Precision Medicine Podcast, Season 4, Episode 55

Dr. Stephen Kingsmore on Scaling Whole Genome Sequencing for Uncovering Genetic Defects in Infants

March 2022

Karan Cushman, Producer:

Welcome to season four of the Precision Medicine Podcast, sponsored by Trapelo. This is the podcast where experts come to discuss the problems oncologists, reference labs, and payers face as precision medicine grows and consider solutions for advancing the quality of patient-centered cancer care. Be sure to subscribe at precisionmedicinepodcast.com to get the latest episodes delivered straight to your inbox.

Jerome Madison:

Welcome to the Precision Medicine Podcast. I'm Jerome Madison and before we get into our content I've got to thank our loyal listeners, our subscribers from all over the world for your feedback and engaging the content because today's guest and our topic were all recommendations from those who listen to the podcast on a routine basis. Today, we are privileged to have Dr. Stephen Kingsmore, president and CEO of the Rady Children's Institute for Genomic Medicine in San Diego, California. Dr. Kingsmore, thank you so much for being a guest on the Precision Medicine Podcast.

Dr. Stephen Kingsmore:

Thanks for inviting me, Jerome. I'm glad to be here. I'm looking forward to our chat this morning.

Jerome Madison:

Karan, so you know I do my homework on a guest, right?

Karan Cushman:

Oh, yes.

Jerome Madison:

And I don't think 24 hours is enough time to talk about some of the interesting things that I've learned about Dr. Kingsmore and his background, but Dr. Kingsmore being from Ireland, your first stop in the US was Duke University. And I saw an interview where you said that it was there at Duke where you learned to think, and I thought that was really profound. Tell us about your experience at Duke, which at the time that you came there, it was the pre-dawn of molecular genetics. And what eventually attracted you to the work you're doing today at Rady with critically ill newborns?

Dr. Stephen Kingsmore:

Yeah. It was a unique marvelous time. So I had wanted to come to the states for years. Unfortunately, the United States didn't want to let me in immediately. So I had to go through all that green card thing that you see movies about, which is kind of tough to be on the far side of, but finally the day came and I got my green card, and my wife and I we were newly weds. We'd gotten married in Columbia, South Carolina, the day after I got my green card. A few months later we arrived in the US and I got to see my first college basketball game at Duke and my first football game at Duke.

And this was a time that was kind of unique in the world because a lot was known about genetics and some was known about genomics, but honestly it wasn't based on studying DNA. It was because people had studied proteins and biochemical molecules to make sense of genetic diseases. And suddenly we started to have some new technologies and techniques that allowed us to start for the very first time to really study the human genome and it was fascinating. I was amazed. I had a medical undergraduate, I had been exposed to most genetics at the Weizmann Institute in Israel and so I was very into that, but suddenly now we could do an awful lot of experiments that just prior to that had not been possible. And it was a unique time when every week there was a startling discovery that changed our whole view of the world.

Jerome Madison:

Sorry. I had to mute. My phone was ringing and my computer was ringing at the same time. So Cameron Indoor Stadium. Coming from Ireland and watching football, what the rest of the world calls football, how was your first experience watching the Duke basketball game, coach Krzyzewski?

Dr. Stephen Kingsmore:

It took me a while to understand why people were so passionate about pounding a ball across a piece of wood. I really did. I got to tell you. We have professional sports in the UK, but we don't really have college sports and we certainly don't pay for them. They're more away of getting an exercise. So this whole thing to me was very new, but we had four final fours and three national championships in the six years I was there. So I certainly got quite an experience and quite a learning curve in terms of following basketball.

Jerome Madison:

That's awesome. So genetic disease, as you were mentioning, for childhood diseases, a genetic disease is a leading cause of infant death in the United States. And for infants that live severe genetic disease that goes undetected or is inappropriately treated can lead to irreversible disabilities. And to impact this in a positive way, one of the goals of your program at Rady is doing rapid genome sequencing for newborns and you mentioned at scale. Can you explain what that means?

