ERN ITHACA

Webinar 2025

# REPRODUCTIVE GENETIC CARRIER SCREENING (1/2)

Tuesday, 21 oct 2025 - 17h00 - 18h30 CEST

Chair by

Dr Eva VAN STEIJVOORT & Leuven, Belgium

Dr Laurent PASQUIER, Rennes, France

European Commission

EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases



European Reference Networks ITHACA

# Welcome – Technical points

- **We are please to be numerous  156 registrations**
- **Webinar being recorded**

- **Thank you to**
  - Turn off your microphone and disconnect your camera
  - Use the Chat for your questions or Raise your hand at the time of the discussions (end)
  - A satisfaction survey will be sent to you :

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- **https://ern-ithaca.eu/documentation/educational-resources/**

- **Anne Hugon Project Manager ERN ITHACA - anne.hugon@aphp.fr**

# Welcome and Introduction

*”Genomic sequencing technology allows for identification of reproductive couples with an increased chance, as compared with that in the general population, of having a child with an autosomal recessive or X-linked genetic condition. As reproductive genetic carrier screening is being implemented inconstantly throughout Europe, it is time to review and discuss where the main medical, technical and ethical stakes stand”.*

*Chaired by : Laurent PASQUIER & Eva VAN STEIJVOORT*

*A few words on our speakers.*

- *Pr. Stylianos ANTONARAKIS, Geneva Hospital, Switzerland*
- *Pr. Borut PETERLIN, Ljubljana, Slovenia*
- *Pr. Pascal BORRY, Leuven, Belgium*

# Agenda

- Welcome and Introduction
  - Dr Laurent PASQUIER & Dr Eva VAN STEIJVOORT  Rennes, France & Leuven, Belgium
- Topic 1- The introduction and evolution of carrier screening in reproductive genetics
  - Pr. Stylianos ANTONARAKIS,  Geneva Hospital, Switzerland
- Topic 2 - Expanded Carrier Screening: clinical utility, limitations, and laboratory challenges
  - Pr. Borut PETERLIN,  Ljubljana, Slovenia
- Topic 3 -Expanding Choices, Expanding Questions: the ethics behind expanded carrier screening
  - Pr. Pascal BORRY, Leuven, Belgium
- Time for questions and discussion
  - Conclusion with speakers and moderator

# Topic 1- The introduction and evolution of carrier screening in reproductive genetics

Pr. Stylianos ANTONARAKIS,  Geneva Hospital, Switzerland

| Test | Samples tested | Positive | Affected fetus | Disorders | PMID |
|---|---|---|---|---|---|
| NIPT | 146314 | 1 in 161 | | T21, 18, 13 | 25598039 |
| Carrier | 346790 | 1 in 162 | 1 in 649<br>Mixed Europeans | 94<br>severe recessives<br>417<br>variants | 27533158 |
| | | 1 in 63 | 1 in 255<br>Ash Jewish | 94<br>severe recessives<br>417<br>variants | |
| | | 1 in 656 | 1 in 2624 | Cystic fibrosis | |
| | | 1 in 2336 | 1 in 9345 | SMA | |

