# 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
- Travere Therapeutics
- Orchard Therapeutics
- Sarepta Therapeutics
- Muscular Dystrophy Association

**Laboratory Partners**

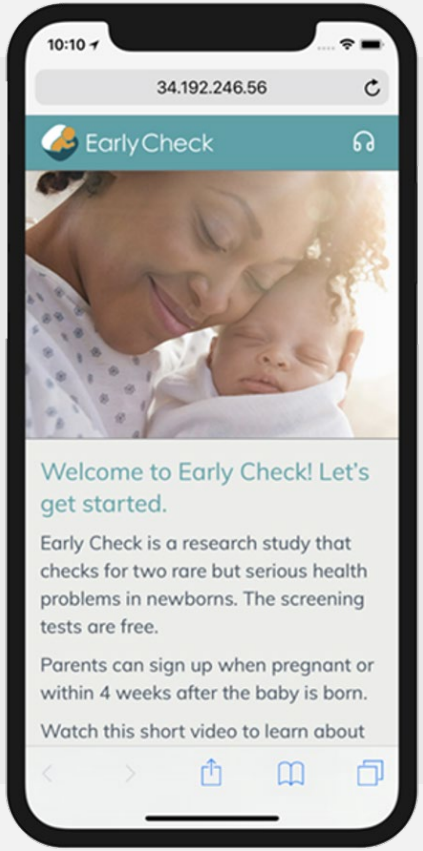
- GeneDx
- Illumina

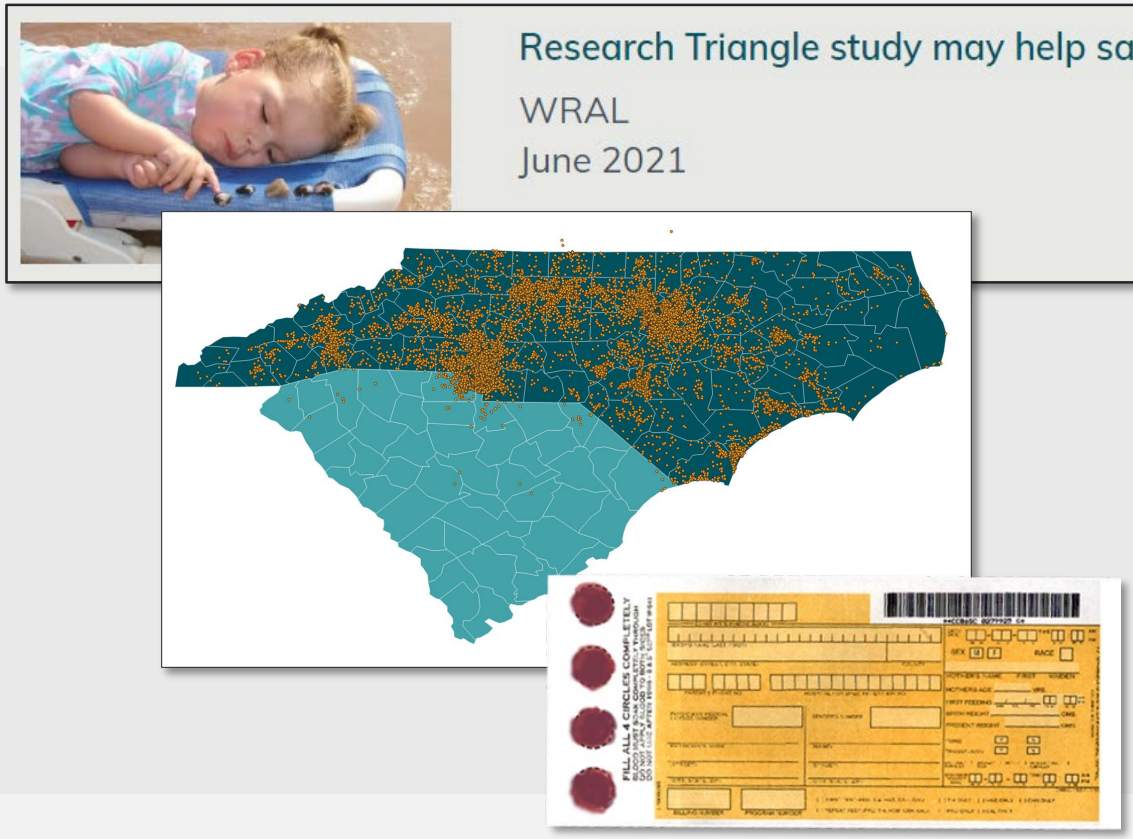
# Agenda

- 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




## 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.

**2**

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

**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

**Treatable Conditions (~180)**

Retinoblastoma
