ETH zürich

Understanding Global Practices on Prenatal Genetic Screening

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CONFLICTS OF INTEREST

- Nothing to disclose

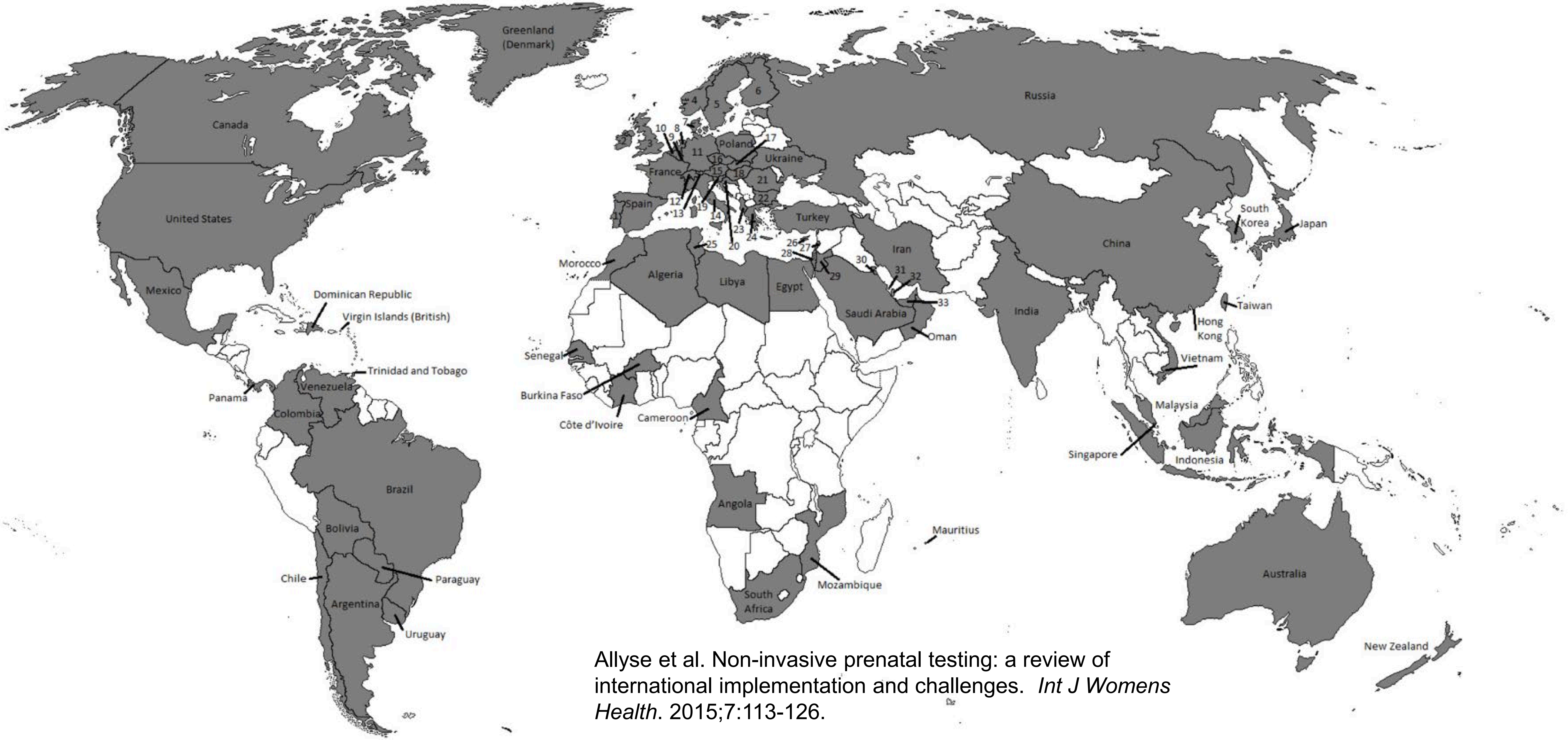
EXCEPTIONALISM OF PRENATAL GENETIC SCREENING

- Standard component of prenatal care in many countries
- Ethically distinct from other tests offered during pregnancy

  → Follow-on decision-making

  → Non-invasive maternal blood draw
- Professional & societal deliberation on its ethical provision

GLOBAL AVAILABILITY OF PRENATAL GENETIC SCREENING



Allyse et al. Non-invasive prenatal testing: a review of international implementation and challenges. *Int J Womens Health*. 2015;7:113-126.

BROAD VARIABILITY IN IMPLEMENTATION

Gadsbøll et al. Current use of noninvasive prenatal testing in Europe, Australia and the USA: A graphical presentation. *Acta Obstet Gynecol Scand*. 2020;99(6):722-730.

