

$$P(A|B) = \frac{P(B|A)P(A)}{P(B)}$$

# How a simple graph improves people' understanding of informed consents and their decision-making in a medical screening setting

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**CSCN**

**CENTRO DE NEUROCIENCIA  
SOCIAL Y COGNITIVA**

ESCUELA DE PSICOLOGÍA · UAI

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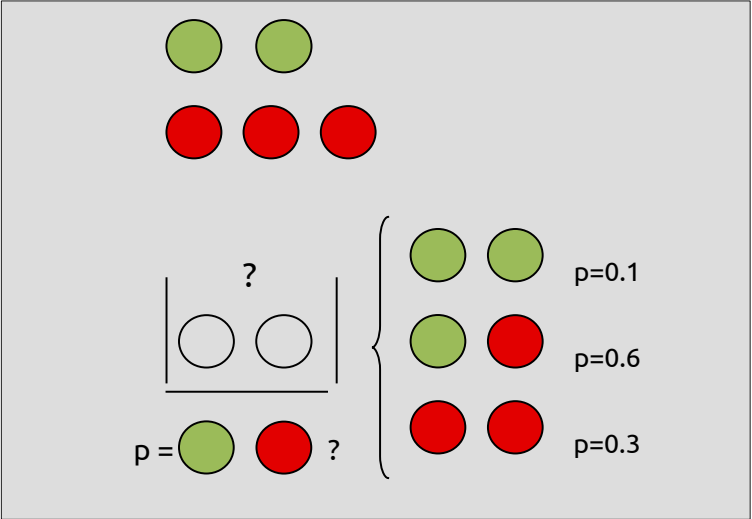
**Fondecyt**  
Fondo Nacional de Desarrollo  
Científico y Tecnológico

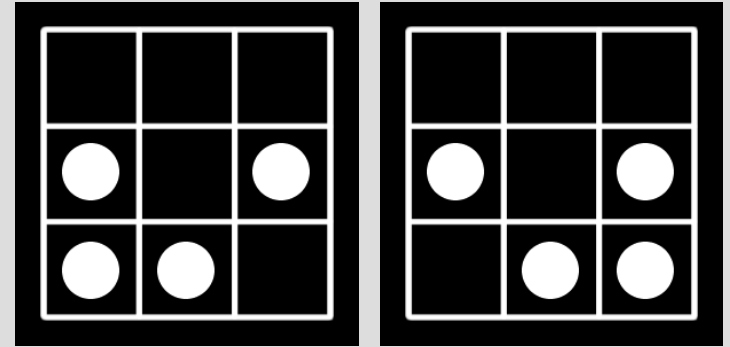
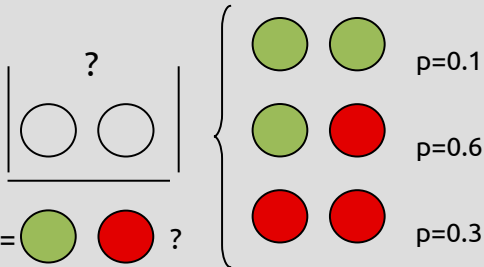
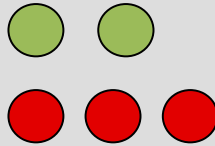
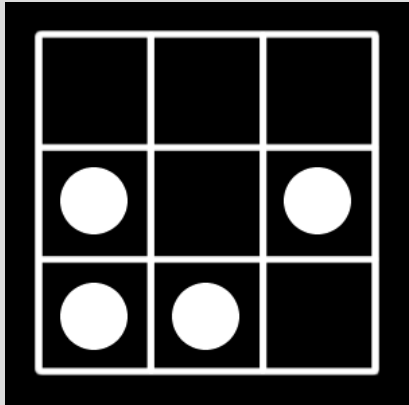
(2015-2016) Fondecyt Regular 1150824. Communication and interpretation of diagnostic tests: behavioral and physiological variables of Bayesian reasoning

(2017-2019) Fondecyt Regular 1171035. Towards a more integrated model of Bayesian reasoning: calculation, understanding and decision making in medical contexts



"Just a darn minute! — Yesterday  
you said that X equals two!"



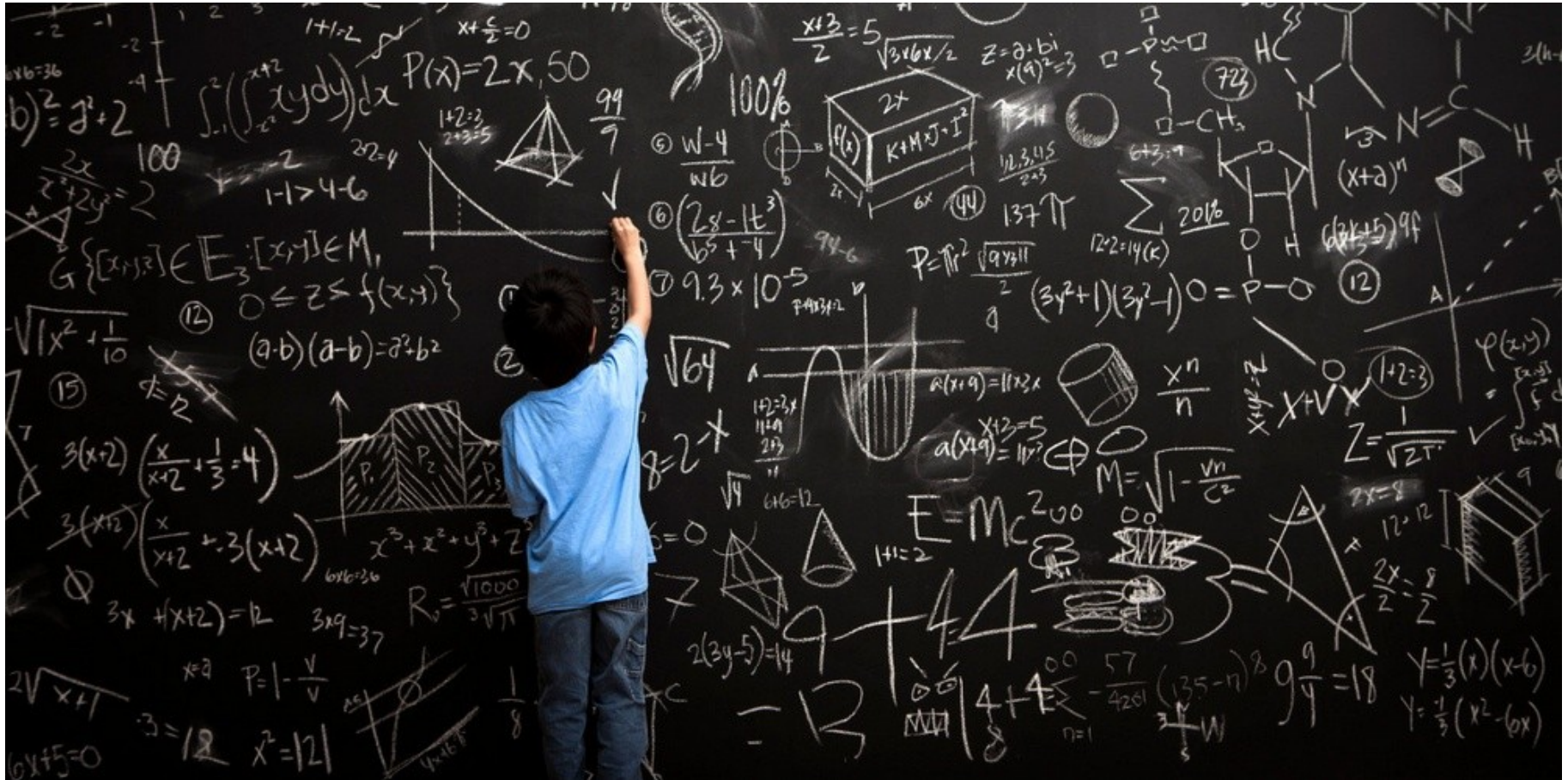


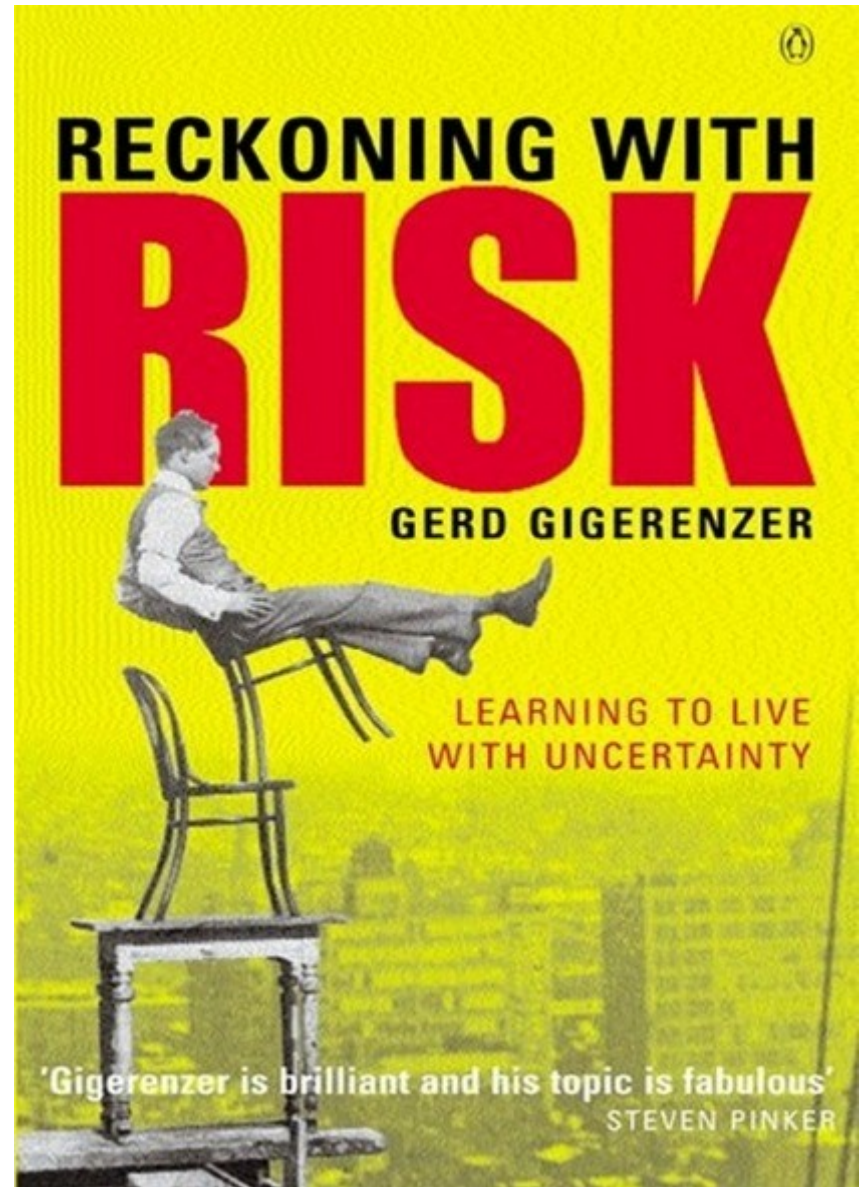
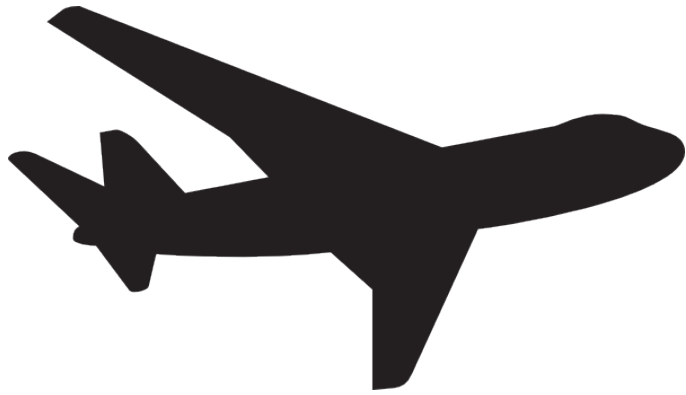


# High complexity

Low confidence (<50%)

High accuracy rate (>80%)





The probability of breast cancer is 1% for women aged forty who participate in routine screening.

If a woman has breast cancer, the probability is 80% that she will get a positive mammogram.

If a woman does not have breast cancer, the probability is 9.6% that she will also get a positive mammogram.

A woman in this age group has a positive mammogram in a routine screening. **What is the probability that she actually has breast cancer? \_\_\_\_\_ %**

# Classical Problem

■ Relative Probabilities



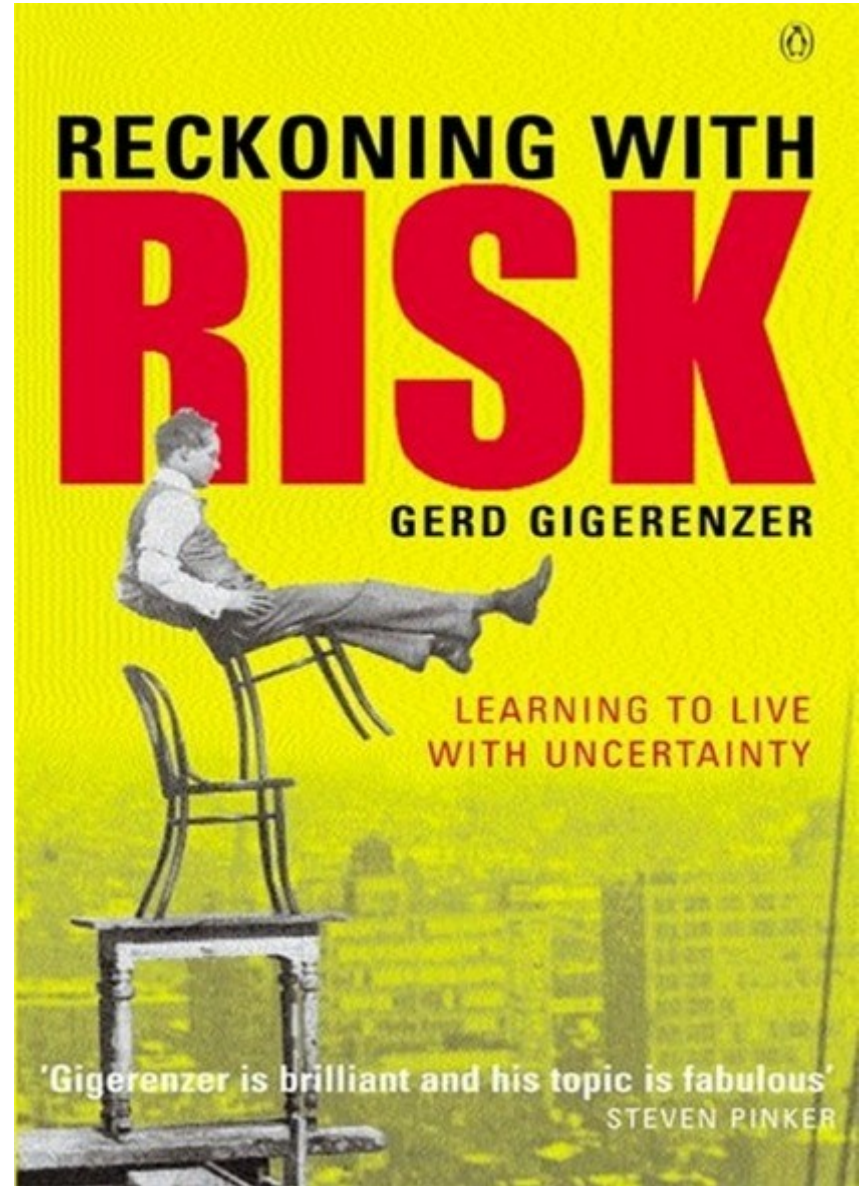
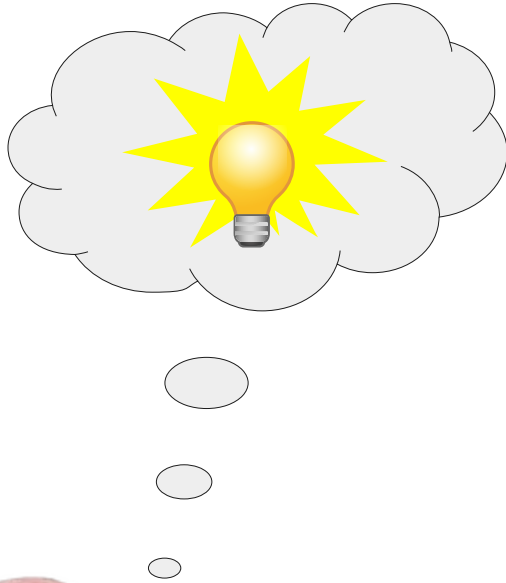
## High complexity

High confidence (>80%)

Low accuracy rate (~5%)







10 out of every 1,000 women at age forty who participate in routine screening have breast cancer.

8 of every 10 women with breast cancer will get a positive mammography.

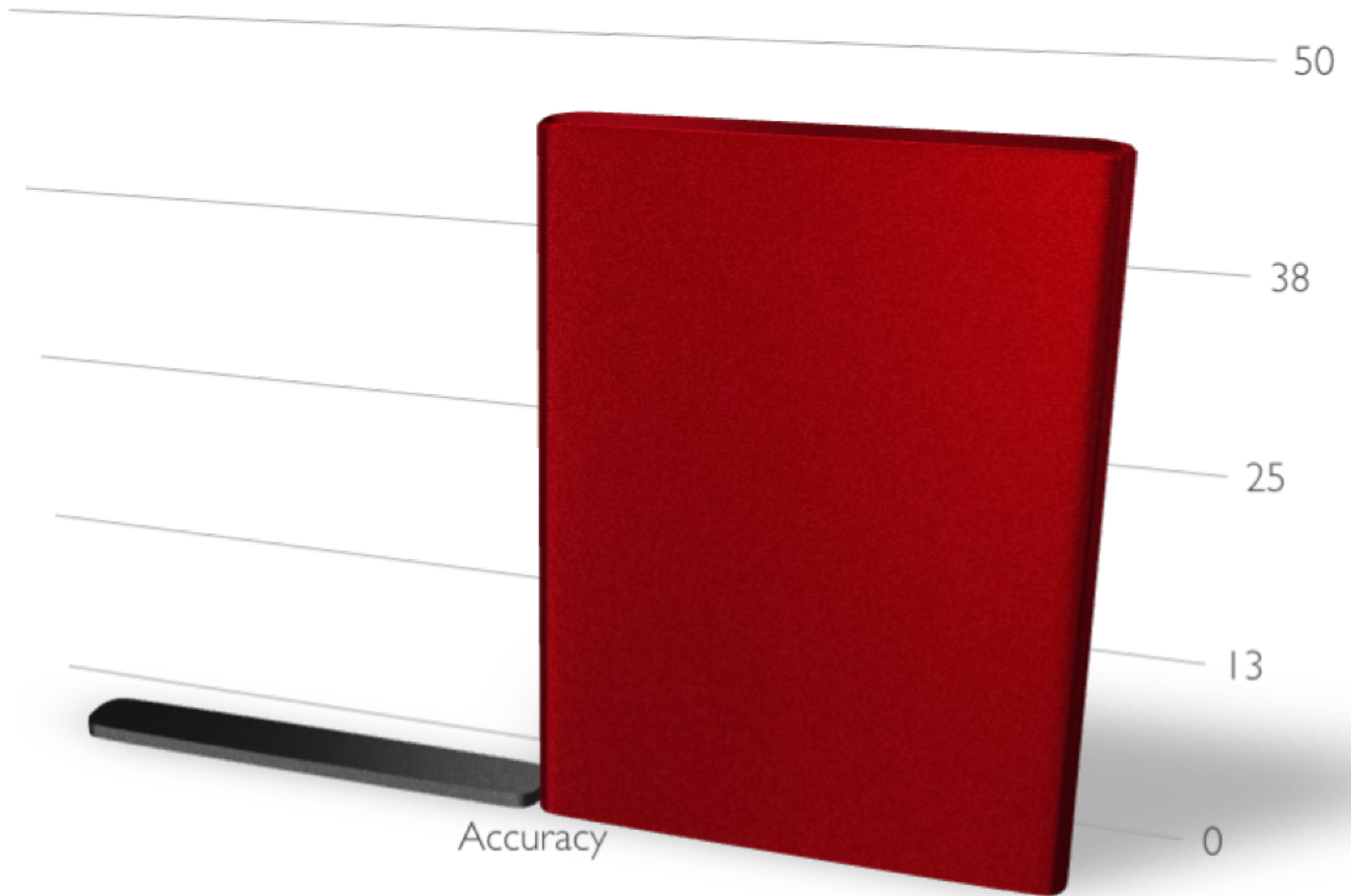
95 out of every 990 women without breast cancer will also get a positive mammography.

Here is a new representative sample of women at age forty who got a positive mammography in routine screening. **How many of these women do you expect to actually have breast cancer?** \_\_\_\_ out of \_\_\_\_

# Classical Problem

■ Relative Probabilities

■ Absolute Frequencies







$$p(H|D) = \frac{p(D|H)p(H)}{p(D)}$$



# Base Rate Neglect

# Unicorn Test

Hit rate: 100% (sensitivity)

False positives: 0,1% (1 - specificity)



# Massive screening

Type 2 diabetes

## Population A



## Population B



# Massive screening

Type 2 diabetes

## Population A



## Population A

Hit rate  
**100%**

False  
positives  
**0,1%**

Prevalence  
**1 out of 1000**

**$p(\text{Disease}|\text{+})?$**

<25%

25-49%

50%

51-75%

>75%

## Population A

Hit rate  
**100%**

False  
positives  
**0,1%**

Prevalence  
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**$p(\text{Disease}|\text{+})?$**

<25%

25-49%

50%

51-75%

>75%

1 out of 1000





Hit rate **100%**

1 out of 1000

False positives  
**0,1%**



PPV = 50%



Hit rate **100%**

1 out of 1000

False positives  
**0,1%**

$$p(H|D) = \frac{p(H) \cdot p(D|H)}{p(H) \cdot p(D|H) + p(\bar{H}) \cdot p(D|\bar{H})}$$

PPV = 50%



Hit rate **100%**

1 out of 1000

False positives  
**0,1%**

$$p(H|D) = \frac{p(H) \cdot p(D|H)}{p(H) \cdot p(D|H) + p(\bar{H}) \cdot p(D|\bar{H})}$$

PPV = 50%



Hit rate **100%**

1 out of 1000

False positives  
**0,1%**

$$p(H|D) = \frac{0.1\% \cdot 100\%}{0.1\% \cdot 100\% + 99.9\% \cdot 0.1\%}$$

PPV = 50%



Hit rate **100%**

1 out of 1000

False positives  
**0,1%**

$$\frac{\text{TRUE +}}{\text{TRUE + + FALSE +}} = \frac{\text{[green bar]}}{\text{[green bar] + [red bar]}}$$



# Massive screening

Type 2 diabetes

## Population B



Population B

Hit rate  
**100%**

False  
positives  
**0,1%**

Prevalence  
**500 out of 1000**

**$p(\text{Disease}|\text{+})?$**

<25%

25-49%

50%

51-75%

>75%

Population B

Hit rate  
**100%**

False  
positives  
**0,1%**

Prevalence  
**500 out of 1000**

**$p(\text{Disease}|+)?$**

<25%

25-49%

50%

51-75%

**>75%**

**(99,8%)**



500 out of 1000

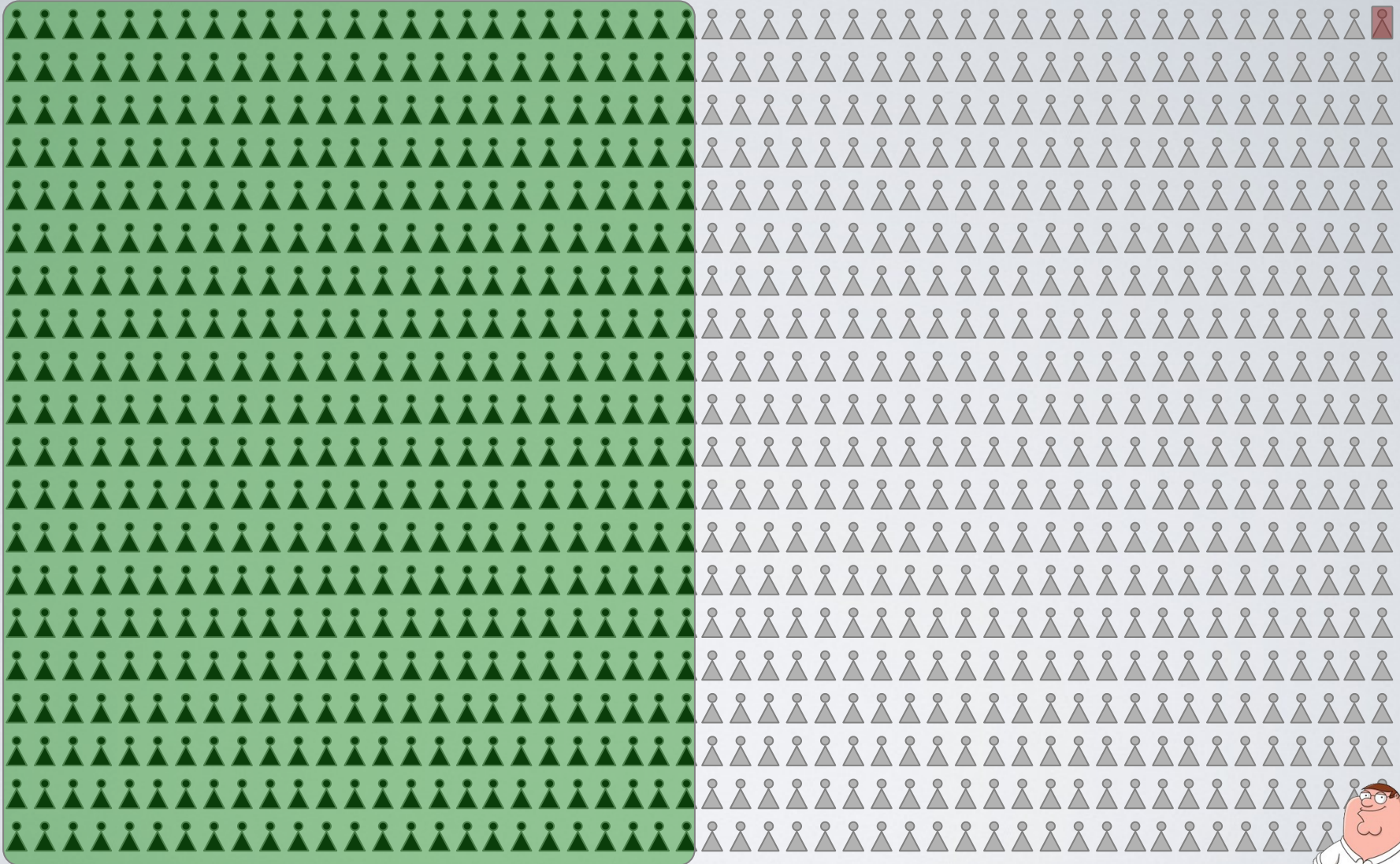


Hit rate **100%**

500 out of 1000

False positives

**0,1%**



PPV = 99.8%



Hit rate **100%**

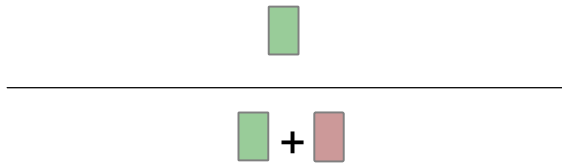
500 out of 1000

False positives  
**0,1%**

$$\frac{\text{TRUE} +}{\text{TRUE} + + \text{FALSE} +} = \frac{\text{[Large Green Box]}}{\text{[Large Green Box]} + \text{[Small Green Box]}}$$



