



# Early Check – A Newborn Screening Research Program in North Carolina

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RTI International

# Disclosures

## **Early Check is supported by:**

- The Leona M. and Harry B. Helmsley Charitable Trust
- JDRF International
- Janssen Pharmaceuticals
- Traverre Therapeutics
- Orchard Therapeutics
- Sarepta Therapeutics
- Muscular Dystrophy Association

## **Laboratory Partners**

- GeneDx
- Illumina



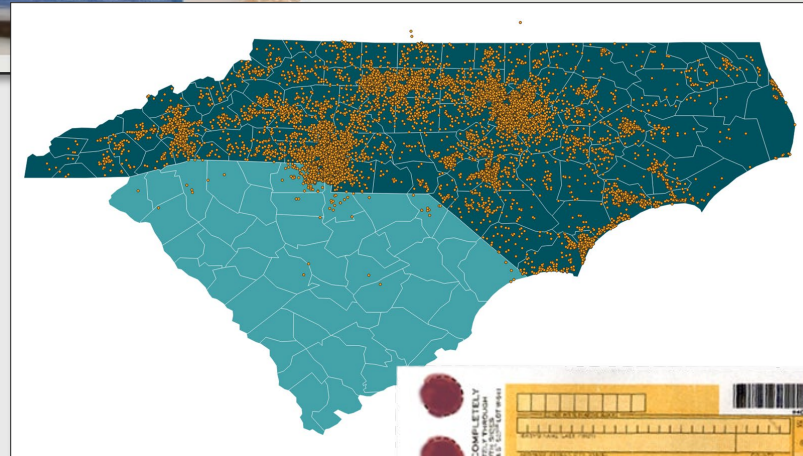
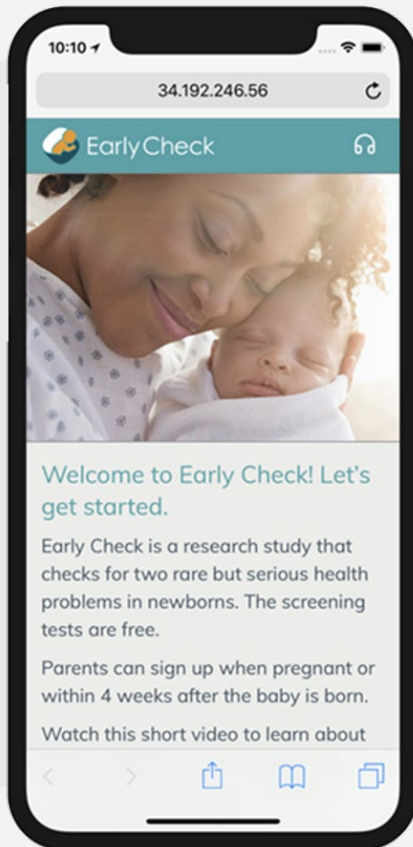
# Agenda

- Early Check study
- Addition of genomic sequencing and T1D genetic risk scores
- How are the lessons learned from the first 5 years of Early Check informing risk for T1D screening?
- Early data






# Early Check: A Voluntary Newborn Screening Program

Screening newborns in N.C. since 2018

A photograph of a newborn screening card. The card is yellow and contains various fields for information such as the baby's name, date of birth, sex, race, and mother's name. There are also checkboxes for 'FILL ALL 4 CIRCLES COMPLETELY' and a barcode at the top right.

### Early Check Panel

-  Duchenne Muscular Dystrophy (DMD) and related conditions
-  Fragile X Syndrome (FXS)
-  Spinal Muscular Atrophy (SMA)





# Objective: Respond to opportunities facing newborn screening



Genome (and 'Omic') technologies



Precision medicine advances, including for common disorders



Rapidly growing pipeline of transformative therapies

## Sequencing study aims (initial 3-year study with plans to continue)

1

Implement and evaluate the use of genome sequencing (GS) to screen up to 10,000 newborns for childhood-onset, monogenic conditions.

