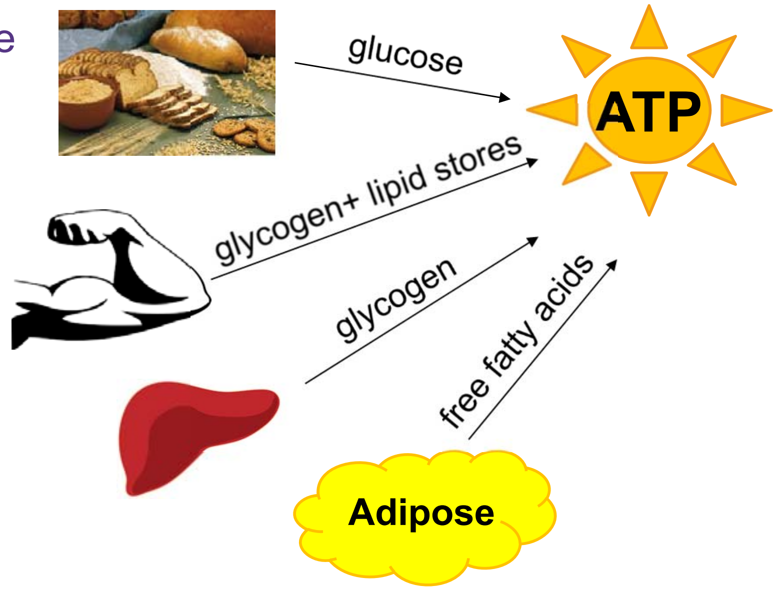
- Energy (ATP) for exercise is provided mostly through a combination of fat and carbohydrate (CHO) utilization
- Minimal energy will be derived from protein <u>unless energy intake and/or CHO intake is inadequate</u>



ATP through:

- Glycogen and lipid stores in muscle
- Blood glucose from dietary CHO and liver glycogen
- Free fatty acids released from adipose tissue

FACTORS DETERMINING FUEL USAGE DURING EXERCISE

- Intensity
- Duration
- Fitness level

Intensity

- High intensity (>80% VO_{2max}) uses primarily glucose via anaerobic glycolysis
- Uses up muscle glycogen quickly, 18x faster than aerobic glycolysis
- Up to about 60% VO_{2max} uses primarily FA aerobically
- At about 65% VO_{2max} using equal amounts of FA and CHO
- Crossover point: use more CHO less FA

Duration

- Phosphagens initially (ATP/CP)
- Muscle glycogen is predominant source of fuel for exercise of 30-60 minutes
- Long term exercise may use 60-70% fat (marathon, ultra events)

Fitness level

- Training improves ability to generate fuel via aerobic metabolism at a higher exercise intensity
- Trained individuals better able to release FA from adipose (LPL) and have more mitochondria and oxidative enzymes for aerobic metabolism

All metabolic systems work concurrently: ATP/PCr, Anaerobic Glycolysis, Aerobic Metabolism. Relative amount of each varies.

Initially and at high intensity, primary CHO source is muscle glycogen.

SPORTS NUTRITION BASICS

Accepted Standards:

AND Evidence Analysis Library

https://www.andeal.org; accessed September 2019

ACSM/AND/CDA Position Stand: Nutrition and Athletic Performance

Thomas DT, Erdman KA, Burke LM. *American College of Sports Medicine Joint Position Statement. Nutrition and Athletic Performance*. Med Sci Sports Exerc. 2016 Mar;48(3):543-68

International Olympic Committee (IOC) Guidelines

Nutrition for athletes : a practical guide to eating for health and performance : based on an international consensus conference held at the IOC in Lausanne in October 2010. By the nutrition working group of the international Olympic committee ; rev. and updated in April 2012 by Ron Maughan and Louise Burke. https://library.olympic.org accessed September 2019