- Public Funding:
  - Limited to T21, 18, 13
  - Inclusion of SCA controversial
  - Addition of selected RATs
  - Genome-wide approach, including microdeletions & RATs

- Screening Workflow:
  - Universal screening
  - Contingent screening for intermediate and/or high-risk patients

SOCIO-POLITICAL CONTEXT DIFFERS ACROSS COUNTRIES

Health Care Systems

Public Prenatal Screening Programs

Laws & Guidelines on Clinical Practice

Legal Status of Abortion

Acceptance & Support for People with Disabilities

Maternal, Perinatal, & Public Health

...impact implementation of prenatal genetic screening to reflect specific societal values

Examine global practices and how ethical values on prenatal genetic screening are operationalized

ETHICAL VALUES OF PRENATAL GENETIC SCREENING

- Informed consent
- Reproductive autonomy
- Disability rights
- Right not to know
- Right to privacy
- Right to an open future
- Human dignity
- Distributive justice
- Health equity

INFORMED CONSENT

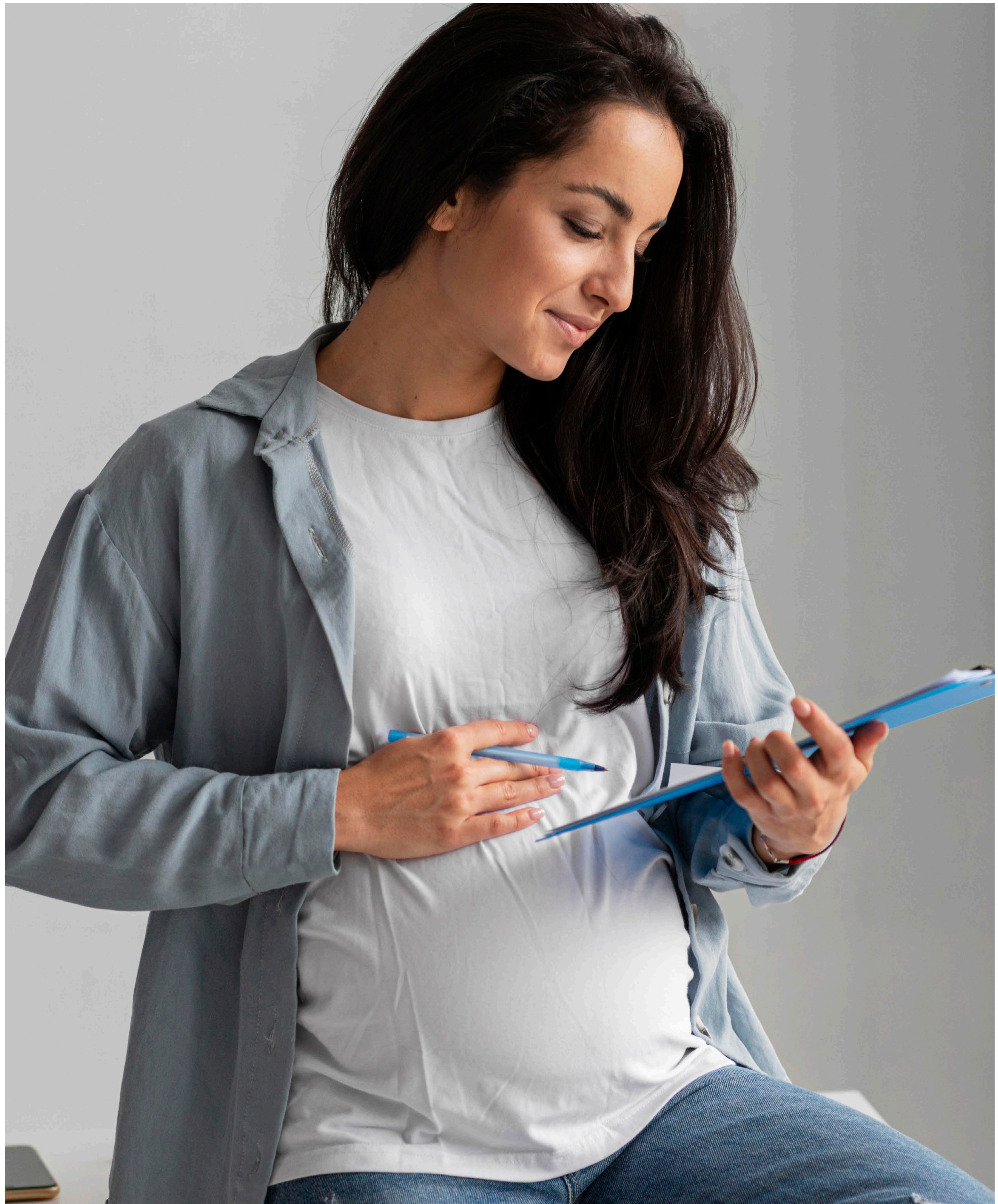
- Counseling for sufficient patient understanding of a test for autonomous decision-making
  - →Process of **moral deliberation** so that the decision is consistent with their values
  - →Potential for harm