2

Determine uptake and acceptability of GS newborn screening among parents of eligible newborns.

3

Implement and evaluate short-term follow-up procedures to inform public health application.

4

Assess 12-month outcomes for children with positive results and their caregivers.



# Type 1 diabetes aims (initial 3-year study with plans to continue)

1

Conduct formative research among parents, primary care providers, specialists, and public health professionals on using genetic risk scores in newborn screening for type 1 diabetes.

3

Determine uptake and acceptability of type 1 diabetes screening among parents of eligible newborns, compared with outcomes from panels of rare monogenic conditions.

2




Prepare for, implement, and evaluate a pilot study to screen at least 5,000 newborns for increased risk for type 1 diabetes.

4

Assess 12-month outcomes for children with increased risk for type 1 diabetes and their caregivers.



# Adding Genome Sequencing to Early Check: Initial Panels

Early Check Panel	
	<b>Treatable Conditions (~180)</b> Retinoblastoma      Hemophilia Neonatal diabetes      Fructose intolerance
	<b>Conditions with Potential Treatments (~30)</b> Duchenne muscular dystrophy Menkes disease
	<b>Risk for Type 1 Diabetes</b>

# The first 5 years informing Early Check's future

**We can screen thousands of infants per year** with a very low touch approach and no study visit.

**State partnership** is essential for large-scale screening to inform public health.

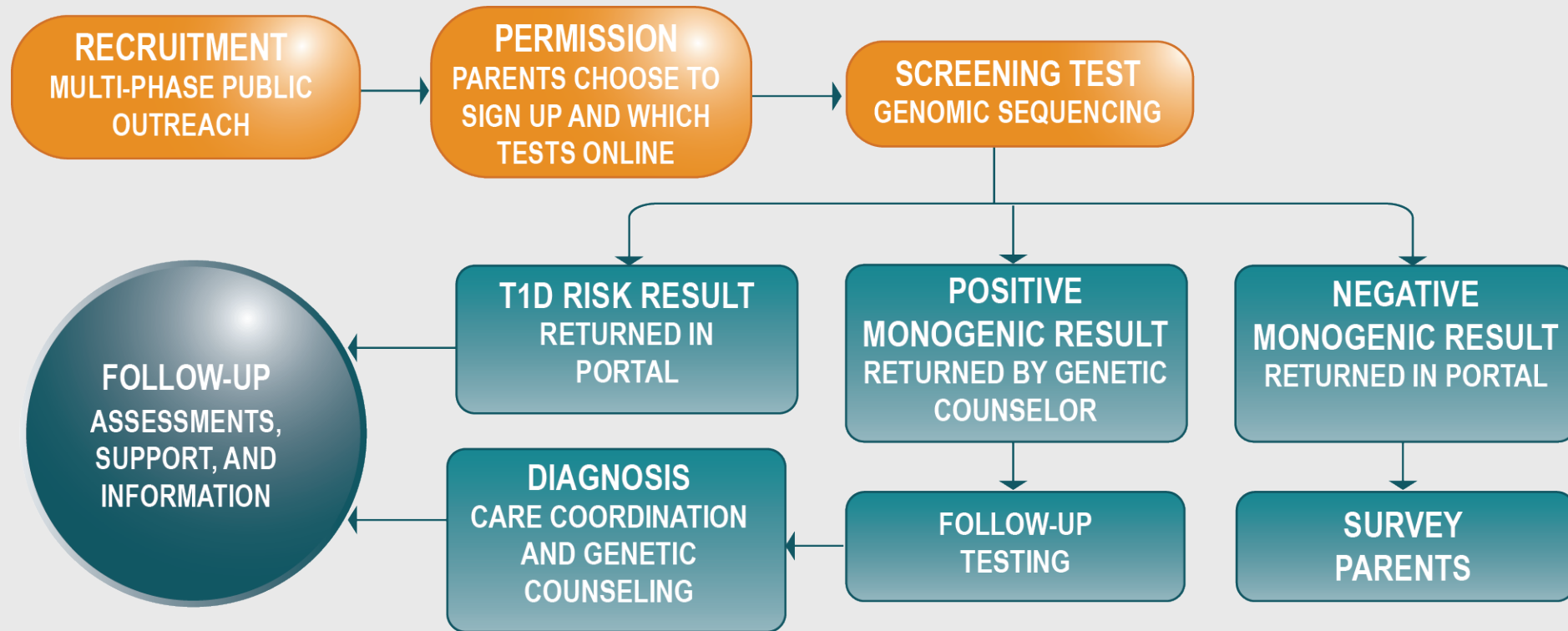
**User-centered electronic informed consent** can successfully support a low-touch informed choice.

