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OPEN PEER COMMENTARIES

Ticket, Roadmap, or Road Not Taken?

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In their essay, MacDuffie et al. lay out a cogent and persuasive set of arguments that the middle ground between clinical and personal utility should be guided, but not rely on, a genomic diagnosis (MacDuffie et al. 2025). They carefully describe the issues implied by their group's on-going work on defining the "middle-ground utility" of genomic diagnoses, and they provide bold recommendations that would serve all young children and families well, even if no genomic diagnosis is made.

One quibble with their formulation is that, for clinicians and educators who focus on developmental and behavioral concerns, what MacDuffie et al. call "utility in community contexts" is in fact the mainstay of treatment. Special education services and developmental therapies have an evidence base, are delivered by trained professionals, and can improve outcomes, just like medications or surgeries. As such, developmental services could be deemed "clinical utility," and access to them should be like access to any medical intervention.

That said, many scholars and practitioners in the genomics world are less acquainted with developmental interventions, so there is value in the authors' calling attention to middle-ground utility. In addition, access to developmental and educational services often occurs through Part C Early Intervention Programs (EIP) and Part B special education services, which as MacDuffie et al. describe, are determined through the implementation of the federal Individuals with Disabilities Education Act (IDEA) and not through traditional health insurance.

"Middle ground" terminology is ok, as long as the concept doesn't become prematurely reified. As MacDuffie et al. note, there is little research to support their supposition that a genomic diagnosis serves as a ticket or a roadmap. Given the tendency of the genomic sequencing industry to proclaim the value of diagnosis, there is the danger that enthusiasts will cite the authors' work on terminology as proof of further benefit to the child and family. MacDuffie et al. recognize this need for more research in their recommendation to build the evidence base for developmental services. They also know that the momentum of implementing genomic sequencing can easily overwhelm calls—such as theirs—for concomitant investment in resources to ensure that developmental services are available and accessible to all families.

In this open peer commentary, I focus even more on the opportunity costs of genomic sequencing, both at the family and societal level. Here's a plea to consider the road not taken, based on my observations during three decades of working with families as a developmental-behavioral pediatrician and as a state and federal government health official.

The way it usually happens is this. Sometime after a child's first birthday, someone notices he's not talking as much as he should. His mother will mention it to the pediatrician, who may say that boys talk late. Or if the family is lucky, the pediatrician will refer them to their state's Part C EIP for an evaluation. Usually though, it takes a few visits to the doctor after other concerns arise: a comment from a teacher, really bad tantrums, or something just doesn't seem right. Mostly it is just a persistent mother, and eventually, maybe through a speech-language pathologist (SLP), the child gets referred to the EIP program.

Once the family gets to that point, federal guidelines require the EIP team to ask the family their goals. They will likely say something like "I want my child to talk" or "I want him to behave better." Depending on the individual state's financial commitment to early intervention, the child will qualify for as little as one or two visits a week from an SLP or an early intervention specialist, even if the child has a serious developmental disorder like autism. Health insurance may cover more developmental or behavior therapies. Depending on how much he

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improves by his third birthday, he may qualify for preschool special education services through Part B of IDEA; and depending on his progress there, he may qualify for special education services in kindergarten and beyond. If he is determined eligible, he will have an Individualized Education Plan detailing the modified instruction and/or related services that he will receive, which could include some speech or occupational therapy to help him access the educational curriculum. There is no cost to families for these public benefits, but often more is needed so families will turn to health insurance for additional therapies.

Along the way, life continues. In addition to the typical caregiver activities of working, running a household, and maybe trying to have some fun, the family will be responsible for finding therapists who are available, well-trained, and accept their insurance. There will be staff turnover, co-pays, insurance denials, and recurring questions about whether this is the right therapy or therapist. When there is a choice to enroll in special education, decision-making gets even more complex. The public preschool is free but available only in the mornings; the private daycare is open the whole day but is expensive and the center director is unsure that they can meet the child's needs. Well-meaning family members cast doubt on the diagnosis, social media offer compelling but unorthodox solutions, and siblings wonder why their brother gets all the attention.

