

Adapting the Wilson and Jungner criteria for the genomic age

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JASP - November 17, 2005

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Research Programme

- **Background**
- **Research question**
- **Objectives**
- **Methods**
 - Three inter-related research projects
- **Results**
 - Framework and synthesized list of criteria (A Andermann)
 - Review of criteria in real life examples (I Costea)
 - Consultation with stakeholders on utility (S Beauchamp)
- **Discussion**
 - Revising the list of genetic screening criteria (I Blancquaert)

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Cette présentation a été effectuée le 17 novembre 2005, au cours de la journée « Dépistage populationnel en génétique : développement, implantation et évaluation » dans le cadre des Journées annuelles de santé publique (JASP) 2005. L'ensemble des présentations est disponible sur le site des JASP, à l'adresse <http://www.inspq.qc.ca/jasp/archives/>.

Background

- **Increasing body of genetic knowledge**
 - Rapid advances in genetic technology
- **Pressure from various stakeholders to apply knowledge**
 - Expand existing screening programs
 - Develop new screening programs
- **Difficult political decisions to be made**
 - Which genetic conditions to screen?
 - How to offer genetic screening?
- **Classic Wilson and Jungner criteria over 30 years old**
 - Large genetic screening literature since then
 - Changing social and medical contexts
- **Need to develop screening criteria for the genomic age**
 - Address complex implications relating to genetic screening
 - Support decision-makers in policy-making

Research question

- What are the **key criteria** to help policy-makers prioritize **which** new genetic screening programs should be introduced, and **how** to introduce them?

Objectives

- **Overall Research Programme**
 - To produce a list of criteria and make recommendations that support decision-makers in developing rational policies for population-based genetic screening
- **Project 1:**
 - To develop a **framework** for genetic screening policy-making
 - To review and **synthesize criteria** previously proposed by academics, professionals, institutions and patient support groups
- **Project 2:**
 - To review the use of criteria in **real life** examples
- **Project 3:**
 - To assess criteria **utility** with stakeholders

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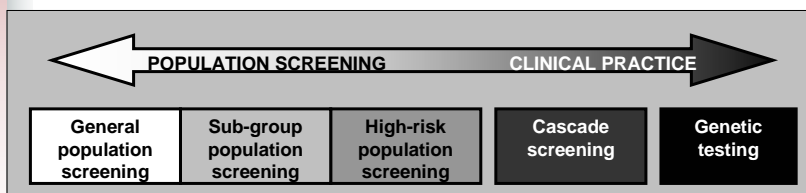
Methods

- Project 1
 - Literature review
- Project 2
 - Literature review
- Project 3
 - Questionnaires and focus groups

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Scope of research

- **Inclusion**
 - All types of population-based genetic screening
 - population, timing, disease, test, risk prediction, intervention
- **Exclusion**
 - Clinical practice
 - Grey zone = Cascade screening



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FRAMEWORK

- See Handout JASP Poster 2004

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Previous frameworks

- Each focuses on a key area:
 - **Natural history**
 - CDC, USA (Lindegren et al, 2004)
 - **Pressure to introduce new programs**
 - The Nuffield Trust, UK (Zimmern and Cook, 2000)
 - **Financial implications**
 - CHEPA, Canada (Morgan et al, 2003)
 - **Process of assessing potential programs**
 - National Academy of Sciences, USA (NRC, 1975)
 - **Approach to evaluating genetic tests**
 - Foundation for Blood Research, USA (Haddow and Palomaki, 2000)