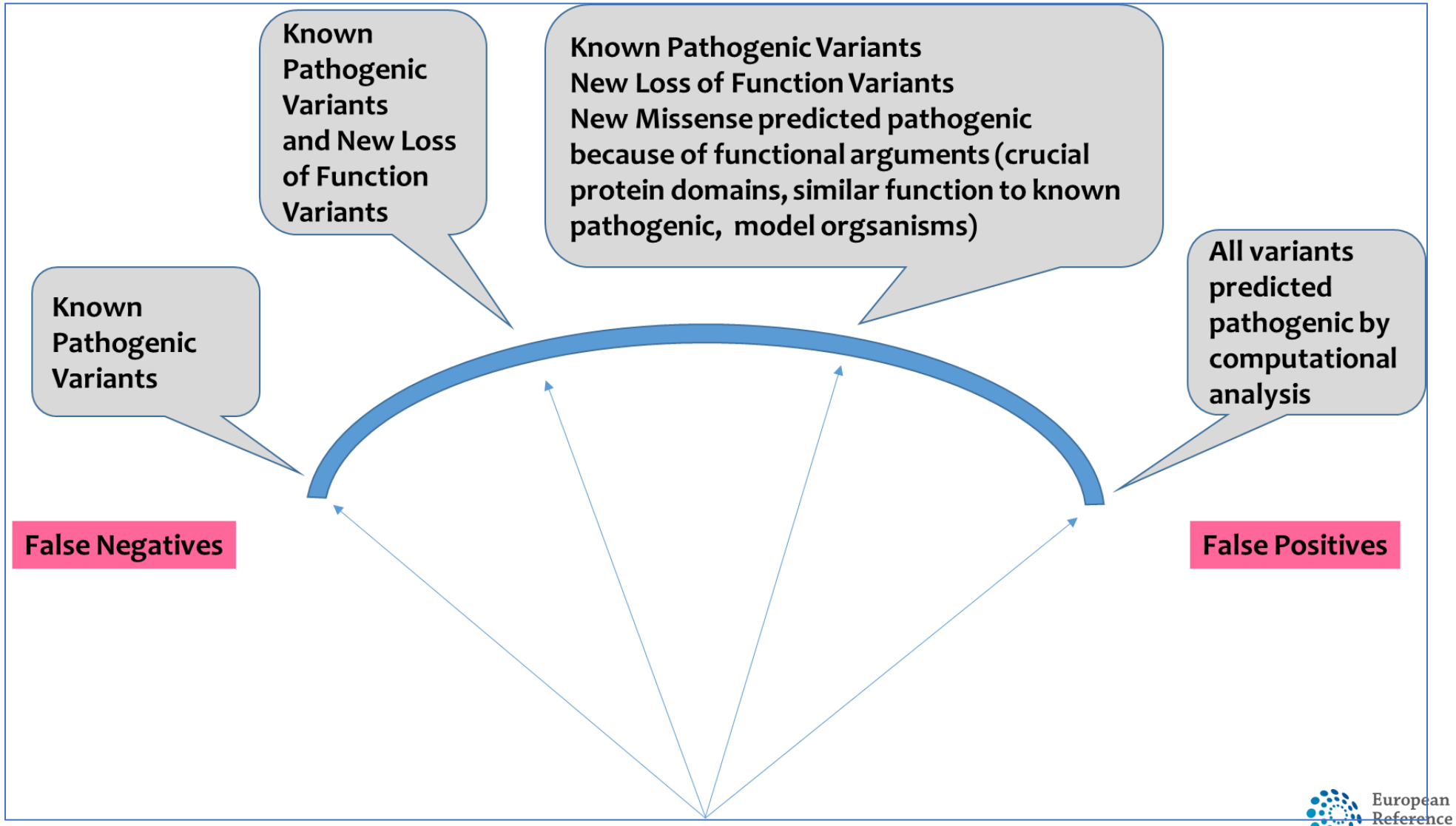
# Carrier Screening

## *Criteria for disclosing results for carrier screening*

- Include genes associated with recessive or X-linked genetic disorders:
  - When prenatal carrier screening is offered *or*
  - That meet one or more of the following criteria in *many* people with a disorder that:
    - Limits the lifespan
    - Requires significant medical involvement and/or medical care cost
    - Causes significant physical, cognitive, or sensory impairment
    - Demands significant daily care and/or care cost to the family
    - Is risky for the mother during pregnancy
    - Can be treated to significantly mitigate or reverse symptoms
- Include genes associated with disorders that meet the aforementioned criteria for only *some* people with the disorder because of variable expressivity, incomplete penetrance, or mild phenotype. Results for these genes will be clearly identified.
- Exclude genes when there is insufficient evidence of pathogenicity.
- Exclude genes in which variants are associated with risk for disease.
- Exclude conditions associated with dominant inheritance except when homozygosity or compound heterozygosity is known to be associated with a specific phenotype.

Himes et al, *Genetics in Medicine* | Volume 19 | | July 201 7

# *Interpretation of variants*



## *Recommendation*

***Establishment of an international database to provide recommendations on carrier screening of individual variants***

Antonarakis SE. *Nature Reviews Genetics* 2019

# Recessive Disorders need two variants in trans

## The complication… of allelic Tango

Mild Disease

No phenotype

Severe phenotype

## *Recommendation*

***Establishment of an international database of combination of variants for autosomal recessive disorders***

Antonarakis SE. *Nature Reviews Genetics* 2019

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# Carrier Screening

## PreviGene

*Screening of 400+ genes in outbred couples*
*New Twist Capture Reagent*
*Known pathogenic variants*
*All additional LOF*


## PreviGene wide

*Screening of all recessive genes in consanguineous couples*
*Twist Whole Exome*
*Known pathogenic variants*
*All additional LOF*

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## *“Safe” match*

## *“Cautious” match*

DOR YESHORIM

מונע דמעות
PREVENTING TEARS

*successfully eliminates the agonizing occurrence of fatal and debilitating genetic diseases in Jewish families worldwide through its premarital genetic screening program*

***30,000 individuals screened every year***
***4,775 "families" have been spared from having children with genetic disease***
***26,000 compatibility requests annually***

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New Technologies

Cheaper, high quality sequencing

Non-protein coding genes

Long-read sequencing for Structural variants

Long-read sequencing for Repeat Expansions

Long-read sequencing for Duplicated and Paralogous genes

Microarrays (known variants) ?

PRS Screening ? 😯

# Topic 2 - Expanded Carrier Screening: clinical utility, limitations, and laboratory challenges

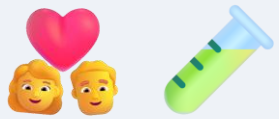
Pr. Borut PETERLIN,  Ljubljana, Slovenia.

# Clinical utility

- Reproductive utility
- Direct benefit to the proband
- Benefits to the relatives
- Public health impact