Neonatal diabetes
Hemophilia
Fructose intolerance

**Conditions with Potential Treatments (~30)**

Duchenne muscular dystrophy
Menkes disease

**Risk for Type 1 Diabetes**

# The first 5 years informing Early Check's future

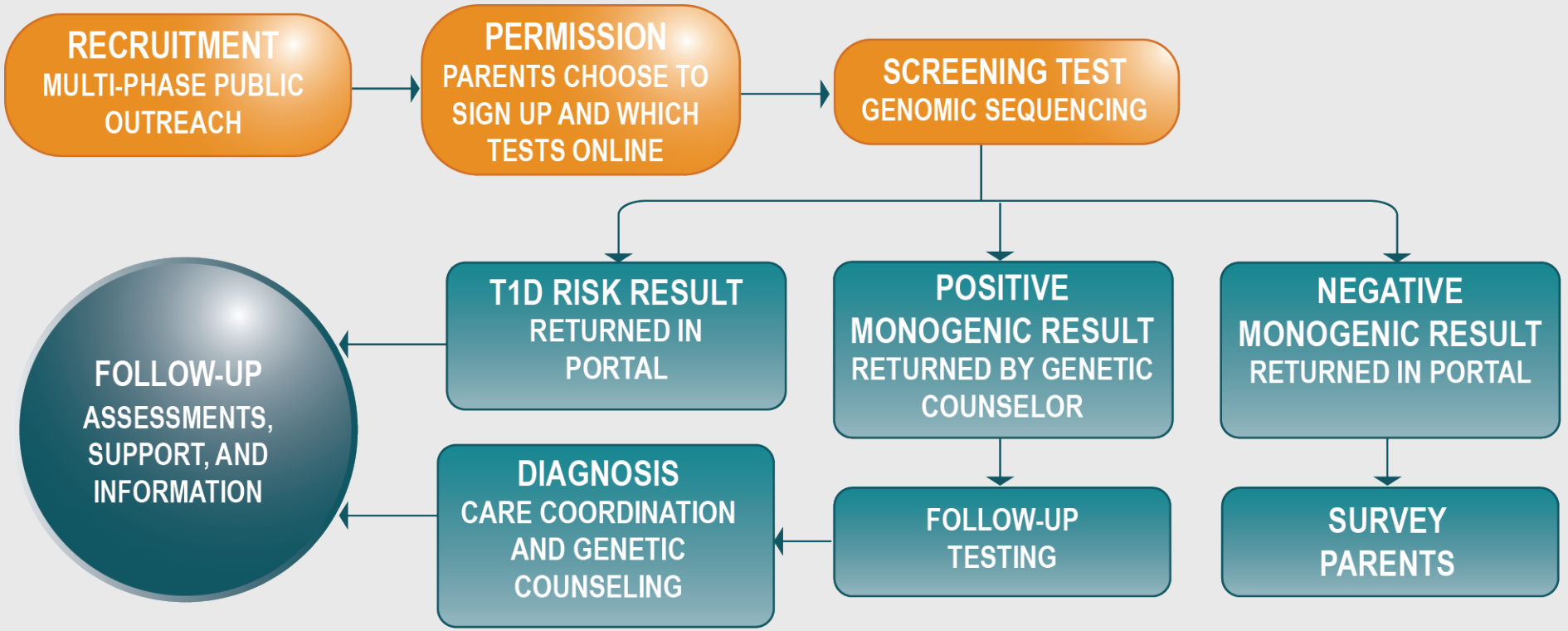
**We can screen thousands of infants per year** with a very low touch approach and no study visit.

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

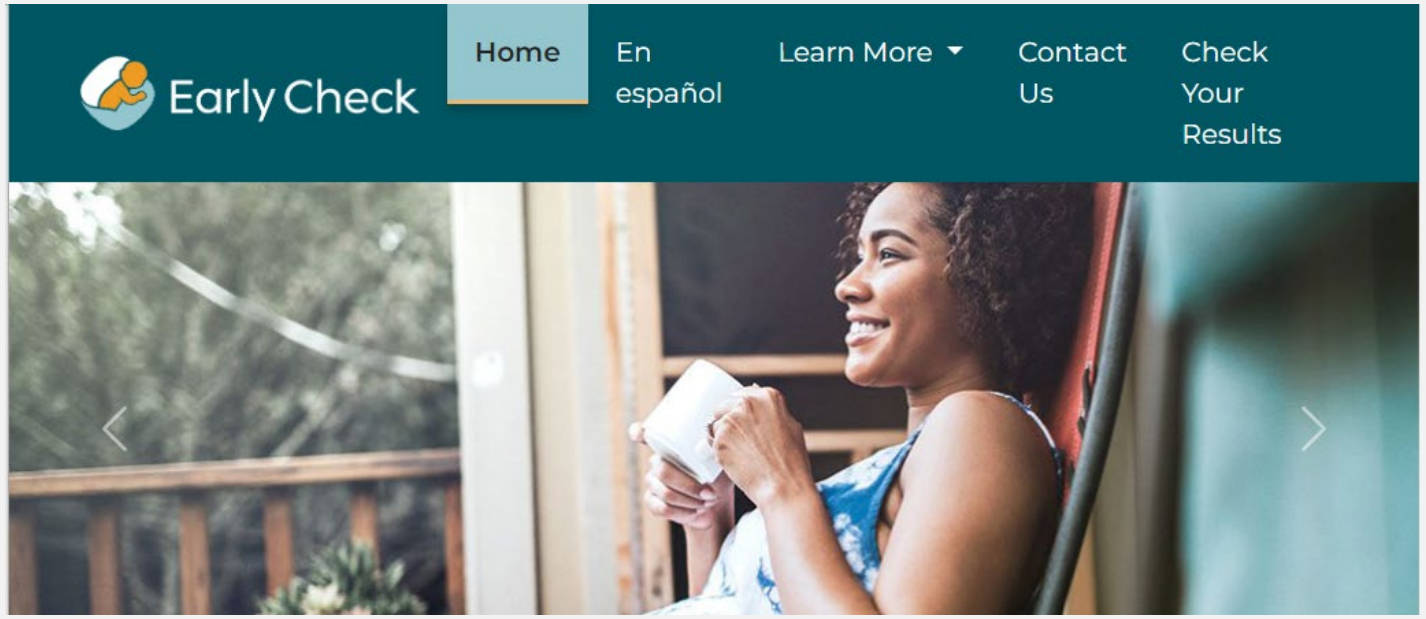
**User-centered electronic informed consent** can successfully support a low-touch informed choice.