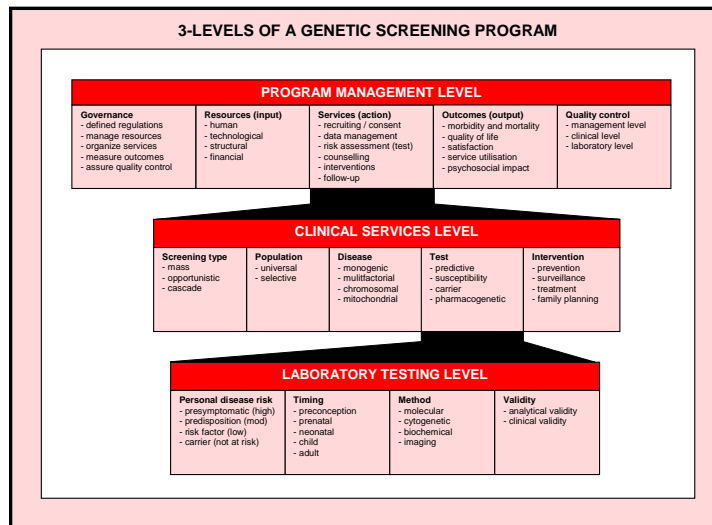
NO COMPREHENSIVE FRAMEWORK

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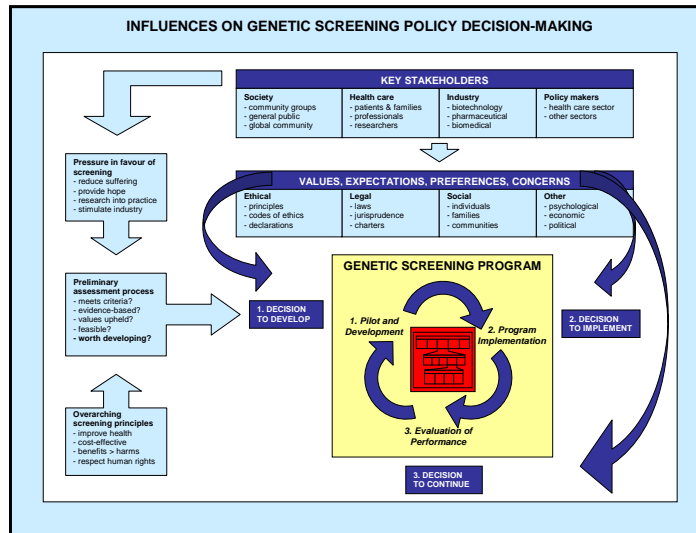
3-part framework

- Three levels of a genetic screening program
 - More than just a screening test
 - Program Management Level
 - Clinical Services Level
 - Laboratory Testing Level
- Genetic screening policy-making process
 - Preliminary assessment process
 - Development, implementation and evaluation cycle
- Genetic screening policy-making context

1) Three-tier program

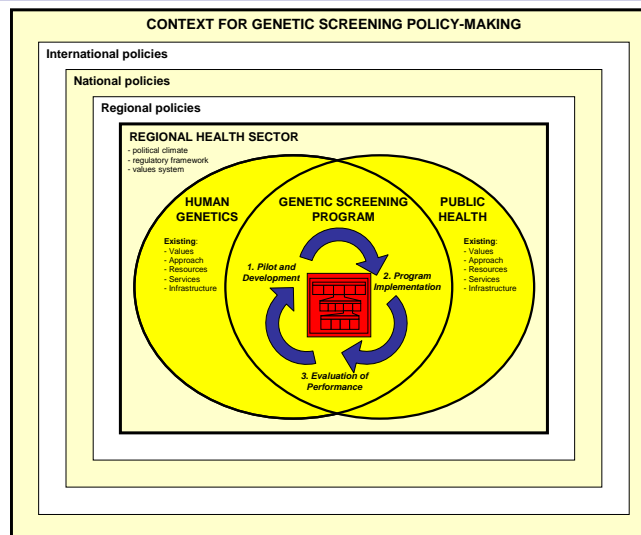


2) Decision-making



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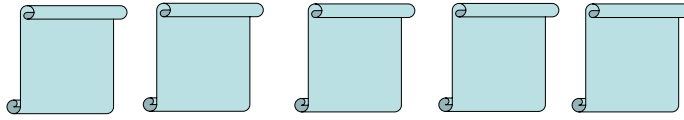
3) Policy context



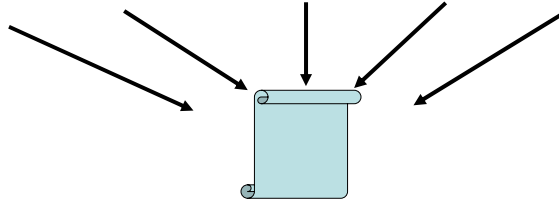
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LIST OF CRITERIA