Dr. Stephen Kingsmore:

Sure. Sure. So one of the big changes that's happened in the last 20 years is we've realized that many, many, many diseases that heather to we thought were complex and we're not really sure what the mechanism, we find out that they were genetically simple. That there's a single gene, a single place in the human genome and if there's a mistake in that genome code it causes disease and it's pretty black and white. And this has been revolutionary. We've had to rewrite textbooks

over and over and over again. There are now over 7000 known genetic diseases and each has its own hormone genome, its own gene, its own protein, its own mistakes and the code of life and by decoding the human genome, we're spelling out that alphabet. It's 3.2 billion DNA letters long. That's a lot. That's a book over 400 feet tall, just to put that in perspective. And that's basically the blueprint for being a human being and all the instructions for human life with all of its complexity.

Dr. Stephen Kingsmore:

And so these days we can do that in less than a day. We can decode and read through the entire genome and we can figure out what is causing a baby to have illness. This is just a staggering idea and yet now it's happening every day across north America and across the 90 odd countries that this podcast is broadcast into. And so scalable means instead to doing it for one or two, we want to do it for every kid who might have a genetic disease. That's what we mean by scalability. It's a difference between making a Ferrari and making a Ford Focus or something like that, where you want to make a million of them because everybody wants one. Everybody needs a genome if they've got so good.

Jerome Madison:

Yeah.

Karan Cushman:

For sure. I love that visualization of the 400 feet.

Jerome Madison:

So Dr. Kingsmore, Project Baby Bear was a monumental initiative that was funded by the state of California that makes a clear case for the routine practice of rapid whole genome sequencing. Particularly for in infants who are hospitalized in the ICU, can you share the details of that project and the outcomes that were recently published?

Dr. Stephen Kingsmore:

Sure. I'd be happy to. So this is an interesting project. So I am a scientist and we do research, but once you done enough research, you then move to something which is kind of a new idea, it's called implementation science. And it's where you move from research studies where you're discovering something for the first time to now saying, "Okay, can we do that in the real world?" It's one thing to say, I can decode genomes at scale and diagnose babies when mom and dad participate in a research study. It's a very different thing to say, "All right, we're going to let this loosen five California children's hospitals and see whether we can still get that same type of amazing change in outcome for the children."

Dr. Stephen Kingsmore:

So we approached the legislature of California, our politicians, and they gave us a grant of about $2 million and said, "Do it. We don't want you to do research. We want you to implement this." And so we partnered up with five children's hospitals across California from the north to the south. A couple of them were really urban and some were really rural. And your listeners might not be aware of that, but California is incredibly diverse and we have vast tracks of California that are fields where you grow produce for the entire country. So we were representing both places

like Los Angeles and then these regions that are very agricultural and equally our population, people probably know this, is also really diverse. We've got lots of Hispanic, Latino folk, lots of Northern European folk like myself and we actually have lots and lots of Asian folk who wind up on this west coast of the US. So we had a lot of diversity represented in the babies that we enrolled.

Dr. Stephen Kingsmore:

We wind up with 178 families participating in this study across the five hospitals. And basically what the parents allowed us to do was to decode the baby's genome. All of these were babies. So they were less than a year of age. All of them were hospitalized and all of them had a disease that the doctors weren't really sure what the trigger was. They were very diverse diseases. Everything from seizures to babies who weren't growing, to babies who had congenital anomalies or other organs that weren't working properly. And overall across the 178, 76 were shown to have a genetic disease causing their symptoms. That's 43%. That's almost one half that actually beat the research studies.

But more importantly than that were two things that the politicians were really interested in because having a diagnosis is important, but they really wanted to know, did it change the treatment for these babies and how much would it cost? So we looked at the proportion of babies whose care was changed as a result of the diagnosis. And we found that 31%, 55 babies had changes in their care because of the results of genome sequencing. Some of those stories were quite amazing in terms of the impact on their outcome. So that also recapitulated what we'd shown in research studies that this really did change how babies were managed.