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

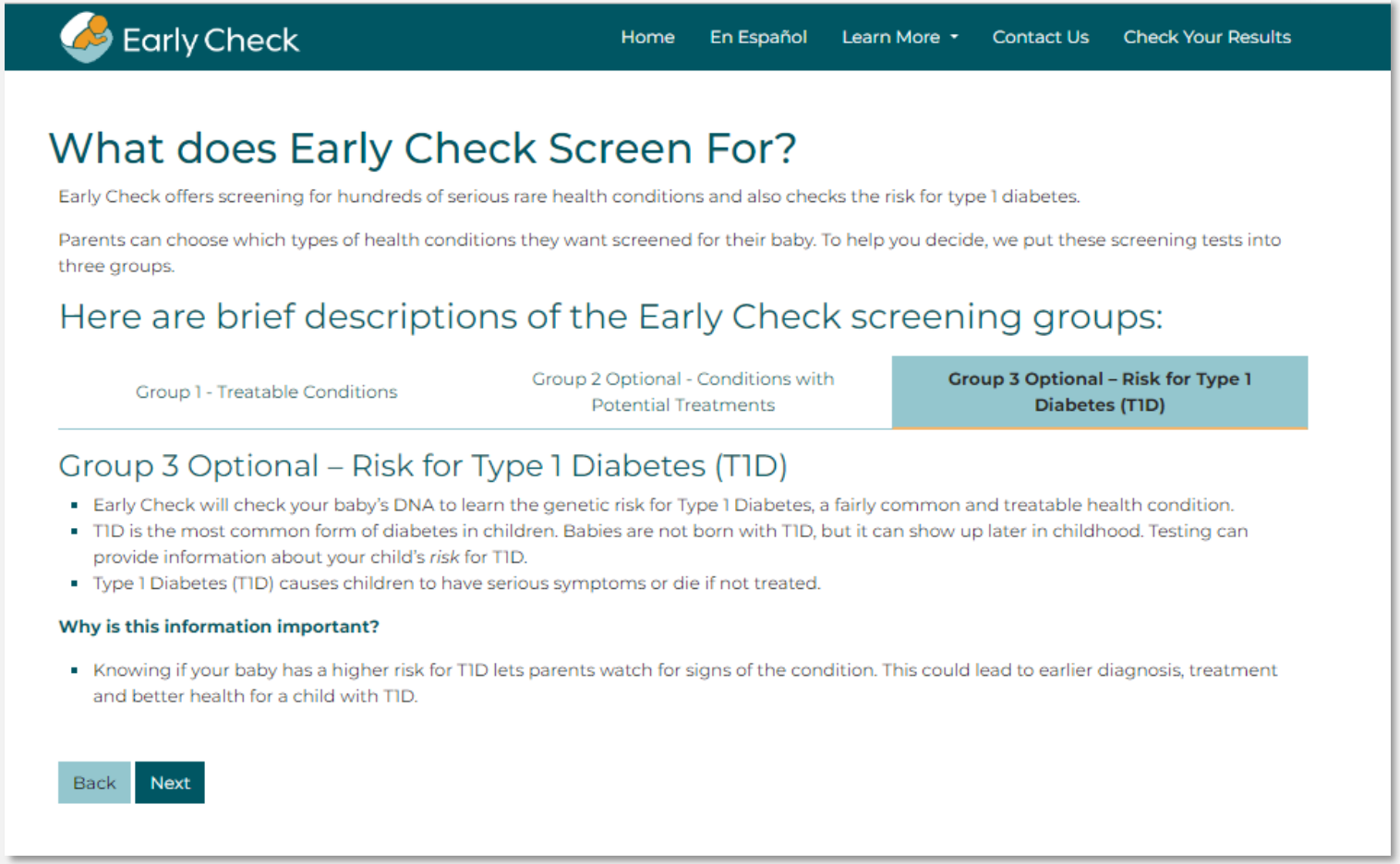
# Process Overview



# Early Check Portal



# Obtain Consent