

# Evaluating the Consequences of Detecting and Reporting Fragile X Carrier Status of Newborns

Don Bailey, Ph.D.

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# Assessing Benefit

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- Although there is no cure for FX, children with the full mutation are likely to experience a range of impairments that could be reduced, delayed, or prevented through early intervention



## What if....

- A relatively inexpensive test existed that could accurately screen newborns for FXS?
- This screening test was DNA based and accurately reported CGG repeat length?
- The test differentiated children with a full mutation from those with a premutation?
- You wanted to know whether disclosing carrier status was a good idea?



# Projected Number of Children Identified: 3500 births per year

<b>FX Status</b>	<b>Incidence</b>	<b>Total Per Year</b>	<b>Total for 5 Years</b>
<b>FM Male</b>	<b>1:2400-1:3500</b>	<b>0-1</b>	<b>2-3</b>
<b>FM Female</b>	<b>1:2400 – 1:6000</b>	<b>0-1</b>	<b>2-3</b>
<b>PM Male</b>	<b>1:290-1:800</b>	<b>2-5</b>	<b>9-26</b>
<b>PM Female</b>	<b>1:129-1:259</b>	<b>6-12</b>	<b>29-58</b>
<b>TOTAL</b>		<b>9-19</b>	<b>45-90</b>

## Would carrier children benefit from screening?

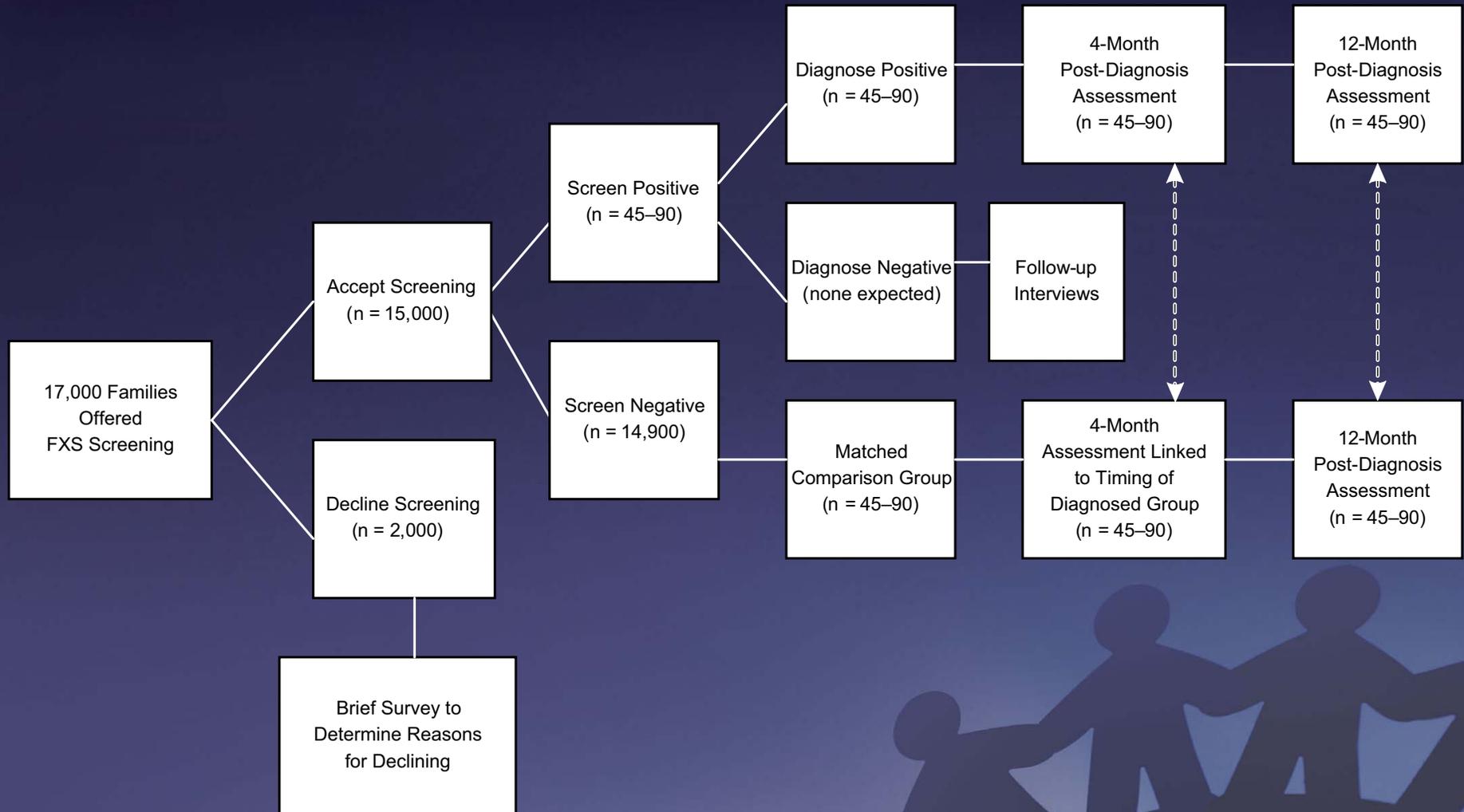
- Some carriers will be at risk for secondary conditions, and an important question is whether early identification could reduce or prevent these risks
- But data such as this would likely be insufficient for evaluating the desirability of disclosing carrier status, because of broader concerns