# Scenarios for ECS

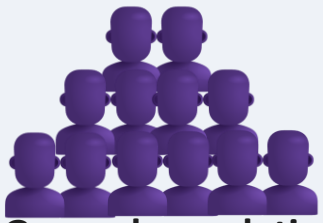
## Preconception



Couples in MAR

Gamete donors

Consanguineous couples



General population

## During pregnancy


ECS may be offered as part of antenatal care

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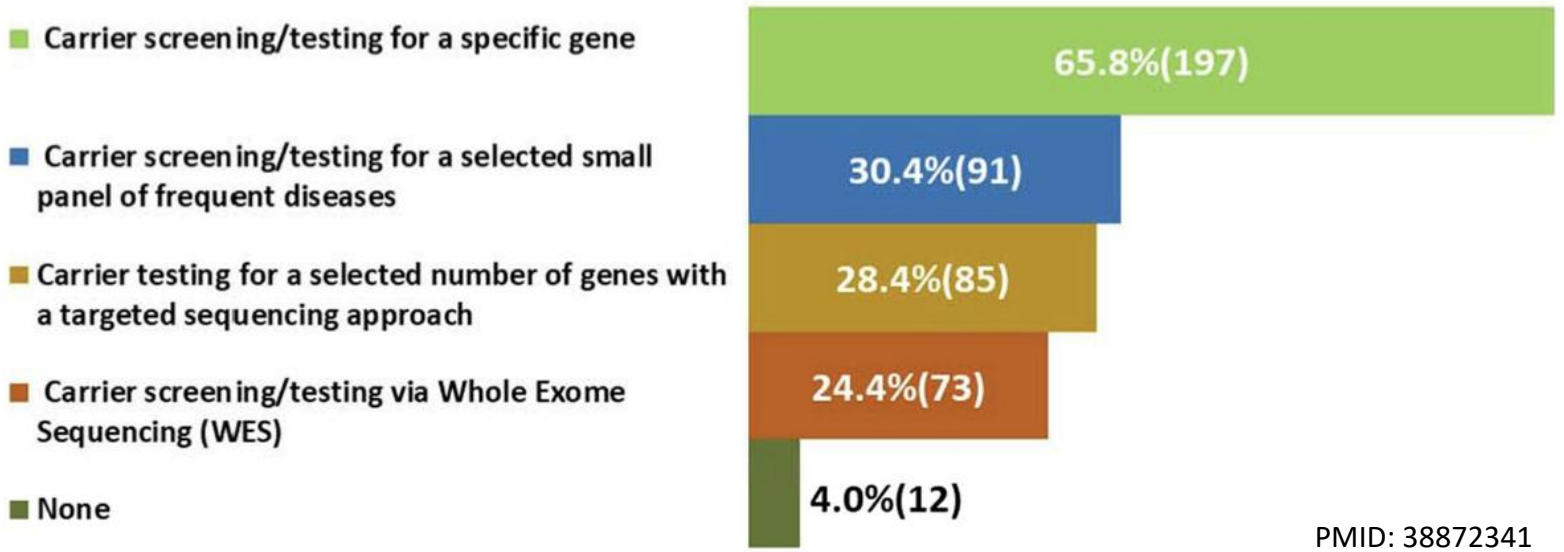
# Scenarios for ECS: couples in MAR

Couples in MAR




PMID: 38872341

**Main reason:** Perceived responsibility to enable prospective parents to make informed reproductive decisions

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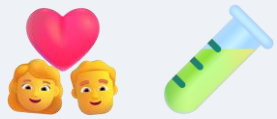
# Scenarios for ECS: couples in MAR



**Couples in MAR**

Increased rate of recessive disorders?

1.2-9.8%

PMID: 36072675, PMID: 34021342, PMID: 37450097

Recessive predispositions for subfertility?

# Scenarios for ECS: gamete donors


Offered by 62.4% of EU MAR centers PMID: 38872341

17.6% of donors rejected PMID: 33842976

2 % of donors with genetic risks (through ECS)

PMID: 38204949

# Scenarios for ECS: consanguineous couples

**Consanguineous couples**

16.5 X increased risk in 1st degree cousins

PMID: 33740458

28% of couples at increased risk

PMID: 33742171

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# Scenarios for ECS: general population


Risk of aneuplodies + CNVs = 0.4%

PMID: 37986093

Risk for recessive disorders = **1-2%**

PMID: 38872341

# Clinical utility

- Reproductive utility
- Direct benefit to the proband
- Benefits to the relatives
- Public health impact

