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THE ETHICS OF SCREENING FOR RARE DISEASE

Seminar presentation: Rare Disease Social Research Center, the Institute of Philosophy and Sociology, Polish Academy of Sciences.

December 15, 2020.

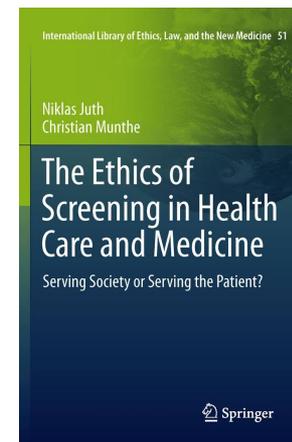
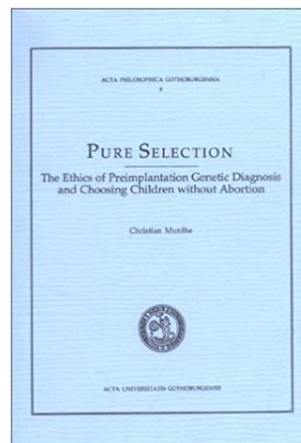
CHRISTIAN MUNTHE, PROFESSOR OF PRACTICAL PHILOSOPHY. EMAIL: CHRISTIAN.MUNTHE@GU.SE



Short presentation of myself

webpage: <https://www.gu.se/en/about/find-staff/christianmunthe>

- Worked on ethics, value and policy issues regarding health science and technology since 1990's
- Three particular concentrations:
 - genetic testing
 - Reproductive (prenatal and preimplantation genetic) testing and counselling
 - screening programs
- Specific policy re. rare disease
 - Scientific council of the Swedish *Rare disease foundation*
 - Priority setting and resource allocation expertise for the Region Västra Götaland County Council
 - Work in Swedish policy councils in the medical Ethical and health policy area.



bioethics

SPECIAL ISSUE PAPER

A New Ethical Landscape of Prenatal Testing: Individualizing Choice to Serve Autonomy and Promote Public Health: A Radical Proposal

Christian Munthe



Volume 29, Issue 1
Special Issue: Prenatal
Screening
January 2015
Pages 36-45

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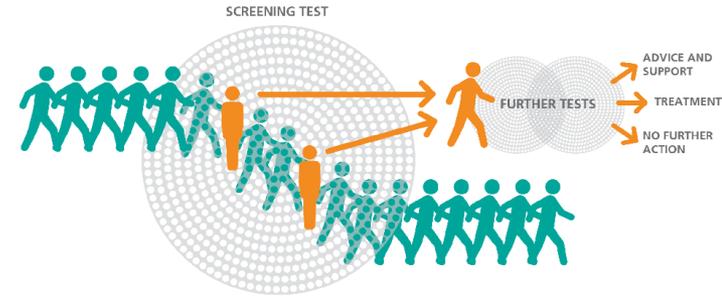
Four ethics/value challenge areas of screening for rare disease

- Specific ethical aspects of *population* screening
- Specific ethical aspects of *condition* screening of individuals
- *Reproductive* screening: prenatal, preimplantation, preconception
- The structural, natural and ethical implications of "rarity"



Population screening

- "Looking for the needle in the haystack", but most of what you touch is hay
- The haystacks didn't ask for it: the big difference between screening and ordinary diagnostics.
 - Aim is often unclear: for society or for patients?
 - More risk of manipulation
 - More difficult to ensure personal autonomy
 - Risk of causing more confusion than clarification
 - Pre and post testing counselling
- Large scale information gathering is very costly and potentially harmful
 - False negatives
 - False positives
 - Weak predictive certainty
 - Stigmas: individual, social, institutional
 - Lack of meaningful interventions
 - Overtreatment
 - Surveillance
- **Population screening programs very hard to justify**, even if single instances of testing or "condition screening" (next slide) seem legit.