in the Portal



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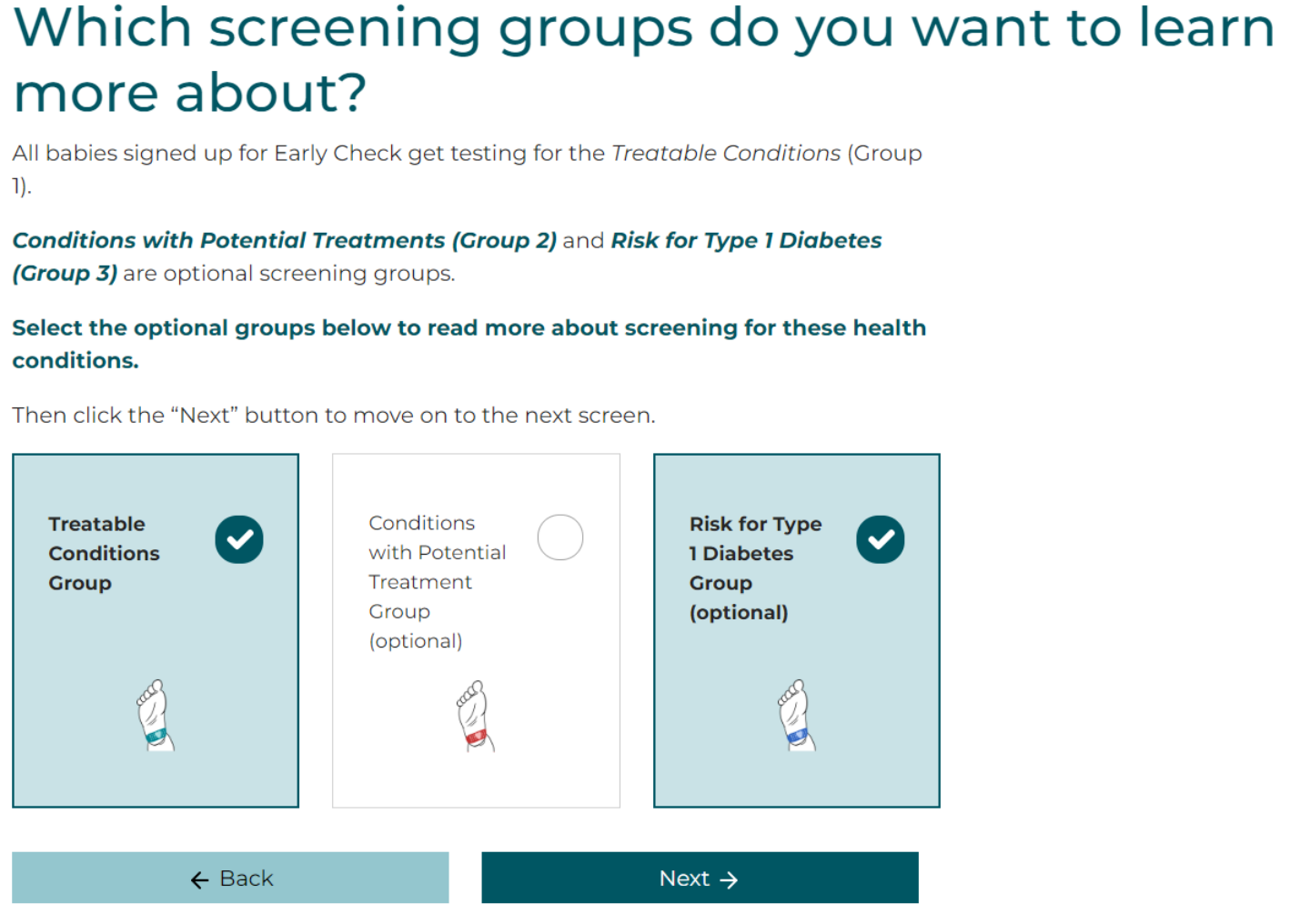
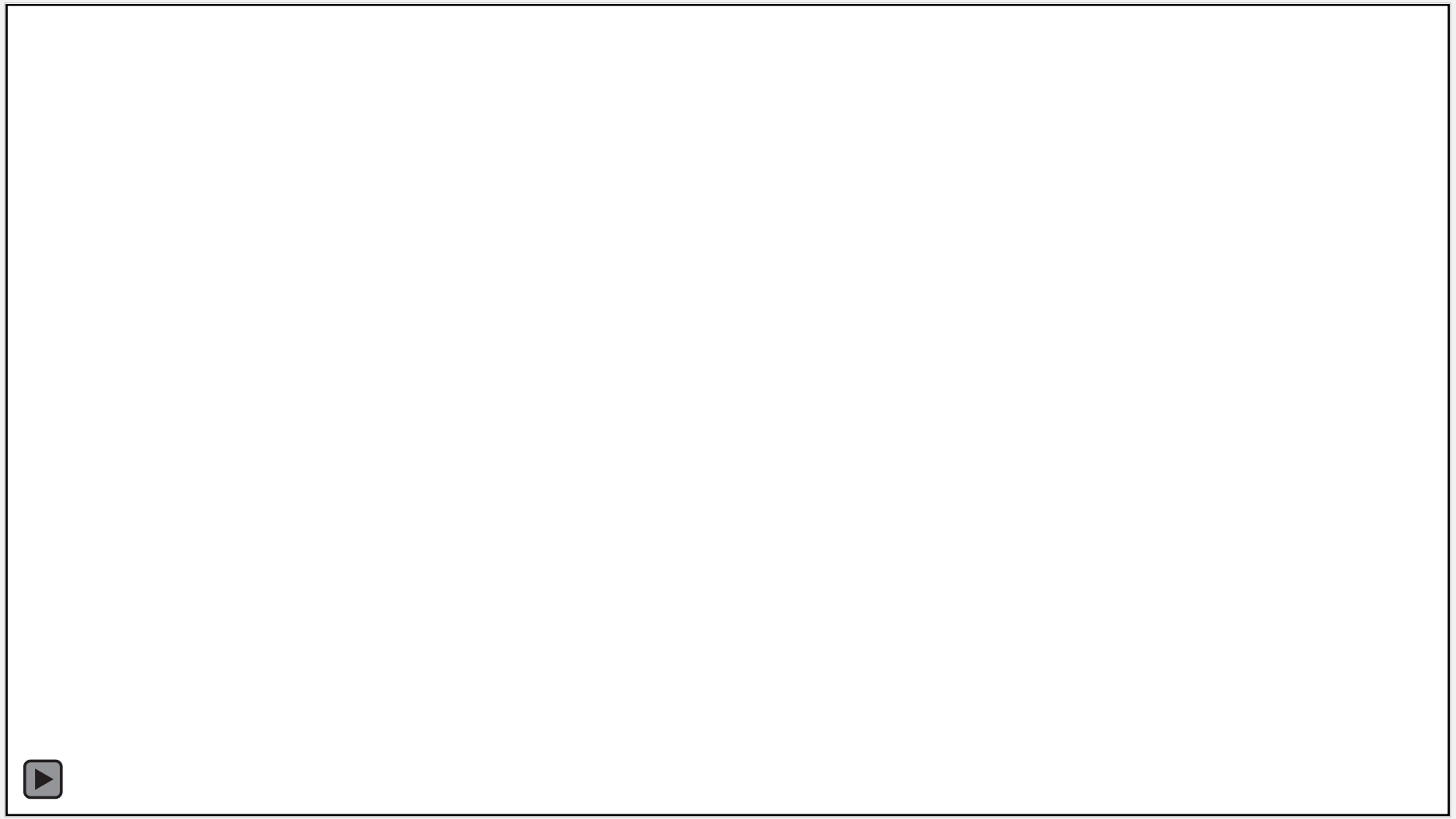