NUTRITION FOR ATHLETIC PERFORMANCE

- **CALORIES to meet energy needs while optimizing weight and body composition**
- **CARBOHYDRATE fuel for working muscles**
- **PROTEIN for growth and repair of tissues, immune function**
- **FLUIDS for hydration, increased blood volume**
- **VITAMINS & MINERALS for optimal functioning**

Nutrients as members of a team: need them all in the proper proportions to win the game

SUMMARY OF EVIDENCE-BASED RECOMMENDATIONS

Energy	Adequate energy must be consumed to maximize training effect
Carbohydrate	3-12 gm/kg based on gender, sport and daily energy expenditure
Protein	1.2-2.0 gm/kg for strength and endurance athletes
Fat	≥20% of total kcals as per public health guidelines
Fluid	Drink to avoid fluid deficit > 2% of body weight
Vitamins and Minerals	Supplements generally not needed

Adapted from Thomas, 2016

The art is in individualizing these recommendations.

Protein 1.2-2.0 gm/kg for strength and endurance athletes

Protein supplements provide no advantage over "real" food

ENERGY = CALORIES

- Most important requirement!
- Must match energy intake from food with energy needs for activity to optimize performance
- Under-eating will limit performance as the body's resources must be used for basic needs
  - Breakdown of muscle tissue
  - Increased risk of injury
  - Premature fatigue

1 hour intense basketball practice can use 600-900 calories

Must be adequately fueled when beginning activity

Restricting calories for purposes of weight loss will hinder athletic performance

Restricting calories alters substrate utilization, more protein used for fuel, less CHO available

CHO: PRIMARY ENERGY SOURCE FOR WORKING MUSCLES

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- Powers contractile proteins of muscle
- Fuels brain and CNS
  - Needed for quick reactions and strategizing
- **Spares protein by minimizing gluconeogenesis**
- Used in fat metabolism

CARBOHYDRATES: ACSM/AND Guidelines

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Intensity	Range from 3-5gm/kg for low intensity to 8-12 gm/kg for high intensity long duration
Duration	60-90 g/h recommended during events of 45 minutes or longer
Recovery	1.0-1.2g/kg for 1st 4 hours after exercise to stimulate replenishment of muscle glycogen stores and reduce protein catabolism

Adapted from Thomas, 2016

Range from 3-5gm/kg for low intensity to 8-12 gm/kg for high intensity long duration

1-4g/kg 1-4 hours before activity to “top off” stores

Inadequate CHO → increased muscle protein breakdown to support gluconeogenesis

PROTEIN: HOW MUCH DO ATHLETES NEED?

- Strength and endurance athletes need more protein than sedentary people
- Inadequate CHO and kcals increase protein requirements
- EAA are key
  - Leucine → mTOR → MPS
- Extra AA's beyond needs are wasted, not used for protein synthesis

Strength and endurance athletes need more protein than sedentary people

How much more is still under investigation

Protein research flawed due to inadequate control of kcals and CHO

Inadequate CHO and kcals increase protein requirements

EAA are key

Leucine turns on mTOR which initiates MPS

All EAAs needed to synthesize new protein therefore BCAA supplementation not recommended

Extra AA's beyond needs are wasted, not used for protein synthesis

Can't oxidize BCAA's with MSUD

PROTEIN: AND/ACSM RECOMMENDATIONS

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Range	1.2-2.0 grams/kg body weight depending on gender, activity, intensity, and calorie & CHO intake
Influencing Factors	Training intensity, energy restriction, stage in training
Supplementation	Not needed; protein needs can be met by diet alone

Adapted from Thomas, 2016

1.2-2.0 grams/kg body weight depending on gender, activity, intensity, and calorie & CHO intake

- **Higher end during intense training or energy restriction**
  - **Increasing protein will minimize loss of lean tissue with energy deficit**

Recommendation often misinterpreted

Trained athletes need less

Higher protein can help preserve muscle with energy deficit, but will not allow for hypertrophy. Must prioritize athlete's goals.