# Population A



# Population B



+



Epidemiology 101:

**Screening tests , PPV and NPV**



**Screening tests** are to discard healthy people:

- NPV very high
- PPV, ... it depends, but who cares?

$$\text{PPV} = \frac{\text{TRUE } +}{\text{TRUE } + + \text{FALSE } +}$$

$$\text{NPV} = \frac{\text{TRUE } -}{\text{TRUE } - + \text{FALSE } -}$$

## We focus on **PPV**:

- Relevance for the individual
- Massive cost of False Positives
  - (e.g. 4 Billion US\$/year Breast cancer)
- Important for the decision:

screening test? → + → diagnostic test?

$$\text{PPV} = \frac{\text{TRUE +}}{\text{TRUE +} + \text{FALSE +}}$$

# Unicorn Test

In the real world



Hit rate: 100% (sensitivity)

False positives: 0,1% (1 - specificity)









Sensitivity = 100 %

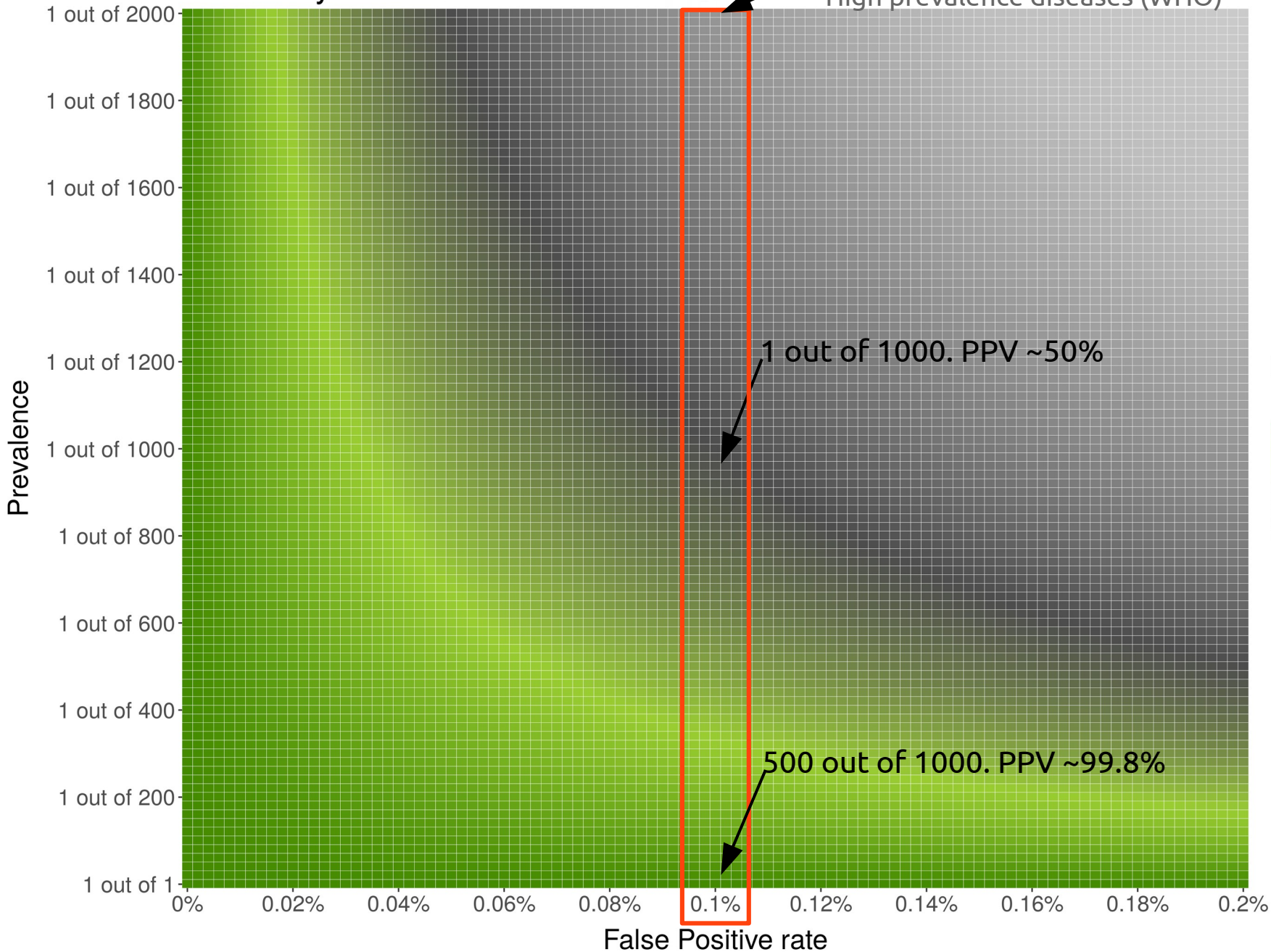
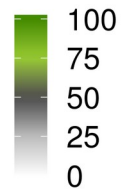


1 out of 2000. PPV ~33%  
High prevalence diseases (WHO)

1 out of 1000. PPV ~50%

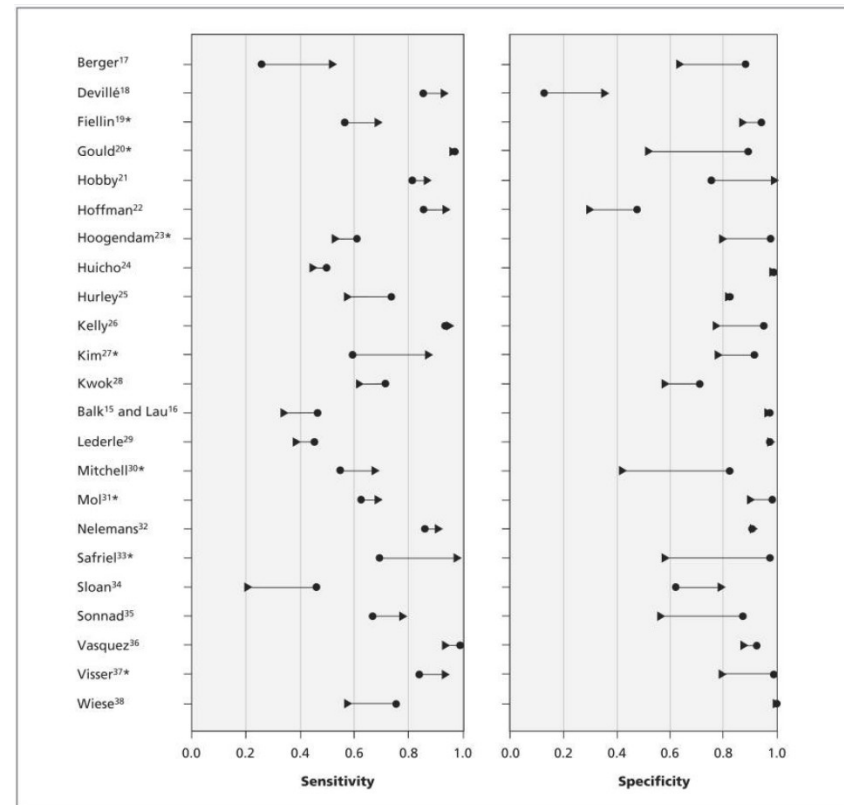
500 out of 1000. PPV ~99.8%

PPV (%)



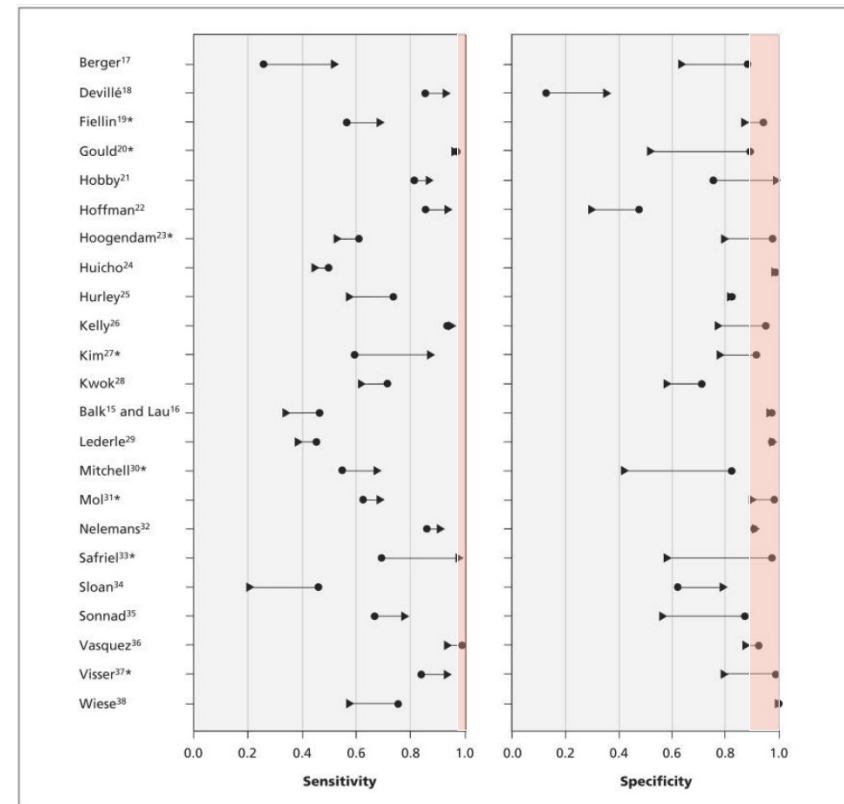
# Real world screening tests

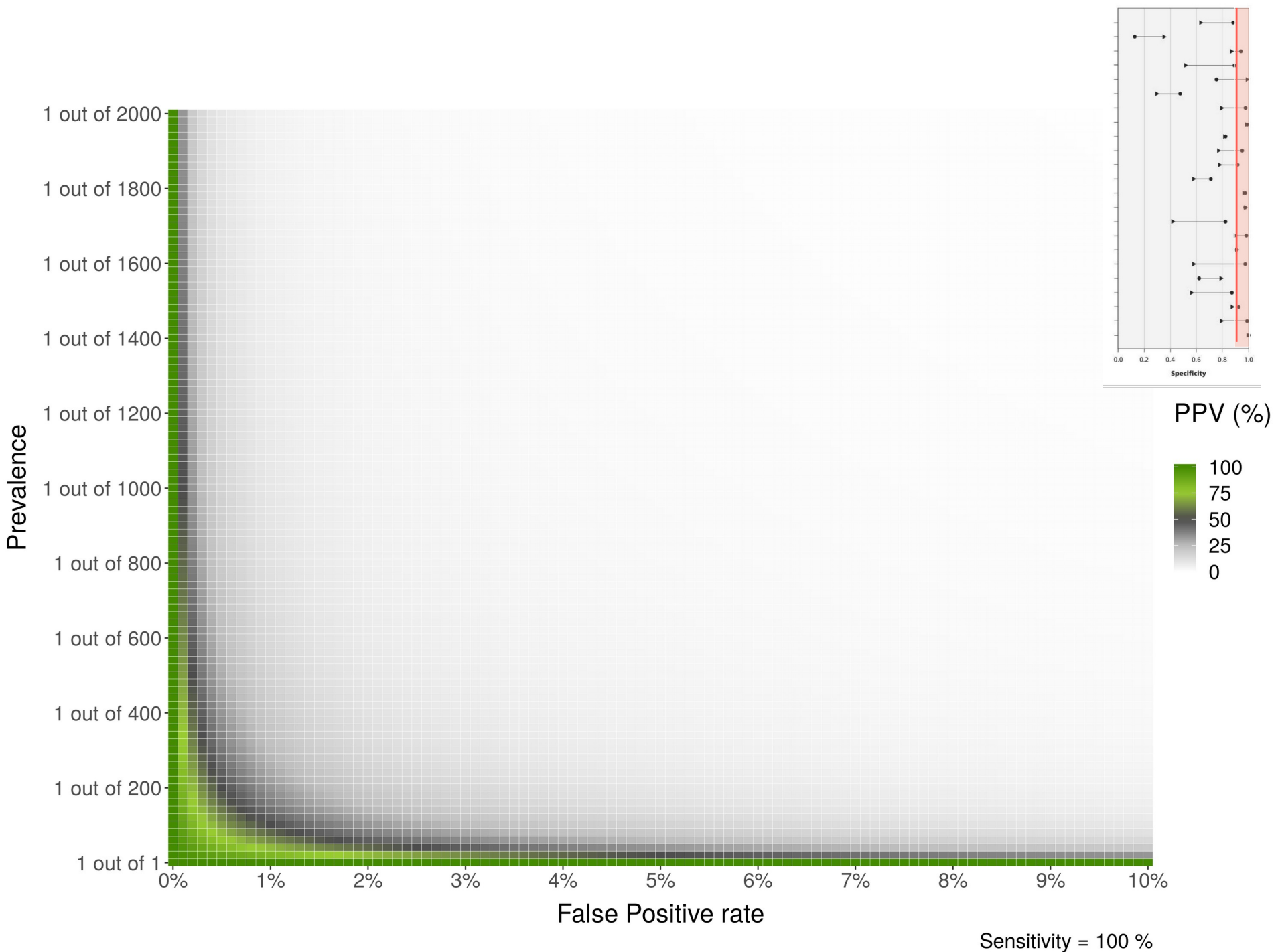
In real world diseases



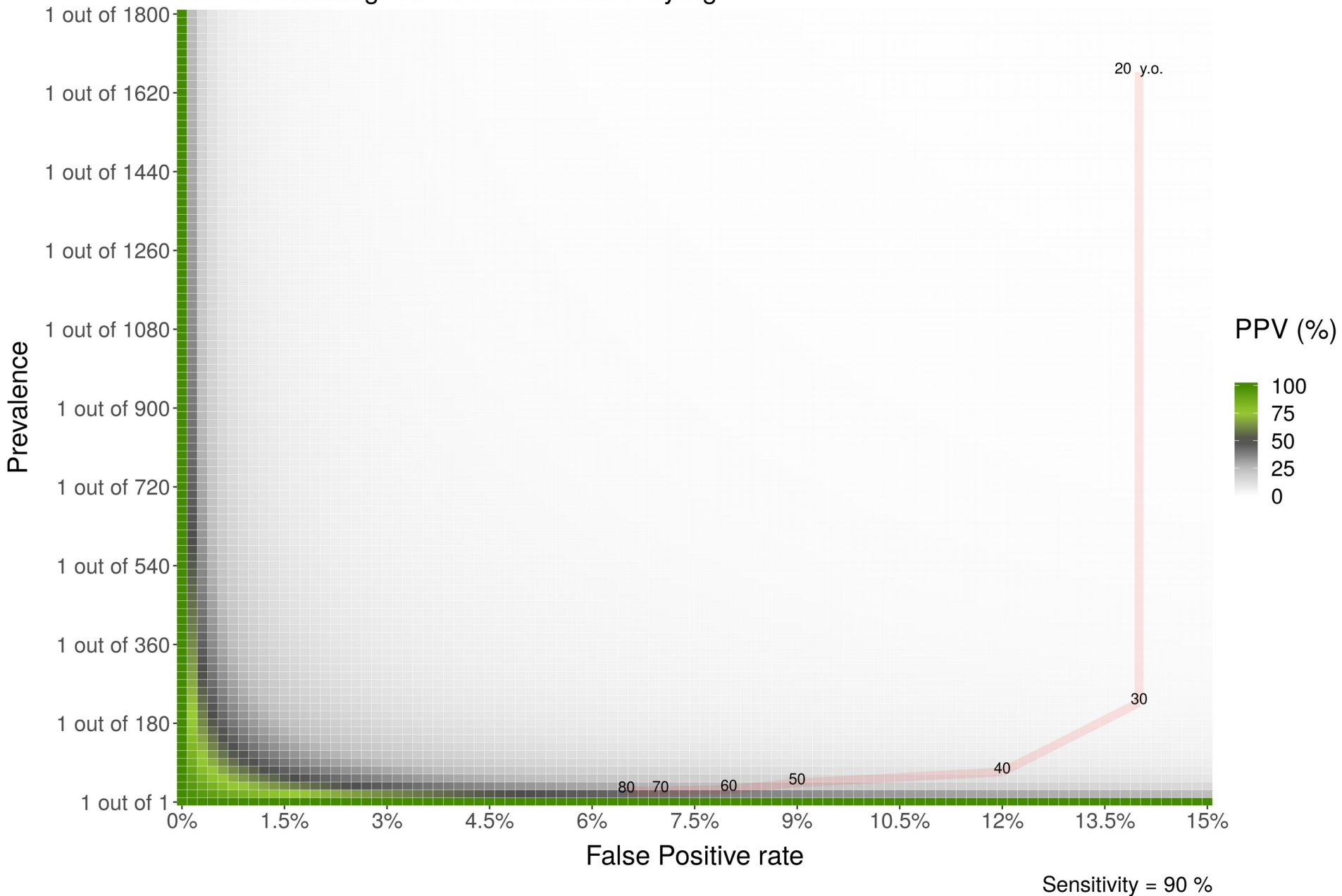
Sensitivity: 100%

False positives: 0 - 10% (1 - specificity)



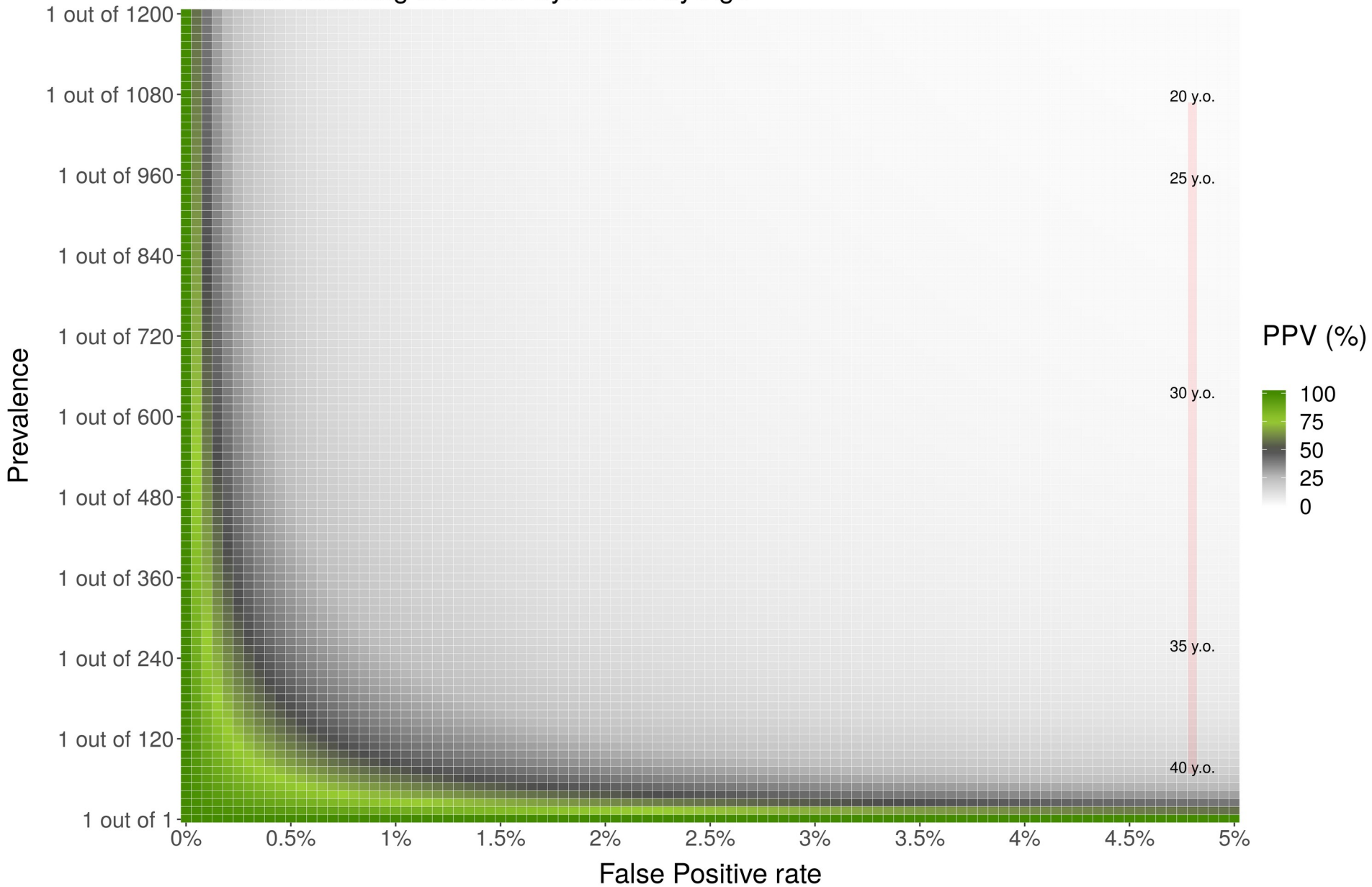


# PPV of Mammogram for Breast Cancer by Age





# Prenatal screening for Down Syndrome by Age



Sensitivity = 81 %





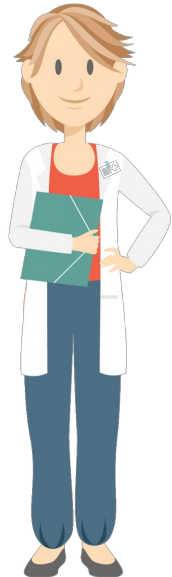
# Physicians

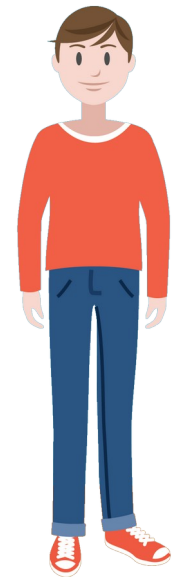
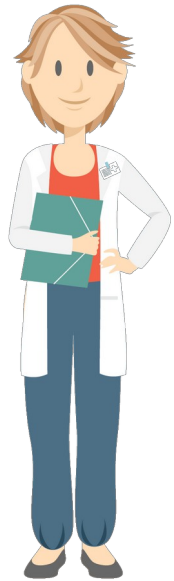
Probabilities

4 to 21% accuracy

Nat. Frequencies

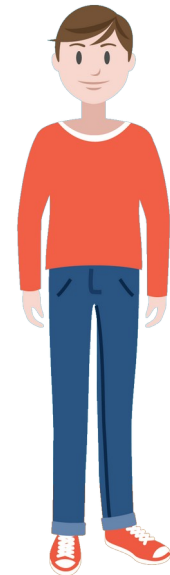
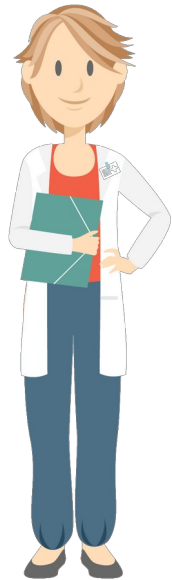
29 to 67% accuracy





Hit rate **100%**

False positives  
**0,1%**



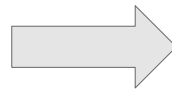
# Consent forms

Shared decision making





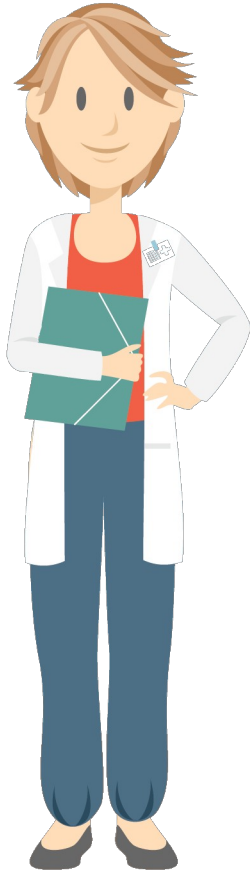
Information  
about the test  
and the medical  
condition



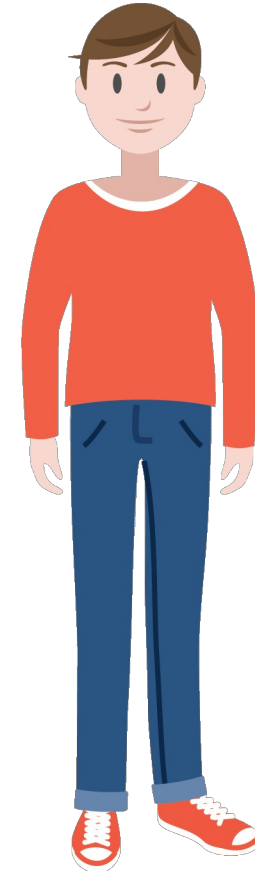
What's the  
meaning of a +  
result?