And then lastly, we looked at cost of care and the concern was this could cost an awful lot of money and it may not be something that we can sustain. So much to our surprise, we found that we saved $2.2 million net of the cost of doing the sequencing and the counseling and the care. So not only does this change outcomes, not only does it save babies lives, but it actually saves cost. And the reason for that is babies don't stay as long in hospital. That we get a prompt answer to what's causing their disease. We put them on effective therapy and they get to go home much quicker and that's the major driver of cost is how long they languish in hospital before the doctors figure it out.

Karan Cushman:

Amazing.

Jerome Madison:

Tremendous impact. I mean, you said it, for healthcare systems, for payers to take note there has to be that bottom line benefit. And you mentioned the cost of the program paid for itself with an estimated $2.5 million in healthcare savings above the state's investment.

Dr. Stephen Kingsmore:

That's exactly right. And that puts it into a very rare class of modern innovations in healthcare that more than pay for themselves.

Jerome Madison:

The Precision Medicine Podcast will continue right after this.

Karan Cushman:

With the explosion of new data and biomarkers and cancer today, how can healthcare professionals keep pace to know which genes will best inform treatment decisions? Trapelo knows. Trapelo is the first single technology platform used by oncologists, labs and payers to resolve the complexity of precision medicine in real time. Trapelo knows which patients to test and when, it knows which tests are most appropriate, which labs are preferred, and which treatments are most likely to be reimbursed. Visit trapelohealth.com to learn how you can give cancer patients the most appropriate evidence-based treatment options when time matters most.

Jerome Madison:

So Karan, another Dr. Kingsmore tidbit here, he's our first official Guinness World Record holder to be on the podcast.

Karan Cushman:

Really?

Jerome Madison:

Yeah. So Dr. Kingsmore you were, I guess first officially recognized in 2016 for the fastest genetic diagnosis accomplished by successfully diagnosing a critically ill newborn in 26 hours, but since then, you've already broken your own record twice. You broke it again a couple of years later in doing rapid genome sequencing and diagnosing a critically ill newborn in 19 and a half hours. But those records absolutely pale in comparison to the work that you and your team did for newborn baby and his parents, which was published in a case study in the new England Journal of Medicine just last year. And you diagnosed his genetic disease in less than 13 hours. Can you tell us about this case and how, I guess ultra, ultra rapid whole genome sequencing helped this family?

Dr. Stephen Kingsmore:

Yeah. Yeah. I'm actually in my office this morning and I'm sitting looking at those two Guinness World Records and they really are kind of a cool thing. And the reason we did that was we wanted something that would help people understand this in the public. Decoding a genome is this kind of mystical fog, obscured concept, but the idea of doing it faster than anybody else is something that people can get their arms around. And the reason is many of these babies are critically ill. They can't wait around. Their organs are failing and if we don't get answers fast, we may be too late. We might not be able to save their organ function. And so the baby that we tested about a year ago using these technologies is a perfect example.

So it was a Sunday night, it was almost midnight and his mom brought him to the emergency department here at Rady Children's Hospital in San Diego. And the complaint was very simple, she couldn't console her baby. He was very irritable and couldn't be consoled. The doctor examined the baby and found that indeed he was extremely irritable and he had mild signs when was examined that suggested he had brain irritation. So he had a scan of his brain done in the emergency department at nearly midnight and it showed really gross abnormalities that looked like birth asphyxia. So babies sometimes when they're born, they lack oxygen. I think we all know this and that's what his brain scan looked like. It had white patches that suggested this. The big name for that is hypoxic ischemic encephalopathy. So the baby was admitted. He was 41 days old at this time. He was admitted to the intensive care unit for monitoring and treatment.