PROTEIN TIMING

- Exercise-induced MPS occurs for at least 24 hours in untrained, less in trained
  - Benefit of protein immediately before or after resistance exercise not supported by research
- Distribute protein throughout the day
  - After exercise and every 3-5 hours
- 15-25 g (or 0.25-0.3 g/k BW) at a time appears optimal

15-25 g at a time appears optimal

0.25-0.3 gm/kg BW

0.40 gm/kg BW in older adults

Include bedtime feeding

FLUIDS AND HYDRATION

FLUIDS AND EXERCISE PERFORMANCE

- As little as 2% loss in body weight due to fluid loss (dehydration) will have a negative impact on performance
  - Rating of perceived exertion is higher
  - Ability to regulate body temperature is impaired
  - Exercise capacity is reduced
- Excess fluid may → hyponatremia and cerebral edema
- MUST KNOW SWEAT RATE! DRINK TO A SCHEDULE!

Many overdrink to avoid dehydration. This can be dangerous.

Handout describes methods of assessing sweat rate

SPORTS DRINKS

- Advantages over water
  - Provide fuel and electrolytes
  - Hydration more effective
  - Flavor may promote fluid intake
  - Added CHO kcals will↓ protein catabolism
- Vitamins and many other items added to beverages do not aid in hydration or improve performance
- Key ingredients: Sugar, Sodium, Potassium

Advantages over water

Provide fuel and electrolytes

Reduce gluconeogenesis

Hydration more effective

enhanced absorption and retention of fluids

Flavor may promote fluid intake

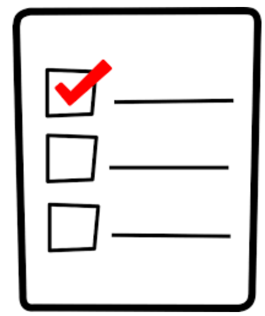
Added CHO kcals will reduce protein catabolism

Guidelines suggest using a sports drink for activities > 1 hour, but may always be beneficial with IEMs to prevent catabolism.

NUTRITION ASSESSMENT OF THE ATHLETE

NUTRITION ASSESSMENT

- Goals: athletic performance vs. fitness vs. weight management
- Height, weight, girth, body composition
- Health history/ lab data
- Estimate energy requirements
- Diet hx (include timing around physical activity, dietary supplements)
- Exercise:
  - Periodization: in-season, off-season, pre-season
  - Variability in workout days
- Knowledge, beliefs, attitudes



ASSESSING ENERGY EXPENDITURE

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- Prediction equations not highly accurate
- Estimating energy expenditure of exercise is difficult
  - Consider intensity, frequency, duration
  - Single activity factor vs weighted
  - METS (metabolic equivalents)
  - PAL (physical activity level)
- Include Non-exercise activity thermogenesis (NEAT)
  - All EE not from eating, sleeping, or primary sport ex leisure + transportation + work

INSTITUTE OF MEDICINE PHYSICAL ACTIVITY LEVELS

Physical Activity Level (PAL) Category	Mean PAL Value (Range)	Example
Sedentary	1.25 (1.1–1.39)	A person with a sedentary occupation who spends his or her entire day sitting
Low level of physical activity	1.50 (1.40–1.59)	An office worker who sits most of the day other than the walking necessary to perform tasks of daily living
Active	1.75 (1.60–1.89)	An athlete who exercises approximately 1 h/d or a person with an active vocation equivalent to walking 6–8 mile/d
Very active	2.20 (1.90–2.50)	A competitive athlete engaging in several hours of vigorous exercise training

Adapted from Karpinski and Rosenbloom (ed.), 2017 (page 203)

Estimates of energy expenditure are rough and reflect a starting point.

Must monitor weight changes and adjust when necessary.