CLINICIAN



PATIENT



**SHARED DECISION MAKING**

Information & Recommendations

Values & Preferences





# How people deal with Consent Forms?



Service, and in exchange for service, all users of this site agree to immediately assign their first-born child to NameDrop, Inc. If the user does not yet have children, this

Service, and in exchange for service, all users of this site agree to immediately assign their first-born child to NameDrop, Inc. If the user does not yet have children, this

**1.7%**  
complained about the Child clause

# Consent forms

## Prenatal screening examples

A) Not enough information

B) Too much information

C) Consequences



A) Not enough information

B) Too much information

C) Consequences



TOSHIBA

20111010.094517.H.U.C.:20111010.094517.H.... F  
HOSP UNIV DE CANARIA... - - Semana 20

10/10/2011  
09:50:00 AM



0  
5  
10



T

20G  
79  
DR  
60

6C1  
15.0  
19 fps

#497  
IP6

HDD:89% Free

CINE REVIEW ▶



ABC



X

Prevalence

90%

Hit rate  
(sensitivity)

1%

False positives  
(1 – specificity)

Positive  
Predictive  
Value  
(PPV)

X

Prevalence

90%

Hit rate  
(sensitivity)

1%

False positives  
(1 – specificity)

Positive  
Predictive  
Value  
(PPV)

$$p(\text{Down syndrome}|+) = \frac{x \times 90\%}{(x \times 90\%) + ((1-x) \times 1\%)} = \frac{x}{x+y} = \sim [0..100]\%$$



A) Not enough information

B) Too much information

C) Consequences



# Harmony™ PRENATAL TEST



An advance in  
non-invasive fetal  
trisomy testing



## What is a Trisomy?

Humans have 23 pairs of chromosomes, which are strands of DNA and proteins that carry genetic information. A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

### TRISOMY 21

Trisomy 21 is due to an extra chromosome 21 and is the most common trisomy at the time of birth. Trisomy 21, also called Down syndrome, is associated with mild to moderate intellectual disabilities and may also lead to digestive disease and congenital heart defects. It is estimated that trisomy 21 is present in 1 out of every 800 births in Canada.<sup>1</sup>

### TRISOMY 18

Trisomy 18 is due to an extra chromosome 18. Trisomy 18, also called Edwards syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 18 often have congenital heart defects as well as various other medical conditions, shortening their lifespan. It is estimated that trisomy 18 is present in approximately 1 out of every 6,000 births.<sup>2</sup>

### TRISOMY 13

Trisomy 13 is due to an extra chromosome 13. Trisomy 13, also called Patau syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 13 usually have severe congenital heart defects and other medical conditions. Survival beyond the first year is rare. It is estimated that trisomy 13 is present in approximately 1 out of every 16,000 newborns.<sup>3</sup>

## Harmony™ Prenatal Test

**Simple, safe and accurate for you and your pregnancy.**

**The Harmony Prenatal Test is a non-invasive test that detects common fetal trisomies in pregnancies of 10 weeks or more, based on directed analysis of DNA in maternal blood.**

### WHAT WILL THE HARMONY PRENATAL TEST TELL ME AND MY HEALTHCARE PROVIDER?

The Harmony Prenatal Test assesses the risk of three fetal trisomies by measuring the relative amount of chromosomes in maternal blood.

### HOW IS THE HARMONY PRENATAL TEST DIFFERENT FROM OTHER PRENATAL TESTS?

The Harmony Prenatal Test is based on the newest advances in non-invasive prenatal testing. It is a simple and safe blood test that has been shown in clinical studies to detect the risk of fetal trisomies with high accuracy.<sup>4,5</sup>

The Harmony Test has been shown to have detection rates of up to 99% and false positive rates as low as 0.1% for trisomy 21, 18 and 13.<sup>4,5</sup> Diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) are accurate for detecting fetal trisomies, but they are invasive and pose a slight risk for fetal loss.<sup>4,5</sup>

<sup>1</sup>Canadian Down Syndrome Society

<sup>2</sup>Ontario Ministry of Health and Long-Term Care

<sup>3</sup>U.S. National Library of Medicine

<sup>4</sup>Norton, M., et al. *Am J Obstet Gynecol.* (2012), doi:10.1016/j.ajog.2012.05.021

<sup>5</sup>Ashoor, G., et al. *ULTRASOUND Obstet Gynecol.* (2012), doi:10.1002/uog.12299.

Harmony

Cost\*

\$795.00

\* Subject to change without notice.

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\* The information provided about sensitivity and specificity are probably an overestimation...



**Harmony™  
PRENATAL TEST**

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<sup>1</sup>Journal Down Syndrome Society  
<sup>2</sup>Journal American Journal of Obstetrics and Gynecology  
<sup>3</sup>Journal Prenatal Diagnosis  
<sup>4</sup>Journal W. et al. in: Obstet Gynecol 2010; 115: 113-119  
<sup>5</sup>Journal S. et al. in: Obstet Gynecol 2010; 115: 113-119



$$p(\text{Trisomy 21} \mid + \text{test}) =$$

$$(A) \frac{1 \text{ out of } 800 \times 99\%}{(1 \text{ out of } 800 \times 99\%) + (799 \text{ out of } 800 \times 0.1\%)} =$$

$$(B) \frac{0.123}{0.123 + 0.0998} = 0.55$$



What's the meaning  
of a + result?

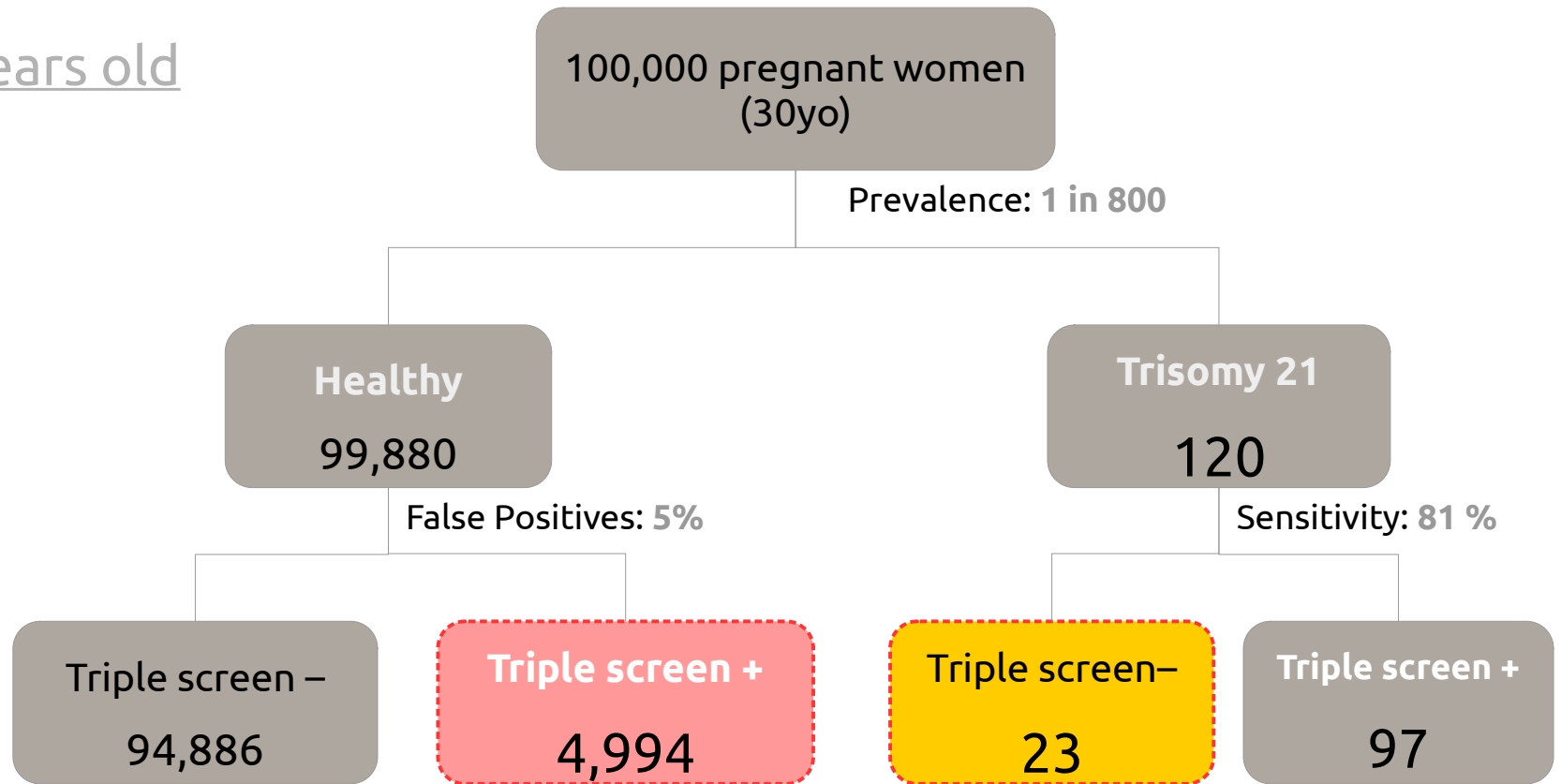
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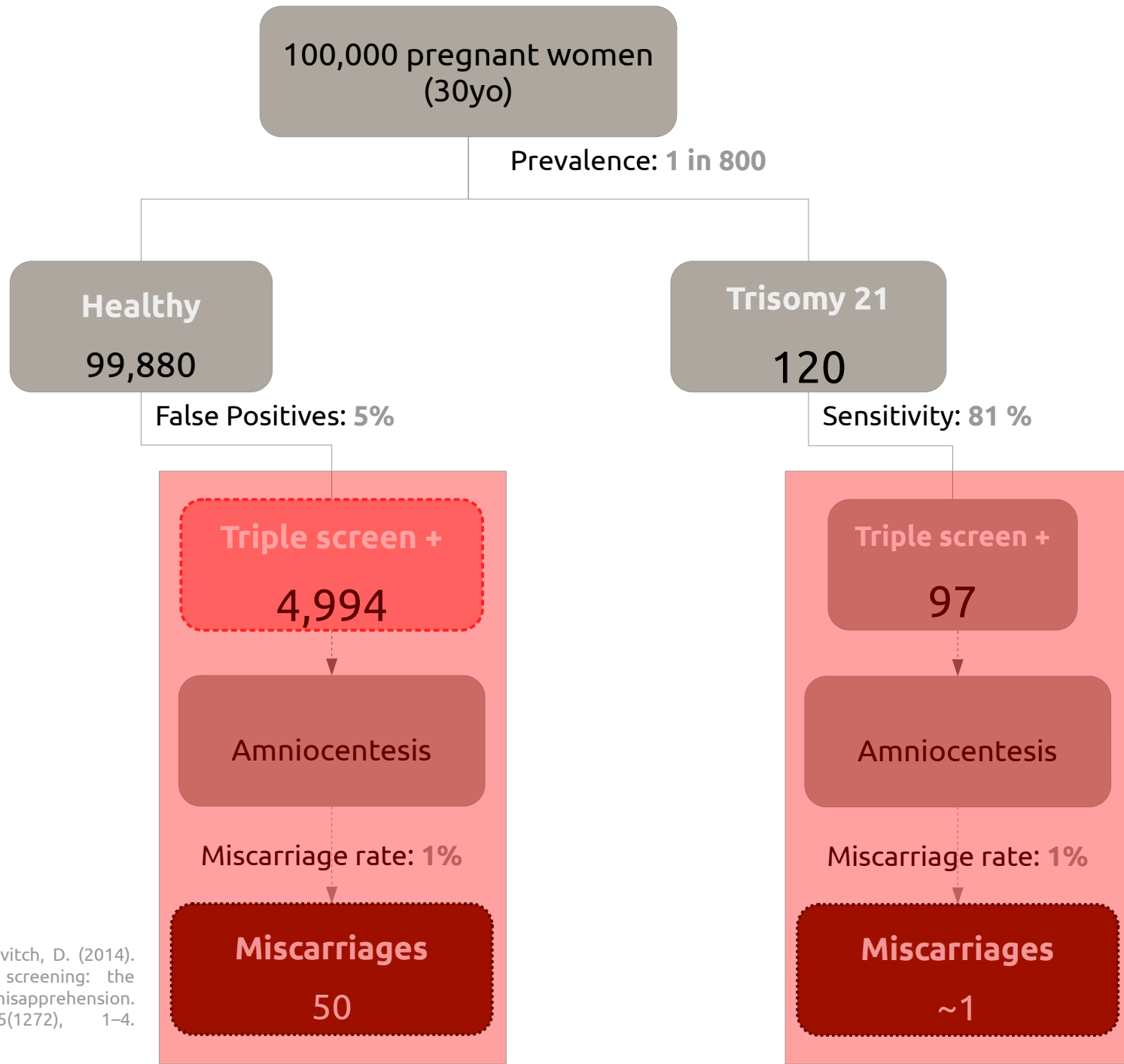


30 years old



PPV (21): ~1.60%  
PPV (18): ~0.48%  
PPV (13): ~0.32%

30 years old









OLD RESEARCH!



# Cognitive mechanisms behind the comprehension of Bayesian reasoning problems

Arithmetic complexity

Individual differences

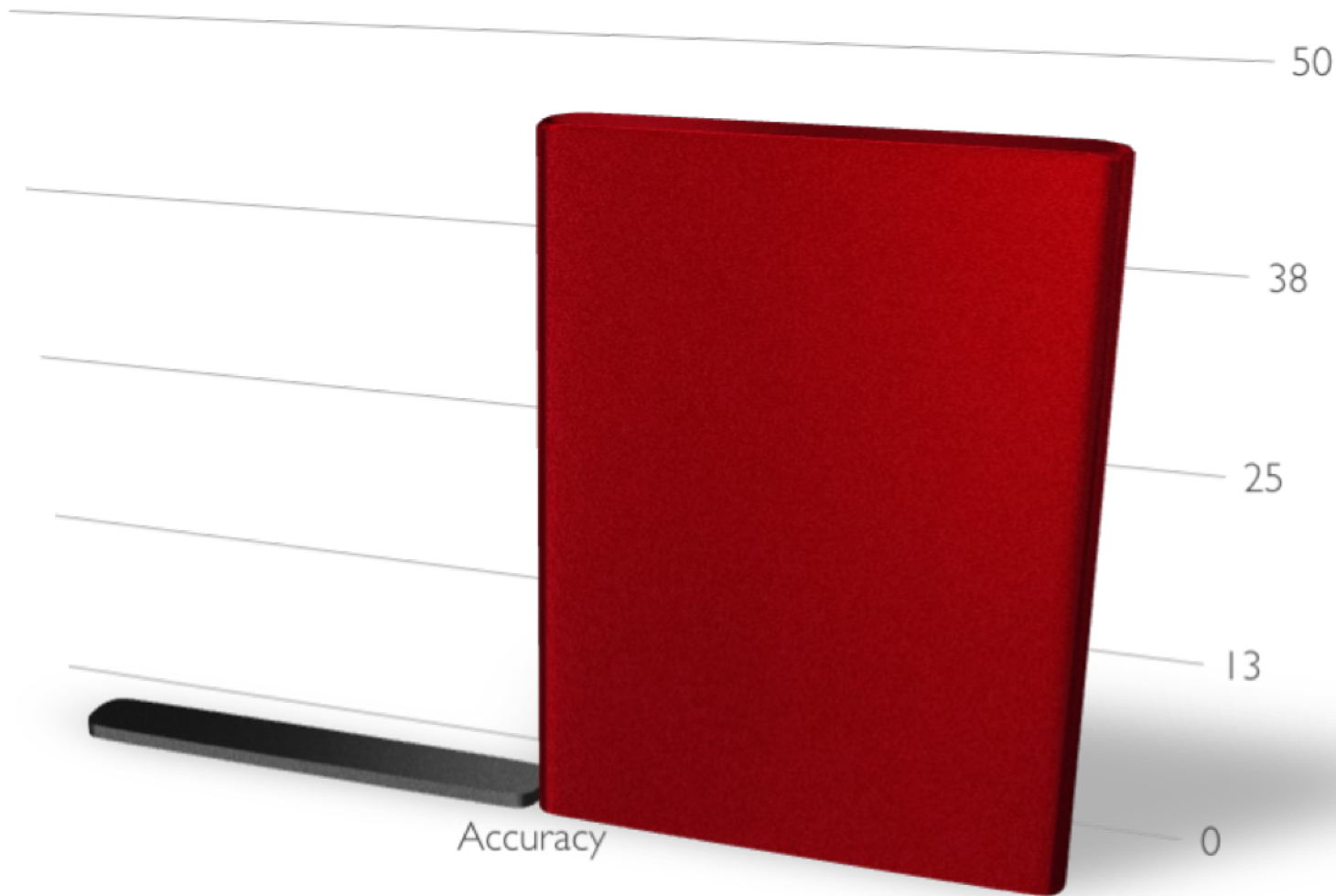
Reference class

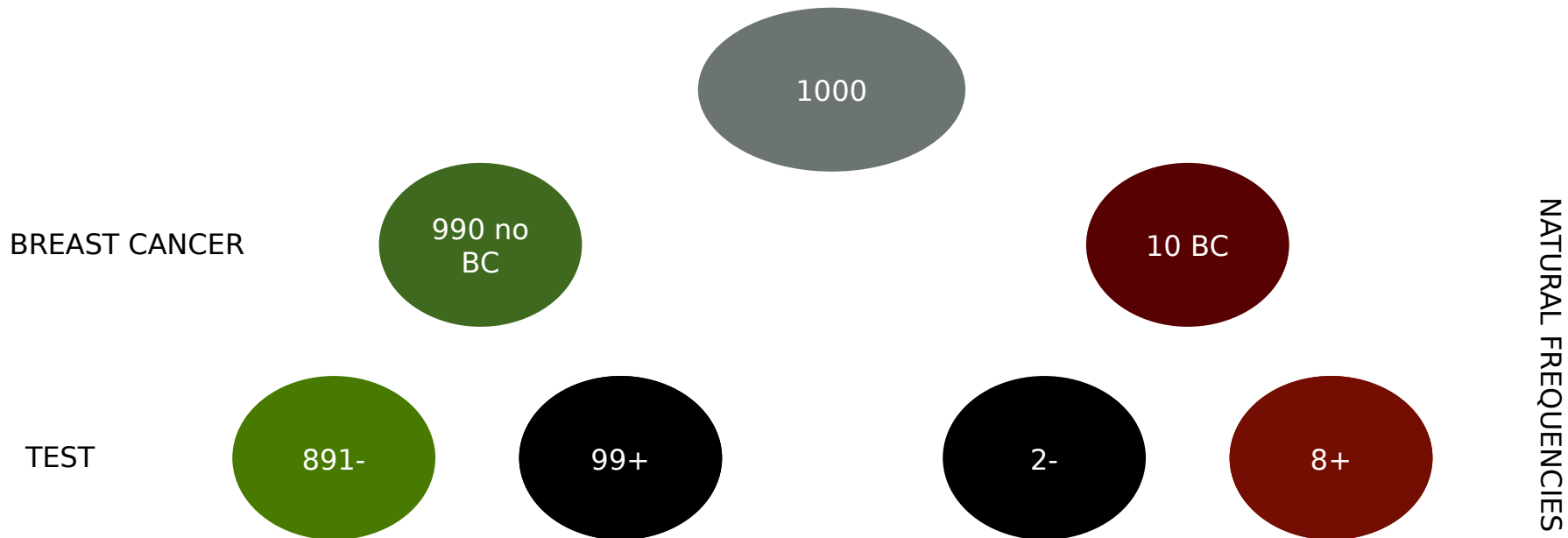
# Arithmetic complexity

# Classical Problem

■ Relative Probabilities

■ Absolute Frequencies





Take, for example, a sample of women who have positive mammograms. How many of these women actually have breast cancer?

$$p(\text{Disease}|\text{Symptom}) = \frac{8+}{99+ + 8+}$$

BREAST CANCER

99% no  
BC

100%

1% BC

TEST

90% -

10% +

20% -

80% +

PROBABILITIES

Take, for example, a sample of women who have positive mammograms. What is the probability that she actually has breast cancer?