**70 %** of couples chose to avoid

PMID: 30310157

**2 %** maternal health implications during pregnancy

PMID: 37562044

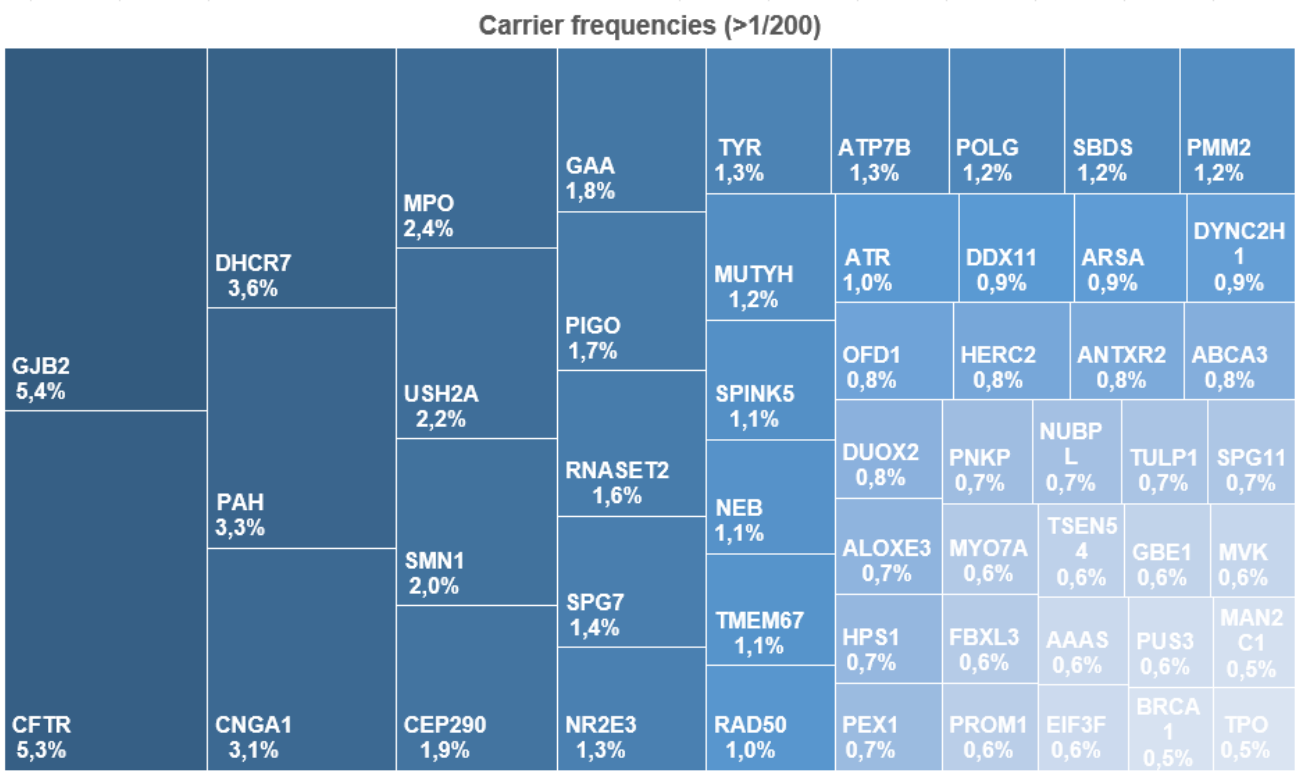
# Limitations of ECS

- Technical and analytical
  - Lack of standardisation – panels
  - Analytical sensitivity / specificity (challenging loci & pseudogenes, repeats)
  - Variants of uncertain significance
  - Residual risk
- Clinical and Counseling challenges
  - Penetrance/expressivity
  - Counselling capacity
- Implementation & Regulatory
  - Guideline variability
  - Real-world heterogeneity

# Panel design

| | ESHG | ACMG American College of Medical Genetics and Genomics | ACOG American College of Obstetricians & Gynecologists |
|---|---|---|---|
| Carrier frequency | Comprehensive | >1/200 | >1/100 |
| Severity | Severe, childhood onset | Detrimental effect on quality of life, cause cognitive or physical impairment, require surgical or medical intervention, early onset in life | Severity that may impact decision-making (moderate, severe, and profound) |

# Risk for recessive disorders in the Slovenian population




52 genes with frequency > 1/200: 91% of RD risk

Cummultaive risk in Slovenians: 1.4%

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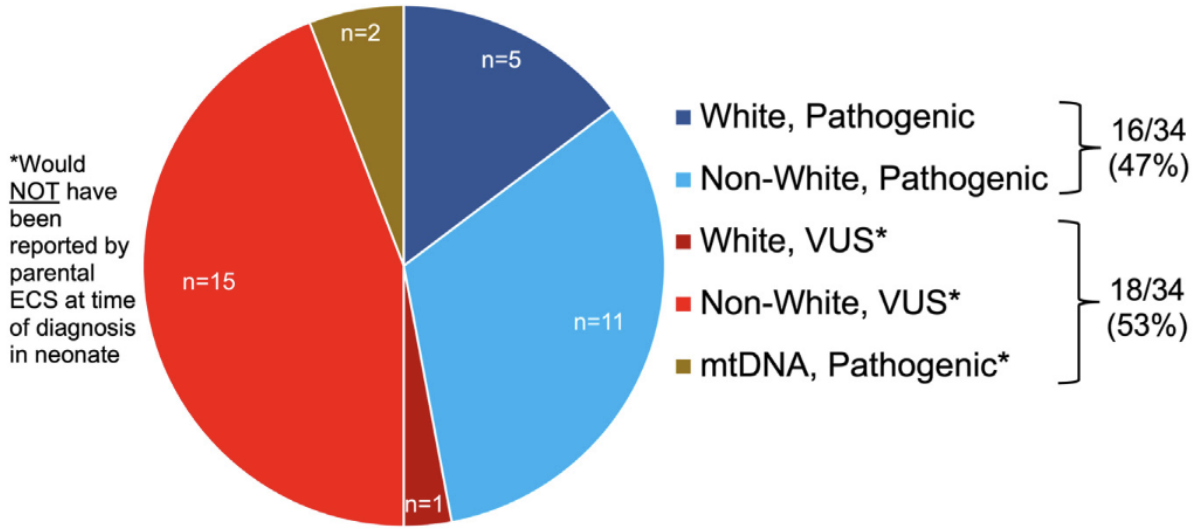


PMID: 38146699

# Limitations of ECS

- Technical and analytical
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  - Real-world heterogeneity

# Topic 3 -Topic 3 -Expanding Choices, Expanding Questions: the ethics behind expanded carrier screening

Pr. Pascal BORRY, Leuven, Belgium .

KU LEUVEN

# Expanded carrier screening: ethical challenges to responsible implementation

Pascal Borry
Centre for Biomedical Ethics and Law, Department of Public Health and Primary Care
KU Leuven

# Carrier testing and screening

- **Two approaches in the identification of carriers:**

**(1) Carrier testing** is defined as the detection of carrier status in persons who do have a higher a priori risk based on their or their partners' personal or family history (Castellani et al., 2010).

