

# Should all newborns undergo genetic screening for type 1 diabetes risk?

## POINT

### **The time to begin screening all newborns for genetic risk is now.**

The idea behind screening is to achieve three goals. The immediate goal is reduction of diabetic ketoacidosis. We know that DKA is a very serious issue with a type 1 diabetes diagnosis. Some studies suggest that up to 60% of children with type 1 diabetes present when they are in DKA. This has short-term and long-term effects on glucose control, which has effects on outcomes and complications. There are a lot of reasons why one would want to avoid a diagnosis in DKA. We know from studies like TEDDY, TrialNet and others that, through screening and monitoring, you can reduce the DKA rates dramatically.



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The midterm goal would be to find more eligible people to participate in clinical trials at a time when more companies are developing therapeutics aiming to slow progression and one day even prevent type 1 diabetes.

Then there is the long-term goal, which would be to match people with a therapy. This has relevance now with teplizumab (Provention Bio), an immunotherapy drug, which has shown in a clinical trial that it can slow the development of type 1 diabetes and recently received an FDA breakthrough designation. This drug could potentially be on the market in 2 years.

With genetic screening, you eliminate all of the children who are at low risk for developing type 1 diabetes, even though some of those children will still go on to develop the disease. So you “miss” some of those children. No screening model is perfect. I would advocate for a hybrid genetic screening model, where newborns are screened at birth, and then there is a second universal screening around age 4 years. There are challenges with this, in terms of cost and access.

If the goals are to treat as many people as possible and avoid the long-term complications of type 1 diabetes, you want to be able to access as many people as possible, both for the prevention of DKA and ultimately for therapies. Some say that screening is not viable now because you cannot do anything to treat the disease. To those people, I respond that we can reduce incidence of DKA in children. The stronger argument is now we can actually modify disease course, which we have never shown before. There will be challenges with payors and clinicians and issues of getting screenings on to a schedule, but certainly the time is now.

We have a real opportunity here to change the course of a disease that has no cure. Research has demonstrated that we can delay the onset of disease, which we know will have a significant impact on long-term health. We are on the path to long-term prevention and, ultimately, to a cure. We must approach it early with genetic screening.

Jessica Dunne, PhD, is senior director of research at JDRE.

## COUNTER

## **Universal genetic screening of newborns for type 1 diabetes risk should not be done unless there is a treatment.**

Genetic screening for type 1 diabetes risk in newborns is controversial for several reasons. Even if an infant has high genetic risk, the chances of going on to develop type 1 diabetes are very low. Second, as of right now, even if we know a person is at increased risk genetically, we have no way to stop disease progression. You are screening for something you cannot do anything about. Third, certainly in the United States, there are concerns that insurance companies could argue that higher genetic risk is a preexisting condition that could be used to potentially deny coverage or increase the cost of coverage. Finally, genetic screening indicating a heightened risk for type 1 diabetes could potentially raise anxiety in families about a child's risk.



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There are good reasons for genetic screenings in a research setting. If we are going to figure out why some children with high genetic risk go on to develop type 1 diabetes while others do not, then we have to find these children. Without screenings, we could not answer that scientific question or test interventions to prevent type 1 diabetes. Scientists are not opposed to screening newborns; we are doing it in the TEDDY study, which I am involved with. Genetic screening should be performed only in the context of a research study, in which children will be properly followed and families of screened children will receive the resources and support they will need.

I am not suggesting screenings should never be performed. From a scientific point of view, that would set us back. In TEDDY, thousands of children were screened in the U.S., Sweden and Finland. We are learning a lot from these children about the environmental factors and other potential “triggers” that could lead to development of type 1 diabetes in this genetically at-risk population.

There are a few diseases in which a person who is screened and discovered to have a certain gene would definitely go on to develop the disease. For most diseases where genetic screening is available, it is not like that. The screening results reflect increased risk, and that concept can be very difficult for people to understand. In TEDDY, we look at this a lot. We have found that families struggle to grasp the concept. This is not unique to diabetes; many studies on genetic testing reflect this.

If you are a parent and type 1 diabetes runs in your family, you may find it comforting to participate in a research study like TEDDY, where newborns are screened and then followed and monitored. Then, if and when a treatment is available, that child is likely to have a chance at participating in a research trial.

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