$p(\text{Disease}|\text{Symptom}) =$

80% +

\*

1% BC

10% +

\*

99% no  
BC

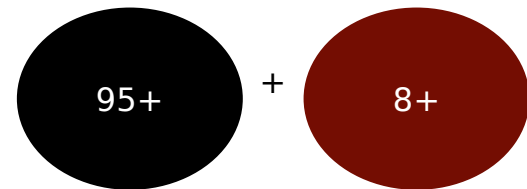
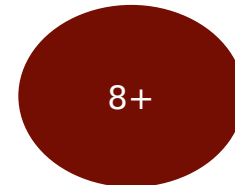
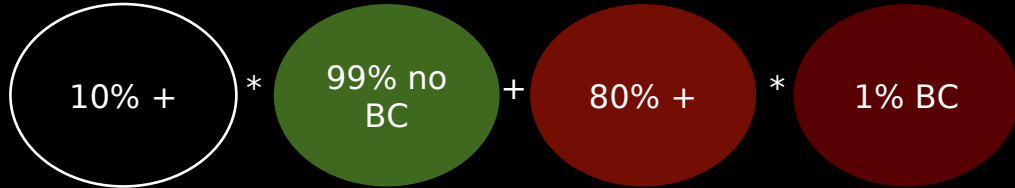
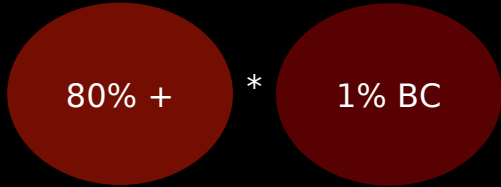
+

80% +

\*

1% BC

NATURAL FREQUENCIES

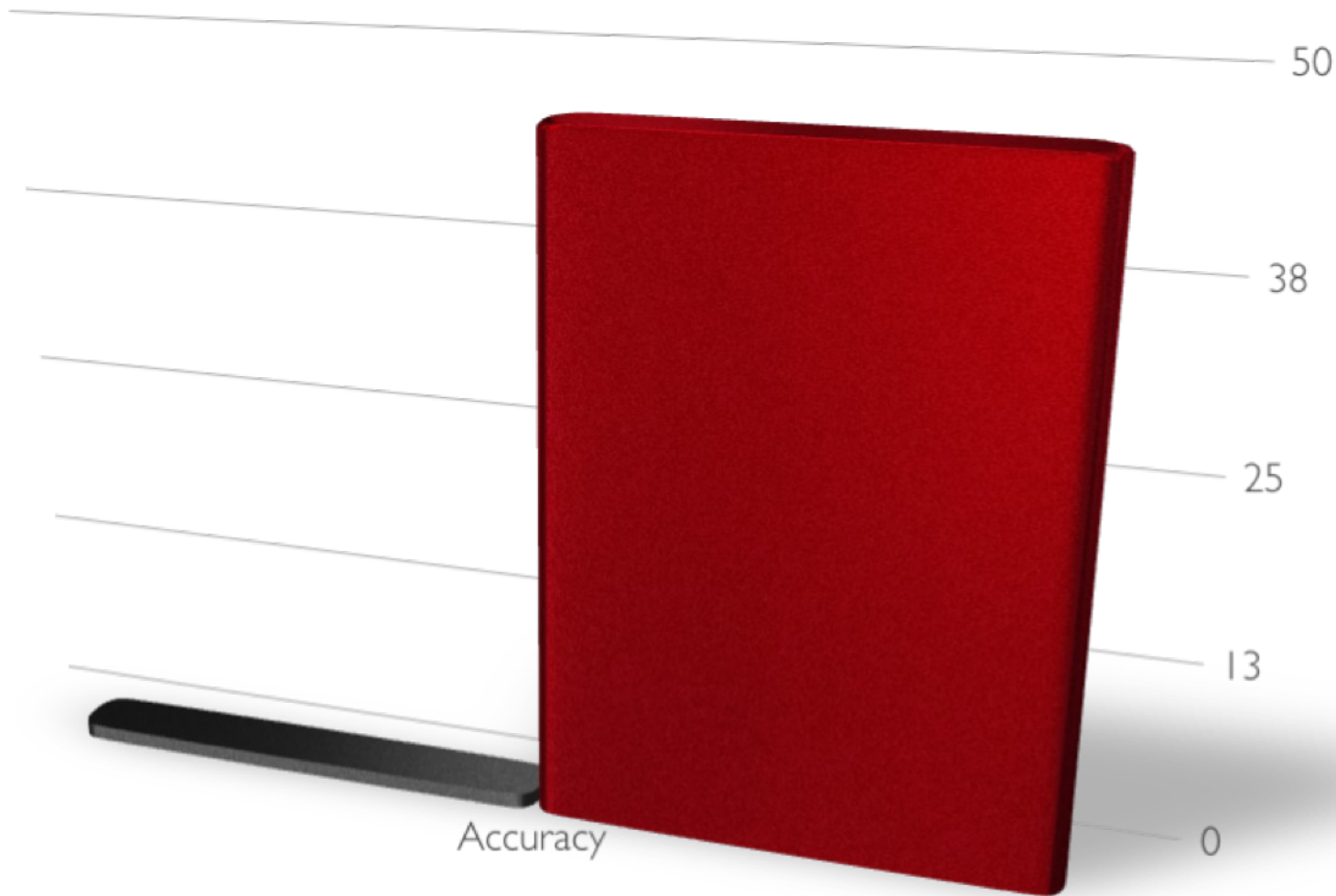


PROBABILITIES

# Classical Problem

■ Relative Probabilities

■ Absolute Frequencies





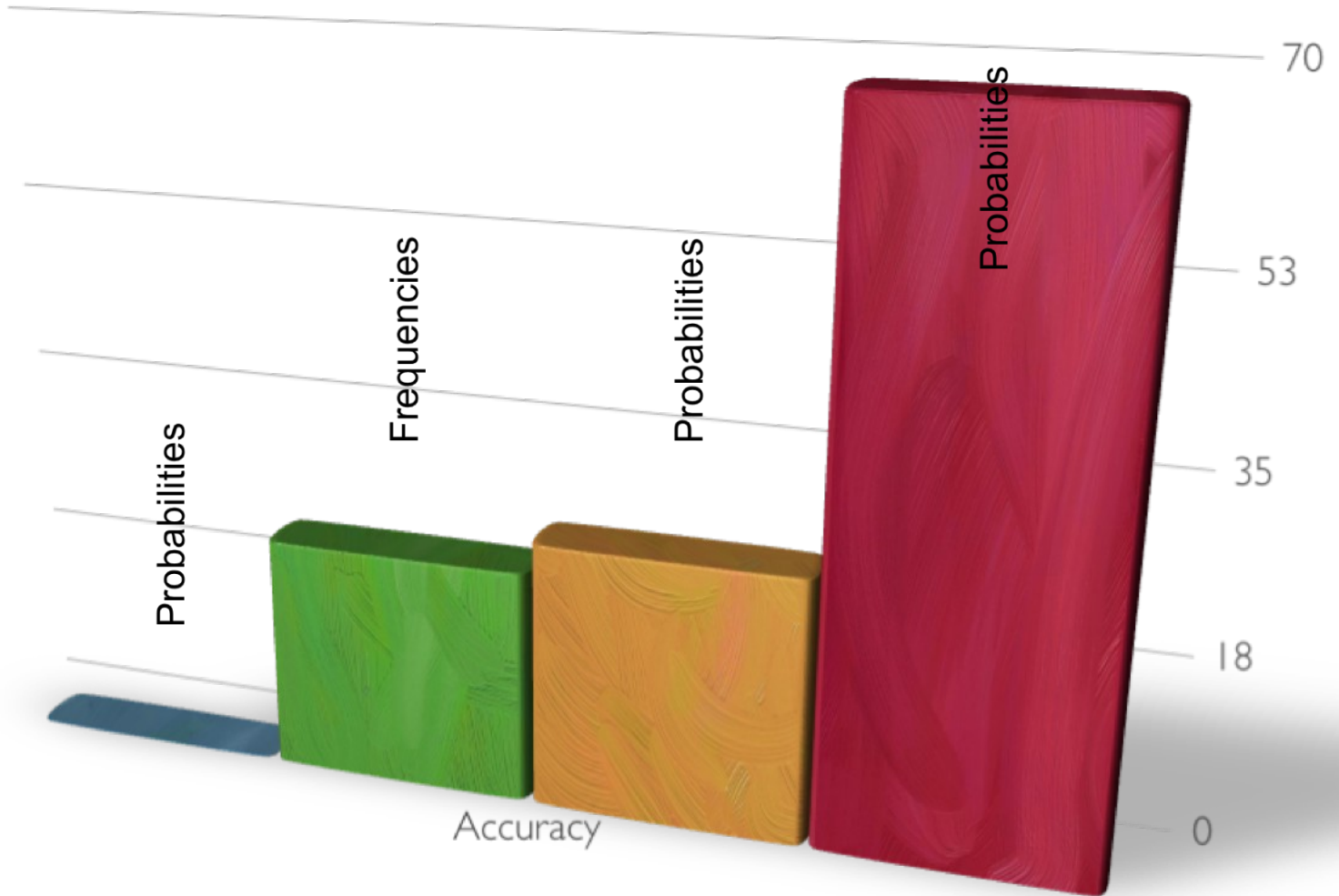
# Arithmetic Complexity

■ AC = 4

■ AC = 2

■ AC = 2

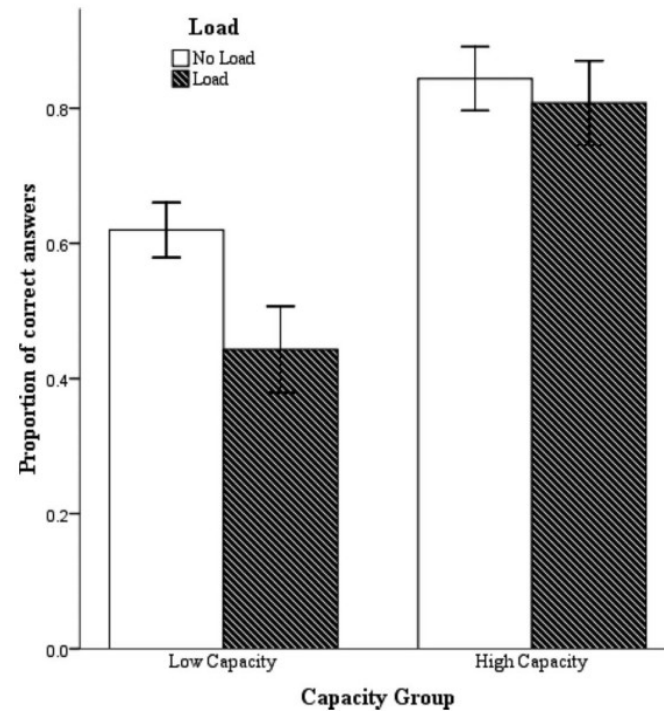
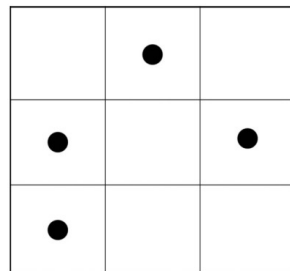
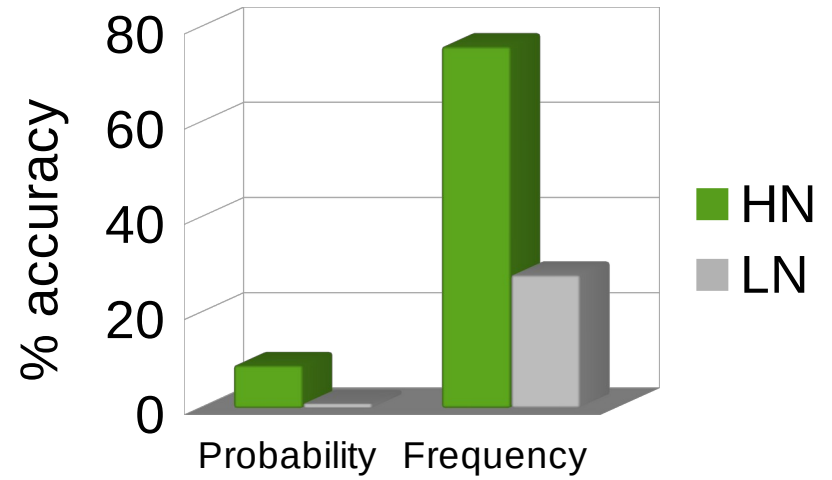
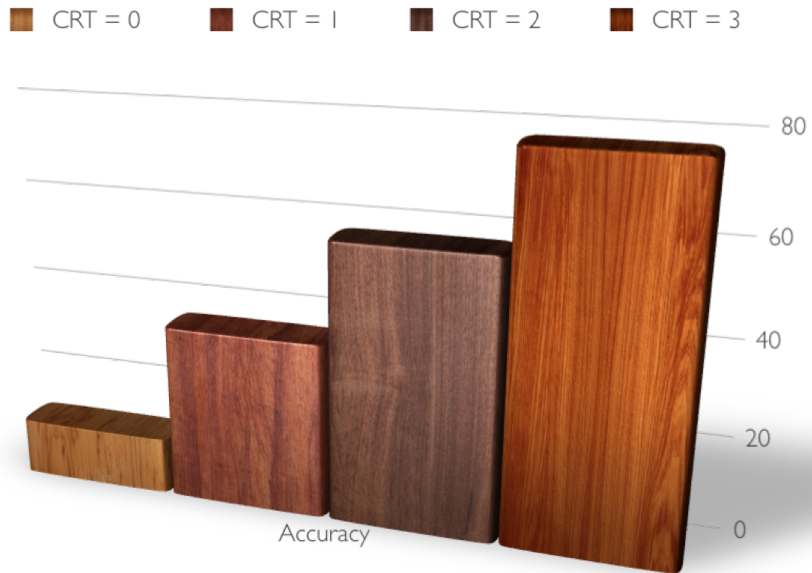
■ AC = 1

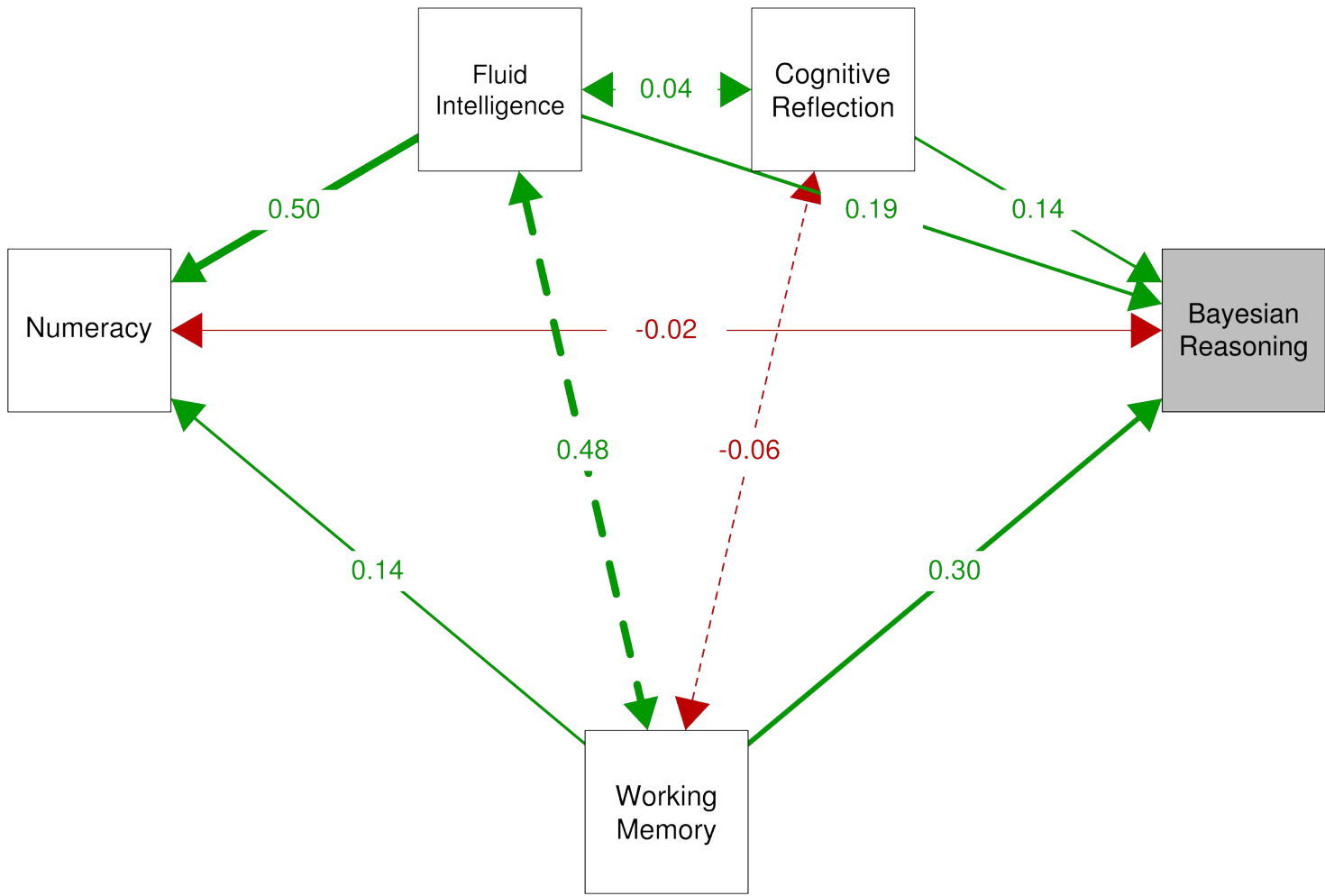


# Individual differences

Cognitive Reflection, Numeracy, WM

Absolute Natural Frequencies





Cognitive mechanisms behind the  
comprehension of Bayesian  
reasoning problems



Improve medical risks communication

# Presentation format

University students

# Presentation format x Response type

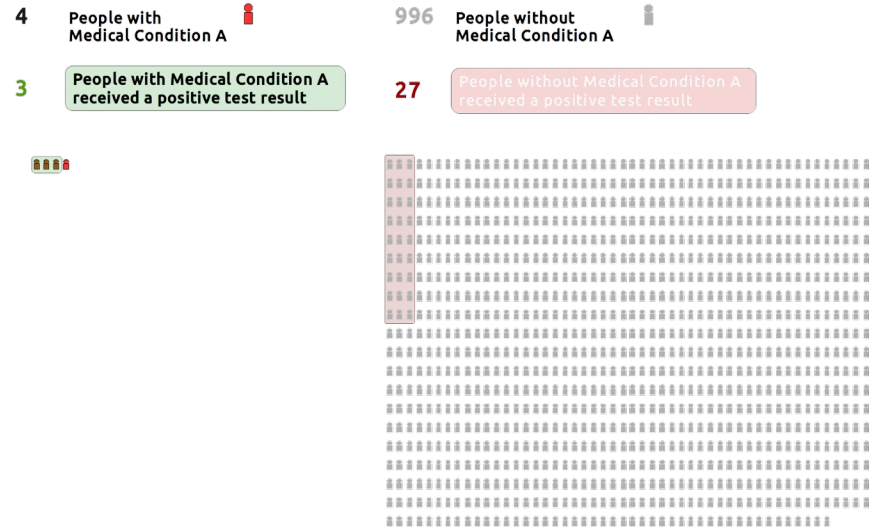
Vulnerable population

# Presentation format

University students

Imagine that a group of 1000 people have participated in a medical screening to detect disease A. The following information is available about the screening.

4 in every 1000 people actually have the disease A.  
 3 in every 4 person with the disease A has received a positive screening test result.  
 27 in every 996 people without the disease A have also received a positive screening test result.



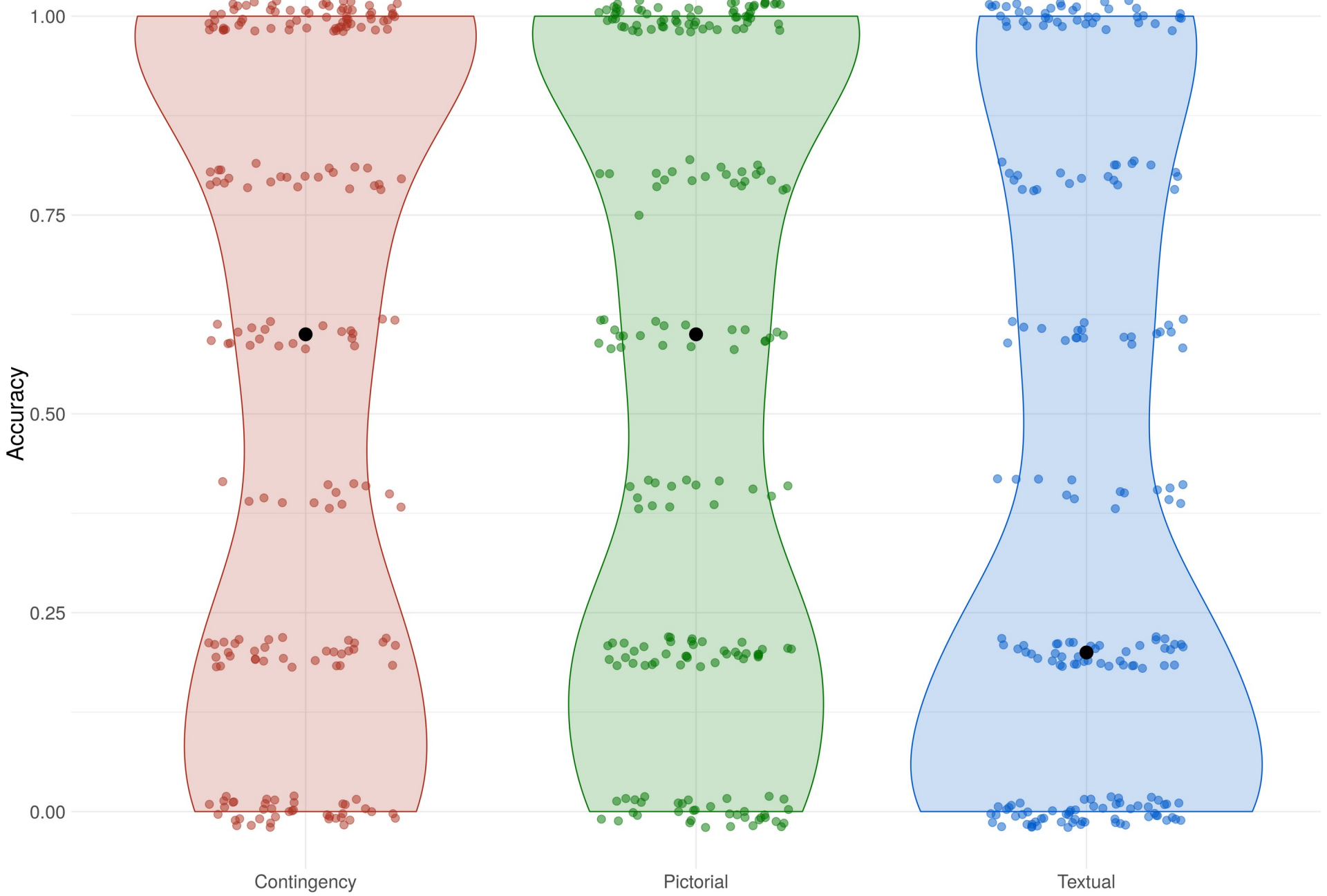
	Received positive screening test result	Received negative screening test result	1000 people
With Disease A	3	1	4
Without Disease A	27	969	996

Of the people who have received a positive screening test result, how many would you expect to have the disease A? \_\_\_\_ out of \_\_\_\_



# Accuracy by presentation type

±5% of correct response

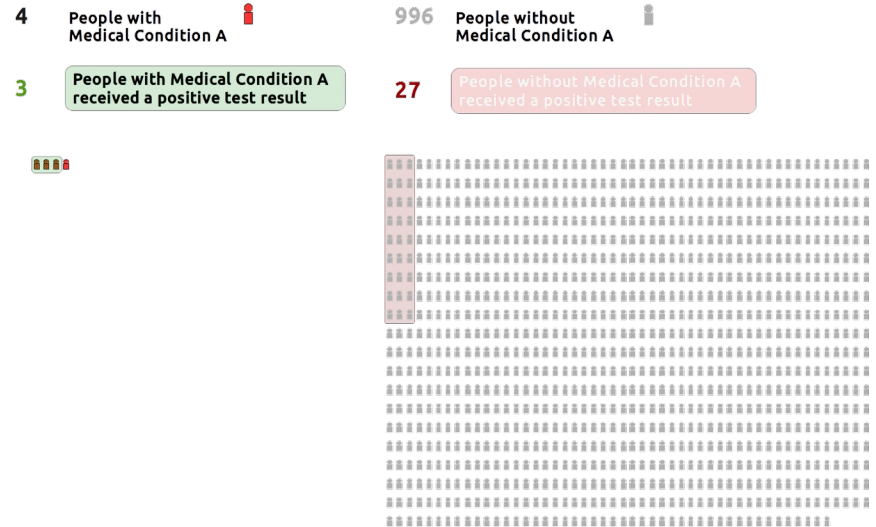


Presentation format x Response type

Vulnerable population

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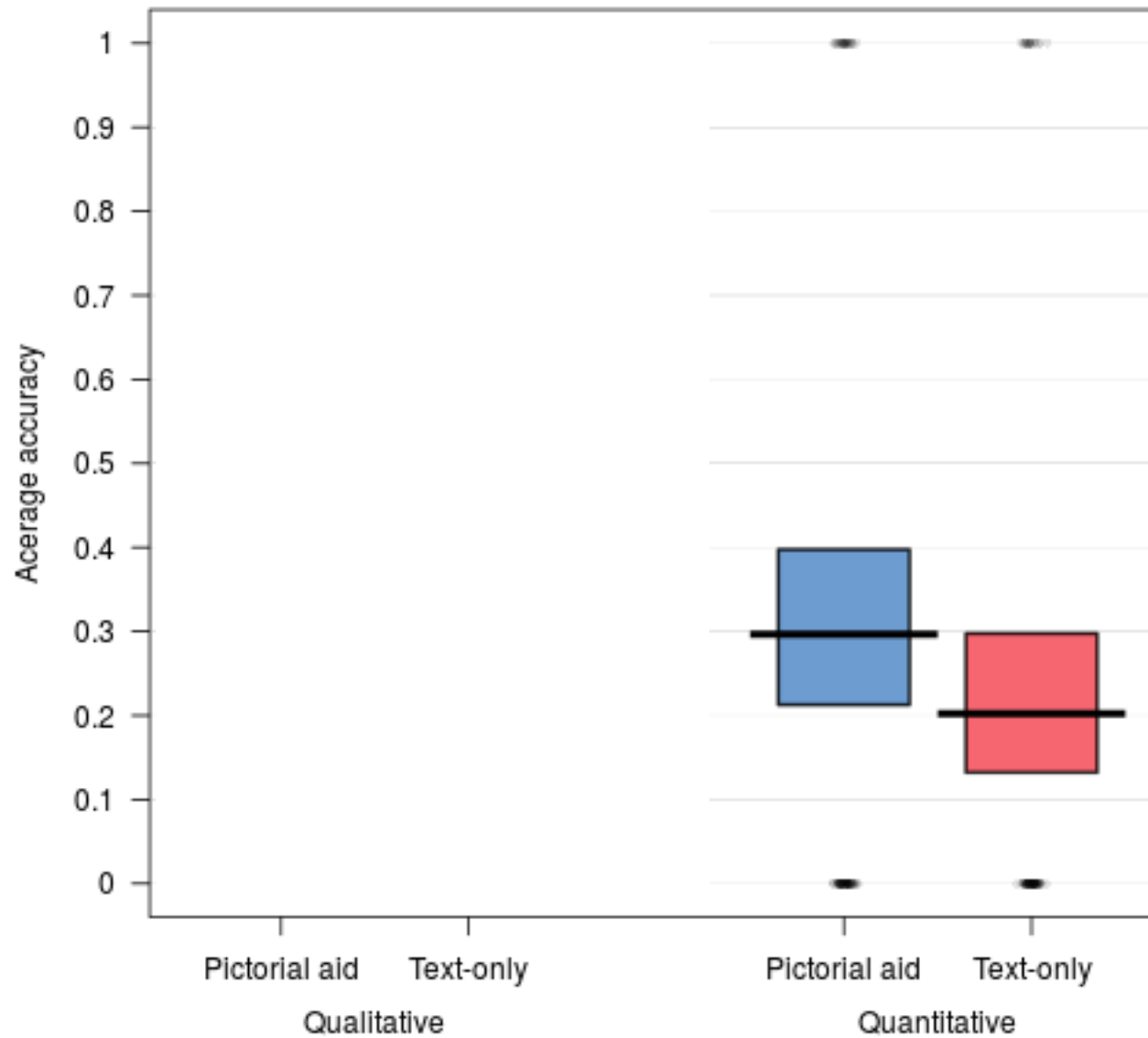


**Of the people who have received a positive screening test result, how many would do you expect to have the disease A?**

