**New Application: Clinical Biochemical Genetics**

**Review Committee for Medical Genetics and Genomics**

**ACGME**

**Sponsoring Institution**

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

1. Medical genetics and genomics [ ]  YES [ ]  NO

If no, please explain:

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1. Laboratory genetics and genomics [ ]  YES [ ]  NO
2. Medical biochemical genetics [ ]  YES [ ]  NO

**Program Personnel and Resources**

**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 post-doctoral fellows. Duplicate table as needed. [PR I.D.1.b)]

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

|  |  |
| --- | --- |
| Name of Laboratory: |  |
| Address: |  |
| Name of Laboratory Director: |  |
| **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 |
| List Other: | # | Method |

2. Concisely describe the office space, meeting rooms, classrooms, laboratory space, and research facilities available at on-site laboratories. [PR I.D.1.b).(1)] (Limit response to 400 words)

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**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.] (Limit response to 400 words)

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1. Summarize the opportunities post-doctoral fellows will have to work with laboratory technicians, laboratory quality officers, laboratory directors, and other laboratory professionals who are involved in the provision of clinical and medical biochemical genetics services. [PR II.D.1.] (Limit response to 400 words)

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1. Summarize the opportunities post-doctoral fellows will have to work with genetic counselors, nurses, dieticians, and other health care professionals who are involved in the provision of clinical and medical biochemical genetics services. [PR II.D.1.] (Limit response to 400 words)

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**Educational Program**

**Patient Care**

1. Describe how and in what settings post-doctoral fellows will demonstrate competence in pre-analytic, quality control, analytic, and interpretive laboratory skills. [PR IV.B.1.b).(1).(a).(i)] (Limit response to 400 words)

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1. Describe how and in what settings post-doctoral fellows will demonstrate competence in post-analytic reporting skills. [PR IV.B.1.b).(1).(a).(ii)] (Limit response to 400 words)

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1. Describe how and in what settings post-doctoral fellows will demonstrate competence in the principles and techniques of specimen selection, sample preparation, analysis, and results interpretation and reporting. [PR IV.B.1.b).(2).(a)] (Limit response to 400 words)

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**Medical Knowledge**

List the activity(ies) (lectures, conferences, journal clubs, clinical teaching rounds, etc.) in which post-doctoral fellows will demonstrate knowledge in each of the following areas. Also indicate the assessment method(s) that will be used to assess competence. [PR IV.B.1.c)]

| **Area of Expertise** | **Settings/Activities** | **Assessment Method(s)** |
| --- | --- | --- |
| *General principles of biology and genetics as related to biochemical genetics [PR IV.B.1.c).(1)]* |
| Biochemical pathways and how they interrelate[PR IV.B.1.c).(1).(a)] | Click here to enter text. | Click here to enter text. |
| Enzyme kinetics[PR IV.B.1.c).(1).(b)] | Click here to enter text. | Click here to enter text. |
| Principles of population-based screening[PR IV.B.1.c).(1).(c)] | Click here to enter text. | Click here to enter text. |
| *Clinical, biochemical, pathophysiologic, diagnostic, and treatment features of biochemical genetic conditions[PR IV.B.1.c).(2)]* |
| Carbohydrate metabolism (e.g., glycogen storage disorders)[PR IV.B.1.c).(2).(a)] | Click here to enter text. | Click here to enter text. |
| Cofactor and metal metabolism and transport (e.g., B12 deficiency)[PR IV.B.1.c).(2).(b)] | Click here to enter text. | Click here to enter text. |
| Complex molecule metabolism (e.g., lysosomal storage disorders and congenital disorders of glycosylation)[PR IV.B.1.c).(2).(c)] | Click here to enter text. | Click here to enter text. |
| Energy metabolism (e.g., mitochondrial myopathies)[PR IV.B.1.c).(2).(d)] | Click here to enter text. | Click here to enter text. |
| Small molecule metabolism (e.g., neurotransmitter disorder)[PR IV.B.1.c).(2).(e)] | Click here to enter text. | Click here to enter text. |
| Treatment options for all metabolic disorders [PR IV.B.1.c).(2).(f) | Click here to enter text. | Click here to enter text. |

**Interpersonal and Communication Skills**

1. Describe the role of the post-doctoral fellows and settings in gaining experience in communicating results to physicians, other health professionals, and health-related agencies. [PR IV.B.1.e).(1).(b)] (Limit response to 400 words)

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**Systems-based Practice**

1. Describe opportunities for post-doctoral fellows to actively participate in laboratory inspections. [PR IV.B.1.f).(1).(h)] (Limit response to 400 words)

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1. Describe how and in what settings post-doctoral fellows will participate in laboratory quality management, including quality control and quality assurance. [PR IV.B.1.f).(1).(i)]

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**Curriculum Organization and Post-Doctoral Fellow Experiences**

1. Will post-doctoral fellows participate in a rotation in laboratory genetics and genomics of at least four weeks in length? [PR IV.C.2.a).(1)] [ ]  YES [ ]  NO
2. Describe how post-doctoral fellows will participate in the development of clinical laboratory methods or tests. [PR IV.C.2.b)] (Limit response to 400 words)

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1. Describe opportunities for direct exposure to the clinical evaluation of patients, medical decision-making, and genetic counseling. [PR IV.C.4.a)] (Limit response to 400 words)

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1. Describe how and in what settings, post-doctoral fellows will have direct exposure to patients with inborn errors of metabolism in inpatient and/or outpatient setting(s). [PR IV.C.4.a).(2)] (Limit response to 400 words)

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1. Briefly describe how post-doctoral fellows will be provided with structured education, including formal coursework, in the following areas: [IV.C.5]

| **Topic Area** | **Structured Education** |
| --- | --- |
| Disorders of amino acid metabolism[PR IV.C.5.b).(6)] | Click here to enter text. |
| Disorders of carbohydrate metabolism[PR IV.C.5.b).(7)] | Click here to enter text. |
| Disorders of fatty acid oxidation[PR IV.C.5.b).(8)] | Click here to enter text. |
| Lysosomal storage diseases[PR IV.C.5.b).(15)] | Click here to enter text. |
| Management of inborn errors of metabolism (IEM) (acute and long-term)[PR IV.C.5.b).(16)] | Click here to enter text. |
| Mitochondrial disorders[PR IV.C.5.b).(18)] | Click here to enter text. |
| Molecular diagnosis [PR IV.C.5.b).(19)] | Click here to enter text. |
| Peroxisomal disorders and other IEM[PR IV.C.5.b).(21)] | Click here to enter text. |
| Population and newborn screening[PR IV.C.5.b).(22)] | Click here to enter text. |

1. Provide a list of the planned seminars, journals clubs, rotations in a clinical chemistry laboratory, and other educational opportunities. Comment on the levels of teaching staff member participation and post-doctoral fellow attendance at these sessions. Provide a list of topics and speakers as appropriate. [PR IV.C.7.)]

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**Supervision and Progressive Responsibilities**

1. How will the members of the faculty ensure that post-doctoral fellows have an opportunity to assume increasing responsibility for result interpretation and reporting as they progress through the program? [PR VI.A.2.f)]

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