**Electronic return of results** can be successful, facilitating large sample sizes.

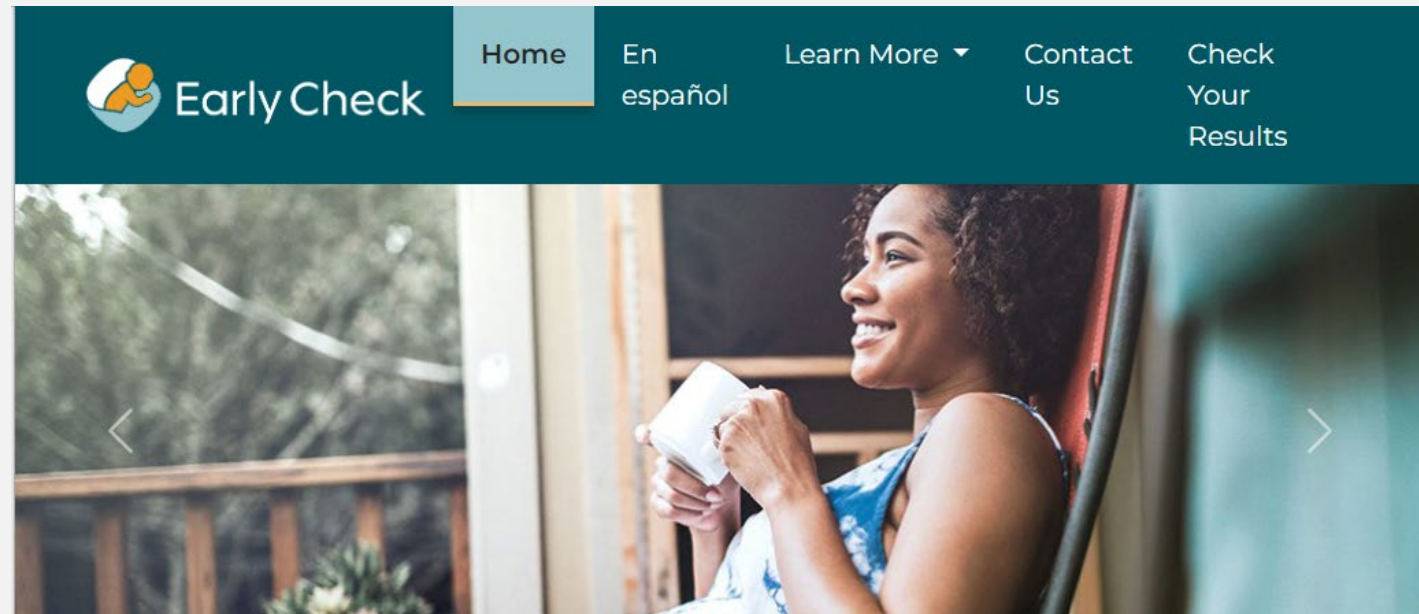





# Process Overview



# Early Check Portal



# Obtain Consent in the Portal

 Early Check

HomeEn EspañolLearn More ▾Contact UsCheck Your Results

## What does Early Check Screen For?

Early Check offers screening for hundreds of serious rare health conditions and also checks the risk for type 1 diabetes.