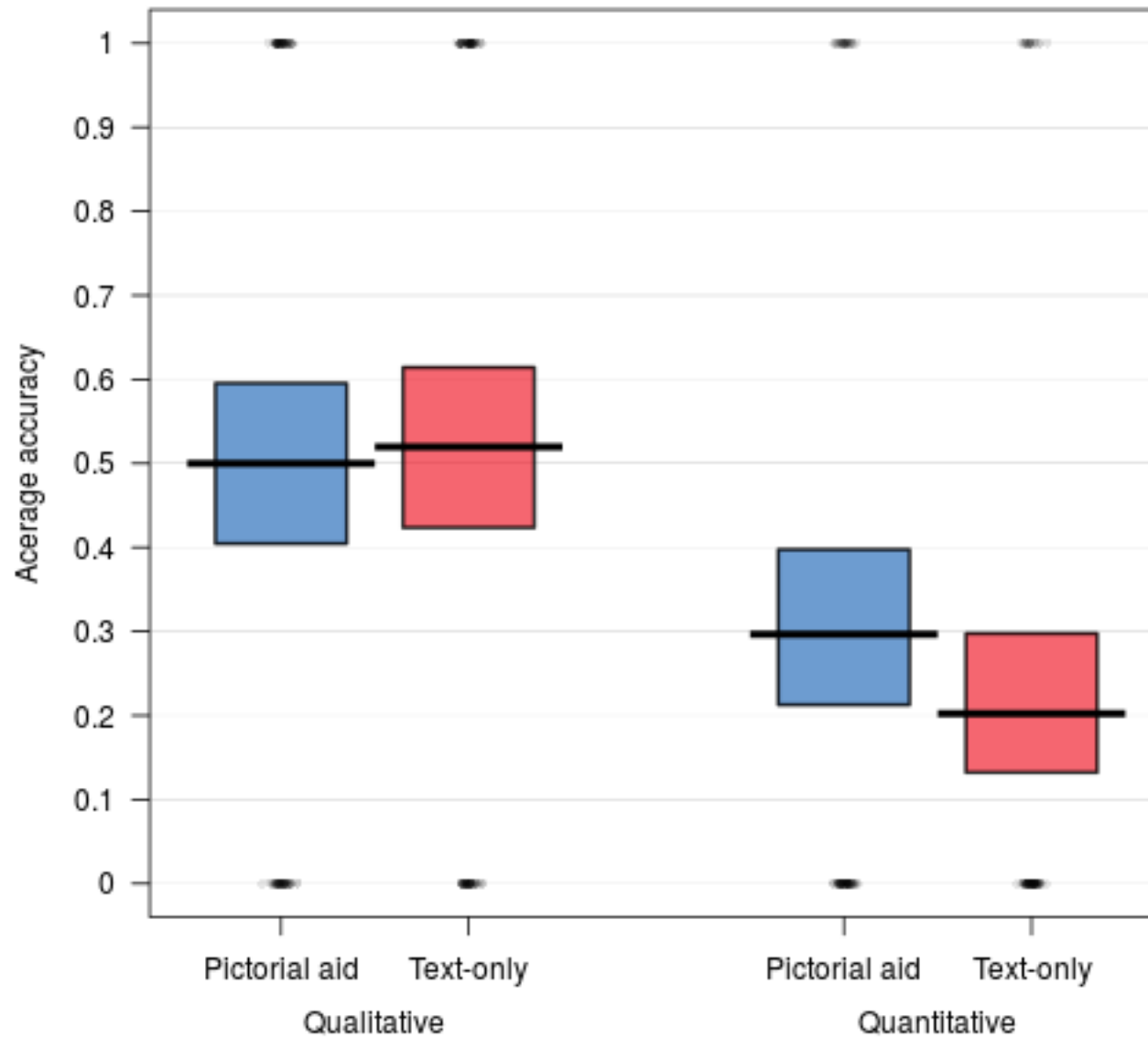
\_\_\_\_\_ out of \_\_\_\_\_

Very few (0-20%)   Few (21-40%)   Half (41-60%)   Quite (61-80%)   Many (81-100%)

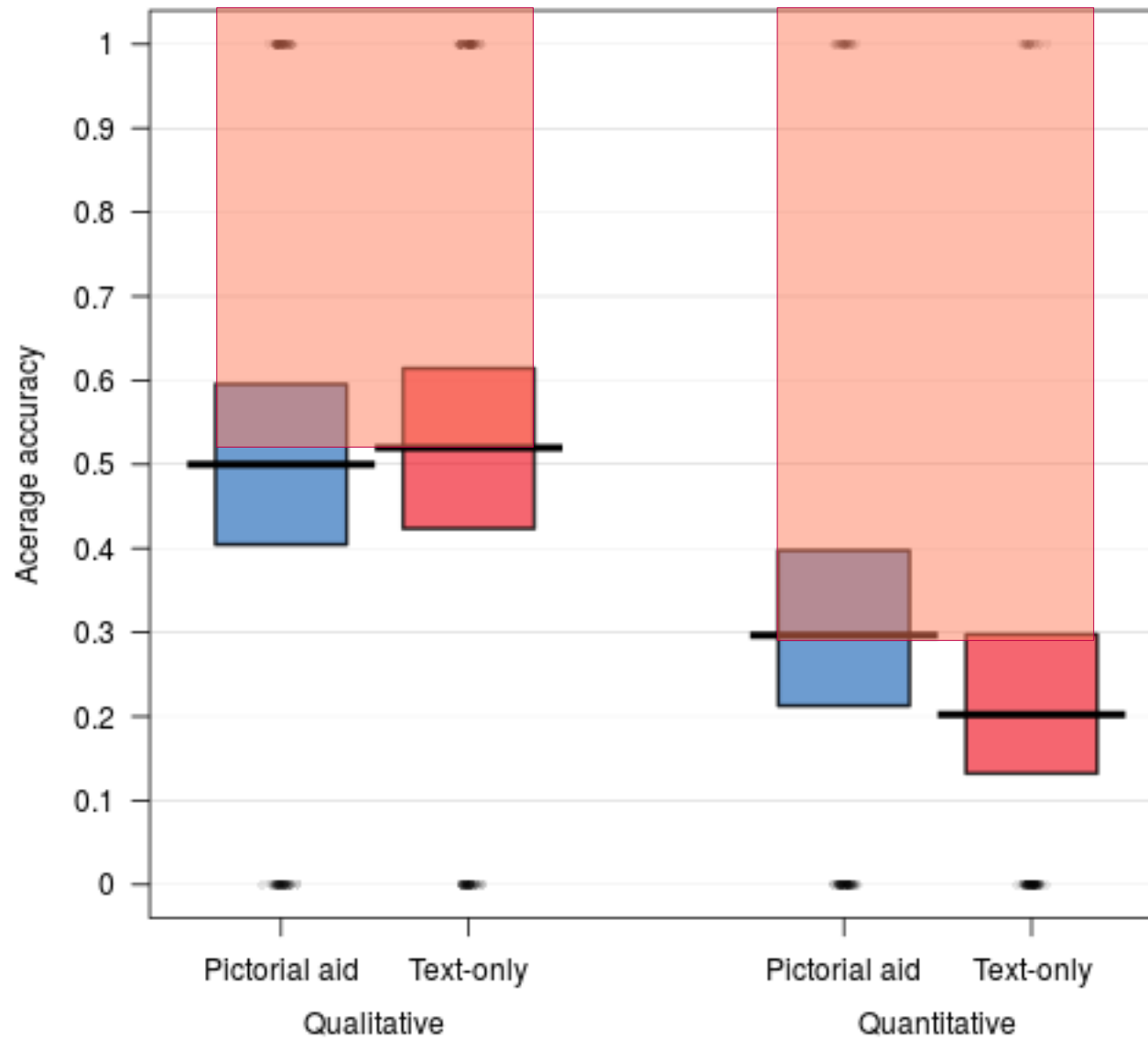
### Accuracy by response type and presentation format



### Accuracy by response type and presentation format



### Accuracy by response type and presentation format

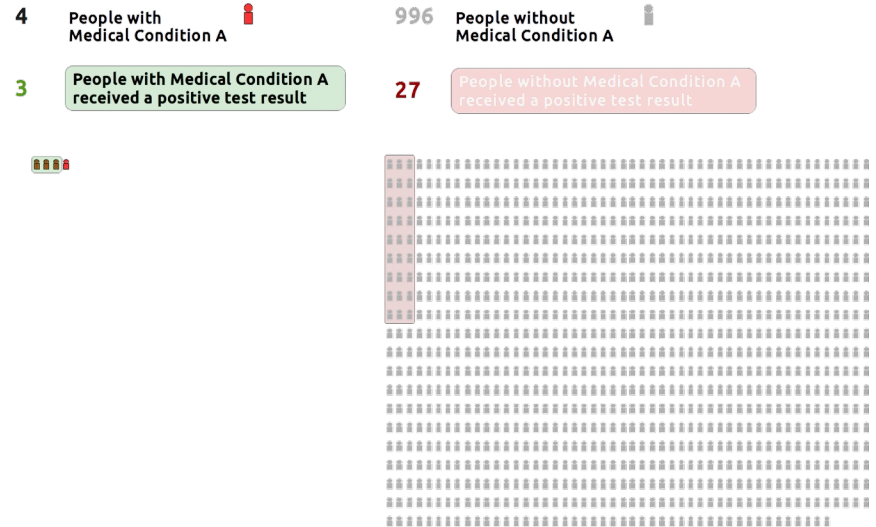


New research



Imagine that a group of 1000 people have participated in a medical screening to detect disease A. The following information is available about the screening.

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~5-10%  
correct  
responses

~50%  
correct  
responses

\_\_\_\_\_ out of \_\_\_\_\_

Very few (0-20%)   Few (21-40%)   Half (41-60%)   Quite (61-80%)   Many (81-100%)



~5-10%  
correct  
responses

## Harmony™ PRENATAL TEST

### What is a Trisomy?

Humans have 23 pairs of chromosomes, which are strands of DNA and proteins that carry genetic information. A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

#### TRISOMY 21

Trisomy 21 is due to an extra chromosome 21 and is the most common trisomy at the time of birth. Trisomy 21, also called Down syndrome, is associated with mild to moderate intellectual disabilities and may also lead to digestive disease and congenital heart defects. It is estimated that trisomy 21 is present in 1 out of every 800 births in Canada.<sup>1</sup>

#### TRISOMY 18

Trisomy 18 is due to an extra chromosome 18. Trisomy 18, also called Edwards syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 18 often have congenital heart defects as well as various other medical conditions, shortening their lifespan. It is estimated that trisomy 18 is present in approximately 1 out of every 6,000 births.<sup>2</sup>

#### TRISOMY 13

Trisomy 13 is due to an extra chromosome 13. Trisomy 13, also called Patau syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 13 usually have severe congenital heart defects and other medical conditions. Survival beyond the first year is rare. It is estimated that trisomy 13 is present in approximately 1 out of every 16,000 newborns.<sup>3</sup>



An advance in  
non-invasive fetal  
trisomy testing



Integrated  
GENETICS  
Laboratory Speciality Testing Group

### Harmony™ Prenatal Test

Simple, safe and accurate for you and your pregnancy.

The Harmony Prenatal Test is a non-invasive test that detects common fetal trisomies in pregnancies of 10 weeks or more, based on directed analysis of DNA in maternal blood.

WHAT WILL THE HARMONY PRENATAL TEST TELL ME AND MY HEALTHCARE PROVIDER?

The Harmony Prenatal Test assesses the risk of three fetal trisomies by measuring the relative amount of chromosomes in maternal blood.


HOW IS THE HARMONY PRENATAL TEST DIFFERENT FROM OTHER PRENATAL TESTS?

The Harmony Prenatal Test is based on the newest advances in non-invasive prenatal testing. It is a simple and safe blood test that has been shown in clinical studies to detect the risk of fetal trisomies with high accuracy.<sup>4,5</sup>

The Harmony Test has been shown to have detection rates of up to 99% and false positive rates as low as 0.1% for trisomy 21, 18 and 13.<sup>6,7</sup> Diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) are accurate for detecting fetal trisomies, but they are invasive and pose a slight risk for fetal loss.<sup>8</sup>

<sup>1</sup>Canadian Down Syndrome Society  
<sup>2</sup>Ontario Ministry of Health and Long-Term Care  
<sup>3</sup>U.S. National Library of Medicine  
<sup>4</sup>Harmon, M, et al. *Prenatal Genetics*. (2012). doi:10.1016/j.preg.2012.05.012  
<sup>5</sup>Harmon, G, et al. *ULTRASOUND Obstet Gynecol*. (2012). doi:10.1002/ulq.12399





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**An advance in non-invasive fetal trisomy testing**

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$$p(\text{Trisomy 21} \mid + \text{test}) =$$

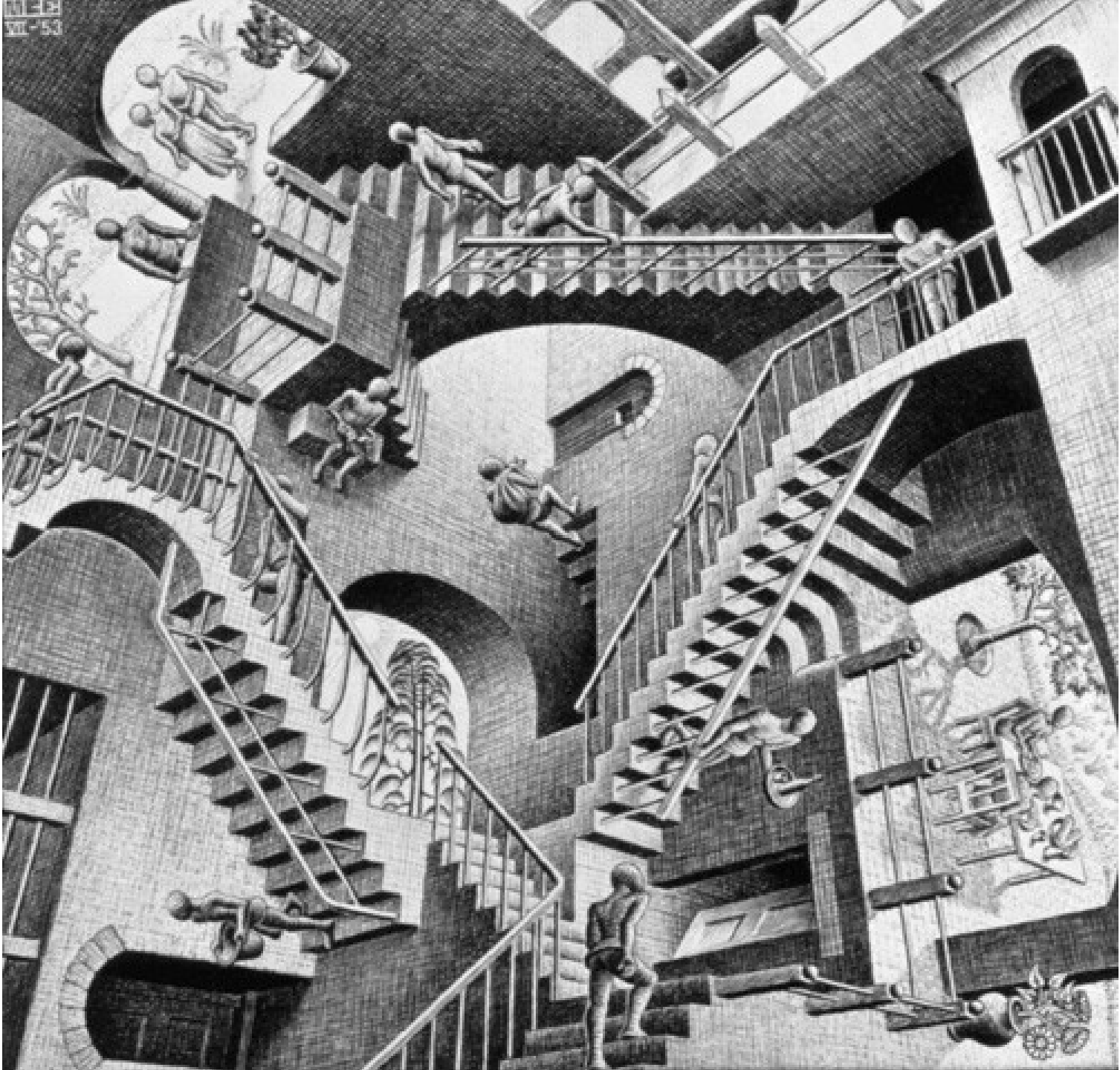
$$(A) \frac{1 \text{ out of } 800 \times 99\%}{(1 \text{ out of } 800 \times 99\%) + (799 \text{ out of } 800 \times 0.1\%)} =$$

$$(B) \frac{0.123}{0.123 + 0.0998} = 0.55$$



What's the meaning of a + result?



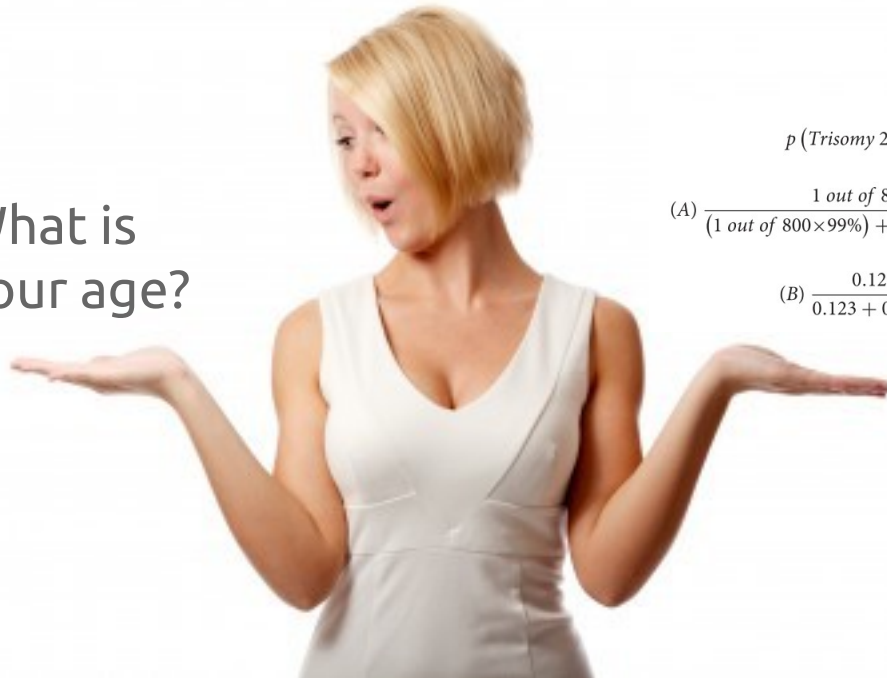


# New paradigm

Ask questions people know how to answer



What is  
your age?

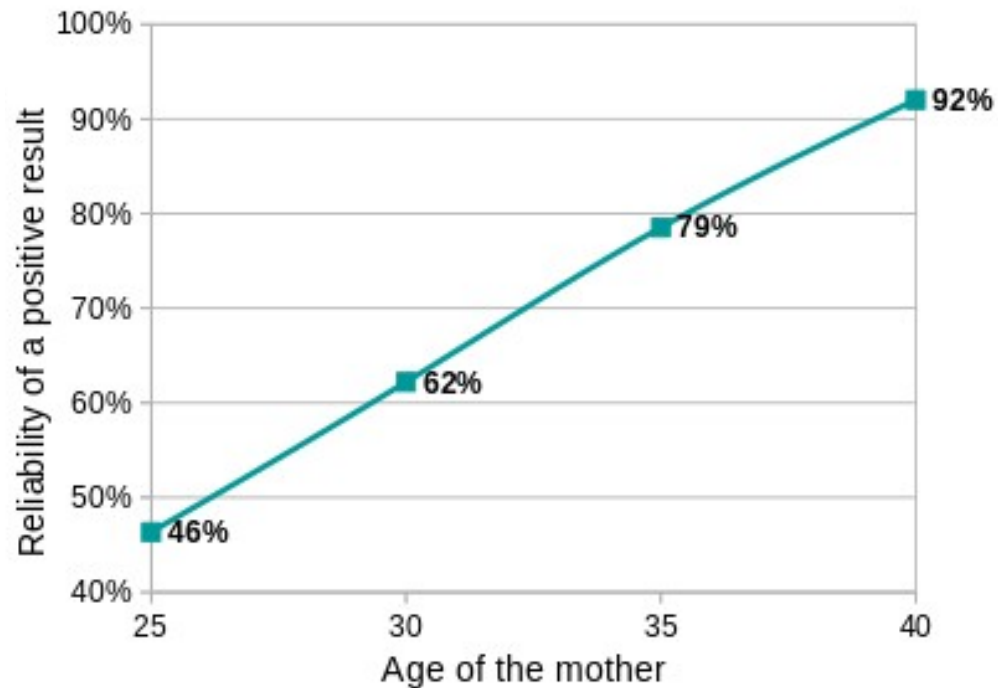


$$p(\text{Trisomy 21} \mid + \text{test}) =$$

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# Ask questions people know how to answer



Reliability of a positive test result depending on the mother's age.





Do people know the information they need?

Can we improve calculation/comprehension?

Can we improve decisions?



Do people know what they need?

All vs Missing information

# All information

## What is a Trisomy?

Humans have 23 pairs of chromosomes, strands of DNA that carry genetic information. A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

### Down syndrome

Trisomy 21, also called Down syndrome, is caused by a third copy of chromosome 21. It is the most common trisomy at time of birth. Down syndrome is associated with mild to moderate intellectual disabilities and congenital heart defects.

**It is estimated that, when a mother is aged 40, Down syndrome is present in 1 out of 60 births.**

### Prenatal Test

When you are pregnant, your blood contains fragments of fetus DNA. A Prenatal Test is a new type of test that samples your blood and analyzes the fetus DNA to determine the risk of Down syndrome. It is used for early detection of Down syndrome in all pregnant women.

### Clarity Early

The Prenatal Test requires a single blood sample and can be done as early as 10 weeks or later in pregnancy. Results are available within one business day.

### A More Accurate Test

The Prenatal Test, a new DNA-based blood test, has been extensively tested. It was developed to be a more accurate prenatal screening. The Prenatal Test has been shown in clinical testing to identify 90% of Down syndrome cases and to have a false-positive rate of less than 0.4%.

A positive result does not necessarily mean that Down syndrome is present.



Clear ANSWERS to Questions that Matter

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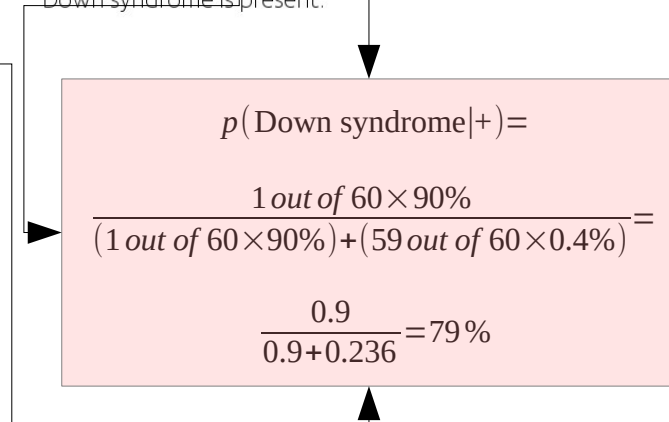
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Clear ANSWERS to Questions that Matter

# Missing information

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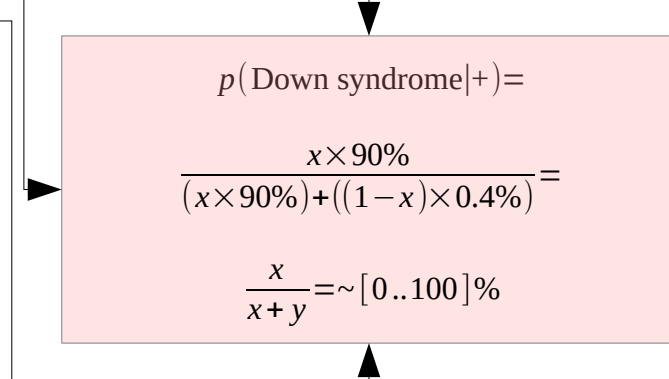
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Clear ANSWERS to Questions that Matter

Did the brochure include enough information to solve the problem?