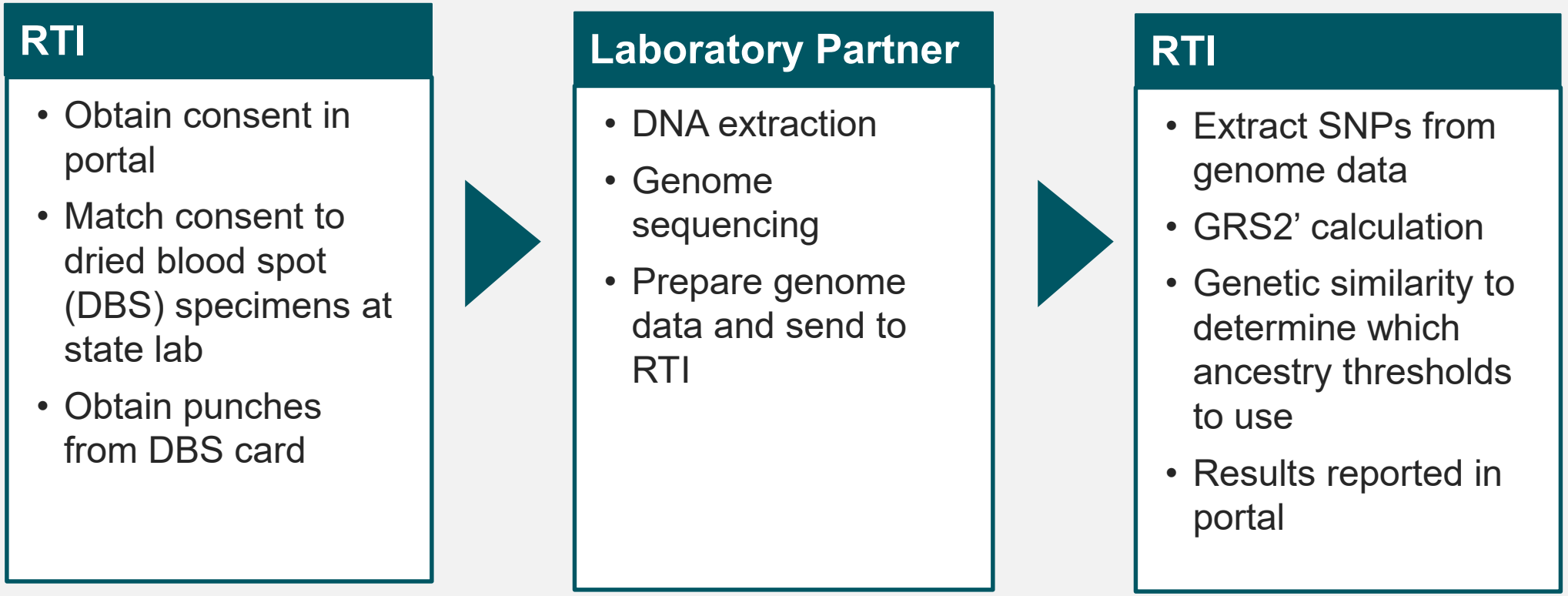
# Video: How is Early Check screening done?



