NEW YORK STATE DEPARTMENT OF HEALTH

CHILDREN WITH SPECIAL HEALTH CARE NEEDS PROGRAM
WADSWORTH CENTER NEWBORN SCREENING PROGRAM

Criteria for Approval of Inherited Metabolic Diseases Specialty Centers

An Inherited Metabolic Diseases (IMD) Center is defined as an Article 28 general hospital that has at least one board certified/eligible pediatric specialist in Inherited Metabolic Diseases. In addition to meeting the requirements specified for a general hospital in New York Codes, Rules and Regulations (NYCRR) Title 10, Part 405, an IMD Center will meet the criteria specified below. The center is able to treat, but is not restricted to, phenylketonuria, branched-chain ketonuria, galactosemia, homocystinuria, biotinidase deficiency, Krabbe disease medium-chain acyl-CoA dehydrogenase deficiency and related disorders. If the Inherited Metabolic Disease Center will operate at more than one location, each sub site must be identified.[1]

I. Hospital Administration

Administration shall be responsible for:

- general operation of the center in accordance with written policies and procedures;
- employment or availability of qualified personnel, who maintain privileges at the hospital;
- compiling of statistical data, and review and revision of reporting systems and data collection;
- providing an annual update of all changes in personnel;

II. Specialty Center Facilities

Specialty Center facilities must include:

- inpatient and outpatient facilities available according to the needs of the individual patient;
- dedicated pediatric inpatient beds, neonatal intensive care unit as part of a Level III perinatal program[2] and pediatric intensive care unit;[3]
- onsite facilities for standard clinical-pathological studies, radiological studies (i.e. X-rays, sonogram, CT, MRI) electrocardiogram studies, electroencephalogram studies, brainstem auditory evoked response (BAER) studies, visual evoked response (VER) studies, nerve conduction studies and nuclear medicine studies.
- laboratory capability for all necessary core studies either onsite or by referral to another New York State approved laboratory as specified by the New York State Newborn Screening Program and the Wadsworth Laboratories.

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[1] The Permanent Facility Indicator (PFI) will identify the Inherited Metabolic Diseases Specialty Center in the application. If the Inherited Metabolic Diseases Specialty Center will operate in more than one location, each sub site must be specified by address and PFI number in the application. Each sub site will: a) be on the operating certificate of the Article 28 facility that is the main site; b) be identified by a PFI number; c) have physician specialists that have admitting privileges to the main site; and d) assure access to all services at the main site specified in these criteria.

[2] If the hospital does not provide maternity services, then a neonatal intensive care unit is not required.

[3] If the hospital does not have a pediatric intensive care unit (PICU), then a protocol must be described for emergent care, appropriate pediatric transport, and continuity of IMD care.

In addition to standard clinical studies, the laboratory shall have the capability to perform the following core tests either onsite or by referral to another New York State approved laboratory.

1. Carnitine (blood and urine) free and total; acylcarnitine (blood)
2. Plasma amino acids, inclusive but not limited to:
   - alanine
   - alloisoleucine
   - arginine
   - argininosuccinic acid
   - citrulline
   - cystathionine
   - cysteine/cystine
   - glutamine
   - homocysteine/homocystine
   - isoleucine
   - leucine
   - methionine
   - ornithine
   - phenylalanine
   - tyrosine
3. Plasma free fatty acids
4. Urine organic acids:
   - acylglycine
   - galactitol
   - urine biopterin
5. Quantitative enzyme assays, galactose-1-PO_4 transferase, galactose-1-PO_4 galactocerebrosidase and biotinidase
6. Fatty acid B-oxidation enzymes in cultured fibroblasts
7. Starch gel electrophoresis/isoelectric focusing
8. Molecular genetic testing
9. Urine reducing substances

III. Specialty Center Personnel

A. Core Personnel:

The core personnel consist of medical director and any additional board certified/eligible pediatric specialist in IMD.

A.1. Medical Director, Inherited Metabolic Disease Center

The medical director shall:

- be a board certified/eligible pediatric endocrinologist with training and experience in IMD, board certified/eligible clinical geneticist with training and experience in IMD, or board certified/eligible pediatrician with training and experience in IMD; and
- assure that all children referred to the center are seen by the director or another board certified/eligible pediatric specialist in IMD.

A.2. Pediatric Specialist in IMD

The board certified/eligible pediatric endocrinologist, board certified/eligible clinical geneticist, or board certified/eligible pediatrician with training and experience in IMD will assure:

- each child referred to the center receives an initial comprehensive evaluation, on an inpatient or outpatient basis, as indicated. The comprehensive evaluation includes a medical, psychological and social history, physical exam and appropriate diagnostic studies;
- appropriate treatment of the child is initiated and assure the development and implementation of a plan for ongoing management with parental involvement;
- the child receives appropriate developmental evaluation and follow-up;
- supportive services are in place to assist families with identifying resources and applying for benefit programs related to the financial aspects of care, education about the disorder and training about self-care for patients and their families;
- the child has a primary care provider;
- adequate and prompt reports on clinical evaluations, recommendations for treatment and follow-up and other necessary records are provided to referring physicians and, when requested, to the screening laboratory; and
- guidance is provided for transitioning to adult care; and
- appropriate evaluation for patients already under treatment at the time of referral.

B. Other Core Personnel

The following personnel shall be available onsite for consultation:

- Board certified/eligible pediatric neurologist, or a board certified/eligible developmental pediatrician;
- Medical social worker;
- Registered dietician or certified dietician/nutritionist with experience in working with pediatric patients;
- Registered nurse coordinator; and
- Laboratory director who meets the qualifications specified in Part 19 Clinical Laboratory Directors, Section 19.2-19.4, Title 10 Official Compilation of Codes, Rules and Regulations of the State of New York. The Laboratory Director's Certificate of Qualification as issued by the NYS Clinical Laboratory Evaluation Program (CLEP) must include:

  Clinical Chemistry or Biochemical Genetics

C. Additional Personnel

Consultation shall be available onsite or by referral in all specialties related to the care of the patient, including:

- board certified/eligible clinical geneticist;
- genetic counselor who is board certified or an active candidate for certification;
- licensed psychologist;
- board certified/eligible pediatric radiologist;
- board certified/eligible pediatric ophthalmologist;
- board certified/eligible MD and/or PhD biochemical geneticist;
- board certified/eligible plastic surgeon;
- licensed audiologist (experienced in working with pediatric patients);
- licensed occupational therapist (experienced in working with pediatric patients);
- licensed physical therapist (experienced in working with pediatric patients); and
- licensed speech language pathologist (experienced in working with pediatric patients)

Reference: U.S. Newborn Screening System Guidelines II: Follow-up of Children, Diagnosis, Management, and Evaluation; Statement of the Council of Regional Networks for Genetic Services (CORN). The Journal of Pediatrics. October, 2000, 137:4.