INFORMED CONSENT – PRE-TEST COUNSELING

- National clinical guidelines & laws stress importance of informed consent
  - Specific components:
    - Legally mandated
    - Professional guidance
  - Specific training

→ Goal: Standardization of practice

→ But challenges with implementation

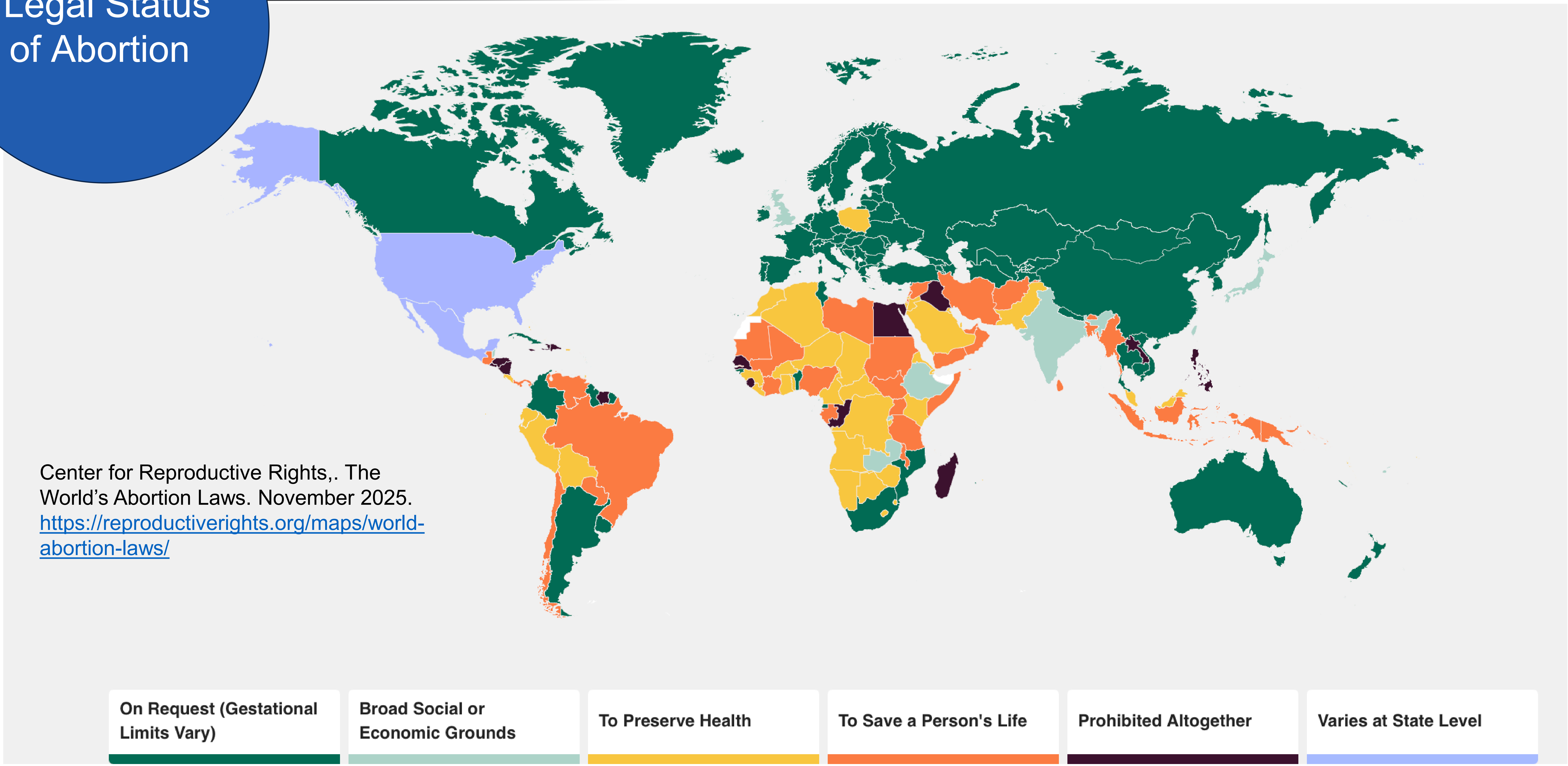


REPRODUCTIVE AUTONOMY

- Individual's right to make informed choices regarding reproductive health without coercion or interference
- Prenatal Genetic Screening:
  - → Right to know information about fetal health
  - → Right *not* to know
  - → Autonomous decisions about the pregnancy

Legal Status of Abortion

GESTATIONAL LIMITS; PERMITTED REASONS



Center for Reproductive Rights,. The World’s Abortion Laws. November 2025. https://reproductiverights.org/maps/world-abortion-laws/