Parents can choose which types of health conditions they want screened for their baby. To help you decide, we put these screening tests into three groups.

Here are brief descriptions of the Early Check screening groups:

Group 1 - Treatable Conditions	Group 2 Optional - Conditions with Potential Treatments	Group 3 Optional – Risk for Type 1 Diabetes (T1D)
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### Group 3 Optional – Risk for Type 1 Diabetes (T1D)

- Early Check will check your baby's DNA to learn the genetic risk for Type 1 Diabetes, a fairly common and treatable health condition.
- T1D is the most common form of diabetes in children. Babies are not born with T1D, but it can show up later in childhood. Testing can provide information about your child's *risk* for T1D.
- Type 1 Diabetes (T1D) causes children to have serious symptoms or die if not treated.

**Why is this information important?**

- Knowing if your baby has a higher risk for T1D lets parents watch for signs of the condition. This could lead to earlier diagnosis, treatment and better health for a child with T1D.

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# Obtain Consent in the Portal

## Which screening groups do you want to learn more about?

All babies signed up for Early Check get testing for the *Treatable Conditions* (Group 1).


***Conditions with Potential Treatments (Group 2)*** and ***Risk for Type 1 Diabetes (Group 3)*** are optional screening groups.

Select the optional groups below to read more about screening for these health conditions.

Then click the “Next” button to move on to the next screen.


Treatable  
Conditions  
Group

☒




Conditions  
with Potential  
Treatment  
Group  
(optional)

☐



Risk for Type  
1 Diabetes  
Group  
(optional)

