

INTRODUCTION		
0 :04	Eleonor Angeles	Welcome to Talking Healthcare: Insights from global leaders produced by the International Hospital Federation. The Talking Healthcare Podcast series features healthcare leaders and executives from the IHF's global community sharing their expertise, knowledge and insights into cutting edge topics in healthcare. In this episode, members of the Global Rare Paediatric Disease Network discuss the challenges and opportunities of using A.I. in the management of rare disease.
0:38	Barbara Walczyk-Joers	<p>Welcome. I am Barbara Walczyk-Joers and I have the distinct pleasure to serve as the President and chief executive officer at Gillette Children's Specialty Health Care. Gillette Children's is over 125 years old in the making and is the United States first public hospital dedicated to children who have physical disability. Today, we are the preeminent independent institution to detect, diagnose, deliver and influence the standard of care for children with muscular, skeletal and neurological conditions that are medically complex and or rare.</p> <p>One of five of our active patients' lives with a diagnosed rare disease. Today, I have the honour as founding member of the Global Rare Paediatric Disease Network to speak with two remarkable IHF members from around the globe about today's rare disease topics of interest. So first, I will share a little background and how we got to today. During the 2022 IHF meeting in Barcelona, we connected with various leaders on the importance of paediatric networking, especially for rare diseases where children face inequities in detection, diagnosis, and intervention for many reasons, regardless of their geography.</p>
1:51	Barbara Walczyk-Joers	<p>Soon after, the IHF leadership produced a concept paper to develop and maintain a network of members providing care and or research for paediatric rare disease for us to share ideas and learn from each other. But the initial focus on administrative and leadership topics. We aspire not to duplicate other existing networks, which often share clinical information, but instead our focus is on international collaboration, not in existence today that will ultimately result in better care for patients with rare disease.</p> <p>In 2023, we conducted a survey of membership hospitals and then convened a discussion group at the 2023 Lisbon meeting to overview findings and generate next steps. Currently, we are planning our topics for Rio this coming September and some of today's content will be in that work. So please stay connected over the coming months to learn more and to engage with us.</p> <p>Now I ask our incredibly talented guests to share a little bit about themselves.</p>
2 :56	Heini Kanervo	Hi, everyone. I am Heini Kanervo, I am the rare diseases coordinator at University Hospital Brussels, one of the seven academic hospitals in Belgium. And I am also a member of the Steering Committee of the Global Rare Paediatric Disease Network. As a rare diseases coordinator, I oversee all activities in our hospital related to rare diseases, and I help coordinate care for children and adults living with the rare disease or who are in search of a diagnosis.

3:25	Ee Shien Tan	Hi everyone. It's really my pleasure to be on this podcast. My name is Ee Shien Tan. I'm the head of Clinical Genetics in Care Women's and Children's Hospital in Singapore. And this area of rare disease is really close to my heart because I care for patients with rare disease. I'm grateful for this chance to learn from the experiences from everyone in this network. Thank you.
3 :47	Barbara Walczyk-Joers	So welcome to both of you, Heini and Ee Shien Tan. And thank you for spending your time with us today. We know all too well that children with rare disease or suspected rare diseases and their families face numerous challenges, including late diagnosis and misdiagnosis and proper, and no response to therapies and a lack of accurate diagnostic and monitoring tools in so many unfortunate scenarios. The list is many to solve. Families often face what we define as a diagnostic odyssey, but so many obstacles along the way. Those obstacles can be financial, emotional, social, and other. rare diseases affect millions of children worldwide and create a substantial economic burden across the health care continuum, as well as poor patient outcomes. As children enter into adulthood.
4 :37	Barbara Walczyk-Joers	Earlier, more efficient diagnosis, improve treatment and monitoring and better management of the patient journey are vital. So we ask, is artificial intelligence A.I. a potential solution for the challenges we are facing together? Today we will discuss some of the opportunities AI offers for paediatric disease management. Also looking at the risks and challenges that we face. So to that start Ee Shien Tan can you give us an idea of how A.I. is currently being used to support paediatric rare disease management in Singapore at your hospital?
5 :14	Ee Shien Tan	Thanks, Barbara. So one of the key challenges in rare disease is that the diagnostic odyssey, I think due to the rarity of conditions and lack of awareness of these conditions, patients often have a long journey to obtaining a diagnosis. So some of our efforts have been to see if there are ways to show up in this journey one such effort in the research space, in both using a 3D facial analysis software to analyze facial features, enabling a diagnosis of a rare condition known as heredity and edema. But you can imagine that there's potential of its use in other genetic conditions who have characteristic facial features. And I look forward to more applications of this software and as another effort in the research space as well, again, the ability to identify cases through mining or electronic health. So looking through massive amounts of data in our electronic health records to see if we can pick up patients who fit the phenotype of a rare disease. So there shouldn't be so many potential uses. And these are some of our examples.
6 :20	Barbara Walczyk-Joers	That's incredible. And the way that you're leading for the rest of us to learn gives us a lot to look forward to. Heini, a little bit about your work over in Belgium.
6 :30	Heini Kanervo	In our Children's Hospital, an example of AI software that we use is bone expert. It's an application designed to analyze bone health using advanced imaging technologies and A.I. and we use it in our children with rare hereditary bone disorders. Also, our hospital in