## How would you determine if disclosing FXS carrier status was a good idea?

- Ask parents who have children with FXS
  - Ask prospective parents and/or the general public what they think about it
  - Review the policy statements of professional organizations or the recommendations of various committees
  - Ask ethicists or state screening programs what they think about it
  - Examine research with other conditions
  - Take the accumulated inventory of benefits and risks and make a policy decision
  - Conduct a prospective study
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- A decorative graphic in the bottom right corner of the slide shows silhouettes of several people standing in a circle, holding hands. The silhouettes are dark blue against the lighter blue background of the slide.

# Study Design: Family Adaptation to NBS for FXS



# Specific Aims

- Aim 1: Determine the extent to which families from diverse cultural and ethnic groups consent to newborn screening for FXS, the reasons parents accept or decline screening, and family or socio-cultural factors related to screening decisions



## Specific Aims

- Aim 2: Assess the extent to which parents of identified children believe they were adequately informed about the possible results from screening, are initially satisfied with their decision to participate, and whether and how their views about screening change over time.



# Specific Aims

- Aim 3: Determine whether families experience adverse mental health outcomes following a diagnosis, and how adaptation varies as a function of child, family, and support variables
  - Stress
  - Depression
  - Anxiety
  - Hope
  - Quality of Life



# Specific Aims

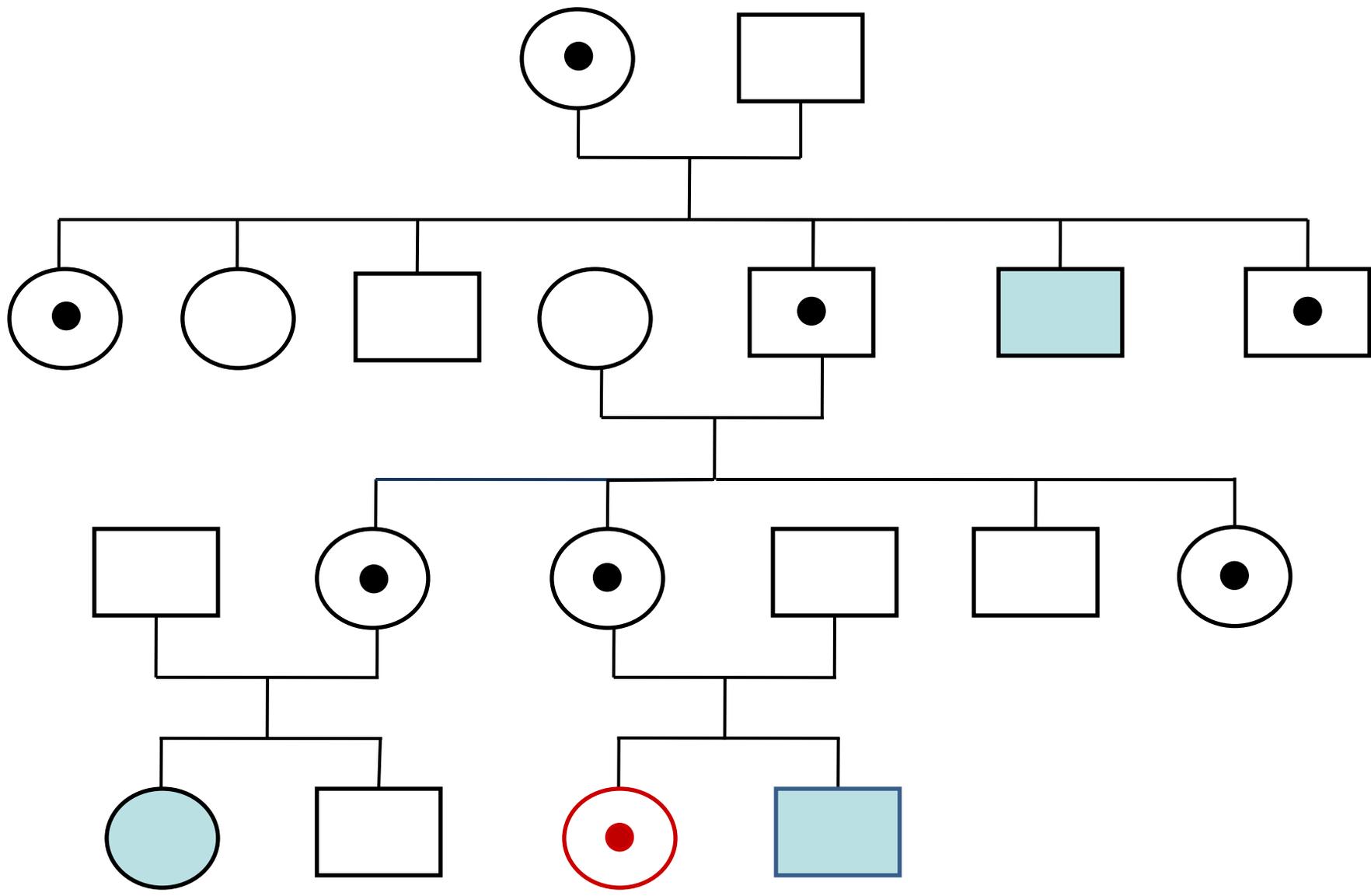
- Aim 4: Describe and explain variation in the quality and evolution of parent-child relations following the diagnosis of FXS
  - Expressed emotion and warmth
  - Positive affect
  - Maternal responsiveness