☒



← Back

Next →



## Video: How is Early Check screening done?



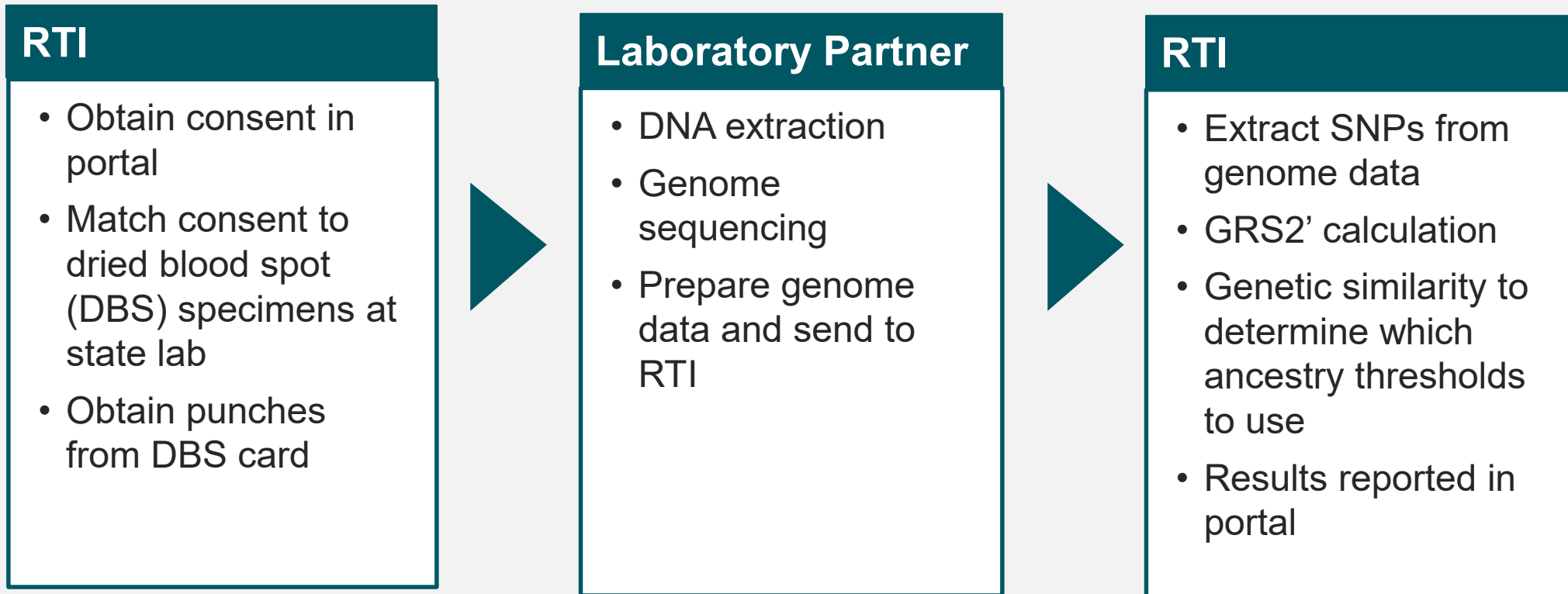


# T1D GRS screening

- Public health unprepared for screening or follow-up if GRS results were included in newborn screening
- Model screening in a public health-like setting




# T1D Screening Process Overview




Consent portal opened for enrollment on September 28<sup>th</sup>



# Return of Results in the Portal

 Early Check

En español Forms & Fact Sheets Results page Log out

 Welcome back, Angela!  
We're glad you're checking in.

Results

Test	Date ▾
HISTORICAL EARLY CHECK	Mar 20, 2020
GROUP 1	Aug 15, 2023
GROUP 2	Aug 15, 2023
T1D	PENDING

Questions?  
Whether you signed up for Early Check 4 hours ago or 4 months ago, you might have some questions. See Answers to [Frequently Asked Questions](#) or here or [Contact Us](#).

Group	T1D risk
Low concern	<2%
Moderate concern	≥ 2 to <5%
Higher concern	≥ 5 to 10%
	≥ 10 %