THE SPORTS NUTRITION PRESCRIPTION

1. Start with energy needs
2. Assess protein needs
3. Maximize CHO within energy requirements
4. At least 20% of kcals from fat
5. Assess sweat rate to determine fluid requirements

Example:

- 25 year old cyclist training for 2 hours/day
  - 80 kg BW
- HB x 1.2 = 2600 kcals
- Energy cost of cycling: 600 kcals/hour x 2= 1200 kcals
- Total energy needs: 3800 kcals
- Protein 1.5 g/kg = 120 gms = 480 kcals protein
- 25% kcals fat = 80 gm fat = 720 kcals
- 3800 – 480 – 720 = 2600 kcals = 650 gm CHO or 8 gm/kg BW

KEY POINTS

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- Avoid energy deficit to optimize performance
- Food and fluid intake must be timed around activity
- Avoid under- or over-hydration
- Distribute protein evenly throughout the day

Avoid energy deficit

- Estimate exercise energy expenditure
- Provide CHO fuel during activities to avoid catabolism
- Weight loss during off-season only

Food and fluid intake must be timed around activity

Avoid under or over-hydration

- Assess sweat rate
- Drink to a plan
- Consider sports drink for all activity of moderate intensity and duration

Distribute protein evenly throughout the day

- Extra kcals for anabolism should come primarily from CHO

SPORTS NUTRITION RESOURCES

- **AND Sports Nutrition Care Manual**
  - New updates published 2/18
  - https://sports.nutritioncaremanual.org/sports-nutrition-care
- **Sports Nutrition: A Handbook for Professionals 6th Edition © 2017 AND**
- **AND/ACSM/CDA Position Stand: Nutrition and Athletic Performance**
  - http://www.eatrightpro.org/resource/practice/position-and-practice-papers/position-papers/nutrition-and-athletic-performance
- **USOC Sports Nutrition Tip Sheets**
  - https://www.teamusa.org/nutrition
- **Sports, Cardiovascular and Wellness Nutritionists (SCAN) DPG**
  - www.SCANdpg.org
- **Australian Institute of Sports Nutrition**
  - https://www.ausport.gov.au/ais/sports_nutrition
- **Physical Activity Guidelines for Americans**
  - https://www.hhs.gov/fitness/be-active/physical-activity-guidelines-for-americans/index.html

APPLYING SPORTS NUTRITION TO AMINO ACIDOPATHIES

Phenylketonuria
Tyrosinemia
Homocystinuria
Maple Syrup Urine Disease

A blend of nutrition and exercise physiology

Requires some "art" to apply scientific principles to humans

Aim is to provide the nutrients needed to support specific level of exercise

Must understand exercise physiology to understand nutrient needs

WHAT THE RESEARCH TELLS US

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- Limited number of studies published
- All studies are acute studies and are not designed to inform the impact of exercise training on intact protein tolerance or metabolic control

GRÜNERT ET AL, 2013

Diurnal variation of phenylalanine and tyrosine concentrations in adult patients with phenylketonuria: subcutaneous microdialysis is no adequate tool for the determination of amino acid concentrations

- First study to assess impact of physical exercise on Phe and Tyr levels
- Results related to exercise
  - No change in Phe concentration within two hours
  - Changes in Tyr within one hour
- Limitations
  - Short duration
  - Intensity of exercise was not explained

HELM, 2014

The effects of an acute bout of moderate-intensity exercise on plasma amino acid concentrations in adolescent boys with phenylketonuria

- Objective: Determine effects of moderate intensity and sedentary activity on amino acid oxidation in 2 male teenagers
- Results
  - Phe levels variable between subjects
    - Participant 1, increased with activity
    - Participant 2, decreased with activity
- Limitations
  - Only 2 subjects with inconclusive data

MAZZOLA ET AL, 2015

Acute exercise in treated phenylketonuria patients: Physical activity and biochemical response

- Objective: Investigate acute effect of acute aerobic exercise on pAAs, glucose & cholesterol, measured pre-exercise vs. immediately post-exercise + meal
  - Controlled for exercise intensity
- Results: PKU vs Control
  - No difference in VO_2, (prescribed and during exercise) and RER (during exercise)
  - Pre-exercise: ↑ Phe and Phe/Tyr; ↓ BCAA and Total Cholesterol
  - Post-exercise + meal: No change in Phe from pre-exercise;Tyr ↑ in Control
- Limitations
  - Subjects were untrained and diet may not have been appropriate for all participants
  - Phe level assessed immediately after exercise and eating

JANI ET AL, 2017

Protein intake and physical activity are associated with body composition in individuals with phenylalanine hydroxylase deficiency.