Into this mix comes the long-awaited appointment with the child neurologist, developmental pediatrician, or other pediatric subspecialist. This clinician may provide a general developmental diagnosis such as autism or global developmental delay. Their plan will include recommendations that the family start (or continue) individual therapies and early intervention programs. National clinical guidelines recommend a diagnostic algorithm (Rodan et al. 2025), however, and this search for an underlying medical explanation for development concerns often becomes the focus of the visit. As the specialist recommends a diagnostic evaluation, including whole genome or whole exome sequencing, another adventure begins for the family. The specialist is offering the tantalizing notion that there may be a medical treatment that solves their child's problem—which makes fighting with the health insurance company or paying the out-of-pocket costs worth it.

The outcome of the diagnostic search, however, is often disappointing. The chances that the result will directly benefit the child's behavior or development through medical intervention are exceedingly small. For some conditions, like hypothyroidism, a special diet or medication is essential, but these conditions are extremely rare, and many should have been revealed through universal state newborn screening programs. A more likely result of the diagnostic work-up is that everything is negative. With rapid advances in genomics, this is happening less often, and families are sometimes learning about a genetic difference in their child that may or may not explain their child's condition. It will usually be something rare, so information will be limited. And it comes with the possibility of increased risks for other health issues in the future, like cancer, that will require lifelong monitoring.

Some families will find value in a genomic diagnosis, however uncertain. Armed with new knowledge, they will connect directly with other families who have a child with a similar diagnosis, search the nation for a clinical team with condition-specific expertise, or simply feel reassured that there is a medical explanation beyond a vague developmental label. Knowing about future risks can be empowering, and some families feel greater control because they know what to look for. A genomic diagnosis can also be helpful regarding the risk of recurrence or to other family members.

Other families will be less enthusiastic about the results. Many families will say something like, "we saw the neurologist and they couldn't find anything." Others will be able to name the syndrome or specific genetic difference, then wonder what it means. Whatever the specific result, their main concern remains that their child still doesn't talk or behave or learn the way they hoped he would. Their therapeutic odyssey continues (Brosco 2018).

Here's the first road *not* taken. Did the family understand that they could say no to the diagnostic evaluation? Shared decision making is the standard for pediatric health care, especially for children with developmental disabilities (Adams and Levy 2017). That means that clinicians should work with families to learn their goals for their child, then devise a treatment plan to meet those goals. A genetic diagnosis may or may not be high on the family's list, and their decision usually depends on the likelihood that the result would change that child's developmental trajectory. How well did the subspecialist inform the family of the risks, benefits, and likely results of genomic sequencing? Given limited time, energy and other resources for families in their everyday lives, are we really helping them by routinely pursuing a diagnostic evaluation?

Here's the second road not taken: this one is at the policy level. MacDuffie et al. implicitly recognize the opportunity costs of pursuing a genomic diagnosis at a societal level, and they recommend increased funding for early childhood services, improved family navigation skills and support, and research to build the evidence base for developmental services. We should notice that they make these sound recommendations in the context of the potential value of a genomic diagnosis. That early childhood development services are not their primary focus is on-going evidence that we are all living through the latest chapter of our nation's long history of seeking to improve health through technology (Brosco 2012).

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Digital Life Models and the Genomic Knowledge Paradox: A Proposal for AI-Assisted Reflection in Genetic Decision-Making

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PRÉCIS

As genomic screening expands globally, individuals increasingly confront an epistemic challenge: they cannot know whether their future self will benefit from—or prefer versus regret having received—genetic risk information without first receiving it. This commentary explores how digital life models (DLMs), or personalized AI systems trained on individual values and decision patterns, might help navigate this dilemma. DLMs offer a novel approach for anticipatory ethical reflection, potentially enabling individuals to simulate their responses to genetic information before irreversible disclosure.

THE KNOWLEDGE PARADOX IN GENETIC SCREENING

Jeff is 30 years old. His mother was diagnosed with early onset Alzheimer's disease at 55, which in some

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