**New Application: Medical Biochemical Genetics**

**Review Committee for Medical Genetics and Genomics**

**ACGME**

**Oversight**

**Participating Sites**

Will the Sponsoring Institution also sponsor ACGME-accredited residencies in the following specialties? [PR.I.B.1.a)-b)]

1. Medical genetics and genomics  YES  NO
2. Pediatrics  YES  NO
3. Internal medicine  YES  NO

Explain any NO responses.

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**Resources**

1. Provide the data requested below, including the number of analyses and the analytic method, for each biochemical genetics laboratory that will contribute significantly to the education of fellows. Duplicate table as needed. [PR I.D.1.a)]

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| 12-Month Period Covered by Statistics | From: Date | To: Date |

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| Name of Laboratory: | Click here to enter text. | |
| Address: | Click here to enter text. | |
| Name of Laboratory Director: | Click here to enter text. | |
| **Test** | **Number of Analyses** | **Analytic Method** |
| Newborn Screening | # | Method |
| Amino Acid Analysis | # | Method |
| Organic Acid Analysis | # | Method |
| Acylcarnitine profile | # | Method |
| Mucopolysaccharide Screening | # | Method |
| Enzyme analyses | # | Method |
|  | # | Method |
|  | # | Method |
|  | # | Method |
| Other small molecule analyses | # | Method |
|  | # | Method |
|  | # | Method |
|  | # | Method |
| Prenatal Diagnosis: Disorders | # | Method |
|  | # | Method |
|  | # | Method |
|  | # | Method |
| List Other: | # | Method |

Provide the patient visit or consult numbers below for each site/clinic based upon the prior academic or calendar year. Note: A single patient may be counted only up to three times, irrespective of the number of visits/encounters; for inpatients, each admission is a single encounter (i.e., each daily visit during a single admission is NOT counted). (Copy table as necessary.) [PR I.D.1.b)]

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| 12-Month Period Covered by Statistics | From: Date | To: Date |

| **Site/Clinic Name** | Click here to enter text. | | | | | |
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| **Types of New Patients** | **0-5** | **5-10** | **10-20** | **20-30** | **> 30** | **Total** |
| Hyperammonemic crisis |  |  |  |  |  |  |
| Metabolic or lactic acidosis |  |  |  |  |  |  |
| Newborn screen |  |  |  |  |  |  |
| **Total** |  |  |  |  |  |  |
| **Types of Established Patients** | **0-5** | **5-10** | **10-20** | **20-30** | **> 30** | **Total** |
| Fatty acid oxidation disorders |  |  |  |  |  |  |
| Galactosemia |  |  |  |  |  |  |
| Glycogen storage diseases |  |  |  |  |  |  |
| Lysosomal storage disease |  |  |  |  |  |  |
| Mitochondrial/energy metabolism disorders |  |  |  |  |  |  |
| Organic acidemias |  |  |  |  |  |  |
| PKU |  |  |  |  |  |  |
| Urea cycle disorders |  |  |  |  |  |  |
| **Total** |  |  |  |  |  |  |

3. Concisely describe the meeting rooms, classrooms, office space, research space, and facilities for record storage and retrieval utilized by the program. [PR I.D.1.c).(1)]

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4. Concisely describe the office and laboratory space provided for fellows for both patient care work and participation in scholarly activities. [PR I.D.1.c).(2)]

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**Personnel**

**Other Program Personnel**

1. Concisely summarize the technical, clerical, and other non-physician personnel who will provide support for the administrative and educational conduct of the program. Is the support of the program in this area satisfactory at all program sites? [PR II.D.]

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1. Working with Other Health Care Professionals

Summarize the opportunities fellows will have to work with genetic counselors, nurses, and nutritionists who are involved in the provision of medical biochemical genetics services. [PR II.D.1

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**Educational Program**

**Medical Knowledge**

1. Describe how (including specific role), and in what settings, fellows learn about the acute management of inborn errors of metabolism (IEM). [PR IV.B.1.c).(1)]

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1. Describe how (including specific role), and in what settings, fellows learn about enzyme replacement and organ transplant therapies. [PR IV.B.1.c).(1).(a).(iv).(c)]

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1. Describe how (including specific role), and in what settings, fellows learn about newborn screening for metabolic disorders. [PR IV.B.1.c).(1).(a).(iv).(d)]

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1. Describe how (including specific role), and in what settings, fellows learn about the genetic epidemiology of inborn errors of metabolism and the application of that knowledge to newborn screening. [PR IV.B.1.c).(1).(a).(v)]

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**Curriculum Organization and Resident Experiences**

1. Will fellows have the opportunity to develop the ability to diagnose IEMs, counsel patients, and manage the broad range of clinical problems encompassed by biochemical genetics?   
   [PR IV.C.7.)]  YES  NO
2. Describe the role of the fellow and settings for learning about care of adults with IEMs.   
   [PR IV.C.7)]

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1. Briefly describe how fellows will be provided with structured education, including formal coursework, in the basic sciences and clinical areas pertinent to biochemical genetics, to include population and newborn screening, disorders of amino acid metabolism, disorders of fatty acid oxidation, mitochondrial disorders, galactosemia, glycogen storage diseases, lysosomal storage diseases and lipidoses, peroxisomal disorders and other IEMs, acute management of IEMs, enzyme replacement therapy, long-term nutritional management, and molecular diagnosis. [PR IV.C.9.b).(1)-(10)]

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1. Will fellows be provided with mentored clinical education in the practice of biochemical genetics in both outpatient and inpatient settings? [PR IV.C.9.c)]  YES  NO
2. Will fellows be provided with basic instruction in medical biochemical genetic laboratory testing? [PR IV.C.9.d)]  YES  NO
3. Will fellows be provided with basic instruction in clinical research? [PR IV.C.9.e)]  YES  NO
4. Will fellows be provided with advanced instruction in the interpretation of biochemical laboratory test results? [PR IV.C.9.f)]  YES  NO
5. What is the planned nature and extent of fellow experience during their assignments to the biochemical laboratory? Describe planned fellow participation in the working conferences of these laboratories and the ongoing discussions of laboratory data during other clinical conferences.   
   [PR IV.C.10.a)]

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8. Provide a list of the planned lectures and other didactic sessions which meet the requirement for a course in biochemical genetics. (Such a course must be more advanced than the genetics course given to medical students.) [PR IV.C.11]

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**Fellows’ Scholarly Activities**

Summarize program research activity and list the staff member(s) who will provide support and supervision of clinical or laboratory research activity by fellows and identify their particular area(s) of expertise. [PR IV.D.3.a)]

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