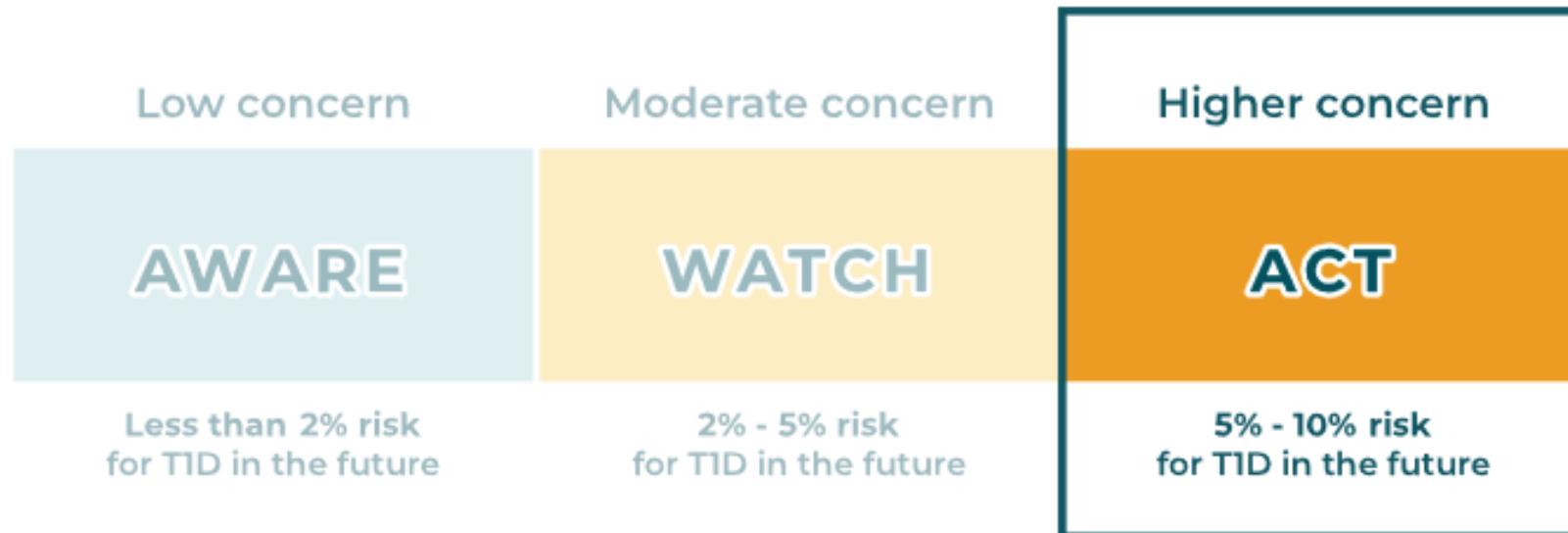
# Return of Results in the Portal

## What's my baby's result?

### Higher Concern for Type 1 Diabetes (T1D): Act

Based on the Early Check DNA testing, your baby's risk for getting type 1 diabetes later on is **higher than most**. Most babies with this result will not get T1D, but some will.

Early Check recommends additional testing.



# Return of Results in the Portal

For babies with this **Higher Concern** result, what is the risk for getting type 1 diabetes during their lifetime?

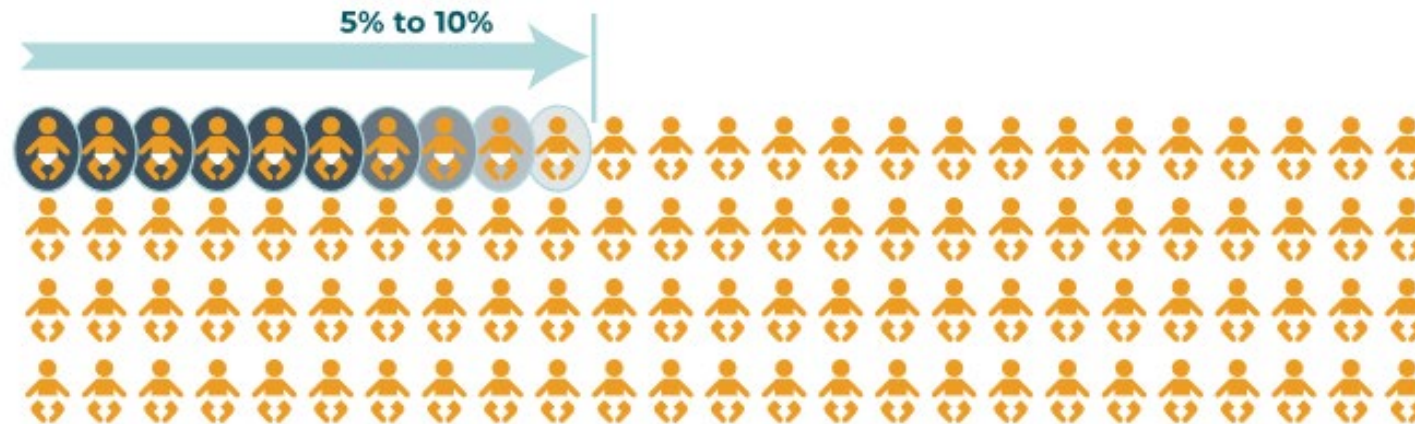
**Risk for  
type 1 diabetes**



Your baby may be more likely to get T1D than usual.  
5%-10% of babies with this result will develop type 1 diabetes during their lifetime.