YES!

$$p(\text{Down syndrome}|+) =$$

$$\frac{1 \text{ out of } 60 \times 90\%}{(1 \text{ out of } 60 \times 90\%) + (59 \text{ out of } 60 \times 0.4\%)} =$$

$$\frac{0.9}{0.9 + 0.236} = 79\%$$

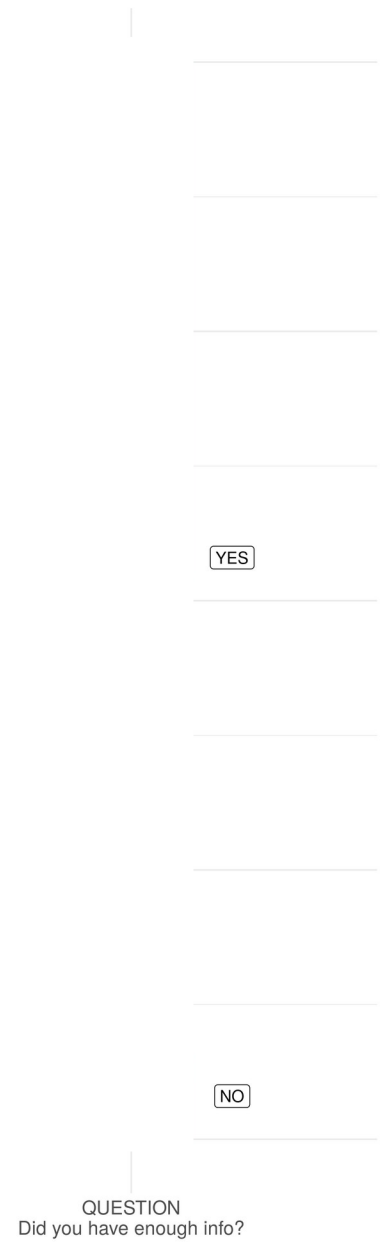
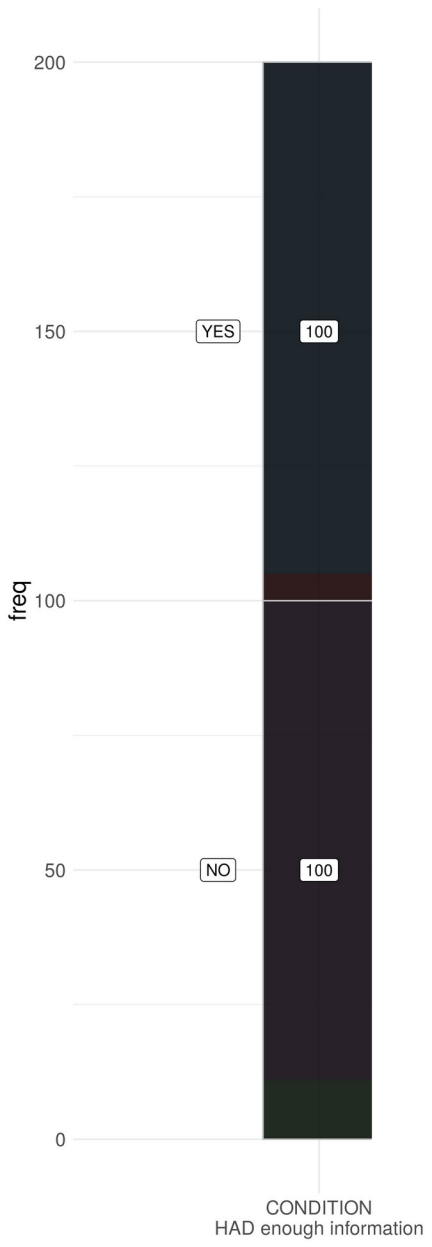
NO

$$p(\text{Down syndrome}|+) =$$

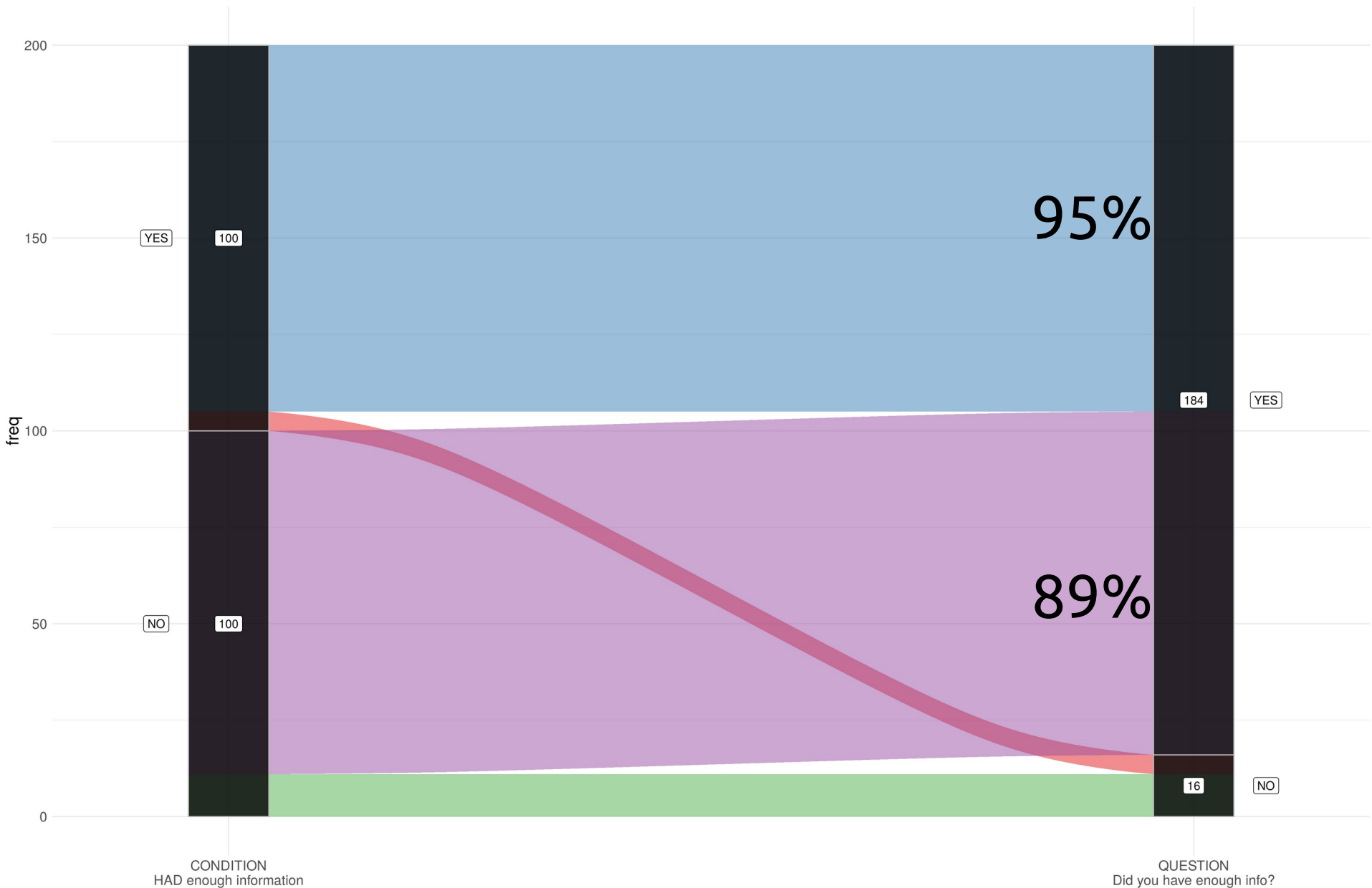
$$\frac{x \times 90\%}{(x \times 90\%) + ((1-x) \times 0.4\%)} =$$

$$\frac{x}{x+y} = \sim [0..100]\%$$





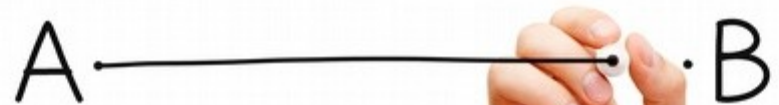
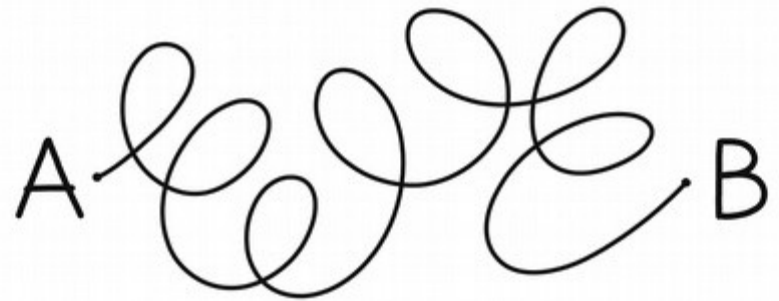
# 89% of those without Enough info said YES



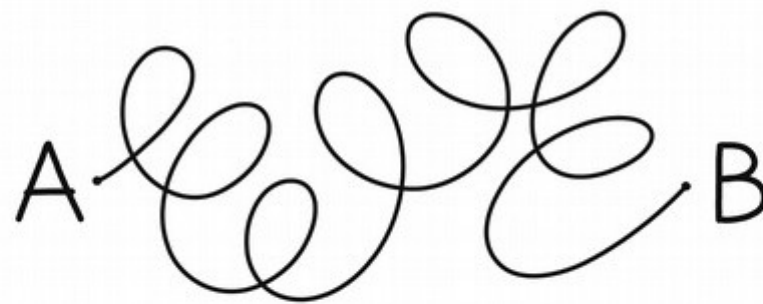


# Can we improve calculation/comprehension?

OLD vs NEW paradigm



OLD paradigm



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**It is estimated that, when a mother is aged 40, Down syndrome is present in 1 out of 60 births.**

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A positive result does not necessarily mean that Down syndrome is present.

Clear ANSWERS to Questions that Matter

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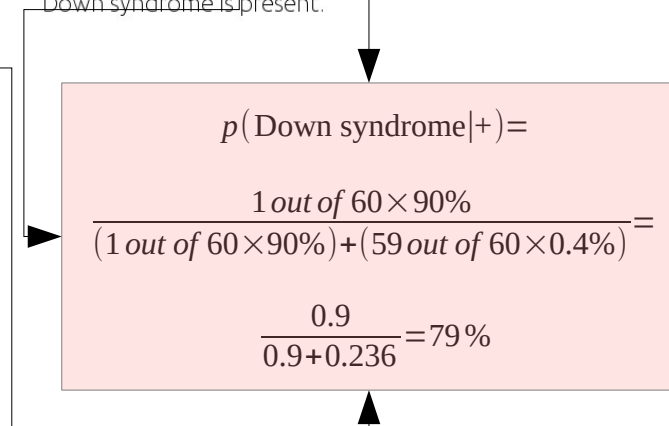
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Clear ANSWERS to Questions that Matter

NEW paradigm



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Clear ANSWERS to Questions that Matter

Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. Birth Defects Res A Clin Mol Teratol. 2010;88:1008–1016y.

### Clarity Early

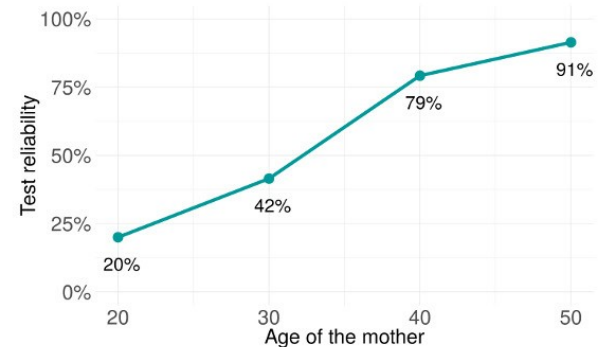
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A positive result does not necessarily mean that Down syndrome is present.

**In the graph below, you can see the probability that a positive prenatal test result actually means the presence of Down syndrome. This probability is derived from risk factors associated with age in women.**





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Clear ANSWERS to Questions that Matter

Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. Birth Defects Res A Clin Mol Teratol. 2010;88:1008–1016y.

### Clarity Early

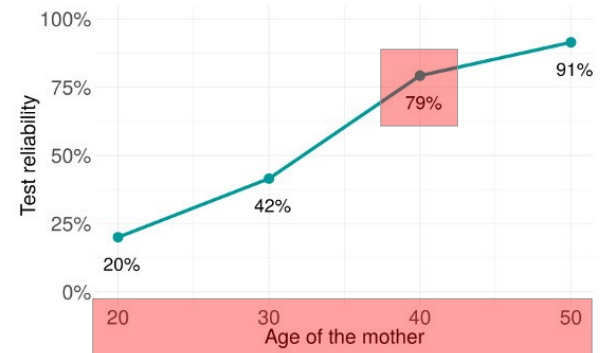
The Prenatal Test requires a single blood sample and can be done as early as 10 weeks or later in pregnancy. Results are available within one business day.

### A More Accurate Test

The Prenatal Test, a new DNA-based blood test, has been extensively tested. It was developed to be a more accurate prenatal screening. The Prenatal Test has been shown in clinical testing to identify 90% of Down syndrome cases and to have a false-positive rate of less than 0.4%.

A positive result does not necessarily mean that Down syndrome is present.

**In the graph below, you can see the probability that a positive prenatal test result actually means the presence of Down syndrome. This probability is derived from risk factors associated with age in women.**







What is a Trisomy?

Women have 23 pairs of chromosomes, known as autosomes. Each pair consists of two chromosomes. A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

Down syndrome

Trisomy 21, also called Down syndrome, is caused by a third copy of chromosome 21. It is the most common chromosomal condition that causes intellectual disability and physical disabilities and congenital heart defects.

It is estimated that, when a mother is aged 40, Down syndrome is present in 1 out of 100 births.

Prevalent Test

When you are pregnant, your blood contains a mixture of proteins, lipids, and hormones. The level of each of these substances in your blood can indicate the risk of Down syndrome in a pregnant woman.

**Clarify Early**  
The prenatal test requires a single blood sample and can be done as early as 10 weeks to date. All pregnancy results are available within one business day.

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The prenatal test, using DNA-based blood test, has been extensively tested. It was developed to be a more accurate prenatal screening. The prenatal test has been shown to have a 99.9% sensitivity and a 99.9% specificity.

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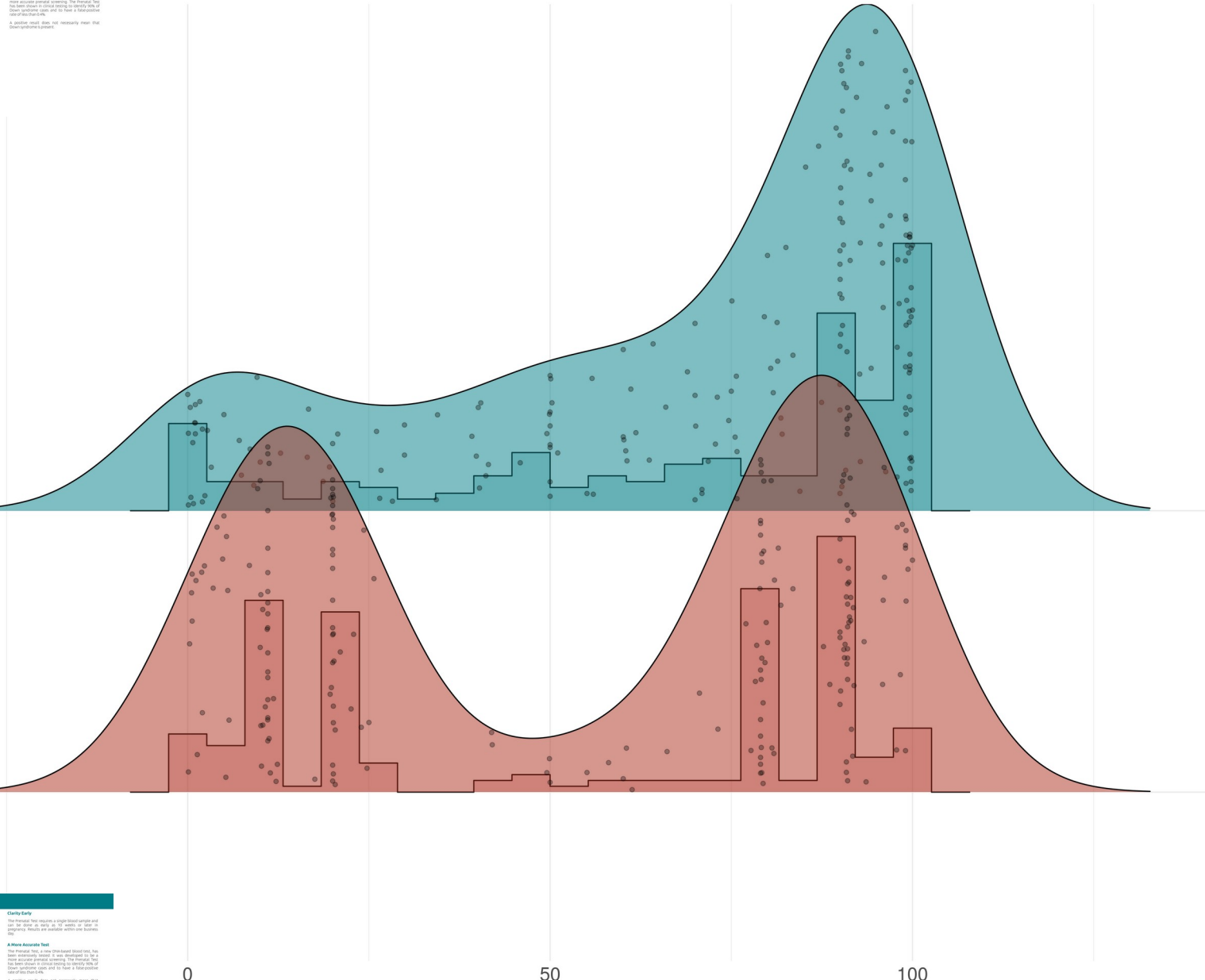
brochure  
OLD

OLD

NEW

brochure

NEW  
OLD



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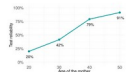
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Clear ANSWERS to Questions that Matter

PPV\_screening



## Good vs bad test

**40 years old**

Prevalence 1 out of 60

PPV~75%

Good test

**20 years old**

Prevalence 1 out of 900

PPV~25%

Bad test

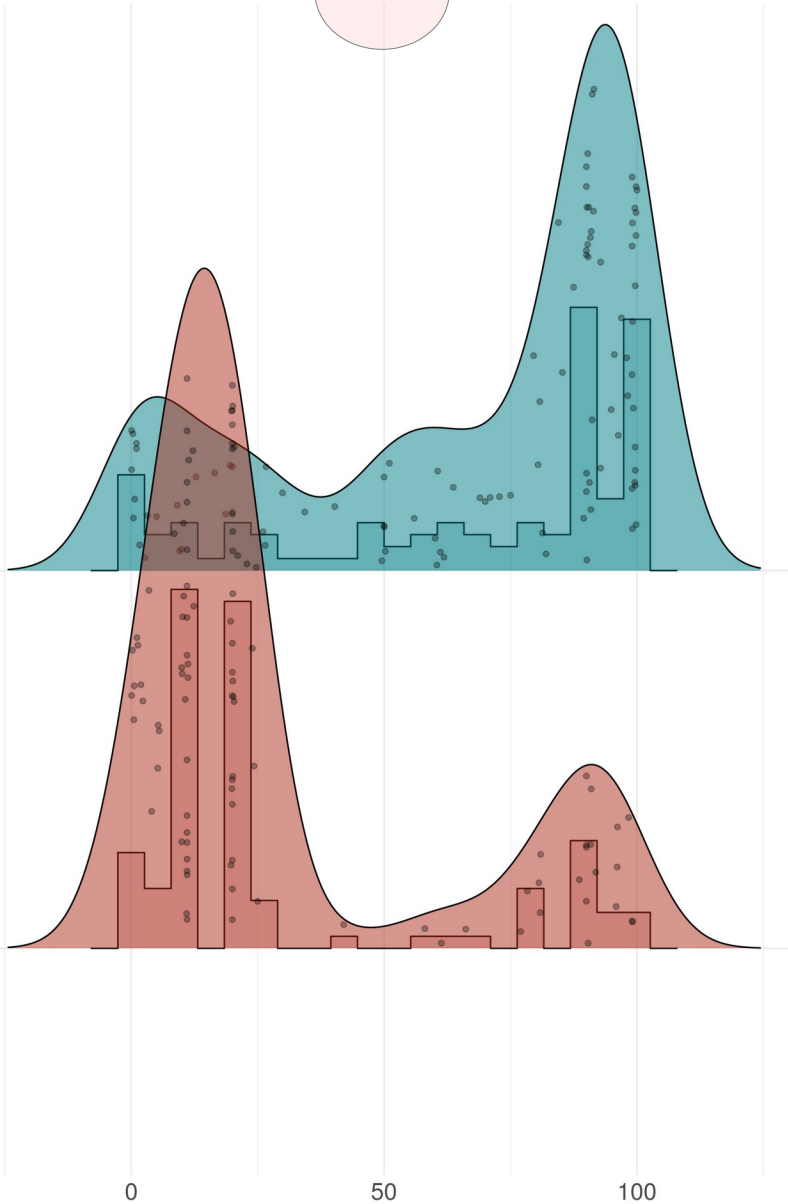


20

brochure

OLD

NEW



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Trisomy 21, also called Down syndrome, is caused by a third copy of chromosome 21. It is the most common trisomy at time of birth. Down syndrome is associated with mild to moderate intellectual disabilities and congenital heart defects.

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Clear ANSWERS to Questions that Matter

Photo: iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes

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Photo: iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes; iStock.com/Robert\_Holmes

Clarity Early

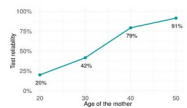
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brochure  
OLD  
NEW

20

40

brochure  
NEW  
OLD

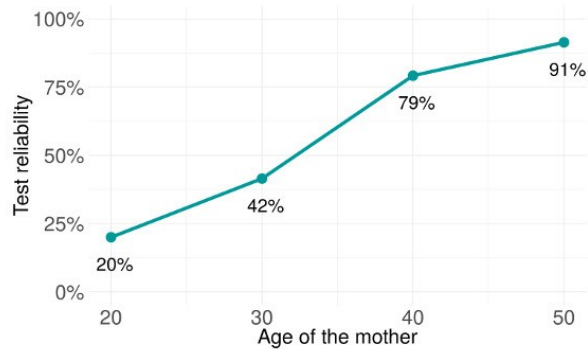
NEW

PPV\_screening



16%  
average absolute  
error

40%  
average absolute  
error



$$p(\text{Down syndrome}|+) = \frac{1 \text{ out of } 60 \times 90\%}{(1 \text{ out of } 60 \times 90\%) + (59 \text{ out of } 60 \times 0.4\%)} = \frac{0.9}{0.9 + 0.236} = 79\%$$

# Can we improve decisions?

OLD vs NEW paradigm

# Screening test

# Follow-up test

Would you recommend this **screening test**?

Imagine the result is

+

Would you recommend a **Follow up test** with 1% chance of a procedure-related miscarriage?

YES / NO

YES / NO

**What is a Trisomy?**

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A positive result does not necessarily mean that Down syndrome is present.



**Clear ANSWERS to Questions that Matter**

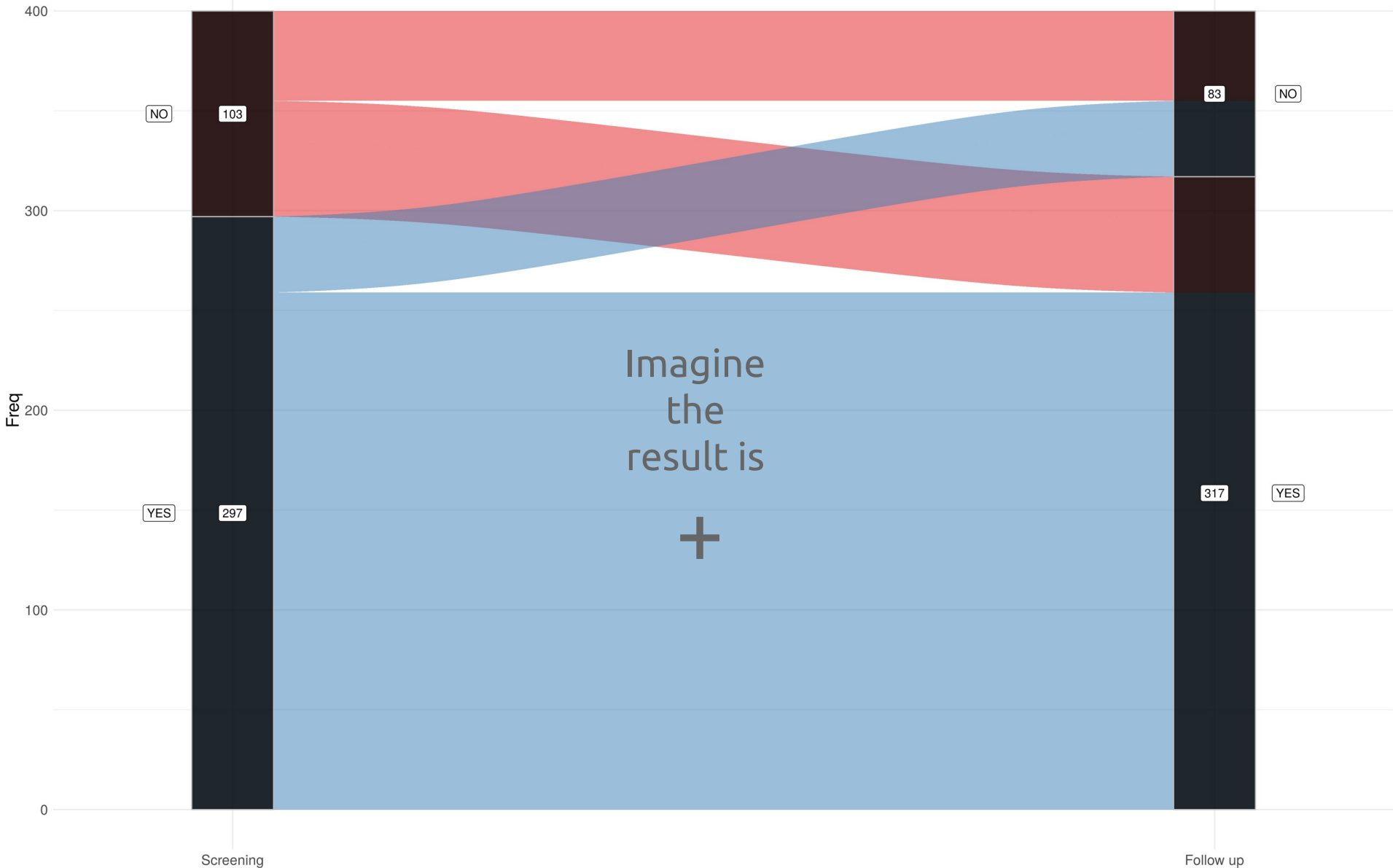
Photo by iStockphoto.com/Scott Jones/Thinkstock.com. All rights reserved. No part of this publication may be reproduced, stored in a retrieval system, or transmitted, in any form or by any means, electronic, mechanical, photocopying, recording, or by any information storage and retrieval system, without permission in writing from the publisher.