Condition screening (of individuals)

- More like diagnostics, only without a clear set of suspect conditions: trawling a person for an explanation to an incomprehensible health problem
- Increasingly common strategy in "precision medicine" setups to solve super rare disease cases in the clinic
- Facilitated by wide or whole genome sequencing + big data bioinformatics
- **Uncertain benefits:** is there an effective intervention at the end of the tunnel?
- **Grey area between experimental research and clinical medicine:** therapeutic misconception
- How to deal with **unforeseen information?**
- How to deal with **unclear information?**

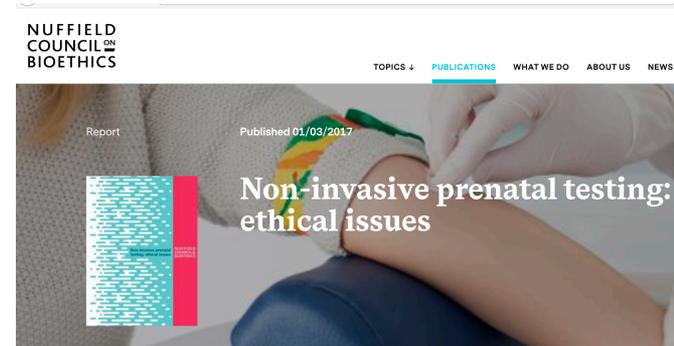


Risks of causing harm, manipulation and confusion rather than benefit, emancipation and clarification



Reproductive screening

- "But at least we can improve reproductive counselling"
- Not at all certain that population (prenatal) screening programs accomplish this (patient demand is not proof of actual benefit).
- Criticism 1: **geared towards avoiding people** with rare diseases rather than helping people with rare diseases
- Criticism 2: given the lack of support in society for families with rare diseases, these programs create a **structural pressure to avoid having (certain) children.**
- Criticism 3: Expresses an **eugenic societal concern** with the "quality" of future population
- Criticism 4: **Difficult to justify resource allocation** if the only aim is some improvement of reproductive counselling.
- **Ordinary individual (reproductive) diagnosis (on request based on medical reasons) might avoid such criticism.**



Volume 29, Issue 1
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Pages: ii-iv, 1-57
January 2015



Ethical imperatives of timely access to orphan drugs: is possible to reconcile economic incentives and patients' health needs?

[R. Rodriguez-Monguio](#) , [T. Spargo](#) & [E. Seoane-Vazquez](#)

[Orphanet Journal of Rare Diseases](#) 12, Article number: 1 (2017) | [Cite this article](#)

The ethical relevance of "rarity"

- Structural natural and economic barriers to improved medical management in general
- May create reasons to accept more expensive and less certified interventions, but this is controversial and complex
 - Helping an already burdened group is important, but so is not to additionally harm such a group
 - May lead to unsustainable economic consequences due to industry exploitation
 - Problematic ties between industry and patient advocacy groups
- Adds to known challenges of population screening programs
 - (Much) worse prospect of precision
 - More often lack of proven effective interventions
 - Higher risks of misleading expectations
- If unknown, only good reason for condition screening
 - But important to mind the risks!
 - Especially: mind the risk of patients becoming unwitting guinea pigs in genomic research!

Health Care Anal (2017) 25:1–20
DOI 10.1007/s10728-014-0284-5



ORIGINAL ARTICLE

For the Sake of Justice: Should We Prioritize Rare Diseases?

Niklas Juth

Feature article

Sustainability principle for the ethics of healthcare resource allocation

Christian Munthe ,^{1,2} Davide Fumagalli,^{1,2} Erik Malmqvist ^{1,2}

Munthe C, et al. *J Med Ethics* 2020;0:1–8. doi:10.1136/medethics-2020-106644



Background considerations

- The individual, the family and the societal structures: very complex web of tensions
- Minding the change of values, stakes and prospects/risks when individual diagnostic testing is scaled up
- Minding power relations, dependencies and potentially exploitative relationships

THANK YOU!