**(2) Carrier screening** is defined as the detection of carrier status in persons who do not have an a priori increased risk for having a child with a certain disease.

# Carrier screening

- 7028 with suspected Mendelian inheritance
- 1139 are recessive
- Analysis for 7717 regions from 437 target genes (448 severe recessive childhood diseases)
- Average genomic carrier burden 2,8 (range 0-7)

(Bell et al. 2011)

# Expansion of carrier screening

- Ethnicity based screening would be replaced

**A comparison of ethnicity-based carrier screening guidelines between ACOG and ACMG.**

| Disease | Recommended by ACOG | ACMG |
|---|---|---|
| Ashkenazi Jewish | | |
| Tay-Sachs | X | X |
| Canavan disease | X | X |
| Familial dysautonomia | X | X |
| Bloom syndrome | | X |
| Gaucher disease | | X |
| Fanconi anemia type C | | X |
| Mucolipidosis IV | | X |
| Niemann-Pick disease type A | | X |
| African | | |
| Sickle cell disease | X | |
| Asian | | |
| Alpha thalassemia | X | |
| Beta thalassemia | X | |
| Panethnic | | |
| Cystic fibrosis | X | X |
| Spinal muscular atrophy | | X |

*Ready. Expanded genetic carrier screening. Fertil Steril 2012.*

Reproductive BioMedicine Online (2010) 21, 537–551

ELSEVIER

www.sciencedirect.com
www.rbmonline.com

ARTICLE

**A universal carrier test for the long tail of Mendelian disease**

Balaji S Srinivasan [a,b,c,*,1], Eric A Evans [a,1], Jason Flannick [a,d], A Scott Patterson [a], Christopher C Chang [a,e], Tuan Pham [a], Sharon Young [a], Amit Kaushal [a,f,g,h], James Lee [a,i,l], Jessica L Jacobson [a,j], Pasquale Patrizio [a,k]


KU LEUVEN

# (Preconceptional) (expanded) carrier screening

- Can increase reproductive decision-making:
  - Becoming aware of possible genetic risks to future offspring
  - Strenghthen reproductive autonomy and informed decision-making
  - In case of preconceptional carrier screening, less time constraints, less pressure, and less emotional stress than when a test is performed during pregnancy
- Could lead to reduction of disease incidence
- Could enable perinatal diagnosis and treatment, which can diminish disease severity

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Starting a family is an important choice. Knowing your risk of having a child with a serious disorder is important, too. Make the choice to know, with Good Start — the right choice for carrier screening.

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- Extensive Validation
- Proven Clinical Performance
- Industry Expertise

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CARRIER STATUS DNA INSIGHT™

Pathway Genomics' carrier status test provides relevant genetic insights to inform physicians about the health of their patients' future children. By following the American College of Obstetricians and Gynecologists (ACOG) recommendations, we offer a comprehensive preconception and prenatal carrier screening test.

offers more ACOG-recommended conditions than most other carrier tests on the information from a single source.

Hum. Reprod. Advance Access published February 28, 2011

Human Reproduction, Vol.0, No.0 pp. 1–6, 2011

doi:10.1093/humrep/der042

human reproduction OPINION

# Preconceptional genetic carrier testing and the commercial offer directly-to-consumers

Pascal Borry[1,2,3,*], Lidewij Henneman[2], Phillis Lakeman[2], Leo P. ten Kate[2], Martina C. Cornel[2], and Heidi C. Howard[1]

[1]Centre for Biomedical Ethics and Law, Katholieke Universiteit Leuven, Kapucijnenvoer 35 BOX 7001, 3000 Leuven, Belgium [2]Department of Clinical Genetics, VU University Medical Center, and EMGO Institute for Health and Care Research, P.O. Box 7057, 1007 MB, The Netherlands [3]Department of Medical Humanities, VU University Medical Center, and EMGO Institute for Health and Care Research, P.O. Box 7057, 1007 MB, The Netherlands

# Analysis of carrier screening offers (2017)

To analyze and compare the characteristics of ECS panels across providers

- Size of ECS panels
- Nature of disorders included
- Approaches used (mutation panel/sequencing gene)
- Adherence to professional guidelines regarding ECS

DOI: 10.1002/pd.5109

PRENATAL **DIAGNOSIS**

SPECIAL TOPIC ISSUE ON ADVANCES IN THE DIAGNOSIS OF SINGLE GENE DISORDERS

## Expanded carrier screening for monogenic disorders: where are we now?

Davit Chokoshvili* , Danya Vears and Pascal Borry

Centre for Biomedical Ethics and Law, Department of Public Health and Primary Care, University of Leuven, Leuven, Belgium