- Objective: Identify the relationship between dietary protein, physical activity (PA), and PAH genotype on lean mass and other body composition parameters in a large subject sample with PAH deficiency
- Results
  - Adults: ↑ intact protein intake associated with ↑ fat-free mass index (FFMI)
  - Children: ↑ modified protein intake associated with ↑ FFMI
  - All subjects: less intense (vs. high-intensity) PA associated with ↑ fat mass index (FMI)
  - No association of genotype on activity or body composition
- Limitations
  - No direct exercise intervention

ROCHA ET AL, 2019

A series of three case reports in patients with phenylketonuria performing regular exercise: first steps in dietary adjustment.

- First publication with recommendations for PKU patients re: exercise
- 3 Case Studies
  - Strength Training
  - Intermittent Exercise
  - Aerobic Exercise
- Recommendations:
  - Optimization of Phe-free L-amino acid supplements
  - High quality protein given post-exercise

EFFECT OF EXERCISE ON METABOLIC CONTROL

- Most studies show that exercise has no effect on Phe levels
  - Current Literature has measured acute response (30min-9 hours)
    - Muscle protein synthesis is optimized at 24 hours post exercise (Tipton, 1999)
- Muscle breakdown may cause an increase amino acid levels, due to release of amino acids from muscles related to catabolism (MacDonald, 2019)

PROTEIN, SPORTS AND AMINO ACIDOPATHIES

- Protein needs increase with exercise
- Timing of intake
  - Intake should be distributed throughout the day
- High quality proteins contain all essential amino acids in appropriate concentrations
  - Metabolic Formulas
    - Amino acid-based formulas: May be oxidized and absorbed more rapidly than whole protein
      - Some studies have shown lower protein utilization with free amino acids (Gropper, 1991)
    - Glycomacropeptide (GMP)-based: more research needed to show a benefit over amino acid-based formulas for protein utilization
  - Consider consumption of metabolic formula with intact protein-containing meal
    - To obtain all essential amino acids
    - Leucine initiates muscle protein synthesis (MPS)

Formula/Medical Food is the main source of protein in diet for patients with Amino Acidopathies, without providing phe

Many Formula's contain calories, Phe free (or low Phe) protein, fats, carbohydrates, vitamins, and minerals.

Traditional formulas are made from Free Amino Acids (which are the building blocks of proteins) but do not contain Phe.

Glycomacropeptide (GMP) formulas are made of whole protein molecules and contain some Phe.

0.25-0.3 g/kg (or 15-20g protein) should be consumed within 4 hours of activity for enhanced muscle protein synthesis

It is important to replenish amino acids/protein to recovering muscles

LEUCINE CONTENT (in mg) OF METABOLIC FORMULAS COMPARED TO COW'S MILK (per 10 g PE)

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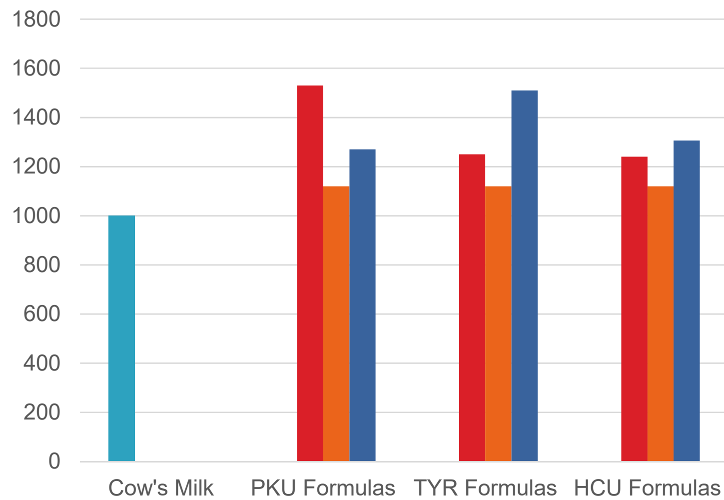