REPRODUCTIVE AUTONOMY

- Value of reproductive autonomy highly stressed in HIC
  - Debate whether basing access on risk status is an infringement
  - Emphasis on right not to know

→ Patient misinformation on voluntariness

→ Repeated emphasis on legal right to TOP perceived as coercive

REPRODUCTIVE AUTONOMY

- Concerns about ensuring TOP only for “serious” reasons
  - Limit test to T21, 18, 13 only
  - Prohibitions on disclosing fetal sex prior to TOP limits
  - TOP for sex selection a “grey area” and a “particular ethical concern” when within legal limits

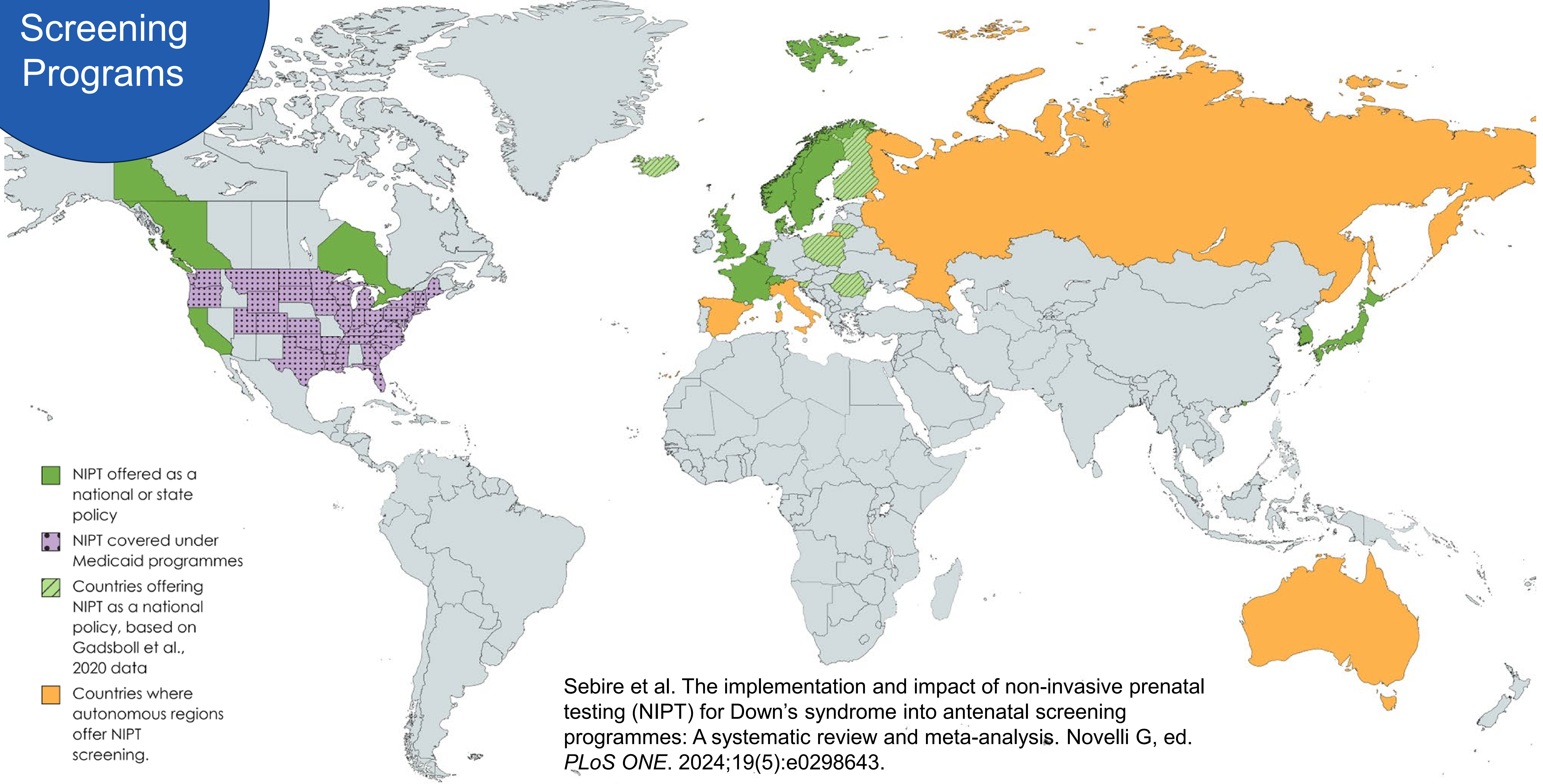
DISABILITY RIGHTS

- Equal opportunities and rights for people with disabilities, including accessibility, non-discrimination, and inclusion in society
- Prenatal Genetic Testing:

→ Critique: Promotes stigma against people with disabilities
→ But, could offer parents a chance to prepare
→ Accurate, up-to-date information on lived experience of a genetic condition

Public Prenatal Screening Programs

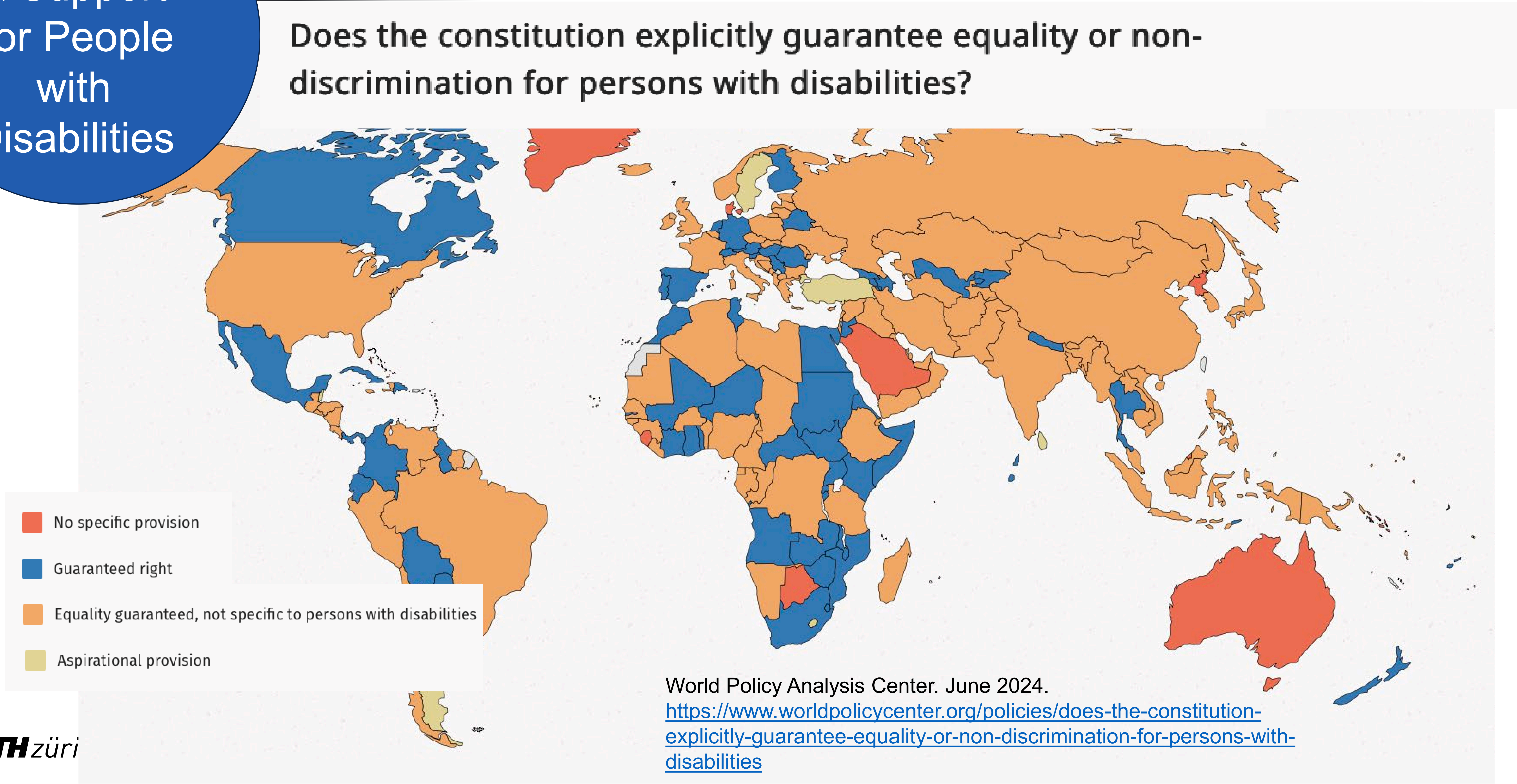
GLOBAL AVAILABILITY OF SCREENING PROGRAMS



Sebire et al. The implementation and impact of non-invasive prenatal testing (NIPT) for Down’s syndrome into antenatal screening programmes: A systematic review and meta-analysis. Novelli G, ed. *PLoS ONE*. 2024;19(5):e0298643.

Acceptance & Support for People with Disabilities

STIGMA AGAINST PEOPLE WITH DISABILITIES COMMON; NATIONAL RESOURCES VARIABLE



Does the constitution explicitly guarantee equality or non-discrimination for persons with disabilities?