		<p>general just started a project in which natural language processing or NLP is used to identify possible persons with a rare disease by screening clinical notes in the electronic health record.</p> <p>And by identifying these patients, we can register them after validation, collect relevant health data for future research and measure quality of care. For example, by researching the time from first contact with our hospital to the time of diagnosis. This project is still ongoing, so maybe in a year or so I will be able to share some results with you regarding its reliability and efficacy and NLP piece also used in measuring patient satisfaction.</p> <p>So patients are asked to fill in a questionnaire after hospitalization or after ambulatory consultation, and NLP is used on the unstructured notes of the patients to interpret their experience.</p>
7 :48	Barbara Walczyk-Joers	<p>That's wonderful. Thank you. And for those of us in healthcare, when we look at our data, unfortunately we know that even with the electronic health records, almost 80% of all our clinical data is unstructured. So the examples you both gave really begin to open up what's possible. If we can structure unstructured and unstructured structured data to better serve our patients with artificial intelligence. So, Heini, when you continue your exploration of what you and your colleagues are doing, what impacts have you seen in terms of patient care, workload for staff or financial cost and burden? What are you seeing generating out of this work?</p>
8 :31	Heini Kanervo	<p>Well, in the first example, I gave the bone expert, we all know that a software applications can enhance diagnostic capabilities and overall clinical workflow. So radiologists save time by using bone expert because they are able to identify issues more quickly than they would doing everything themselves. So they only need to validate the results and they have more time for other tasks.</p> <p>And then in the NLP driven applications we use can ultimately improve patient care because we are and will hopefully be able to measure quality of care effectively and we can facilitate research which relevant data that is automatically then being extracted from the electronic health records, which also saves us a lot of time instead of having to manually input all the relevant data.</p>
9 :20	Barbara Walczyk-Joers	<p>So wonderful science like we might get proactive versus retroactive, which we're known for in health care. Ee Shien Tan anything to add on the impacts in terms of patient care, staff, workload or workflow?</p>
9:35	Ee Shien Tan	<p>Well, I spend a moment most of our AI endeavour assigned the research space, but I think potentially if it can reduce the diagnostic odyssey, I mean, we know the impact on patients would be tremendous.</p> <p>Not only does it allow for better patient care and reduce the psychological and financial burden on the patients, as well as a burden on the health care system. And I think for myself, I'm thinking that I could explain it to the physician because it's virtually impossible for a physician to memorize all the information about these 7000 rare diseases. And I can think that AI can extract they use this information in a meaningful way to support us. It would be really beneficial and it would reduce our work.</p>
10:20	Barbara Walczyk-Joers	<p>The speed to value, I think is the theme that we're hearing, which is fantastic when we think what is it? And you can correct me 70% or</p>