Dr. Stephen Kingsmore:

The next morning, his mom and dad were there when doctors were rounding and they got the full picture. And it turned out that he had had an older sister, nine years earlier who'd presented at exactly the same age, 42 days of age, with exactly the same symptoms and exactly the same findings on her brain scan. Now the cause of her illness was never discovered and in fact, she was hospitalized almost her entire life until she died just shy of a year of age. We also found out that mom and dad were actually related and you may or may not know this, but that makes us think about certain genetic diseases that are more common if mom and dad are related.

So a genome was ordered that afternoon and by about six o'clock, we had the blood sample in our genome center and some of our staff stayed overtime. And we had been prepping for about a year to set another world record. And we looked at one another and we really felt this was the right baby to be the first in the world to get this. Because of the picture in his sister, we knew that he was likely to go downhill within just a couple of days and that we only had a couple of days to get it right to know what was the cause of his illness. So amazingly enough, we had an answer, as you said in about 13 hours. And we found that what he had was a genetic disease, it was a recessive genetic disease. So he had inherited it both from his mom and his dad. We were correct about that, but the amazing thing was that this fatal disease he had was treatable with two vitamin supplements, biotin and thymine.

And so at 8:30 in the morning, so he'd been admitted close to midnight, so this is a day later, 8:30 in the morning, we were able to order those two vitamin supplements. He got them around lunchtime and by 6:00 PM, he was back to baseline. He was alert. He was calm and he was bottle feeding. So he's now a year old and he's doing well. He's never had a seizure again. In the interim he had started to seize and his brain had shown increasing signs of encephalopathy, but we were able to reverse those. And it's our hope that he will have a pretty normal lifespan. And so you just think about those two different children, same family, same disease, one a decade ago, one now, and it's black and white. It's nice [inaudible 00:20:33]. It's the difference about genomes versus no genomes in terms of changing outcomes.

Karan Cushman:

I think I just found your hobby, which is you need to give yourself also some credit for being an Olympic athlete and an incredible marathoner, not just a beach runner.

Dr. Stephen Kingsmore:

Well, thank you. I'll take it.

Karan Cushman:

That was amazing.

Jerome Madison:

I'll take a moment for a proud dad moment, Dr. Kingsmore and Karan. I was so fascinated by this case. I was talking about it with my family and my 13-year-old daughter, Marlo aspires to be a physician. She was asking so many questions and she asked to read the link and I sent it to her and she read it. And you talk about sticking your chest out. I didn't do nothing, but just the fact of just reading this and her getting interested in it, it is just incredible. For our listeners out there, Rady Children's Institute for Genomic Medicine, very much a leader in whole genome rapid sequencing for newborns. Can you tell our listeners, you also do this for a number of other

hospitals around the country and there are other states that have started similar programs to Baby Bear. If there are people out there listening physicians, family members who want more information, can you point them in the right direction?

Dr. Stephen Kingsmore:

Yeah, absolutely. So first of all, no hero worship, okay? The only way you can ever do this is to have a really dedicated team. It takes about 50 people to do this in the background. So I get to talk on podcasts, but they're busy actually running genomes now, right?

Jerome Madison:

Yeah.

Dr. Stephen Kingsmore:

They're the heroes, not me, but yes, this has gone from being something that was really rare and only done at a few places to now being broadly available across North America and in fact, around the world it's increasingly becoming available. So in the US, we have 80 children's hospitals who send us samples every day of the week from children in their intensive care units and other countries like Australia and England and Wales, they also now have national programs to provide this to children in hospitals and intensive care units.

Basically it's any baby under a year of age who has an illness that has the doctor stumped. And if that's the case, the recommendation now, the guidance, is to get a rapid genome sequence to rule in or rule out a genetic disease as the cause of their condition. And we're all finding pretty much the same thing, which is one in three of the babies we test has indeed a genetic condition that's causing their symptoms. And many of your listeners will know about this thing called the diagnostic odyssey, which is what used to happen, where it could even take decades for doctors to understand what was the cause of a baby's illness or a child's illness. Sometimes the children were fully grown adults by the time they actually were able to put a label on their disease. So just imagine that now we can do it in a day.