Egg and Whey both have high Protein Equivalency ratio and net protein utilization

Cows milk is considered ideal protein for muscle recovery.

Medical foods provide leucine contents in excess of that in cows milk.

MSUD formulas are not included b/c they don't contain LEU

PRE- AND POST-ACTIVITY SNACKS FOR AMINO ACIDOPATHIES

Pre-Activity

- Increase glycogen levels
  - Fruit
  - Coconut milk yogurt
  - Low protein granola/snack bar
  - Low protein bread/grains

Post-Activity

- Replenish fluid, carbohydrate, and protein stores
  - Formula/Medical Food
  - Consider carbohydrates

Pre Activity

Goal to increase Glycogen levels before exercising

Low-fat, low-fiber, and low-to-moderate protein snacks

Post-Activity

Goal is recovery of energy, replace glycogen stores, and repair muscles.

Formulas can provide necessary amounts of protein and carbohydrates

Can work with type of formula and modular for ideal CHO and Protein based on activity

Grains (traditional or low protein) may be needed dependent on medical food and activity

FUELING AN ATHLETE WITH AN AMINO ACIDOPATHY

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- Consuming adequate protein, carbohydrates and calories
  - Natural protein optimization (according to tolerance)
  - Utilize Formula to meet protein needs
  - Properly dosed Formula will not limit athletic potential
- On-the-go snack/meal options
- Hydrate appropriately
- Participating in team meals could be an area of insecurity for patients

SPECIAL CONSIDERATIONS FOR TYR, HCY & MSUD

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TYROSINEMIA

- Similar to PKU
- Liver Dysfunction should be considered
  - May be advisable to avoid impact sports to prevent possible trauma to liver

HOMOCYSTINURIA

- Risk of thromboembolism
- Skeletal concerns may increase risk of injuries
  - Scoliosis
    - Muscular imbalances
  - Joint Hypermobility
    - Sprained ligaments

MAPLE SYRUP URINE DISEASE

- May be at risk for metabolic crisis
  - Avoid catabolism
    - Higher energy and protein needs
  - Hydration needs to be monitored carefully
    - Hyponatremia

CASE STUDIES

CASE STUDY 1

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- 8 year old male with PKU
  - 34 kg, 136 cm, 80-85% BMI for Age (CDC)
  - Phe levels: 200-250 uMol/L
  - Diet: 400 mg Phe and 45 g Protein Equivalent (PE) from Phe Free L-Amino Acid Supplement
    - 1.3 g/kg protein from medical food taken 3x/day
- Joined competitive baseball team as shortstop
  - Practice/Games increased from 60 min, 3 days/week to 90 min, 5-6 days/week
  - Phe levels increased to >400 uMol/L
    - Increased formula to 60 g PE/day (1.8 g/kg) and distributed formula over 4-5 servings/day (with a serving following practices/games)
      - Phe levels decreased to <150 uMol/L
        - Increased diet to 500 mg Phe/day → Phe levels normalized to 200-250 uMol/L