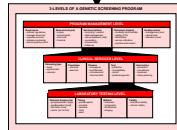
Collect



Synthesize



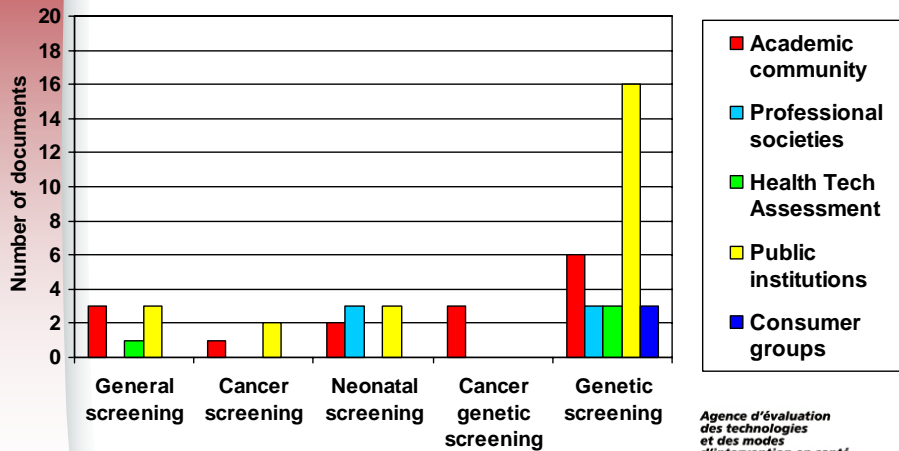
Categorize



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52 documents retrieved



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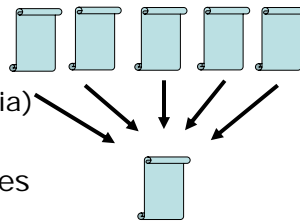
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Sample screening criteria references

- **Wilson and Jungner 1968**
- US National Academy of Sciences 1975
- Laberge and Knoppers 1991
- UK National Screening Committee 1998
- WHO Human Genetics Programme 2000
- European Society of Human Genetics 2003

Synthesis of documents

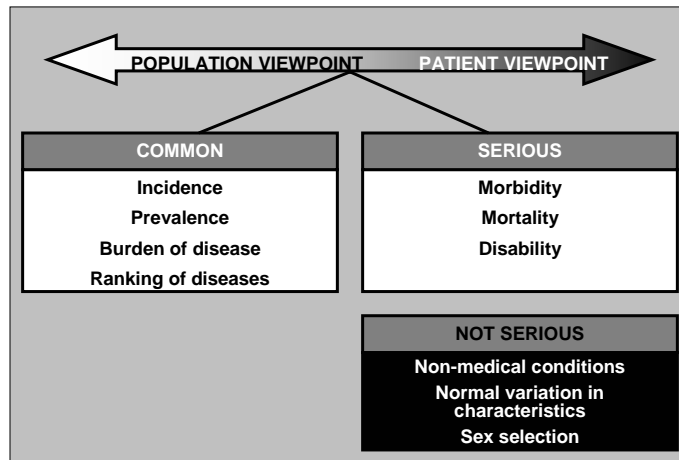
- Each list of criteria broken down
- Grouped according to 20 themes
 - Theme 1-10 (Wilson and Jungner criteria)
 - Theme 11-20 (Emerging criteria)
- Synthesis of contents for all 20 themes
 - Across all 52 documents
 - Original content conserved in tables
 - Identification of sub-themes
 - Summary
 - Labels



Sample criteria summary and label

BEFORE: The condition sought should be an **important** health problem

AFTER: The condition sought should be a **common and/or serious** health problem



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Adapting the Wilson and Jungner criteria

ORIGINAL CRITERIA	ADAPTED CRITERIA
1. important health problem	1. common and/or serious health problem
2. accepted treatment	2. accepted intervention (ex. prevention, treatment, family planning)
3. facilities	3. infrastructure for screening, including education, testing, clinical services and program management
4. latent or early symptomatic stage	4. or increased level of genetic risk
5. suitable test or examination	5. same
6. test should be acceptable	6. screening test and the entire screening program should be acceptable

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Adapting the Wilson and Jungner criteria