So one of the things that is most exciting to me is that this is starting to become what we call standard of care. That means that it's part and parcel of mainstream frontline medicine. And the very first state in the US to do this is Michigan. We had a project there called Project Baby Deer, deer being the kind of state animal of Michigan. Here in California our project that I talked about was Project Baby Bear, for our state animal. So Project Baby Deer was very similar. And as a result of it, it's now the standard of care in Michigan for babies to receive this testing and it's paid for by the state.

It's part of the state Medicaid program so that a baby who needs this will get it in hospital in Michigan. They are the first to do that. Hats off to them. They beat California and it's super exciting to watch this start to ripple out across the states and to realize that in five or 10 years, we just won't have those stories anymore. These tragic stories of moms and dads who couldn't get an answer for their child's disease.

Jerome Madison:

Yeah. You just mentioned bringing the diagnostic odyssey to an end, but I've heard you mentioned, and I'm hoping I'm not misquoting you that 90% of the infant genome sequenced and diagnosed do not move to an effective treatment. Can you talk about why that is and what can be done? What's the opportunity for those patients in now the treatment journey?

Dr. Stephen Kingsmore:

Yeah. It's a great, great point. So we live by our successes, by these amazing stories where it's a simple as 5 cents of over the counter diet supplements that save a baby's life. Those are amazing, but tragically today, they are in the minority. You're right, it's probably closer to 95% of the genetic diseases don't yet have effective therapies. And you'd say to yourself, why is that? Well, part of the reason is many of these diseases were only discovered in the last five or 10 years. So there hasn't been time to make effective treatments. Second of all, doctors couldn't pick them up, right? So we have just invented the first way as a community of actually getting an answer for a child who has one of these illnesses.

Unfortunately, up until now, we didn't know that they had the illness early enough to be able to try a therapy on them. They already were likely in organ failure. So we are now moving into a new area, which is ending the therapeutic Odyssey. How do we make sure that every baby that we diagnose gets treatment of some type, whether it's an experimental treatment or a proven treatment and that's the next phase of what we are doing. As diagnosis becomes standard, we now need to move to trying out new treatments for these babies who don't have effective treatments available today.

And we are just starting this, but we are entering a very exciting era now where we can actually make gene therapies for babies with genetic diseases. This has only just become possible in the last five or 10 years and the bottleneck, as I said was, we couldn't find the patients. Now we can find the patients. We're going to be able to innovate at an unparalleled rate to start to prove out which of these gene therapies are effective and when best to give them.

Jerome Madison:

So you make an incredible point talking about individualized gene therapy. Karan, I'm thinking back to Dr. Edward Abraham, who you mentioned, CEO of the personalized medics and coalition. We had him on the podcast and this had to be like three years ago. And he talked about, he talked about the concerns that many payers and others have about the cost of precision medicine. And one of the examples that he gave, and I believe he wrote a news editorial, there is a drug that was developed close to three years ago that was for spinal muscular atrophy. The cost of that drug was $2 million, but the opportunity cost there is for those patients who do not receive this $2 million therapy for this genetic disease. The downstream cost of treating that patient is upwards of $5 million. Is that a barrier, is cost a barrier? Does it require a paradigm shift, Dr. Kingsmore and what are some other barriers that need to be addressed in order to scale the use of rapid whole genome sequencing?

Dr. Stephen Kingsmore:

There's no doubt about it that the cost is an issue and it's not just a matter of insurance companies needing to be profitable. For the government health system, it has to stay solvent. We can't have it go bankrupt and so we really do need to start to think about cost effectiveness. Spinal muscular atrophy, that you mentioned, is a great case and point, if you have spinal muscular atrophy type one, tragically, you'll die by age two and you won't have much of a life quality before that. With this new you therapy, if it's given immediately, right? It has to be given immediately at birth, but if you do, do that, then you can anticipate that your baby will have life, will learn to walk.