		<p>more of the disease began in childhood. So that speed to value to what you've both indicated in giving some more years to childhood and quality of life would be fantastic. So, we've heard some of the opportunities provided through artificial intelligence and as leaders in the rare disease space. What concerns may you have about how patients with rare disease or their families or their care team might be negatively impacted by the use of AI? And this can be from the paediatric population or just in general for rare disease perspective. We all know AI is moving fast. A lot of AI is not even AI. it's just what we've had over the past decades. But it's the new phase. differing governments and governments are still developing policy today. If you had to say to patients and families what to be mindful of, what to watch for with artificial intelligence, what might that be?</p>
11:29	Heini Kanervo	<p>Well, it's like with the Internet, you know, it's great. And it has made our lives a lot easier. But it is important to be mindful of the risks. For example, what I've noticed is that people tend to over rely on AI. They assume that AI is infallible. While they should be aware of the fact that AI is based on large amounts of available data and what is one of the biggest challenges and rare diseases, it's the lack and unavailability of real world data.</p> <p>So we can assume that AI is, especially in the field of rare diseases, does not have all the answers. And patients should understand that AI is a tool to assist health care professionals, not replace them, and that validation is always needed. You know, final decisions should involve human oversight, and overreliance on AI can also lead to a reduction in the human aspect of care, which is vital.</p> <p>You know, we need empathy and we need patient health care provider relationships for personalized treatment. It's important to maintain a balance between AI assistance and human interaction.</p>
12:37	Barbara Walczyk-Joers	<p>So there's a few debates happening and I think regardless of where we live on that human in the loop or the human in control of the loop, and where does that place in that leads to the governance. Ee Shien Tan, what are some of the advice that you would give to those regarding artificial intelligence?</p>
12:56	Ee Shien Tan	<p>I think in the first place there should be transparency and such that patients are aware that they are interacting with AI or if AI is used in the delivery of care, they should know that.</p> <p>And also, I think we all have to be aware of the potential unintended bias. I think seniors mention it depends on the training data set and machine learning models are entirely dependent on the integrity of the training datasets. And so we need to ensure that these datasets are representative to reduce unintended bias in these populations as well. So I think that's a lot of excitement about AI, but that also needs to be some caution.</p>
13:40	Barbara Walczyk-Joers	<p>So went about it as leaders, clinically and research and Ee Shien Tan some of the work that you're doing with your colleagues in Singapore. Some concerns are head's up going into it for everyone in the rare disease community who has data, any advice or insights?</p>
14:00	Ee Shien Tan	<p>No. I mean, I think data protection is necessary. I mean, we do have to emphasize that. sometimes where we are building AI sets, we think that the more data, the better, which is in a way true. But we also need to ensure that data protection is there to safeguard</p>

		<p>personal data from misuse and to ensure patients trust in how the data is managed. I think that's the trust that should not be broken and we do need to consider this even as we train our AI models. So that's something to think about. But I think the other part of it is I do realize as a clinician how little I know about AI and everything that I had to do to try to understand. And I think it's important to have a clinician closely collaborating with AI, scientist or developer. And it's really a collaboration because that in itself will ensure hopefully that the outcome will impact patients and that mission is there in the problem that we want to solve is clear. So I think that's the thing. Probably we do need to upskill some of our physicians clinicians to understand the language at the same time, work closely together the scientists.</p>
15:29	Barbara Walczyk-Joers	<p>And that's wonderful. And I like to say on this podcast that the world of startups and software companies, data set monetization and medical devices that are looking at that next phase of how to use data for their work, that this group is strongly advising about ethical behaviour and quality behaviour because once you enter the loop of artificial intelligence, we all have responsibility and that can be legal in non-legal accountabilities. But I think that one thing as a group we'll be exploring more is how do we set some guide rails and some stretch areas for us as a group across the nation to see what really is a good practice and best practice, especially when it comes to our children and rare diseases and how to use their data for good and to make sure that the data coming forth is to help others on the same journey. So thank you for that.</p>
16:22	Ee Shien Tan	<p>Just maybe too, I was just reading a paper that the Ministry of Health of Singapore came up with a guidance paper of about artificial intelligence in health care and across some of the good practices and the principles that we need to adhere to even as we use AI it in health care. I thought that was something that I would share the group as well. It's quite a nice paper.</p>
16:48	Barbara Walczyk-Joers	<p>Fantastic. I know in the United States, in Washington, D.C., different members of Congress and the administration have a vision of what they would like, but what the actual structure in oversight will be is still being defined. With this, Heini and Ee Shien Tan, it has been a long journey for her diseases and with artificial intelligence coming and emerging so quickly, it gives us hope and it obviously gives us some concern and pause as well. What can you give to hospital leaders in terms of priorities? If you could give three priorities that you would like to see brought in to mitigate these risks and challenges, anything that you would be able to share, we would greatly enjoy and then bring those appropriate priorities into our discussion and rail this coming September. Ee Shien Tan?</p>
17:40	Ee Shien Tan	<p>I think I do hope that hospital leaders will exercise clinical governance and oversight over the adoption and implementation of the AI to ensure that is responsibly used and safely implemented. And I think that's really the role that hospitals leaders could play in this.</p>
18:00	Heini Kanervo	<p>Yeah, I think so too. I think it's important to develop and enforce ethical guidelines for the use of AI in health care, emphasizing patient rights, informed consent, ethical considerations, and also to</p>