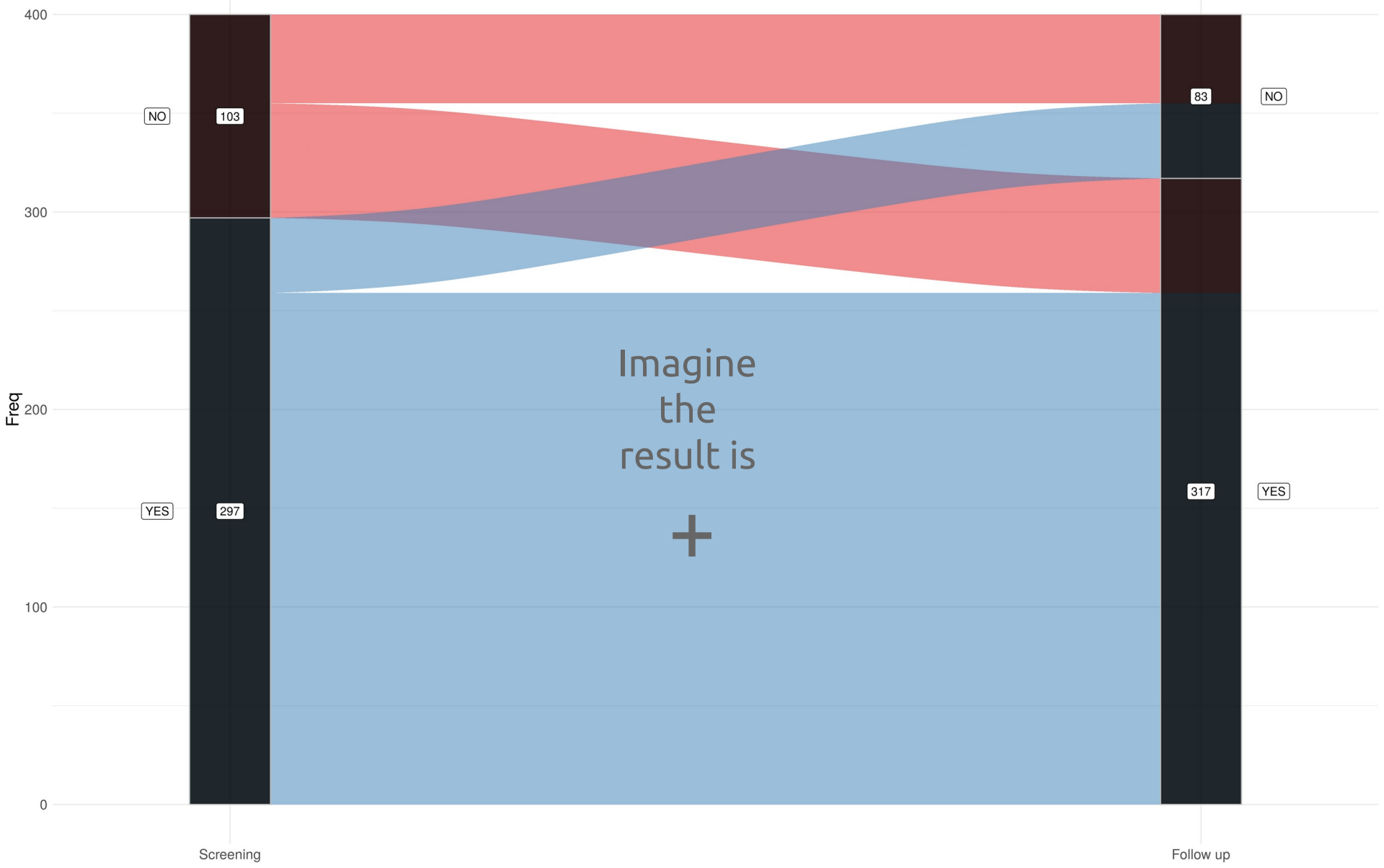
# Screening test

# Follow-up test



# Screening test

# Follow-up test



**20 years old**

Prevalence 1 out of 900

PPV~25%

Bad test

Bad test

Vs

Good test

**40 years old**

Prevalence 1 out of 60

PPV~75%

Good test

Screening test



Follow-up test

OLD

**20 years old**

Prevalence 1 out of 900

PPV~25%

Bad test

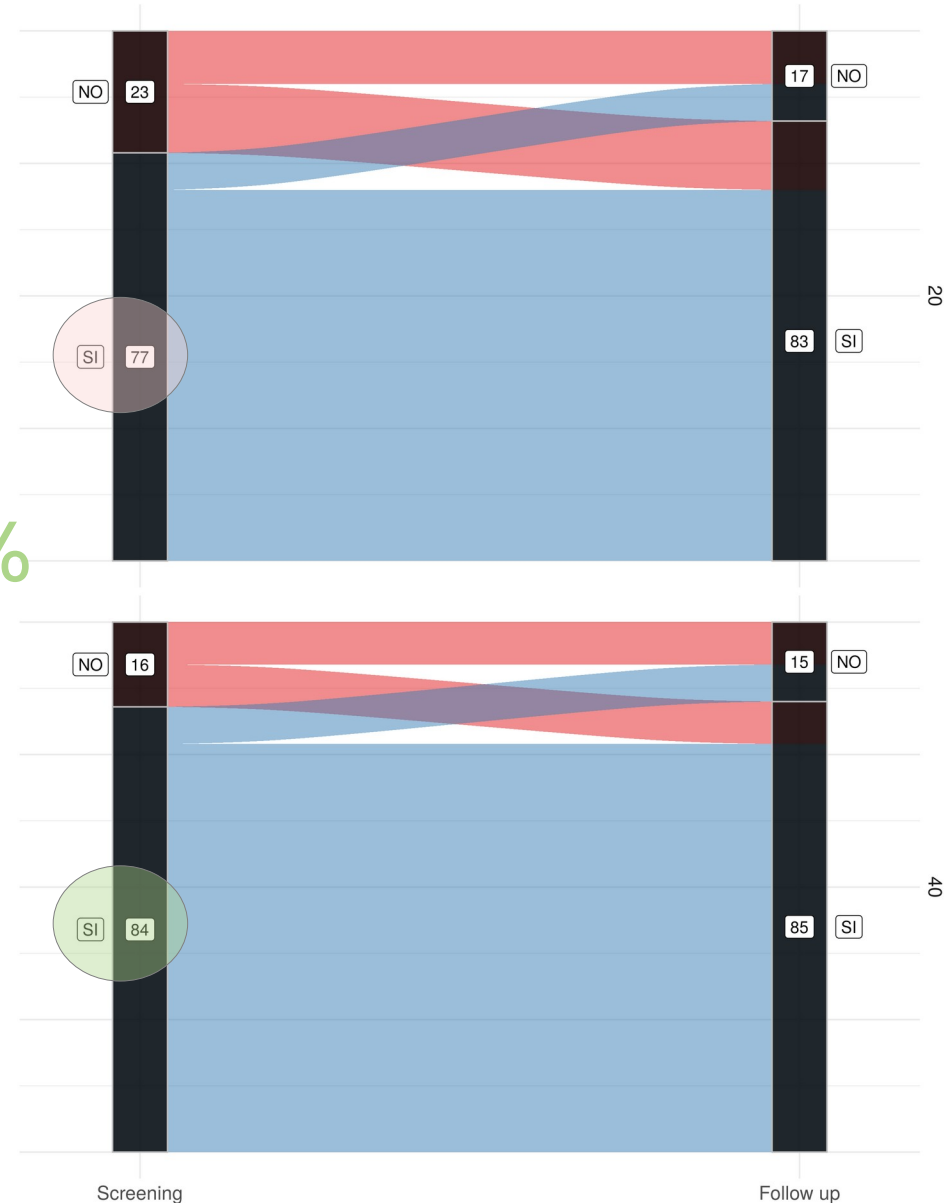
**77% vs 84%**

**40 years old**

Prevalence 1 out of 60

PPV~75%

Good test

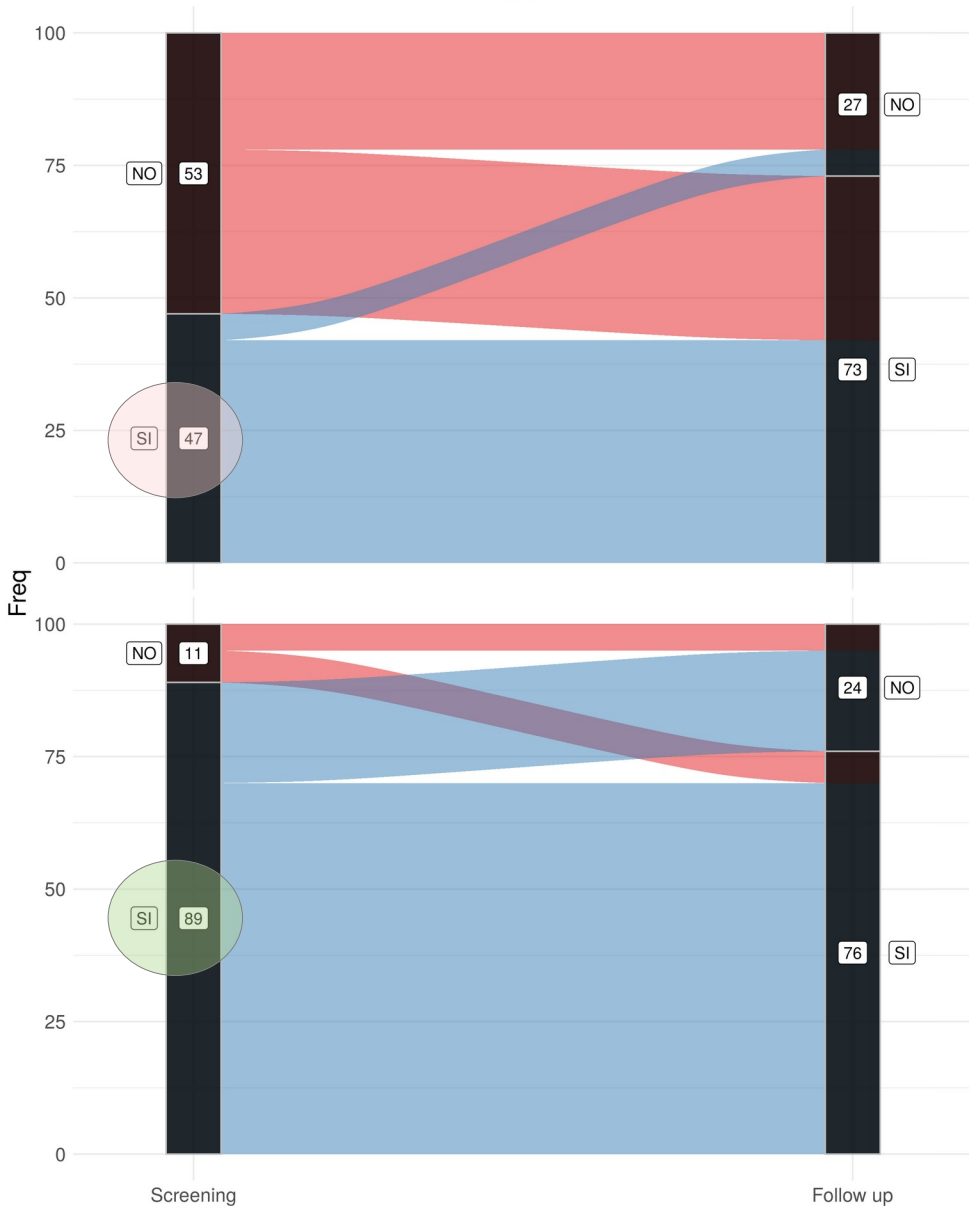


# Screening test



NEW

# Follow-up test



**20 years old**

Prevalence 1 out of 900  
PPV~25%

Bad test

**47% vs 89%**

**40 years old**

Prevalence 1 out of 60  
PPV~75%

Good test

# Screening test



NEW

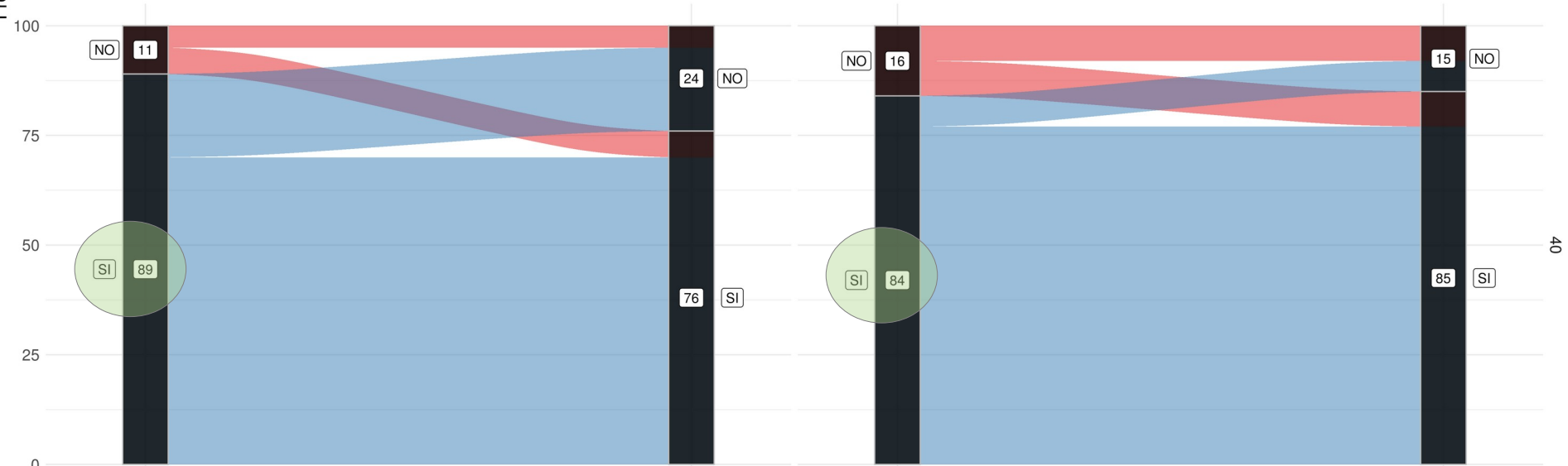
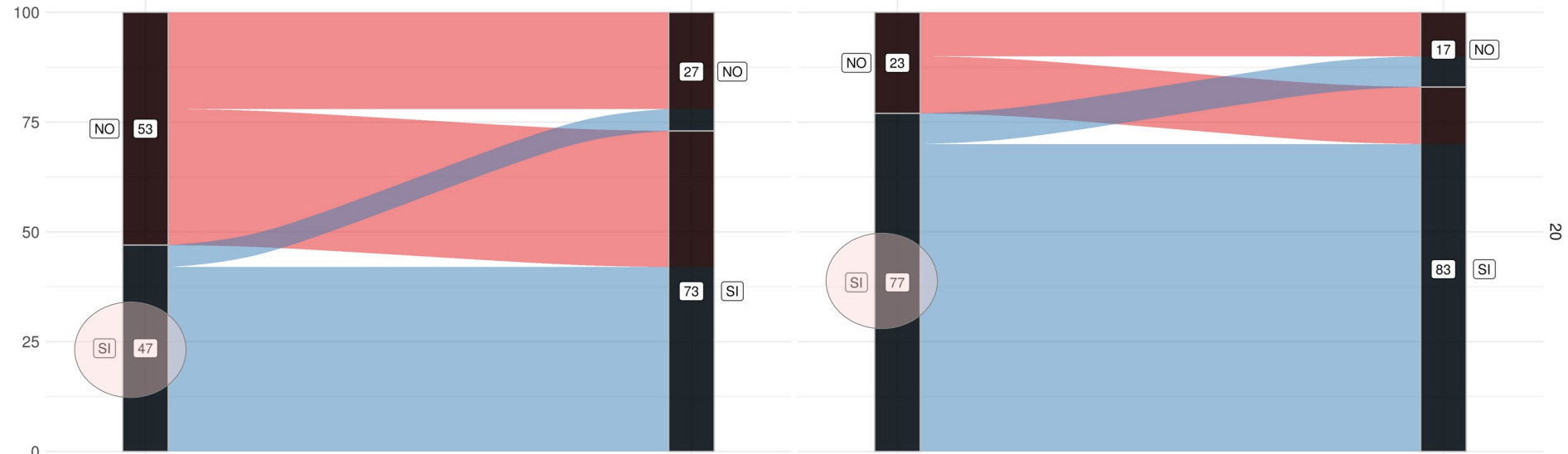
# Follow-up test

# Screening test



OLD

# Follow-up test



Screening

Follow up

Screening

Follow up



# Screening test



NEW

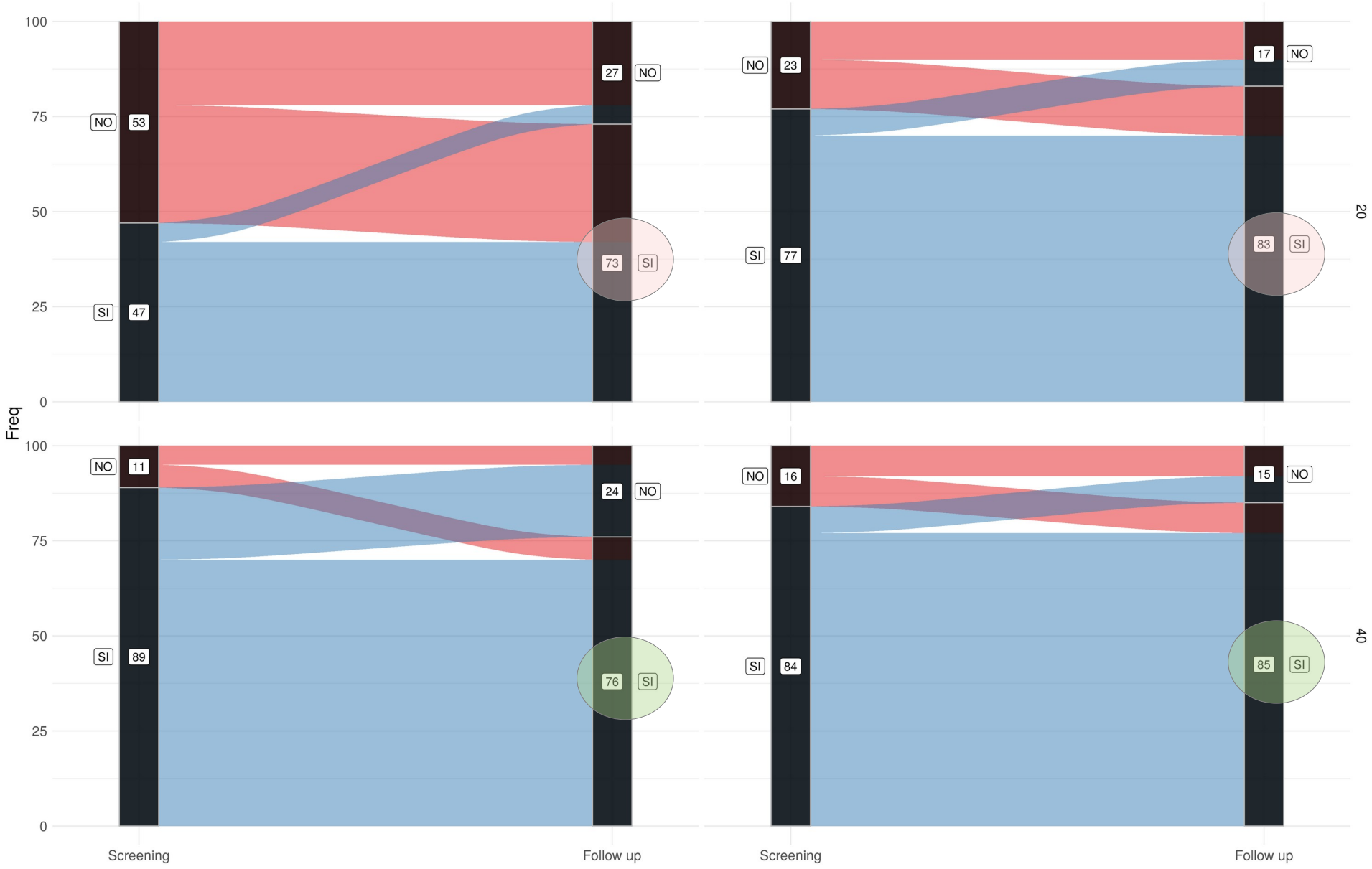
# Follow-up test

# Screening test



OLD

# Follow-up test







# Overview of results

Do people know the information they need?

Can we improve calculation/comprehension?

Can we improve decisions?



# Overview of results

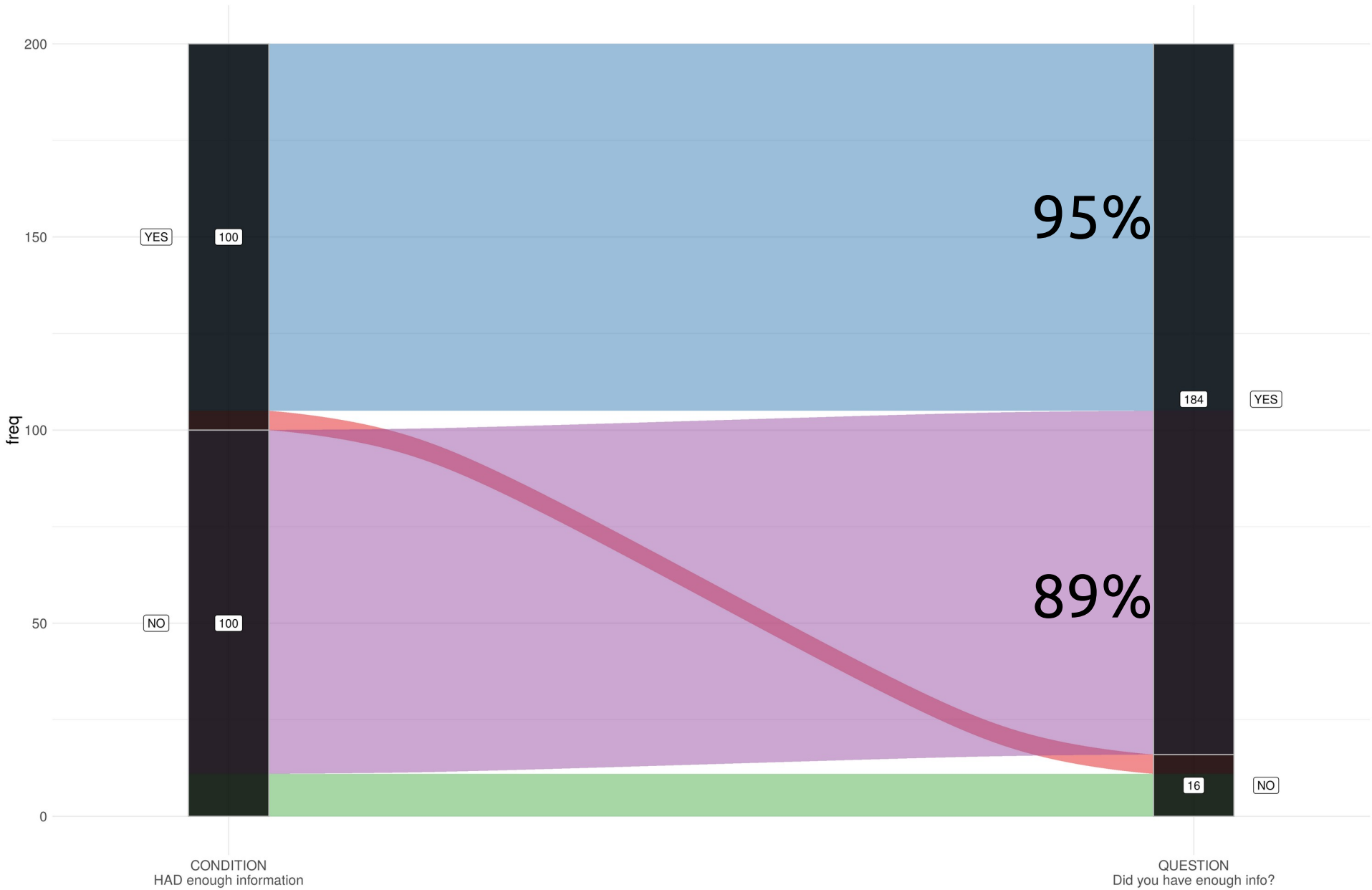
Do people know the information they need?

Can we improve calculation/comprehension?

Can we improve decisions?



# NO



# Overview of results

Do people know the information they need?

Can we improve calculation/comprehension?