You're right, it's only been available for a few years, but the hope is that once we get this perfected, that those baby will grow up to be adults and have very functional lives. So this is just a

huge learning exercise where we want to be good stewards of money. We have great need, we have marvelous new medicines, and so as a society we need to wrestle with this and figure out how to get it right and how to provide healthcare that will be sustainable and also give the best outcomes for these precious babies.

Karan Cushman:

It's exciting that concept, that we're at a place to be good stewards of science. It's happening so fast and now it's up to us to really bring it forward and bring it closer to patients, right?

Dr. Stephen Kingsmore:

Yeah, absolutely. Absolutely.

Jerome Madison:

I understand you at Rady have a Frontiers In Pediatric Genomic Medicine Conference, annual conference that's upcoming, I believe in April of this year. Can you tell us more about that?

Dr. Stephen Kingsmore:

Yes. And now you're absolutely killing me because we just canceled it this morning.

Jerome Madison:

Oh, no.

Dr. Stephen Kingsmore:

I'm going to cry.

Karan Cushman:

Well, we could do it now.

Dr. Stephen Kingsmore:

Yeah. Yeah.

Karan Cushman:

Well, [inaudible 00:32:08].

Dr. Stephen Kingsmore:

Now, Omicron hit. We looked at each other yesterday and we just said, "We're going to put it off. We're going to postpone it."

Jerome Madison:

Well, more reason for people, if you're listening, share this podcast, share it on social media, share it on your LinkedIn, retweet it however you can. One more topic before you get out of here Dr. Kingsmore. We were just talking about kind of the paradigm shift. Currently only a few select private insurances, you talked about the Baby Deer Project in Michigan that has made this

technology standard of care, but very few insurance companies cover this type of testing. So children's hospitals have to rely on grants and philanthropy to afford to underwrite this type of technology.

Jerome Madison:

But in California Assembly member Brian Maienschein has partnered with Rady Children's Hospital to introduce assembly bill 114, the Rare Disease Sequencing for Critically Ill Infants Act. And I believe this is to expand access so this crucial testing by qualifying rapid whole genome sequencing as a Medi-Cal-covered benefit for babies hospitalized in intensive care. And on the federal level, a bipartisan effort was introduced by Senator Susan Collins from the great state of-

Karan Cushman:

Maine. Hello, Susan.

Jerome Madison:

She introduced the Ending the Diagnostic Odyssey Act, which would give states the option of providing federal matching funds for whole genome sequencing for Medicaid eligible children suspected of having a genetic disease. Dr. Kingsmore, how critical is political support in allies and what can our listeners do to help create positive momentum where they are?

Dr. Stephen Kingsmore:

Yeah. So politicians can break log jams. They really can. Sometimes they create log jams and we all know about that, but they can break them as well and those are two great examples. So Brian Maienschein did this. It's now law in California and that means that the Department of Health is writing a policy. And hopefully Senator Collins will be able to do the same at the federal level, because that makes a huge difference. It greatly accelerates what we are able to do otherwise, as you all know, it's a piecemeal exercise where each state, each region, each policy, each plan differs, each hospital, and it can take decades to have something which is life's saving get to all the children who need it.

So what can you do? You can exercise your rights as citizens in a democracy to encourage these folks like Senator Collins, like Brian Maienschein to encourage them and get behind them and support what they're doing because ultimately those politicians are answerable to you and I. And if parents unite, if doctors unite, nurses unite, we will see these things happen much, much faster than if we just let it to happen organically.

Jerome Madison:

Incredible work. Not only from the leadership there at the Rady Children's Hospital, but all of your team. I just commend you for the work that you're doing along with those other hospitals who are participating in your research efforts, testing infants with whole genome sequencing, just an incredible work, dr. Stephen Kingsmore, president and CEO of the Rady Children's Institute for Genomic Medicine. Thank you for being a guest on the Precision Medicine Podcast.

Dr. Stephen Kingsmore:

Thank you and let's just go out with a hand clap to my 90 or so teammate who are actually decoding genomes right now while I just talk about it.