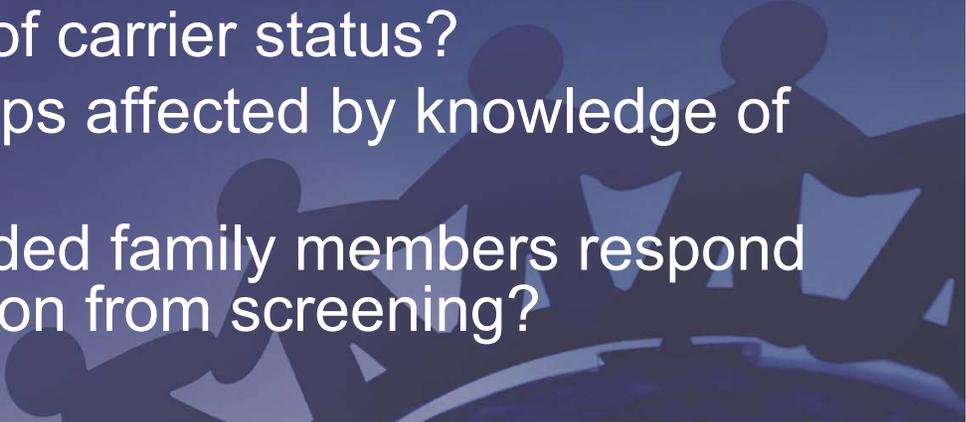
## Specific Aims

- Aim 5: Describe and explain variation in the ways that parents and extended family members respond to, share, and use information gained from a newborn diagnosis of FXS





# Questions to be Answered

- Do parents agree to have their children screened for FXS, knowing that carriers could be detected?
  - What are the reasons parents accept or decline screening, and are these reasons associated with socio-demographic variables?
  - Do families of identified children feel they were adequately informed about possible results?
  - Are families of identified children satisfied with their decision to participate?
  - To what extent do mothers suffer adverse mental health outcomes from disclosure of carrier status?
  - Are parent-child relationships affected by knowledge of FXS carrier status?
  - How do parents and extended family members respond to share, and use information from screening?
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# Summary

- Screening for full mutation FXS would likely benefit children and families
  - Screening for carriers evokes important questions about benefit – for whom and how
  - FXS is a good prototype to use in a prospective study of screening benefits
  - Rather than thinking strictly in terms of cost - benefit, we might need to assess family adaptation to information gained from screening
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- A decorative graphic in the bottom right corner of the slide shows silhouettes of several people of various ages holding hands in a circle, symbolizing a family or community.