Can we improve decisions?



**What is a Prenatal Test?**

**Chorionic Grafting**

**Down Syndrome**

**Amniocentesis**

**Prevalent Test**

20

YES!

40

brochure  
OLD

brochure  
NEW  
OLD

NEW



50

100

PPV\_screening

0

50

100

**What is a Prenatal Test?**

**Chorionic Grafting**

**Down Syndrome**

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# Overview of results

Do people know the information they need?

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# Screening test



NEW

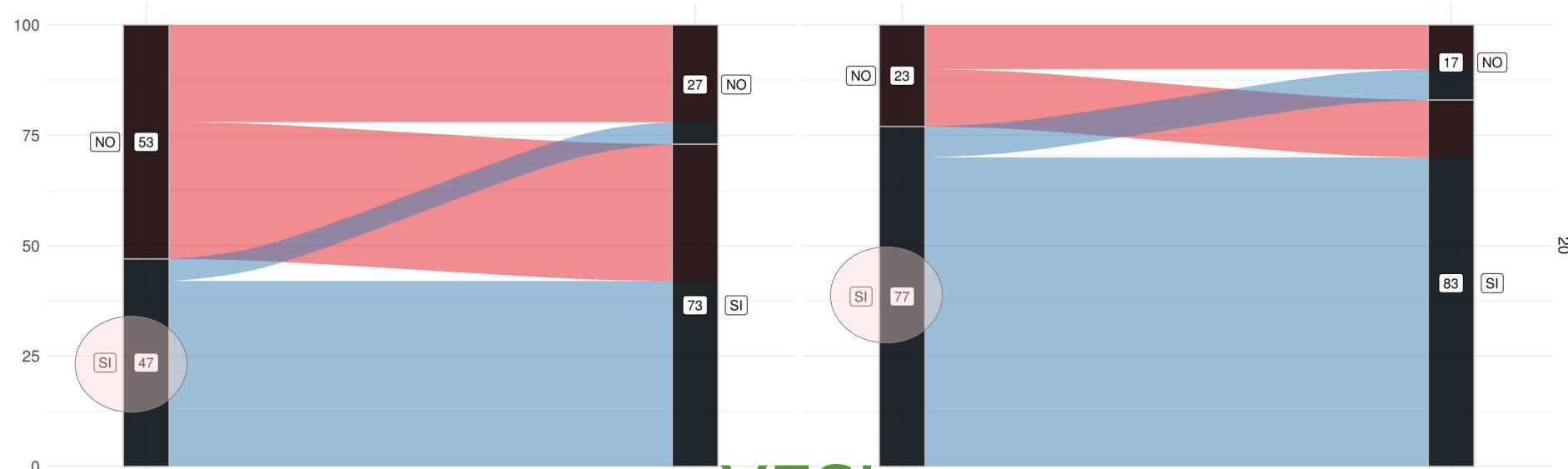
# Follow-up test

# Screening test

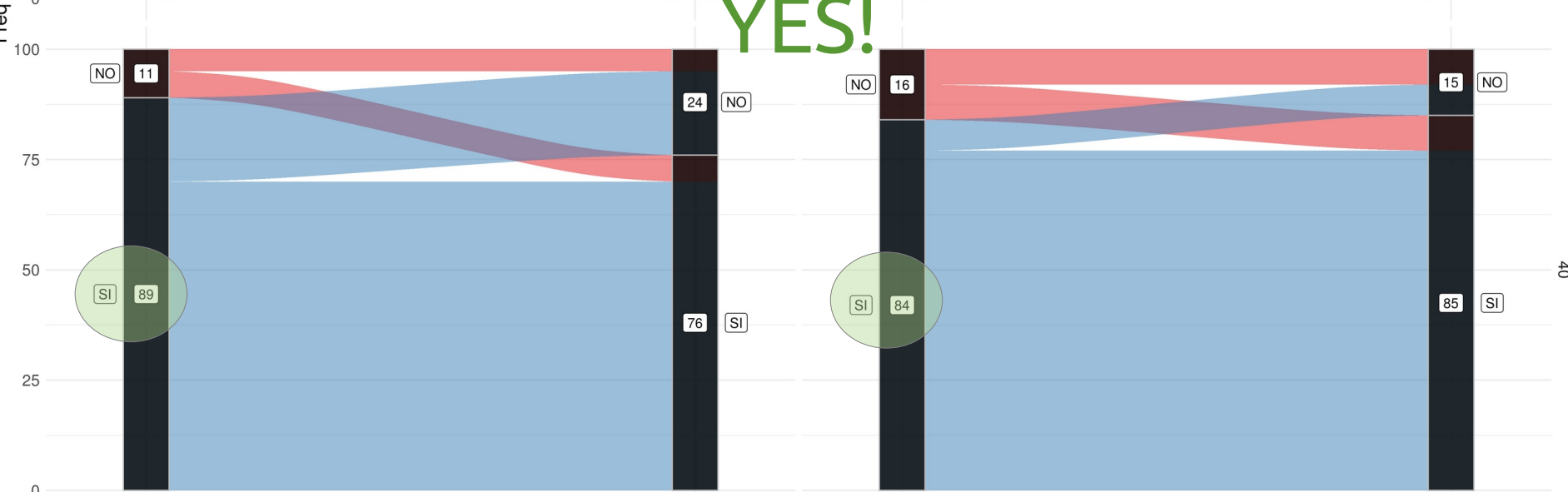


OLD

# Follow-up test



YES!



Screening

Follow up

Screening

Follow up



# Screening test



NEW

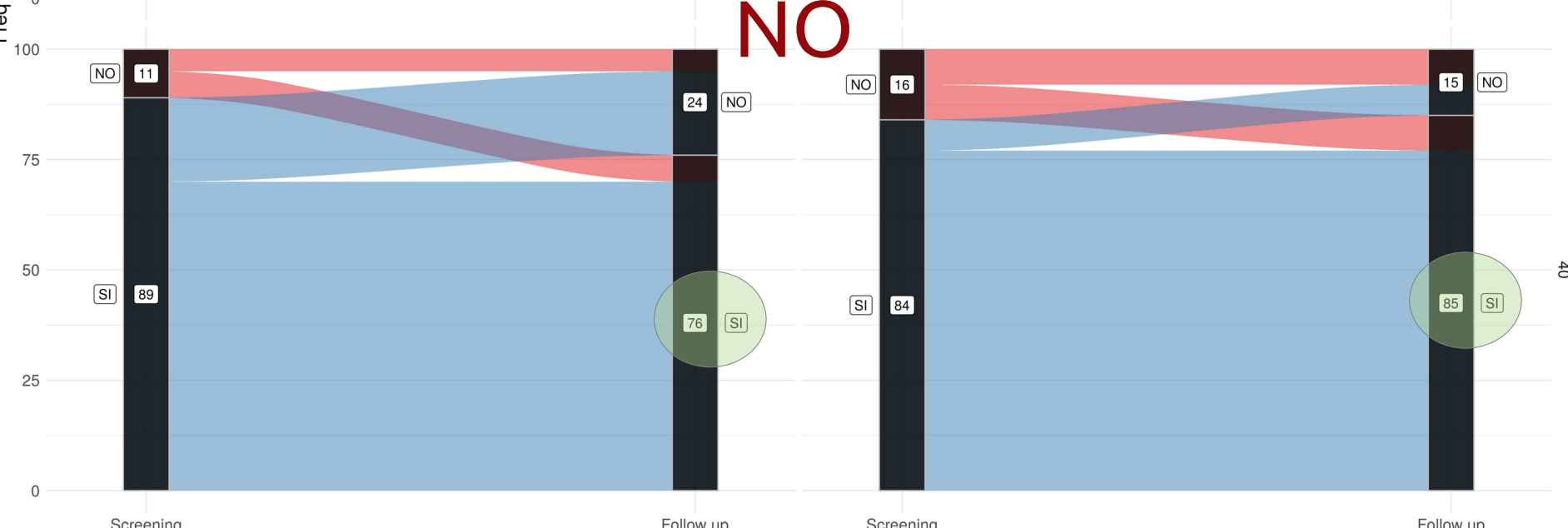
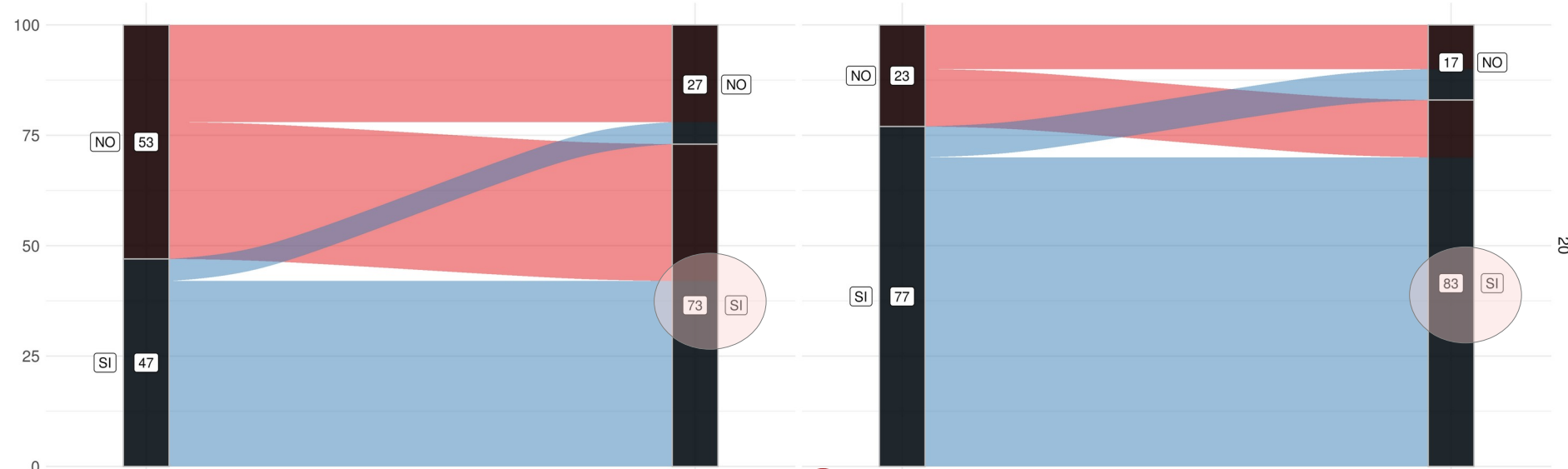
# Follow-up test

# Screening test



OLD

# Follow-up test



NO

Screening

Follow up

Screening

Follow up

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Would you recommend this **screening test**?

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**Clear ANSWERS to Questions that Matter**

Photo by iStockphoto.com/Scott Jones. Photo by iStockphoto.com/Scott Jones. Photo by iStockphoto.com/Scott Jones. Photo by iStockphoto.com/Scott Jones. Photo by iStockphoto.com/Scott Jones.

YES / NO

Imagine the result is +

Would you recommend a **Follow up test** with 1% chance of a procedure-related miscarriage?

YES / NO

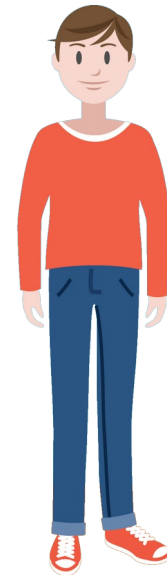
# Take home message

We can do better! And it matters...



We can do better! And it matters...

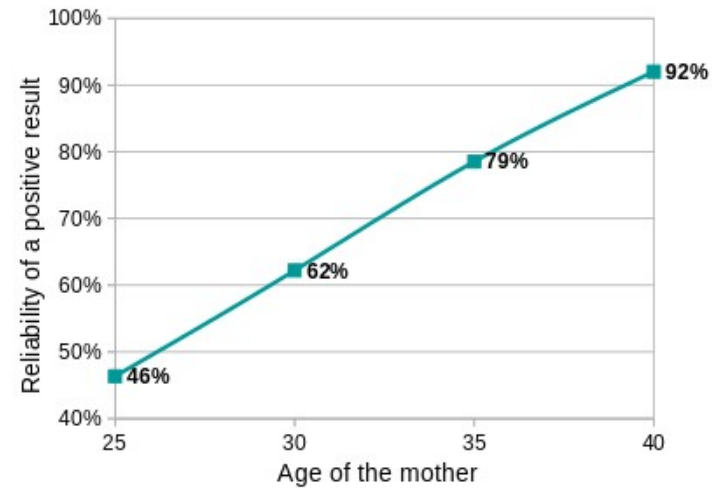
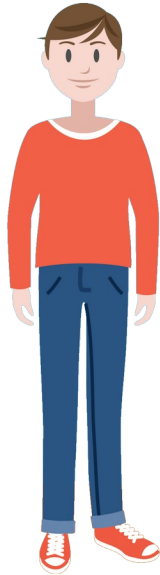
Patients



$$p(\text{Trisomy 21} \mid + \text{test}) =$$

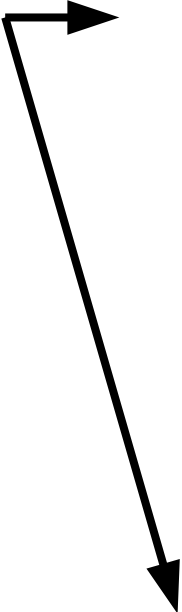
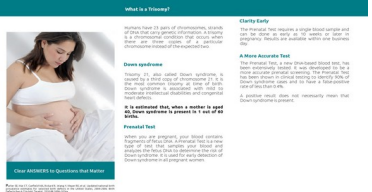
$$(A) \frac{1 \text{ out of } 800 \times 99\%}{(1 \text{ out of } 800 \times 99\%) + (799 \text{ out of } 800 \times 0.1\%)} =$$

$$(B) \frac{0.123}{0.123 + 0.0998} = 0.55$$

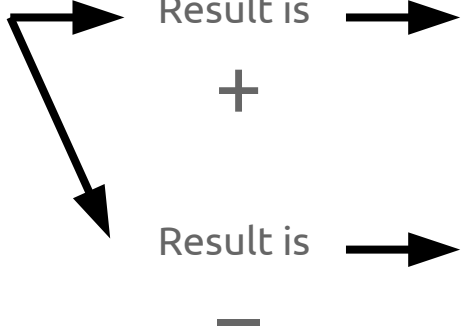


Reliability of a positive test result depending on the mother's age.

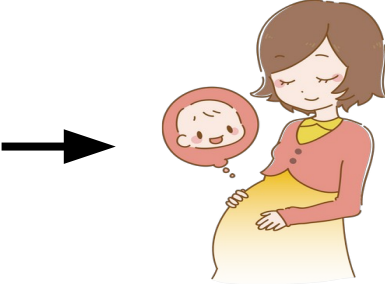
# Would you go through this screening test?



GOOD test



BAD test



We can do better! **And it matters...**

Many **false positives** in low prevalence conditions / populations

This leads to:

unnecessary tests (\$\$\$)

overdiagnosis (loss of health)

miracles!



We can do better! **And it matters...**

When people understand: they may avoid entering the tunnel!

less bad tests → less false positives

This would lead to:

**less** unnecessary tests

**less** overdiagnosis

**less** miracles



We can do better! And it matters...

This is only the first step, but we must be careful



# Next steps...

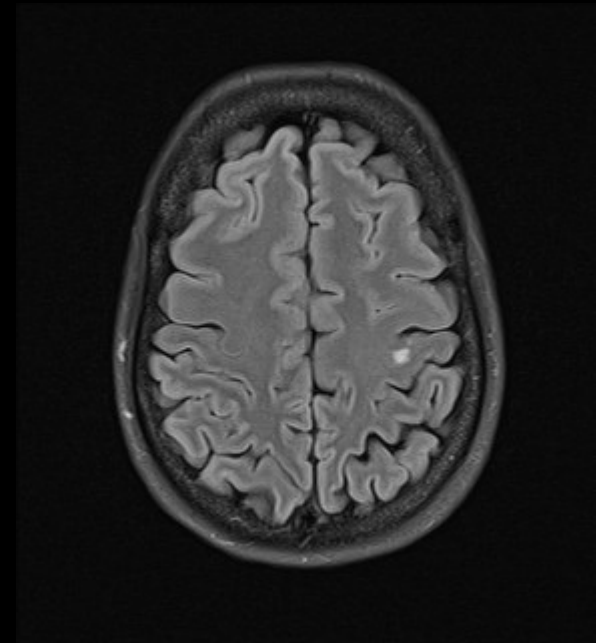
Registered report extending results to  
different risk levels: 1% vs 10%

# Next steps...

Help the ones helping

Screening versus Diagnostic tests

Gender



# Next steps...

Help the ones helping

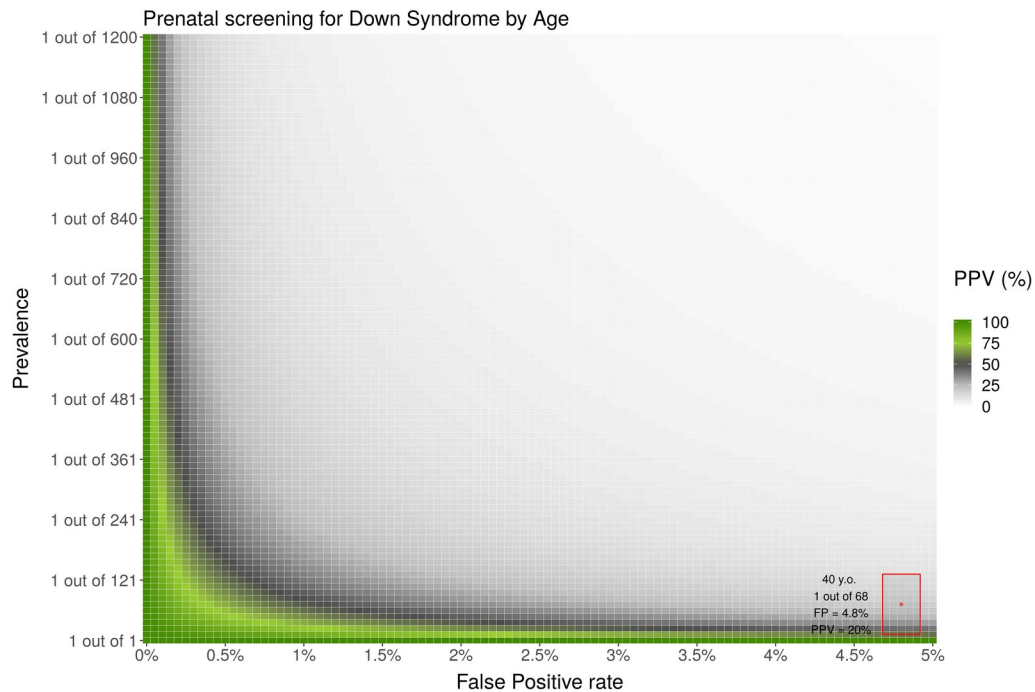
Screening versus Diagnostic tests

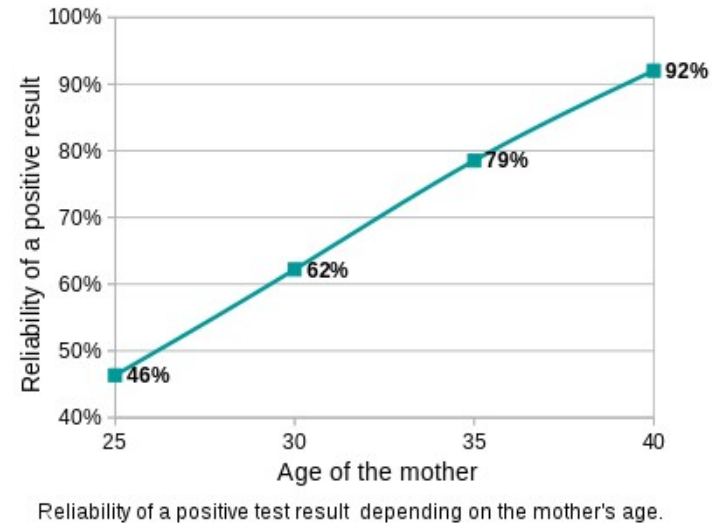
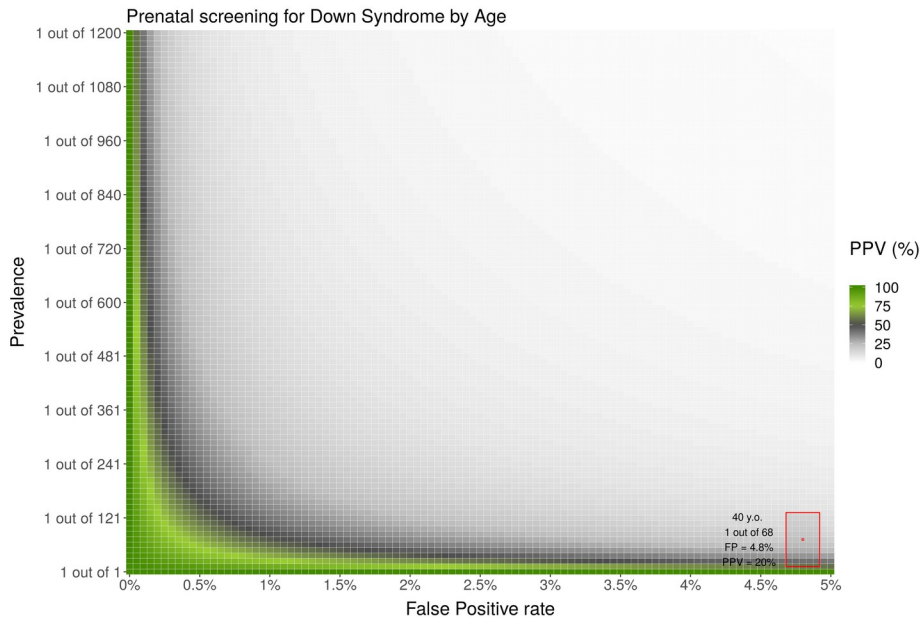
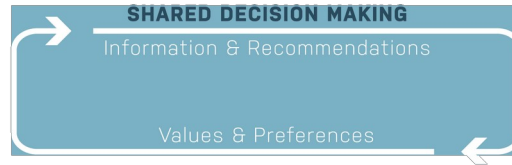
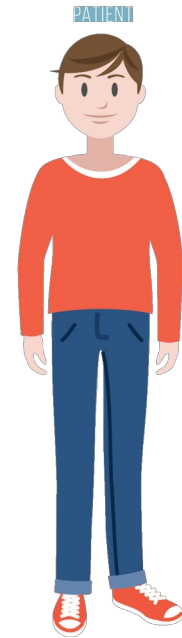
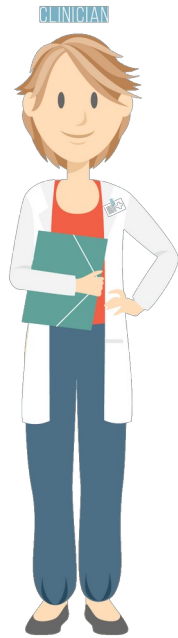
Gender

$$p(\text{Trisomy 21} \mid + \text{test}) =$$

$$(A) \frac{1 \text{ out of } 800 \times 99\%}{(1 \text{ out of } 800 \times 99\%) + (799 \text{ out of } 800 \times 0.1\%)} =$$

$$(B) \frac{0.123}{0.123 + 0.0998} = 0.55$$





# Next steps...

Help the ones helping

Screening versus Diagnostic tests

Gender

## Screening tests

NPV very high

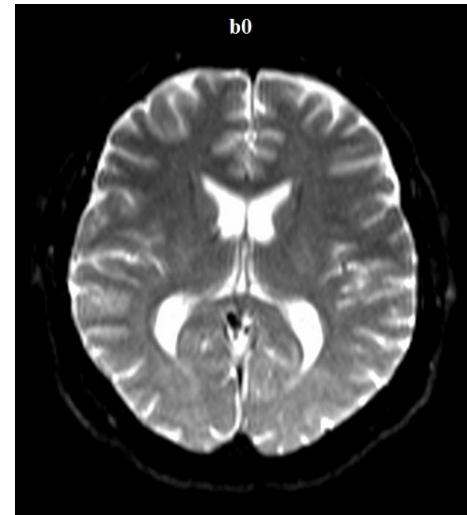
PPV, ... it depends



## Diagnostic tests

NPV, ... it depends

PPV, very high





# Next steps...

Help the ones helping

Screening versus Diagnostic tests

Gender



$$P(A|B) = \frac{P(B|A)P(A)}{P(B)}$$

Imke Hrycyk

Rut Correia

Nicolas Sanchez-Fuenzalida

David Huepe

Esteban Hurtado

Francisca Barriga-Zincke

Miroslav Sirota

Marie Juanchich

Carlos Santamaria

Wim de Neys

Elise Lesage

Dan Froimovitch



**CSCN**

**CENTRO DE NEUROCIENCIA  
SOCIAL Y COGNITIVA**

ESCUELA DE PSICOLOGÍA · UAI

Gorka Navarrete

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@gorkang



**Fondecyt**

Fondo Nacional de Desarrollo  
Científico y Tecnológico

(2015-2016) Fondecyt Regular 1150824. Communication and interpretation of diagnostic tests: behavioral and physiological variables of Bayesian reasoning

(2017-2019) Fondecyt Regular 1171035. Towards a more integrated model of Bayesian reasoning: calculation, understanding and decision making in medical contexts

16% average absolute error

40% average absolute error

Screening - PPV calculation

NEW

OLD