Jerome Madison:

Yes.

Karan Cushman:

Yes. That's [inaudible 00:36:23].

Jerome Madison:

Yes, absolutely. Hey, real quick, Dr. Kingsmore, now San Diego is the considered one of the genome capitals of the United States. It's also considered one of the micro brewery capitals of the United States and I heard you like to partake in a good IPA every now and again.

Dr. Stephen Kingsmore:

I sure do. That's one of the joys of living in San Diego. One is that we've forgotten what winter means and I love that because I grew up in Ireland and second of all, we have beer to celebrate it with every day.

Jerome Madison:

There's also great tequila there, Karan and [inaudible 00:36:58].

Karan Cushman:

Oh, I'm ready.

Jerome Madison:

Yeah. I'm not much a beer fan, but tequila it's less filling and it tastes great. So, I'm all down for that.

Karan Cushman:

That's awesome as I look out at the snow right now. Hey, and I just want to add too, we're going to have to get Marlo and Ella together because just last night, my daughter's 15 and she came to me with this kind of crisis moment where she's really thinking hard about what she wants to do, what she wants to study at college and her top choice right now is in OB-GYN. She really wants to work with infants. So maybe there's something in the science field for her as well. So, thank you, Dr. Kingsmore.

Dr. Stephen Kingsmore:

My pleasure. All the best.

Karan Cushman, Producer:

You've been listening to the Precision Medicine Podcast sponsored by Trapelo. Trapelo is the first clinical decision support tool to align the interests of oncologist, labs and payers to give patients the best chance at beating cancer. To learn more, visit gettrapelo.com. To subscribe to the podcast or download transcripts of any episode. Visit precisionmedicinepodcast.com. We invite you to join the conversation on social media. You can find us on Twitter at PMP, by Trapelo

and on LinkedIn at the Trapelo company page. If you know someone who would enjoy the Precision Medicine Podcast, please share it. They'll thank you and so will we. We hope you'll tune in for the next episode.

About Our Guest

Stephen Kingsmore, MD, DSc

President and CEO of Rady Children's Institute for Genomic Medicine

Stephen F. Kingsmore, MD, DSc, is President and CEO of Rady Children's Institute for Genomic Medicine (RCIGM) where he leads a multi-disciplinary team of scientists, physicians, genetic counselors, software engineers and bioinformaticians who are pioneering the use of rapid Whole Genome Sequencing (rWGS®) to enable precise diagnoses for critically ill newborns. In 2021, he led the RCIGM team to set a new record of 13.5 hours for achieving the fastest molecular diagnosis using rWGS, breaking his previous 2018 world record of 19.5 hours.

Dr. Kingsmore came to Rady Children's in 2015 from Children's Mercy Kansas City, where he was the Director for the Center for Pediatric Genomic Medicine and Executive Director of Medical Panomic, as well as the Dee Lyons/Missouri Endowed Chair in Genomic Medicine at the University of Missouri-Kansas City School of Medicine. He previously served as President and CEO of the National Center for Genome Resources; Chief Operating Officer of Molecular Staging Inc.; Vice President of Research at CuraGen Corporation; Founder of GatorGen; and Assistant Professor at the University of Florida's School of Medicine.

Dr. Kingsmore is board-certified in Internal Medicine and is a Fellow of the Royal College of Pathologists. He received his MB, ChB, BAO and DSc degrees from the Queen's University Belfast in Ireland. He trained in clinical immunology in Northern Ireland and did his residency in Internal Medicine and fellowship at Duke University Medical Center in North Carolina. In 2012, Dr. Kingsmore was named MedScape Physician of the Year and TIME magazine ranked his first record-breaking time of 50 hours for genome sequencing among the top 10 medical breakthroughs for that year. In 2013, he received the Scripps Genomic Medicine Award and the ILCHUN Prize of the Korean Society for Biochemistry and Molecular Biology.