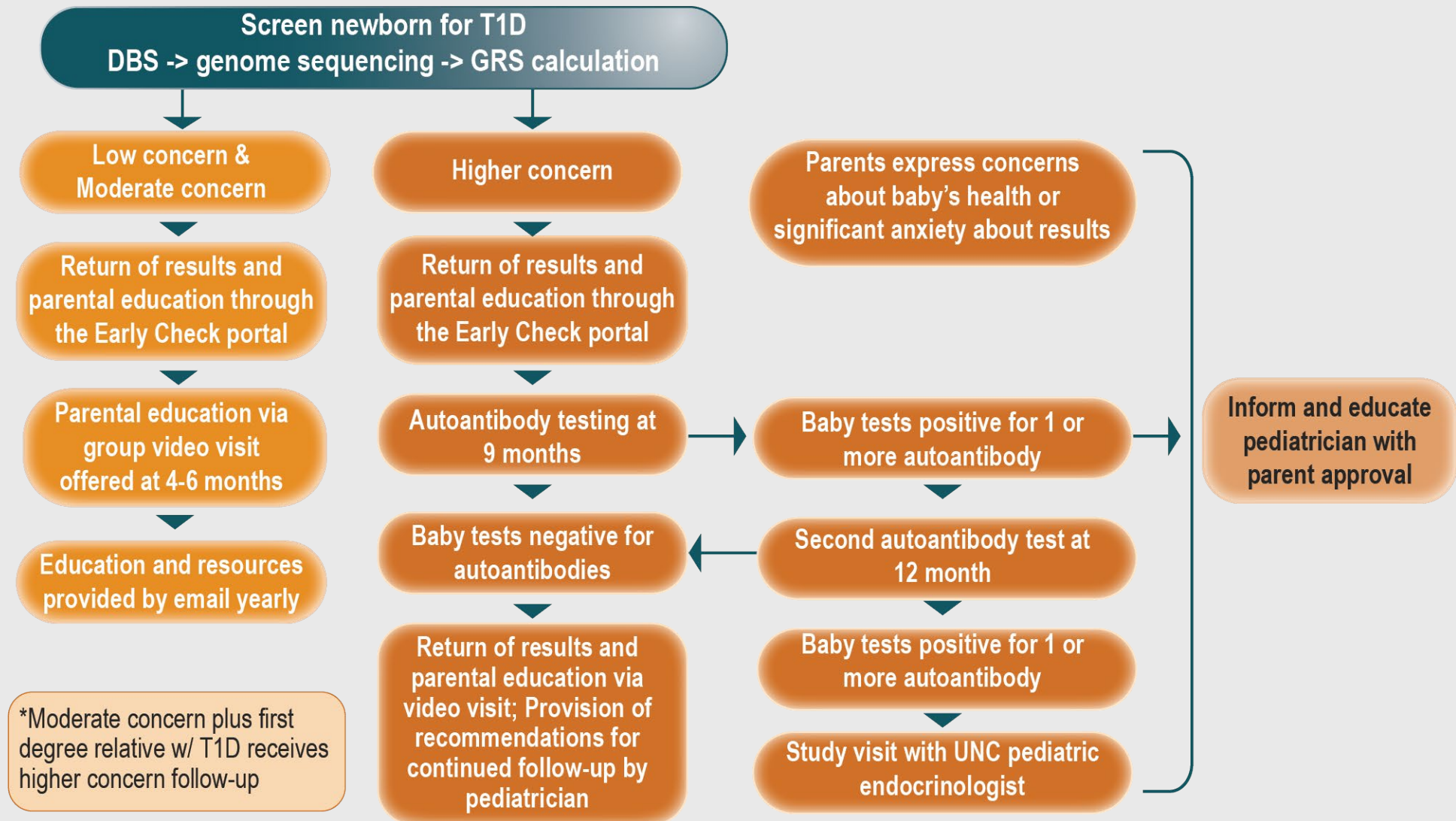
**Here are some things to know:**

- T1D usually starts in childhood.
- Sometimes it doesn't start until adulthood.
- Signs of T1D are rare in the first year of life.