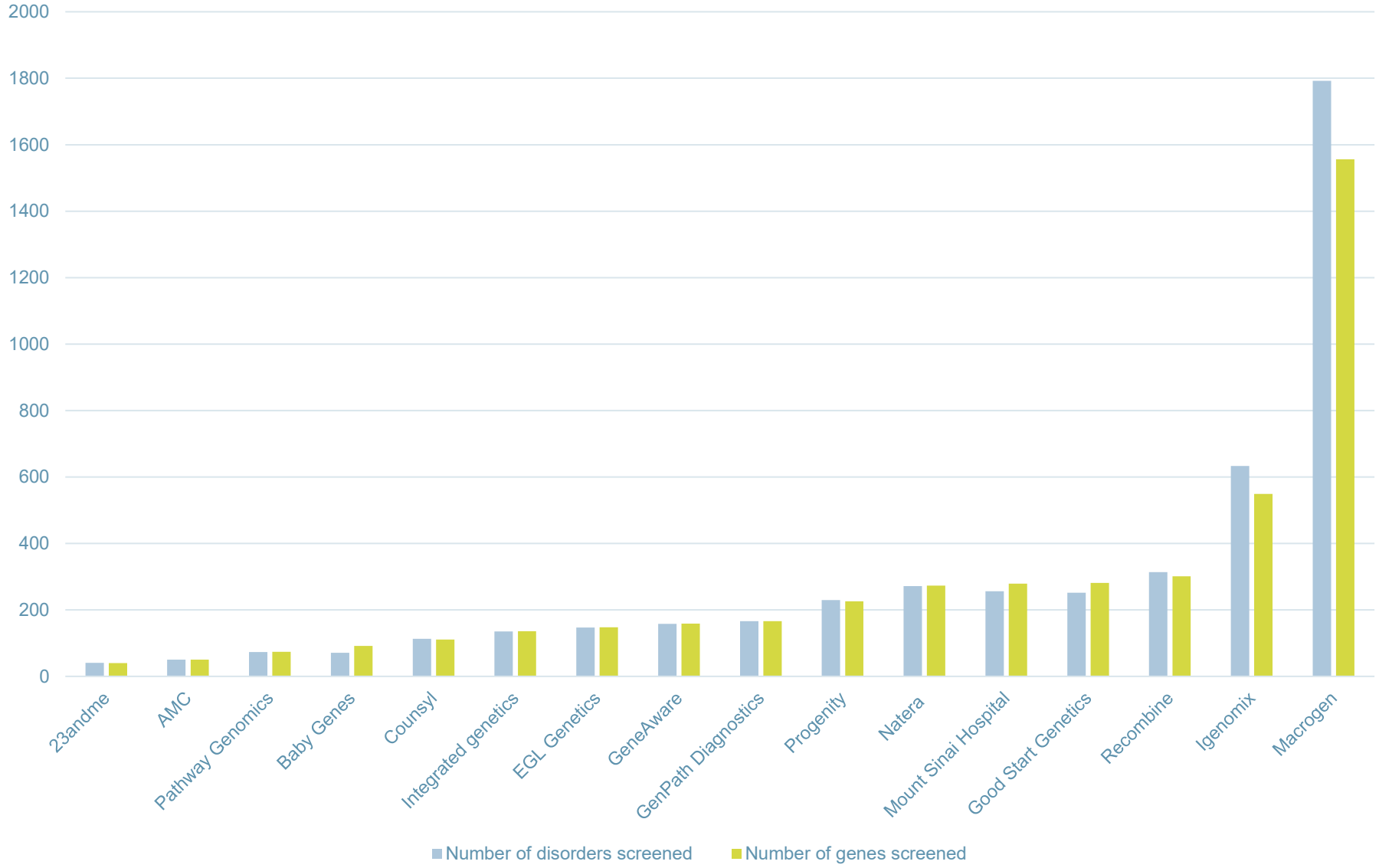
*Correspondence to: Davit Chokoshvili. E-mail: davit.chokoshvili@kuleuven.be

# Results

As of January 2017, sixteen relevant ECS providers were identified

- United States (13), the Netherlands (1), South Korea (1), Spain (1)
- Number of conditions ranged from 41 to >1700
    - Only three conditions (Cystic fibrosis, Maple syrup urine disease 1b, and Niemann-Pick disease) were screened for by all providers.
- Differences in variant inclusion and/or interpretation
- Varying adherence to professional guidelines (e.g. ACMG & ACOG 2015 recommendations on ECS)

# Size of ECS panels across providers



# Differences across mutation panels

Table 4 Screening strategies and the size of mutation panels for the three genes screened for by all 16 providers

| Provider | Maple syrup urine disease type 1B (BCKDHB gene) | Cystic fibrosis and other CFTR-related disorders (CFTR gene) | Niemann–Pick disease A/B (SMPD1 gene) |
|---|---|---|---|
| 23andMe | TG (two variants) | TG (28 variants) | TG (three variants) |
| Baby Genes | Seq. | Seq. | Seq. |
| Baylor Miraca Genetics Laboratories | Seq. | CNV + Seq. | Seq. |
| Counsyl | TG (three variants) + Seq. | TG (99 variants) + Seq. | TG (four variants) + Seq. |
| EGL Genetics | TG or Seq. | TG or Seq. | TG or Seq. |
| GenPath Diagnostics | TG (three variants) | TG (220 variants) | TG (six variants) |
| Good Start Genetics | TG + Seq. | TG + Seq. | TG + Seq. |
| Igenomix | TG (24 variants) | TG (146 variants) | TG (42 variants) |
| Integrated Genetics | TG | TG (609 variants) | TG |
| Macrogen | TG (one variant) | TG (102 variants) | TG (14 variants) |
| Mount Sinai Hospital | Seq. | TG + Seq. | Seq. |
| Natera | TG (21 variants) + Seq. | TG (579 variants) + Seq. | TG (50 variants) + Seq. |
| Pathway Genomics | TG (three variants) | TG (82 variants) | TG (five variants) |
| Progenity | TG (three variants) | TG (656 variants) | TG (four variants) |
| Recombine | TG (six variants) | TG (150 variants) | TG (nine variants) |
| Academic Medical Center Amsterdam | TG | TG | TG |

TG, Targeted genotyping; Seq., (nontargeted) sequencing; CNV, copy number variation analysis.