World Policy Analysis Center. June 2024. https://www.worldpolicycenter.org/policies/does-the-constitution-explicitly-guarantee-equality-or-non-discrimination-for-persons-with-disabilities

DISABILITY RIGHTS

- National vs. siloed conversations on impact of prenatal testing on people with disabilities
  - → Highly variable termination rates
  - → Expansion could lead to less tolerance of people with disabilities

DISABILITY RIGHTS

- Diversity of cultures, religions, & values on children and disability
  - →Value of fertility may encourage birth over TOP
  - → Influence of societal acceptance & visibility of people with disabilities

DISABILITY RIGHTS

- Some efforts to prevent disability bias during post-test counseling:
  - Laws mandating up-to-date information
  - Connection to patient support groups

  →Disability bias has not improved in prenatal counseling over time

HEALTH EQUITY

- Equal opportunity to attain the highest level of health
  - → Reducing barriers to comprehensive prenatal care
  - → Prioritizes addressing health needs of a disproportionately impacted population

HEALTH EQUITY IN LOW RESOURCE SETTINGS

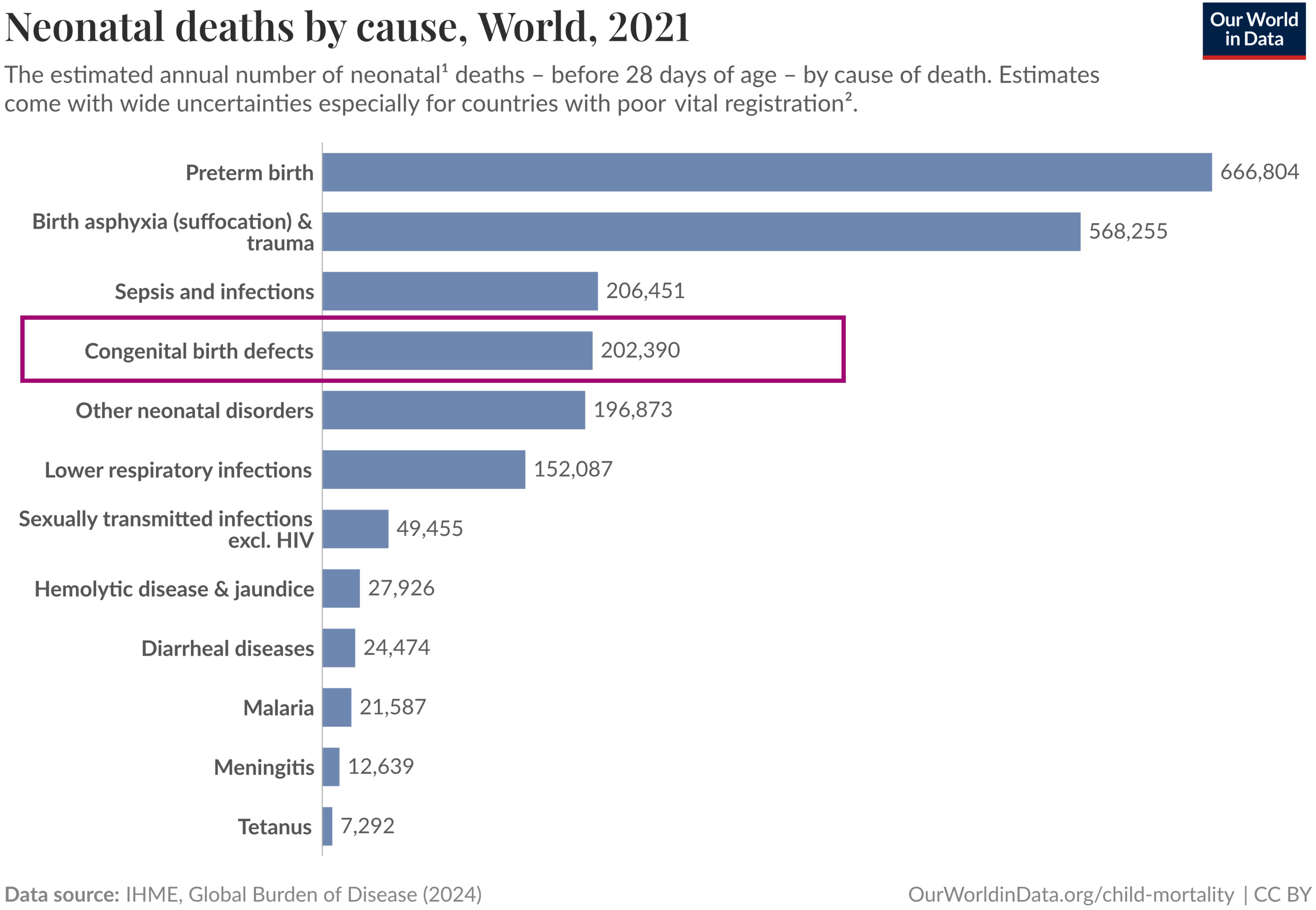
- Context of overall limited availability of prenatal care:
  - → Low uptake & late entry to prenatal care
  - → Diagnosis after legal limits of TOP

- Cost of prenatal genetic screening is prohibitive in LMICs

- Expansion of prenatal genetic testing may not be the priority

Maternal, Perinatal, & Public Health

DISEASE BURDEN SHOULD REFLECT ALLOCATION OF PUBLIC HEALTH RESOURCES



Neonatal deaths by cause, World, 2021

The estimated annual number of neonatal[1] deaths – before 28 days of age – by cause of death. Estimates come with wide uncertainties especially for countries with poor vital registration[2].

