Episode: *Author Interview: “What Matters Ethically About How the UDN Has Changed Since Its Inception”*

Guest: David A. Pearce, PhD
Host: Tim Hoff
Transcript: Cheryl Green

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[bright theme music]

[00:00:03] TIM HOFF: Welcome to another episode of the Author Interview series from the *American Medical Association Journal of Ethics*. I’m your host, Tim Hoff. This series provides an alternative way to access the interesting and important work being done by Journal contributors each month. Joining me on this episode is Dr David A. Pearce, a professor of pediatrics at the University of South Dakota Sanford School of Medicine in Vermillion and the chair of the International Rare Disease Research Consortium. He’s here to discuss his article, coauthored with Dr Elena-Alexandra Tataru, “*What Matters Ethically About How the UDN Has Changed Since Its Inception*,” in the October 2025 issue of the Journal, *Diagnostic Research*. Dr Pearce, thank you so much for being here. [music fades]

DR DAVID PEARCE: Thank you for having me, Tim.

[00:00:51] HOFF: So, what is the main ethics point that you’re making in this article?

PEARCE: I think the main point is, is that these Undiagnosed Disease Networks have really been going for about ten years or more right now. And as you can imagine, the technology has changed considerably with respect to the fact that we can access so much more genetic information in this diagnostic journey that we’re in, that we sort of go on. And what I mean by that is, is that we do genome sequencing. So, we sequence three billion letters, and we really only know about what 5 percent of what our genomes do. So, the message here is, is that we just have to be careful with the overwhelming amount of information that we have, because we may or may not actually reach a diagnosis with that.

[00:01:41] HOFF: And what should health professions students and trainees in particular be taking from this piece?

PEARCE: Yeah, that’s a great question. I think that they should be aware that the technology is changing rapidly as we speak. So, and more importantly, the actual information you can get in terms of if you want to do a genetic diagnosis, if you see something that’s a change or a mutation, or it could be indicative of potential disease, can you actually communicate that information effectively to patients, knowing is it truly pathogenic? And it may actually be in a piece of the genome that we don’t understand. Remember, I just said we don’t understand what 95 percent of our genome does. So be prepared to maybe say this could be the cause of the undiagnosed disease, but you may not be sure. And, more importantly, you may not actually be able to provide too much information in terms of how to put a treatment plan in place, because we don't know what the cause of that particular mutation may be doing.

[00:02:44] HOFF: And finally, if you could add a point to your article that you didn't have the time or space to fully explore, what would that be?

PEARCE: Yeah, I think it's important to emphasize that Undiagnosed Disease Networks is a research protocol. You have to consent people into this for many of the reasons that I've just stated. But the bit that really is missing from the article is, is that I think going back to students and caregivers is, is be prepared to try and actually put a care plan in place. You're going to get an overwhelming amount of information potentially on a pathway or a clinical presentation, and that may not match with the information that we actually have from our genomes right now. So I think you have to be the ultimate caregiver in terms of really trying to figure out what's the best path forwards for these individuals that are living with undiagnosed diseases if you are at the point when you think you are giving them some sort of diagnosis. [theme music returns]

[00:03:41] HOFF: Dr Pearce, thank you so much for your time on the podcast today, and thanks to you and your coauthor for your contribution to the Journal this month.

PEARCE: Yeah. Thanks, Tim. This is a really important topic, so I'm so glad that I had the opportunity to do this.

HOFF: To read the full article, as well as the rest of this month's issue for free, visit our site, journalofethics.org. We'll be back soon with more *Ethics Talk* from the *American Medical Association Journal of Ethics*.