		educate staff on the use of AI and best practices for data security and the importance of protecting patient information. And it's also important to clearly explain to patients and staff how AI systems work, including their benefits and limitations and how decisions are made. Patients should understand what I can and cannot do to manage their expectations and reduce misunderstandings about the capabilities of AI in their care. I think that those are two things that I would put forward as priorities for our hospital leaders.
18:50	Barbara Walczyk-Joers	That's fantastic. Thank you, both of you. And I think from a hospital management and administration perspective, when we think of the impact of AI, as you outlined in the health care journey, what patient rights are, but the changes in informed consent and the ethical considerations and the need is. Heini highlighted that staff truly need to understand this. I think in health care, sometimes that checkbox mentality comes up of saying, we just read it. But the care team's given the privilege of being part of somebody's health care journey really need to understand the risk-benefit of using AI in the care journey and what they are accountable for. And how can we set them up as leaders to have the best structure so that the engagement with the patient and their caregiving team and their family is truly transparent and understandable to all in the process. So thank you for that. Before I close out any last minute thought, either of you would like to add on? I know sometimes those are the best soundbites - if you were going to your government today, what would be the one thing that you would ask for funding for artificial intelligence to help your work? Heini you always have a long list.
20:15	Heini	I know I usually sleep a lot more than this.
20:18	Barbara Walczyk-Joers	We're going to be soul catchers today. What if we all had that magic?
20:24	Heini	Regarding AI to our governments, I think it would be like set up regulations like regulate the use of AI just to make sure that it is used correctly and that all users know how to use it.
20:46	Barbara Walczyk-Joers	Ee Shien Tan?
20:48	Ee Shien Tan	Manpower. I think the relevant expertise, and sometimes we tend to take it all upon ourselves, even in the hospital system to do many and all things. But we really do need expertise and in data science. AI. That's really come alongside us on this journey. So I think manpower would be one big thing that I see. And, and my wish is also that our data would be clean and nice and structured.
21:25	Barbara Walczyk-Joers	Yeah. The dream of structured data 30 years in the making, and of course here where I sit today to not weaponize the data and not make it political and understand that we're at the heart of it all humans. And we have these challenges called rare disease and that we intend and we govern to do good and not to allow others to use it for themselves to create chaos, but really to use in an ethical manner to advance care. Because if we can't come together on A.I. for our children, I don't think any government in the world has a chance. And so I think we need to continue to ask our colleagues around the globe, let's do this the right way. We've done plenty of

		lessons learned in the past with data, but maybe this is the time that we can do the right thing.
22:13	Heini	I also would like to, I'm very curious actually, because we are representatives for United States, Belgium in Europe, but then also Singapore. But what about inequity in AI? We have access to the more innovative AI software and applications. But what about other countries that maybe or other parts of the world that maybe do not have access to them? So it would actually be interesting to hear from people from other parts of the world what their experiences are with AI and what challenges they face. Maybe it's something for another podcast or for IHF, for the world Hospital Congress.
23:10	Barbara Walczyk-Joers	<p>It's fantastic when we think about the coalition that we've put together with the Rare Disease Network, asking colleagues from different countries to share their experience with artificial intelligence so that we can come together and get in front of it as best we can, I think is something we'll all be taking forward from today and to continue helping families across the world. Be mindful, be excited, but also be mindful because we don't want to make that diagnostic odyssey any more challenging than what we're already facing. So, with that, thank you very much for joining us today. And thank you, Heini, and Ee Shien Tan for sharing very practical and intelligent insights to help guide those who can influence the artificial intelligence, impact on rare disease.</p> <p>I hope this podcast motivates our listeners to find out more and also to engage with the global rare Paediatric Disease Network. Please visit us on our website of www.ihf-fih.org for more information. Thank you for tuning in.</p>