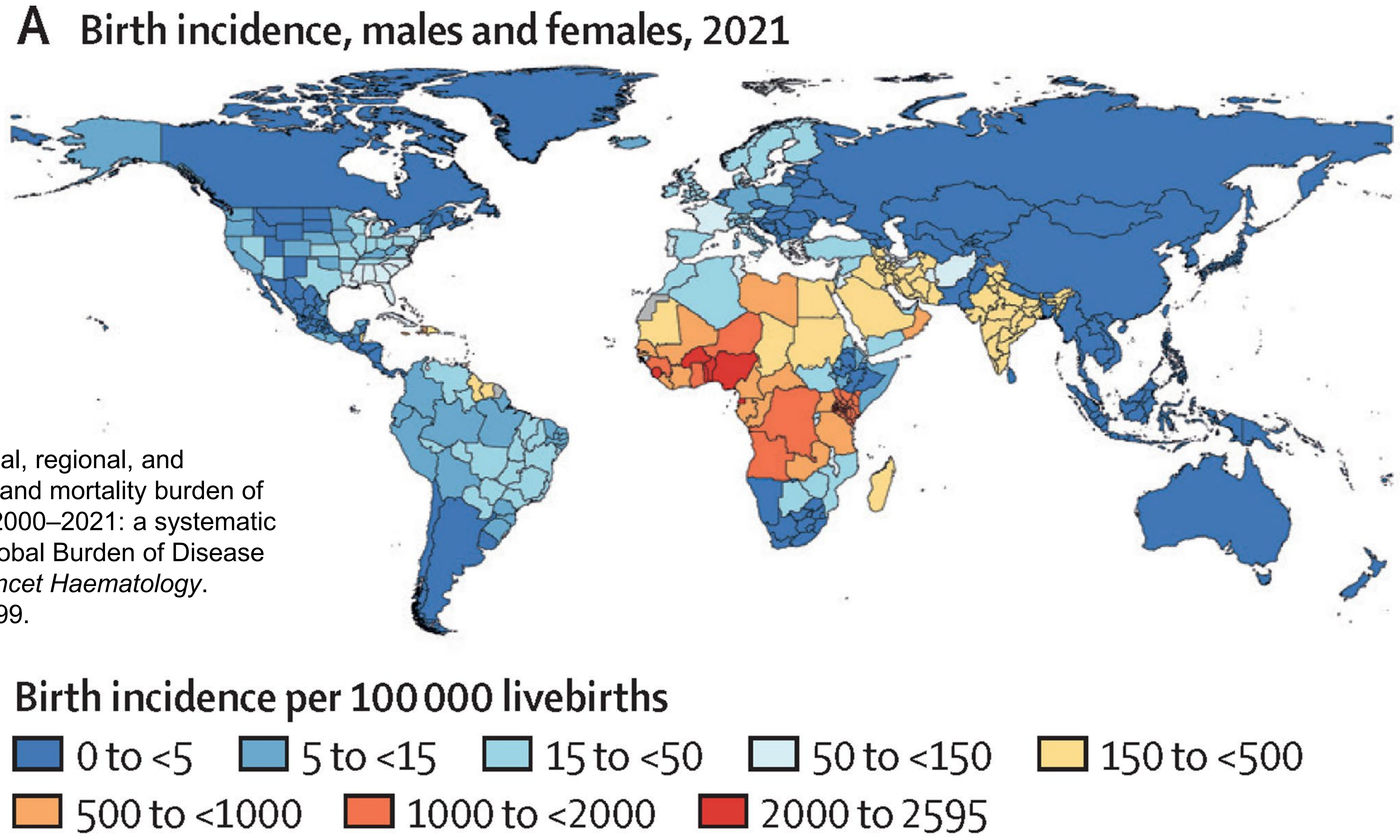
Data source: IHME, Global Burden of Disease (2024)