CASE STUDY 2

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36 year old female with PKU

- 70 kg, 160 cm, 27 kg/m^2
- Phe levels: 200-250 uMol/L
- Diet: 10 g intact protein and 90 g PE from Phe-free amino acid-based medical food
  - 1.4 g/kg (based on 64 kg) protein and 1000 kcal/day from medical food
  - Taken in 3-5 portions/day (dependent on training)
- Triathlete (>60 min training, 6 days per week); diet plan was previously optimized for performance
  - Medical food increased from 1.1 to 1.4 g/kg
    - Dose following training sessions & PE modular added to sports drinks during prolonged sessions
  - Carbohydrate optimization before, during and after training
  - Hydration optimization using electrolytes
- Desire to lose 6 kg gained
  - Energy Balance was +500 kcal/d
  - Added lower-calorie formula modular and ↓ traditional medical food → cut 300 kcals and increased protein to 2.0 g/kg
  - Counseling on appropriate carbohydrate servings
  - **Pt lost weight, kept Phe levels stable, and did not compromise training**

CHO Optimization

Before exercise: 1-3g/kg (2 slices Cambrooke Bread and jelly (~50g protein) or coconut yogurt

During: 50-60 g CHO/hour if exercising for more than 90 minutes (HoneyStingers/GUs/Sports Drinks)

After: medical food and pasta or bread

CASE STUDY 3

20 year old male with PKU

- 84 kg, 187 cm, 24 kg/m^2
- Phe levels: 400-500 uMol/L
- Diet: 250 mg Phe (5 g protein) and 100 g PE from Phe-free amino acid-based medical food
  - 1.2 g/kg protein from medical food taken 2-3x/day
- Weight Training: 75 min, 3-4 days per week with desire to "Get Big"
  - Reported no change in muscle mass with 3 months of training
- Added 20 g PE from GMP product after training sessions (no other changes made to diet) - increased protein intake to 1.4 g/kg)
  - Pt reported 4lb (1.8kg) increase in 2 months and ability to "lift more"
  - Phe levels decreased to 280 uMol/L

CASE STUDY 4

16 year old female with HCY

- 55 kg, 165 cm, 75-85% BMI for Age (CDC) (20.2 kg/m^2)
- Met levels: 40-50 uMol/L, HCY levels: 30-45 uMol/L
- Diet: 18 g Protein/d and 40 g PE from RTD Met-free medical food
  - 1.24 g/kg total protein
  - Medical food taken before school and before bed
- Added Dance Class to school schedule (increased activity from 60 min, 2 days/week to 60 min, 4 days/week)
  - Complained of increased hunger; was filling up on chips, pasta, and other grain-based food; Mom reported weight gain
- Recommended moving evening serving of medical food to after dance practice

→ **Hunger Resolved and weight normalized**

Medications and vitamin C used in addition to diet

POTENTIAL AREAS OF STUDY

- Many unknowns in field of Exercise and Amino Acidopathies….
  - Improved PAA profiles with increased activity?
  - Affect of timing of medical food intake on metabolic control, MPS, athletic performance?
  - Affect of Calories vs Protein on Athletic Performance/MPS?
    - Affect of Protein Quality on MPS?
  - Limitation of MPS in MSUD (due to limitation of BCAAs)?

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ASSESSING SWEAT RATE

Assessing sweat rate will help prevent under and over hydration. Sweat rate must be re-evaluated with environmental changes.

1. Hydrate before exercise session.
2. Warm up 5-10 minutes.
3. Empty bladder.
4. Obtain weight with minimal clothing.
5. Fill a water bottle showing measurements (oz. or ml.)
6. Exercise at typical intensity for a specified duration.
7. Obtain weight with minimal clothing.
8. Determine body weight change.
9. Add fluid consumed.
10. Weight change + fluid consumed = sweat rate.
11. Divide sweat rate by duration to obtain hourly sweat rate.

<u>Example</u>

Pre-exercise weight:	130 pounds
Post-exercise weight:	128 pounds
Weight loss:	2 pounds or 32 ounces
Fluid consumed:	16 ounces
Sweat loss:	32 ounces + 16 ounces = 48 ounces
Duration:	1.5 hours
Hourly sweat rate:	48 ounces/1.5 = 32 oz./hour

*Note: Any urine passed before post-exercise weight must be measured and subtracted.

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