# T1D GRS screening

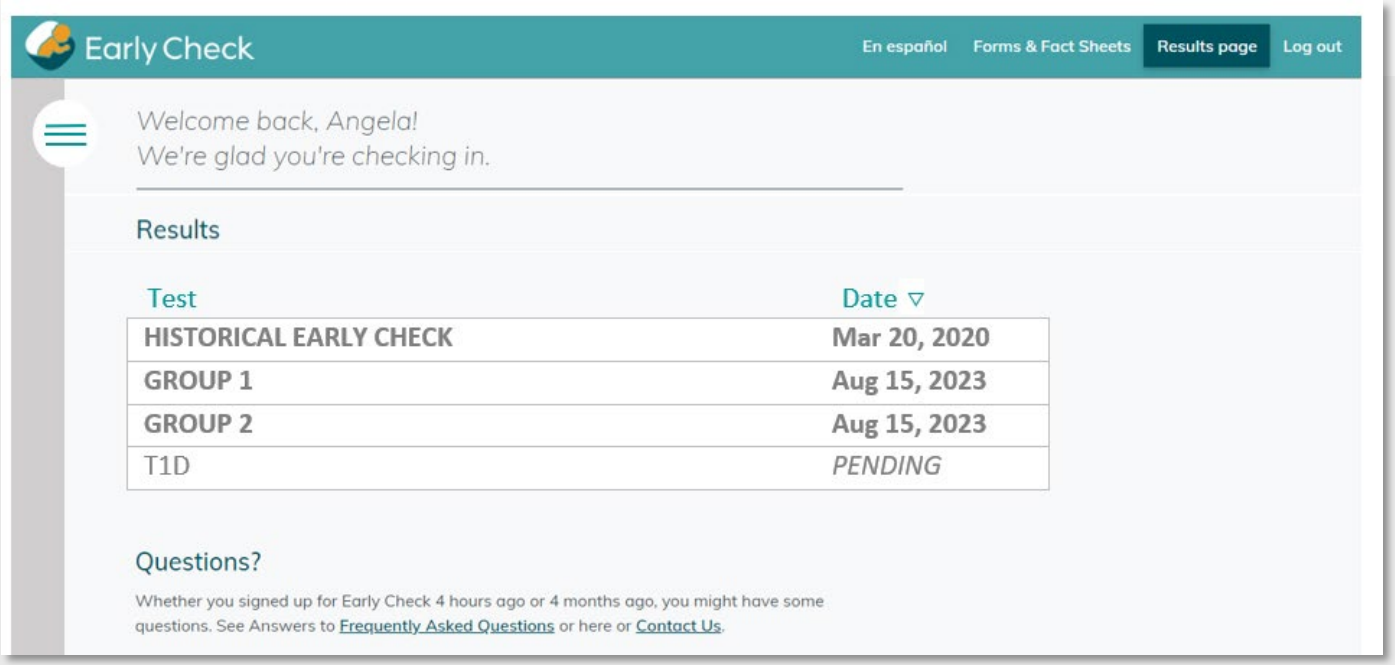
- Public health unprepared for screening or follow-up if GRS results were included in newborn screening
- Model screening in a public health-like setting

