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Big Data Strikes Again: Future Utilization of the UK Biobank as a Resource for Clinical Laboratories

Article:
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Guest: Dr. Joseph T Alaimo is the assistant director of the Clinical Molecular Genetics Laboratory in the Genomic Medicine Center and Department of Pathology at Children's Mercy Hospital, Kansas City, and an assistant clinical professor at the University of Missouri.

Bob Barrett: This is a podcast from *Clinical Chemistry*, sponsored by the Department of Laboratory Medicine at Boston Children's Hospital. I am Bob Barrett. The United Kingdom Biobank is a long-term perspective population-based genomic medicine initiative in which about half a million individuals from across the United Kingdom have undergone an array of health and genetic assessments with the goal of accelerating the discovery of lifestyle, environmental, and genomic factors that influence disease. This unprecedented approach has already yielded an enormous wealth of information that will undoubtedly inform human health behaviors. Areas in which such data will impact the practice of laboratory genomic medicine include variant frequency utilization, population disease risk and novel disease gene discovery. A perspective article appearing is the cover story for the July 2021 issue of Clinical Chemistry examined the UK Biobank and its potential. The authors of that article titled "Big Data Strikes Again: Future Utilization of the UK Biobank as a Resource for Clinical Laboratories" are Dr. Joseph Alaimo and Dr. Carol Saunders, and we are pleased to have Dr. Alaimo as our guest in this podcast. He is the assistant director of the Clinical Molecular Genetics Laboratory in the Genomic Medicine Center and Department of Pathology at Children's Mercy Hospital, Kansas City, and an assistant clinical professor at the University of Missouri also in Kansas City. So, Dr. Alaimo, what is the purpose and importance of such a large-scale genomic study? What's the benefit to society as a whole?

Joseph Alaimo: So, the UK Biobank is a long-term perspective population-based study in which genetic medicine is the focus. And so, this particular study is very unique because it's looking at an entire country and looking at a variety of genomic and also, health and lifestyle assessments. And something like this at this level hasn't really been done before. So, seeing the treasure trove of data that exists or may exist in this type of approach, in terms of understanding our lifestyle choices and how are genetic factors or genetics determine or influences disease. This is something that I think will benefit human health in general and for many, many years to come.

Bob Barrett: Could you review for us who typically receives exome sequencing and why the application of the testing in the Biobank study is unprecedented? And, I guess, can anyone get one?

Joseph Alaimo: Typically, individuals who have some type of physical features usually, at least in my profession, it's in the prenatal setting or in a pediatric setting. When things aren't developing properly, these individuals tend to receive some type of testing whether it be targeted or looking at the entire genome and we commonly refer to this as an exome just looking at regions that are coded into protein that may be the most informative. And so, typically, an individual who is healthy doesn't necessarily need to receive an exome, because it's for somebody who might carry a mutation or a variant in their genome that is causing disease. And so, the approach for the UK Biobank is a little unprecedented in terms of certain individuals are healthy and they're getting some information about their genome. But however, there are also some other individuals who are not so healthy but, like for example: some individuals have asthma and they're looking for genetic determinants for that particular type of manifestation. So, it's kind of a mixed bag and it's somewhat restricted to individuals who medically need one.

Bob Barrett: What are we expecting to learn from genomic medicine initiatives?

Joseph Alaimo: I think we're looking to learn how our particular genetic makeup in combination with the environment and our choices and lifestyles. How those two are married together and can influence a variety of outcomes in our lives. I think most of it could be down to the way we sleep and also to just our longevity, how long we we're able to live disease-free. I think these kinds of initiatives are starting to take a look at and what it is to be a human and how it is to live in your environment.

Bob Barrett: Finally, doctor, how will this data impact laboratory providers?

Joseph Alaimo: Yeah. This is one of the most interesting challenges I think we face and there's so many ways to answer this. I think when you look at the type of testing that is being offered in the clinical setting, so sick children, children who aren't developing properly, individuals who have multiple congenital anomalies were typically looking for monogenic or Mendelian diseases. So, one area that this is going to be very, very helpful is determining which individuals may harbor variants in these disease genes, and specifically, at what frequency in a population. And so if an individual carries such a rare variant and they're walking around healthy and normal in a disease gene that we would suspect to cause some type of

feature in a child. This is really powerful information to rule out that potentially this variant in this child is not necessarily something we should be concerned about. So that's just one example of how this data could be powerful.

Bob Barrett: That was Dr. Joseph Alaimo from the Department of Pathology at Children's Mercy Hospital and the University of Missouri, both in Kansas City. He's been our guest in this podcast on big data and utilization of the UK Biobank as a resource for clinical laboratories. He is a co-author of a perspective article on the UK Biobank that appears in the July 2021 issue of *Clinical Chemistry*. I am Bob Barrett, thanks for listening.