# Adherence to professional guidelines

Table 3 Providers' practices regarding the inclusion of alpha 1 antitrypsin deficiency, MTHFR deficiency, and hereditary hemochromatosis on their expanded carrier screening panels

| Provider | Alpha 1 antitrypsin deficiency (SERPINA1 gene) | MTHFR deficiency (MTHFR gene) | Hereditary hemochromatosis (HFE, HFE2, and TFR2 genes) |
|---|---|---|---|
| 23andMe | Not included | Not included | Not included |
| Baby Genes | Not included | Included | Not included |
| Baylor Miraca Genetics Laboratories | Included | Not included | Not included |
| Counsyl | Included | Not included[a] | Not included[a] (HFE) |
| EGL Genetics | Included | Not included | Included (HFE) |
| GenPath Diagnostics | Not included | Not included | Not included |
| Good Start Genetics | Not included | Included | Included (HFE2, TFR2) |
| Igenomix | Included | Included | Included (HFE, TFR2) |
| Integrated Genetics | Not included | Not included | Not included |
| Macrogen | Not included | Included | Included (HFE) |
| Mount Sinai Hospital | Not included | Included | Included (HFE2,TFR2) |
| Natera | Not included | Included | Included (HFE2,TFR2) |
| Pathway Genomics | Included | Not included | Included (HFE) |
| Progenity | Included | Not included | Included (HFE, HFE2, TFR2) |
| Recombine | Included | Included | Included (HFE2, TFR2) |
| Academic Medical Center Amsterdam | Not included | Not included | Not included |

[a]Not part of the standard screening panel but can be included if specifically requested by the consumer.

Inclusion actively discouraged by professional organisations

# Lack of consensus

Apparent lack of consensus on

- What disorders to include
- What mutations to include and/or how to interpret pathogenicity

Greater harmonization is needed
to reduce heterogeneity of ECS panels
across providers

Research Article

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Personalized Medicine

# Designing expanded carrier screening panels: results of a qualitative study with European geneticists

**Aim:** To explore the views of clinical and molecular geneticists on the inclusion of disorders and specific pathogenic mutations into expanded carrier screening (ECS) tests for reproductive purposes. **Materials & methods:** In-depth semistructured interviews were conducted with 16 European geneticists between April and September 2014. **Results:** All participants supported carrier screening for severe, childhood-onset autosomal recessive disorders with known natural history. Some participants were

Davit Chokoshvili*[,1], Sandra Janssens[2], Danya Vears[1] & Pascal Borry[1]

[1]Centre for Biomedical Ethics & Law, Department of Public Health and Primary Care, University of Leuven, Kapucijnenvoer 35, Box 7001,

# Results: composition of ECS panels

| What should be the criteria for including disorders/phenotypes on ECS panels? | |
|---|---|
| ***Criteria for inclusion*** | 1. Disorders resulting in:<br>- Childhood mortality<br>- Severe childhood disability<br>- Chronic health problems requiring continuous treatment<br>2. Disorders whose natural course can be accurately predicted based on the genotype |
| ***Criteria for exclusion*** | ➢ Disorders considered to be mild<br>➢ Disorders where genotype is a poor predictor of the clinical phenotype |
| ***No clear agreement*** | ➢ Disorders predominantly manifesting in adulthood<br>➢ Discussion on correct interpretation of what is 'severe' or 'mild' |

# Carrier screening

EJHG Open

European Journal of Human Genetics (2016), e1–e12

www.nature.com/ejhg

POLICY

## Responsible implementation of expanded carrier screening

Lidewij Henneman[1], Pascal Borry[2], Davit Chokoshvili[2,3], Martina C Cornel[1], Carla G van El[1], Francesca Forzano[4], Alison Hall[5], Heidi C Howard[6], Sandra Janssens[3], Hülya Kayserili[7], Phillis Lakeman[8], Anneke Lucassen[9], Sylvia A Metcalfe[10], Lovro Vidmar[11], Guido de Wert[12], Wybo J Dondorp[12] and Borut Peterlin*,[11] on behalf of the European Society of Human Genetics (ESHG)

This document of the European Society of Human Genetics contains recommendations regarding responsible implementation of expanded carrier screening. Carrier screening is defined here as the detection of carrier status of recessive diseases in couples or persons who do not have an *a priori* increased risk of being a carrier based on their or their partners' personal or family history. Expanded carrier screening offers carrier screening for multiple autosomal and X-linked recessive disorders, facilitated by new

Superior Health Council

**ADVISORY REPORT OF THE SUPERIOR HEALTH COUNCIL no. 9240**

**Expanded carrier screening in a reproductive context. Towards a responsible implementation in the healthcare system**

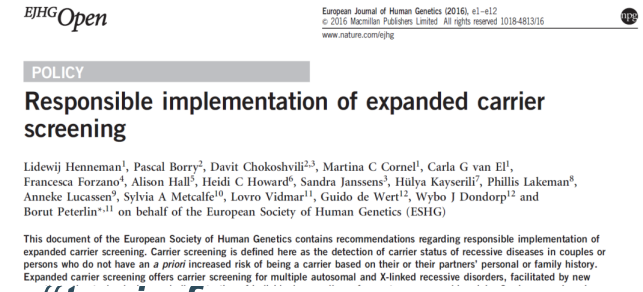
In this advisory report, the Superior Health Council of Belgium provides recommendations on the criteria that need to be applied in preconceptual genetic testing for severe autosomal and X-linked recessive diseases for couples planning a pregnancy.