# T1D Screening Process Overview



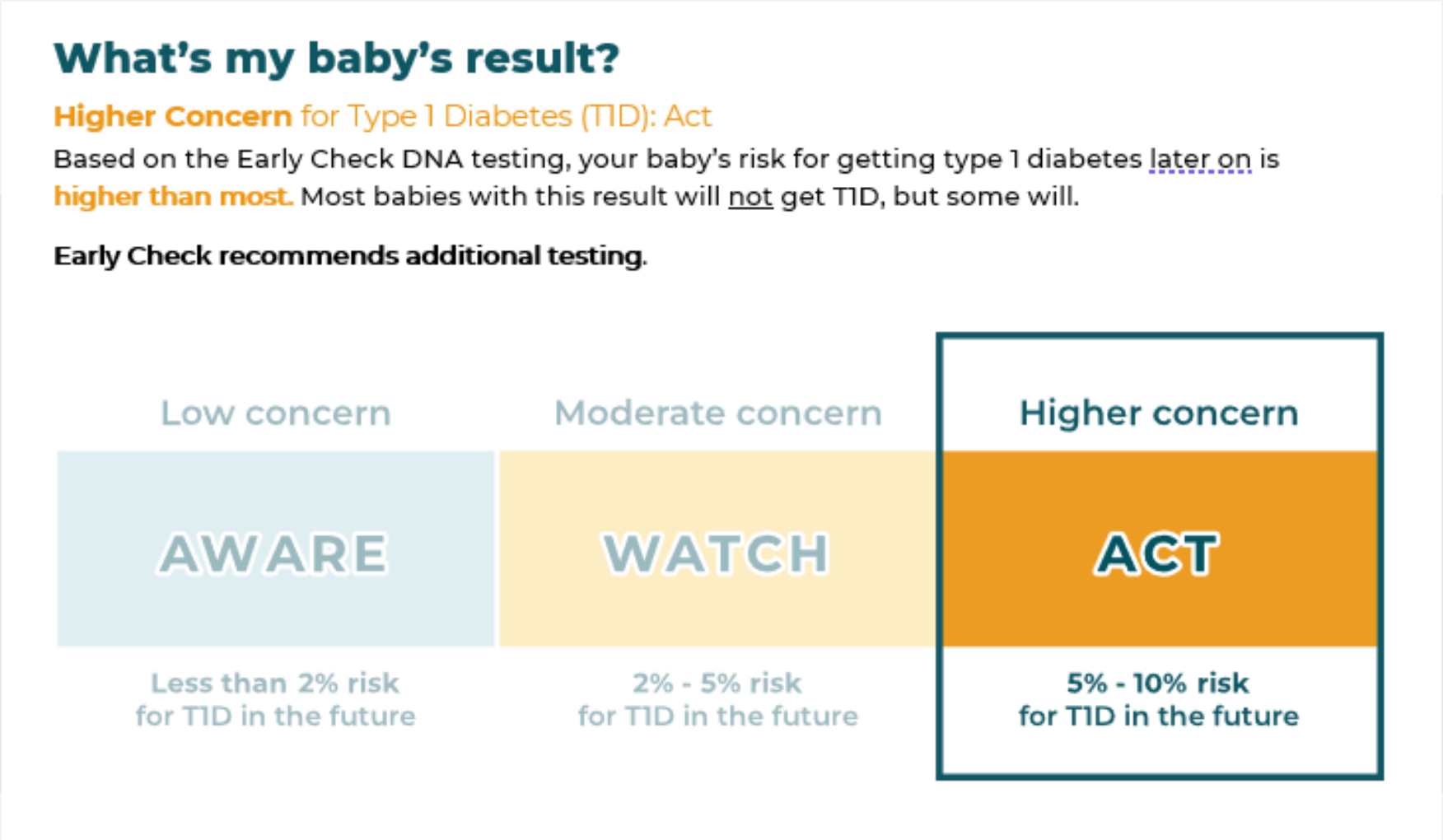

Consent portal opened for enrollment on September 28th

# Return of Results in the Portal



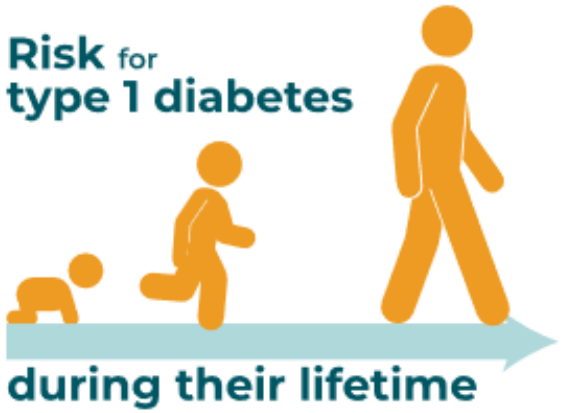

| Group | T1D risk |
|---|---|
| Low concern | <2% |
| Moderate concern | ≥ 2 to <5% |
| Higher concern | ≥ 5 to 10% |
| | ≥ 10 % |

# Return of Results in the Portal



# Return of Results in the Portal

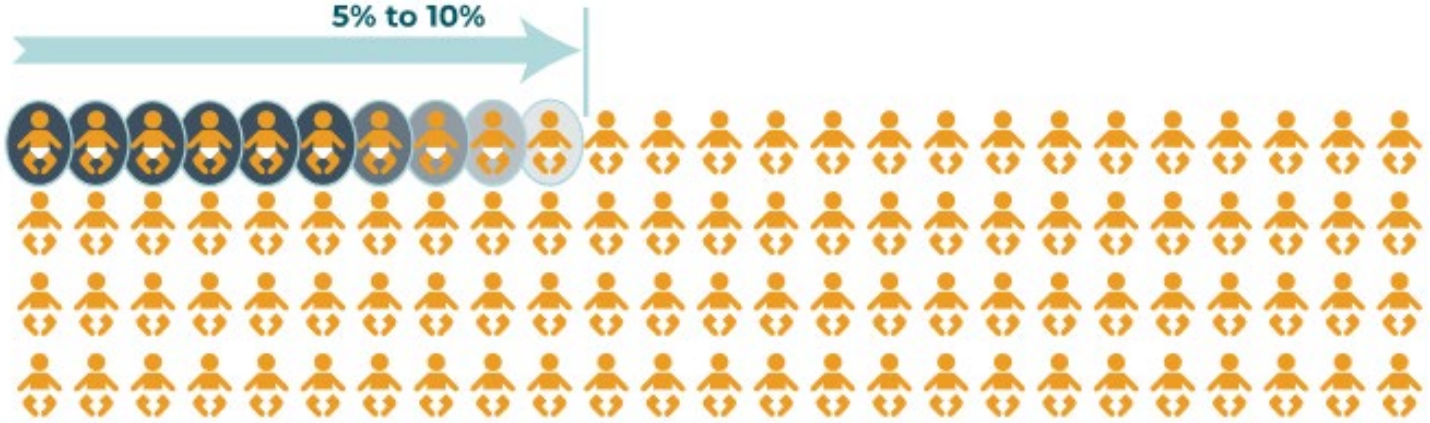
**For babies with this Higher Concern result, what is the risk for getting type 1 diabetes during their lifetime?**