OurWorldinData.org/child-mortality | CC BY

Maternal, Perinatal, & Public Health

FETAL ANEUPLOIDY MAY NOT BE THE MOST PREVALENT GENETIC CONDITION

Maps of total sickle cell disease rates per 100 000 population

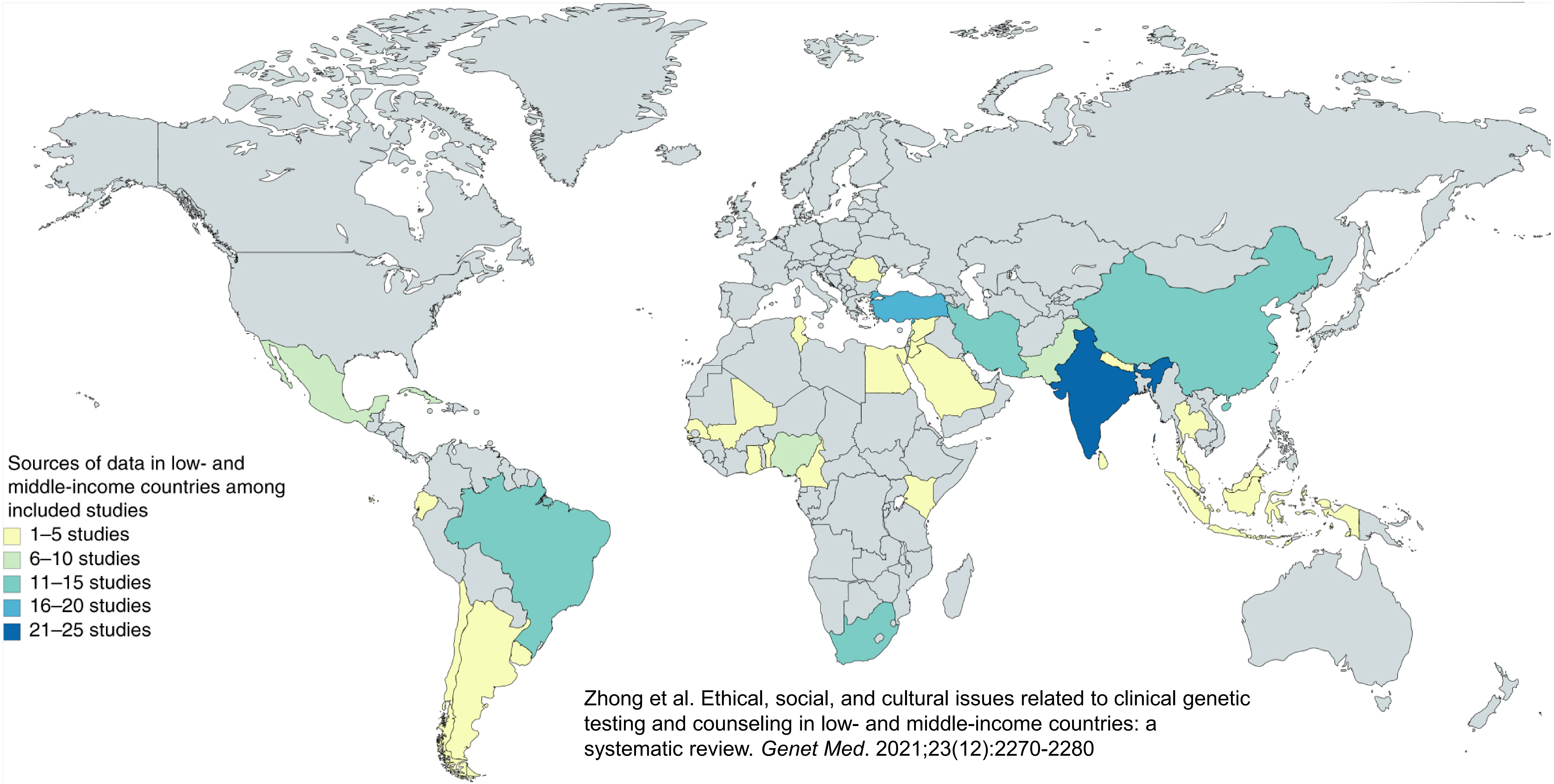


Thomson et al. Global, regional, and national prevalence and mortality burden of sickle cell disease, 2000–2021: a systematic analysis from the Global Burden of Disease Study 2021. *The Lancet Haematology*. 2023;10(8):e585-e599.

HEALTH EQUITY IN LOW RESOURCE SETTINGS

- Follow-on care difficult to access:
  - → Limited genetic workforce
  - → Unequal public insurance support & investment

GUIDANCE DEVELOPED IN HICs MAY NOT REFLECT VALUES AND PRIORITIES IN OTHER CONTEXTS



Zhong et al. Ethical, social, and cultural issues related to clinical genetic testing and counseling in low- and middle-income countries: a systematic review. *Genet Med*. 2021;23(12):2270-2280

CONCLUSION

- Core ethical values on prenatal genetic screening are globally recognized, but differ based on individual national contexts
- Additional focus on ethical values and experiences of LMICs on prenatal genetic screening

ACKNOWLEDGEMENTS

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Thank you!

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