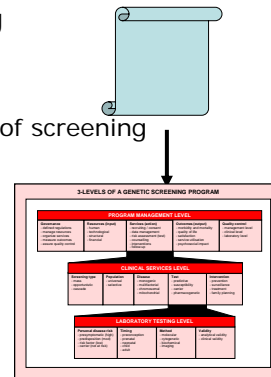
ORIGINAL CRITERIA	ADAPTED CRITERIA
7. natural history of the condition adequately understood	7. and of gene carriers
8. agreed policy on whom to treat as patients	8. categorize as "screen positive", "screen negative" and "screen indeterminate", and a defined process for each group following disclosure of screening results
9. cost of case-finding balanced in relation to possible expenditure of medical care as a whole	9. economic evaluations should add to evidence favouring of screening, but should not be the sole criterion for deciding whether or not to offer screening
10. a continuing process and not a "once and for all" project	10. same

New emerging criteria

11. integrated screening program that incorporates education, testing, clinical services and program management levels	16. quality assurance and program evaluation
12. need for screening, goals and objectives, roles and responsibilities, and financing defined	17. promotion of human rights
13. scientific evidence of screening program effectiveness	18. defined target population
14. overall benefits of screening outweigh potential harms	19. consumers and family members implicated
15. education program and individual risk counselling	20. separate consent for research that differs from clinical

Categorizing criteria

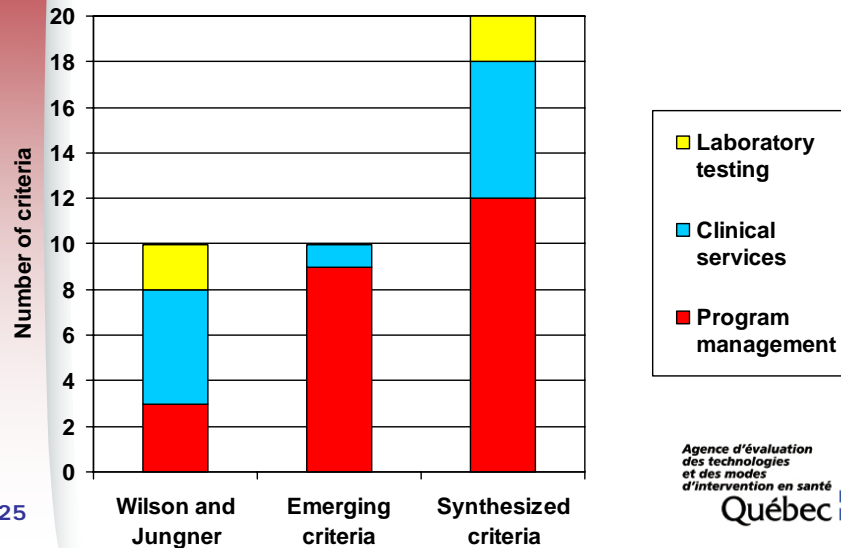
- Adapted Wilson & Jungner vs Emerging
- Genetic vs General
 - Genetic = Specific to genetic screening
 - General = Could be applied to all types of screening
- 3 levels of genetic screening programs
 - Program management level
 - Clinical services level
 - Laboratory testing level



Criteria highly relevant to genetic screening

- The condition sought should be a common and/or **serious** health problem
- Consumers should be included in screening policy-making and **family members** should be implicated in the screening process
- The natural history of the condition and of **gene carriers** should be adequately understood
- There should be a recognizable early symptomatic stage, latent stage or **increased level of genetic risk**


Lab vs Clinical vs Management (handout)



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Programme de recherche

- Cadre conceptuel et liste préliminaire de critères (A Andermann)
- **Recours aux critères : exemples concrets (I Costea)**
- Acceptabilité et utilité des critères pour les détenteurs d'intérêts (S Beauchamp)
- Enjeux entourant l'utilisation de la liste des critères et pistes de solution (I Blancquaert)
- Framework and preliminary list of criteria (A Andermann)
- **Review of criteria in real life examples (I Costea)**
- Consultation with stakeholders on acceptability and on utility (S Beauchamp)
- Revising the list of genetic screening criteria (I Blancquaert)