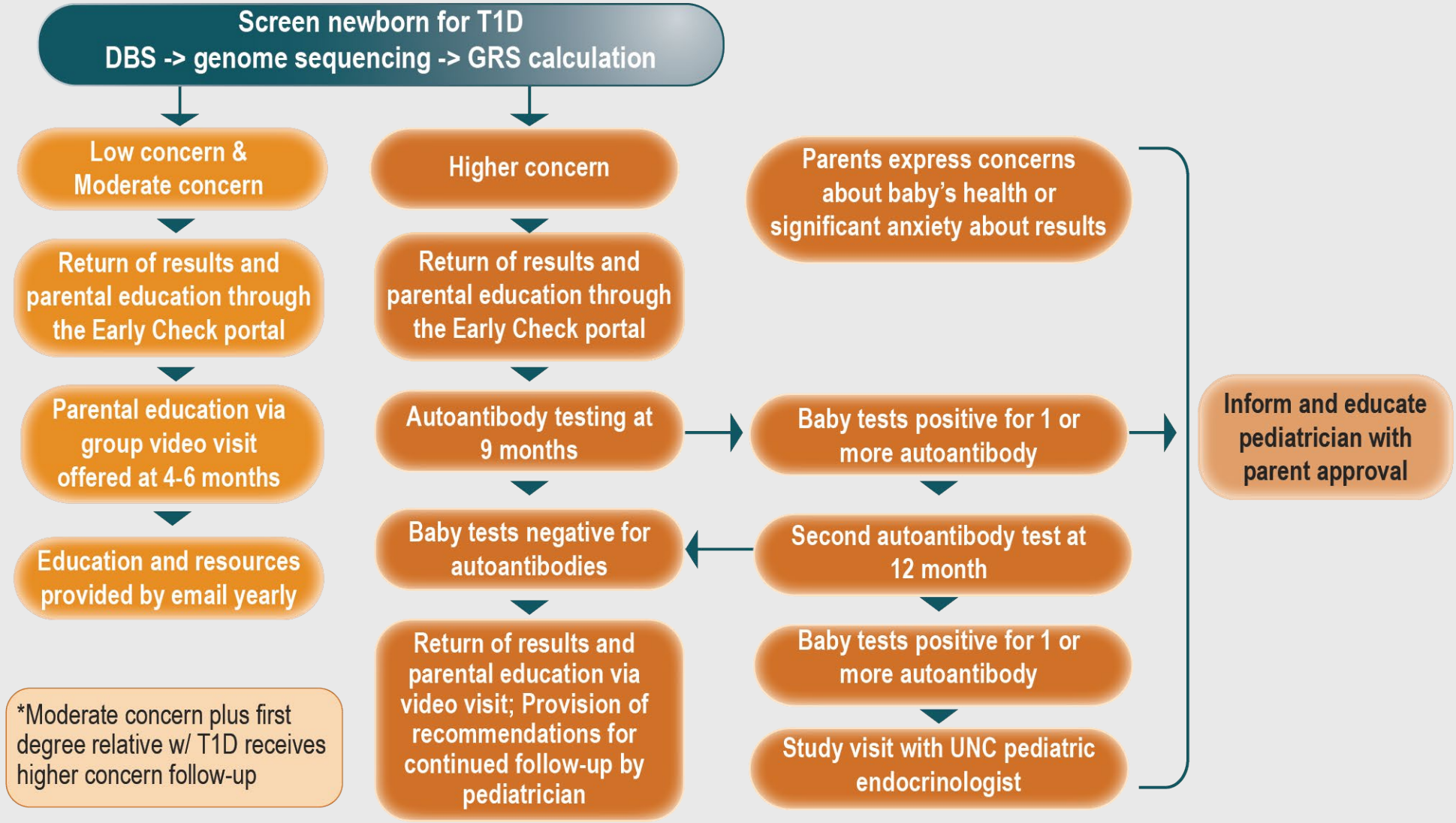

Your baby may be more likely to get T1D than usual. 5%-10% of babies with this result will develop type 1 diabetes during their lifetime.

**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

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Associate
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Lisa Gehtland
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Don Bailey
Senior Advisor

Anne Wheeler
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Methodology Core Lead

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UNC PI, Geneticist

Liz Jalazo
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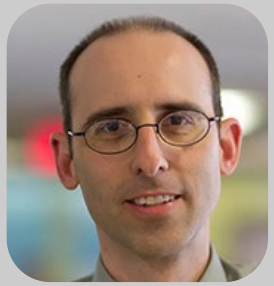

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Jennifer Law
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**NC State Laboratory of Public Health**

# Early Check Team


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Beth Boyea


Javan Carter


Emily Cheves


Heidi Cope


Manisha Dass


Jake Dibble


Anne Edwards


Ana Forsythe


Lisa Gehtland


Angela Gwaltney


Jessica Hunter


Kate Kucera


Colin Mathews


Brooke Migliore


Rebecca Moultrie


Casey Okoniewski


Grier Page


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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