# T1D Follow-up



## Initial Enrollment Data

- Goal: Screen 5,000+ babies for monogenic panels and T1D GRS
- Enrollment opened 9/28/23
- ~95 babies screen/week
- Initial screening results expected early December

	Everyone		T1D Selected	
	n	%	n	%
Babies enrolled	239	100%	175	73%



# Initial Enrollment Data

		Everyone		T1D Selected	
		n	%	n	%
Babies		239	100%	175	73%
Mom's Race/Ethnicity					
	White	153	64.0%	120	68.6%
	Hispanic/Latino or Spanish	25	10.5%	20	11.4%
	Asian	22	9.2%	13	7.4%
	African American/Black	16	6.7%	8	4.6%
	Hispanic/Latino or Spanish,White	5	2.1%	4	2.3%
	Prefer not to respond	5	2.1%	2	1.1%
	African American/Black,White	4	1.7%	1	0.6%
	American Indian/Alaskan Native,White	2	0.8%	2	1.1%
	American Indian/Alaskan Native	1	0.4%	0	0.0%
	Middle Eastern/North African	1	0.4%	1	0.6%
	None fully describe me	1	0.4%	1	0.6%
	Asian,White	1	0.4%	1	0.6%
	American Indian/Alaskan Native,Asian,Hispanic/Latino or Spanish,White	1	0.4%	1	0.6%
	Asian,African American/Black	1	0.4%	1	0.6%



# Initial Enrollment Data

		Everyone		T1D Selected	
		n	%	n	%
Babies		239	100%	175	73%
Dad's Race/Ethnicity					
	White	156	65.3%	124	70.9%
	Hispanic/Latino or Spanish	30	12.6%	23	13.1%
	African American/Black	20	8.4%	12	6.9%
	Asian	17	7.1%	9	5.1%
	Prefer not to respond	4	1.7%	2	1.1%
	African American/Black,White	3	1.3%	0	0.0%
	Hispanic/Latino or Spanish,White	2	0.8%	1	0.6%
	Middle Eastern/North African,White	2	0.8%	2	1.1%
	African American/Black,Hispanic/Latino or Spanish	1	0.4%	1	0.6%
	Asian,African American/Black	1	0.4%	0	0.0%
	American Indian/Alaskan Native	1	0.4%	0	0.0%
	African American/Black,Hispanic/Latino or Spanish,White	1	0.4%	1	0.6%



# Early Check Sequencing Leadership Team



Holly Peay  
Project Director



Heidi Cope  
Associate  
Project Director



Lisa Gehland  
Administrative  
Core Lead



Don Bailey  
Senior Advisor



Anne Wheeler  
Follow Up Core Lead



Melissa Raspa  
Methodology Core Lead



Kate Kucera  
Lab Core Lead

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UNC PI, Geneticist



Liz Jalazo  
Geneticist



Jonathan  
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Geneticist



Jennifer Law  
Endocrinologist

**UNC Chapel Hill**



Scott Shone,  
NCSLPH PI

**NC State Laboratory  
of Public Health**





# Early Check Team



Sara  
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Don  
Bailey



Vesselina  
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Beth  
Boyea



Javan  
Carter



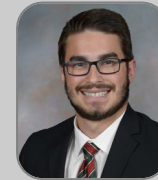
Emily  
Cheves



Heidi  
Cope



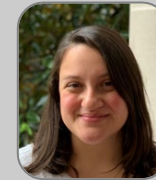
Manisha  
Dass



Jake  
Dibble



Anne  
Edwards



Ana  
Forsythe



Lisa  
Ghtland



Angela  
Gwaltney



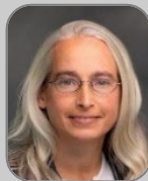
Jessica  
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Brooke  
Migliore



Rebecca  
Moultrie



Casey  
Okoniewski



Grier  
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Holly  
Peay



Melissa  
Raspa



Veronica  
Robles



Samantha  
Scott



Anne  
Wheeler



Becca  
Wright



UNC

Jonathan  
Berg



Liz  
Jalazo



Jennifer  
Law



Laura  
Milko



Cindy  
Powell

NCSLPH



Scott  
Shone



# Thank you

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