This report aims at providing healthcare authorities and healthcare professionals with specific recommendations on the scientific and ethical issues that need to be considered in view of a responsible implementation of preconceptual genetic testing in a reproductive context. The report specifically discusses the framework underpinning the appropriate introduction of such testing and suggests inclusion criteria for diseases that could be targeted by the screening process: (i) severity, (ii) age of onset, (iii) prevalence, (iv) selection of mutations based on clinical significance and (v) treatability.

This version was validated by the Board on February 2017[1]

**Advisory Report 9240 – March 2017**

# ESHG recommendations



EJHG Open

European Journal of Human Genetics (2016), e1–e12
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www.nature.com/ejhg

POLICY

**Responsible implementation of expanded carrier screening**

Lidewij Henneman[1], Pascal Borry[2], Davit Chokoshvili[2,3], Martina C Cornel[1], Carla G van El[1], Francesca Forzano[4], Alison Hall[5], Heidi C Howard[6], Sandra Janssens[3], Hülya Kayserili[7], Phillis Lakeman[8], Anneke Lucassen[9], Sylvia A Metcalfe[10], Lovro Vidmar[11], Guido de Wert[12], Wybo J Dondorp[12] and Borut Peterlin*[,11] on behalf of the European Society of Human Genetics (ESHG)

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- ***(1) Primary purpose of carrier screening**: "to inform them of possible genetic disease risks in future offspring and of the **reproductive options** available in order to enable autonomous choices."*
- ***(2) Focus on "severe childhood-onset disorders."** The main focus should be on "reporting **sequence variants that clearly affect function (with clear clinical significance)."***
- ***(12) Governance**: Governments and public health authorities should adopt an **active role in discussing the responsible introduction** of expanded carrier screening.*

# Moral dimensions

(1) Technology allows for more responsibilities
- (Reproductive) freedom and responsibilities

(2) Individual choices and society

(3) Technology is not neutral
- medicalisation

Questions on desirability and responsible implementation

# Equity with regard to service offerings

- Uncoordinated offerings lead to inequality in access
- Inequality in access (related to financial means, access to information and understanding of information)
- Importance of “accurate, balanced information” (about the test, but also of consequences)
- Importance that information is given “equally to all parents, regardless of their social status, level of education or place of residence”
- Therefore, “urgent need to set guidelines and thus ensure a certain standardization in the practice”

# Perception and integration of persons with disabilities

- Disability right critique
- Message through the screening:
  - “negative event”;
  - “undesirable and incompatible with leading a rewarding life”;
  - “individuals who should not exist”;
  - “increasing our society’s intolerance toward disabled persons”

# Normalcy, difference and the search for the perfect baby

- What is a “normal” child?
- What is a “life worth living”?
- Geneticization (variation or mutation?)


UW KINDJE HEEFT EEN GENETISCHE AFWIJKING...

# Eugenics, discrimination and stigmatization

- Prenatal screening programs: “endorsement of eugenics”
- “While the individual decision to resort to abortion does not in itself constitute eugenics, the cumulative effect of these individual decisions on society raises the question”

- Is this really the case? Eugenics would mean that reproductive choices are not voluntary anymore

# Prevention versus informed decision-making

- Tension between prevention and reproductive decision making
- Interaction between private sphere and society
  - Reproductive decisions are influenced by the context in which they are made
  - Reproductive decisions have an impact on society and general perceptions on the value of human life

# Resources available to future parents

- Financial and psychological support system for handicapped persons, along with the presence of appropriate infrastructures
- Still research being done on disability rather than on the screening procedure?

# Screening is not neutral

- **Initiative is not coming from individual**

  + : people who would otherwise have failed to take initiatives in order to be tested, perhaps due to ignorance, are given an opportunity to gain from the possible benefits of being tested

  - : risk of reducing autonomy and provoking unnecessary anxiety, risk of pressure

# Why screening?

Screening: between hope and hype

- Health Council of the Netherlands (2008)
  - Significant health problem
  - Benefit: ratio of advantages to disadvantages
  - Reliable and valid instrument
  - Respect for autonomy
  - Responsibility in terms of cost-effectiveness

# Benefit: ratio of advantages and disadvantages

- Broader international consensus: “the aim of prenatal screening should not be worded in terms of prevention of health gain, but as giving those concerned worthwile options from which to choose.”
- “practical courses of action for participants” (“zinvolle handelingsopties”)

# Respect for autonomy

- Screening trap: potential participants must be informed of the possible subsequent developments and their implications at the start of the process
- Balance between sufficient information and information overload
- Concept of ‘Generic’ consent

# Potential concerns

- Potential harmful effects of uncoordinated offers
- Lack of consensus on the scope of carrier screening
- Reproductive autonomy versus prevention
- Social pressure
- Pre-test information and post-test information and counselling

# Potential concerns

- Impact on psychological well-being
- Impact on perception of health
- Impact on relationships
- False reassurance among non-carriers (residual risk); small fraction of disorders that could be screened
- Stigmatisation and discrimination

# Time for questions and discussion

- Time for questions

- Satisfaction Survey :

https://forms.office.com/e/CUYyYda3ES

- Website :
  - https://ern-ithaca.eu
  - https://ern-ithaca.eu/webinars/

***Thank you for your participation***