

EUROPE BIOBANK WEEK

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Peggy Manders: "How to deal with unsolicited findings in research?"



www.europebiobankweek.eu

How to deal with unsolicited findings in research?

Peggy Manders

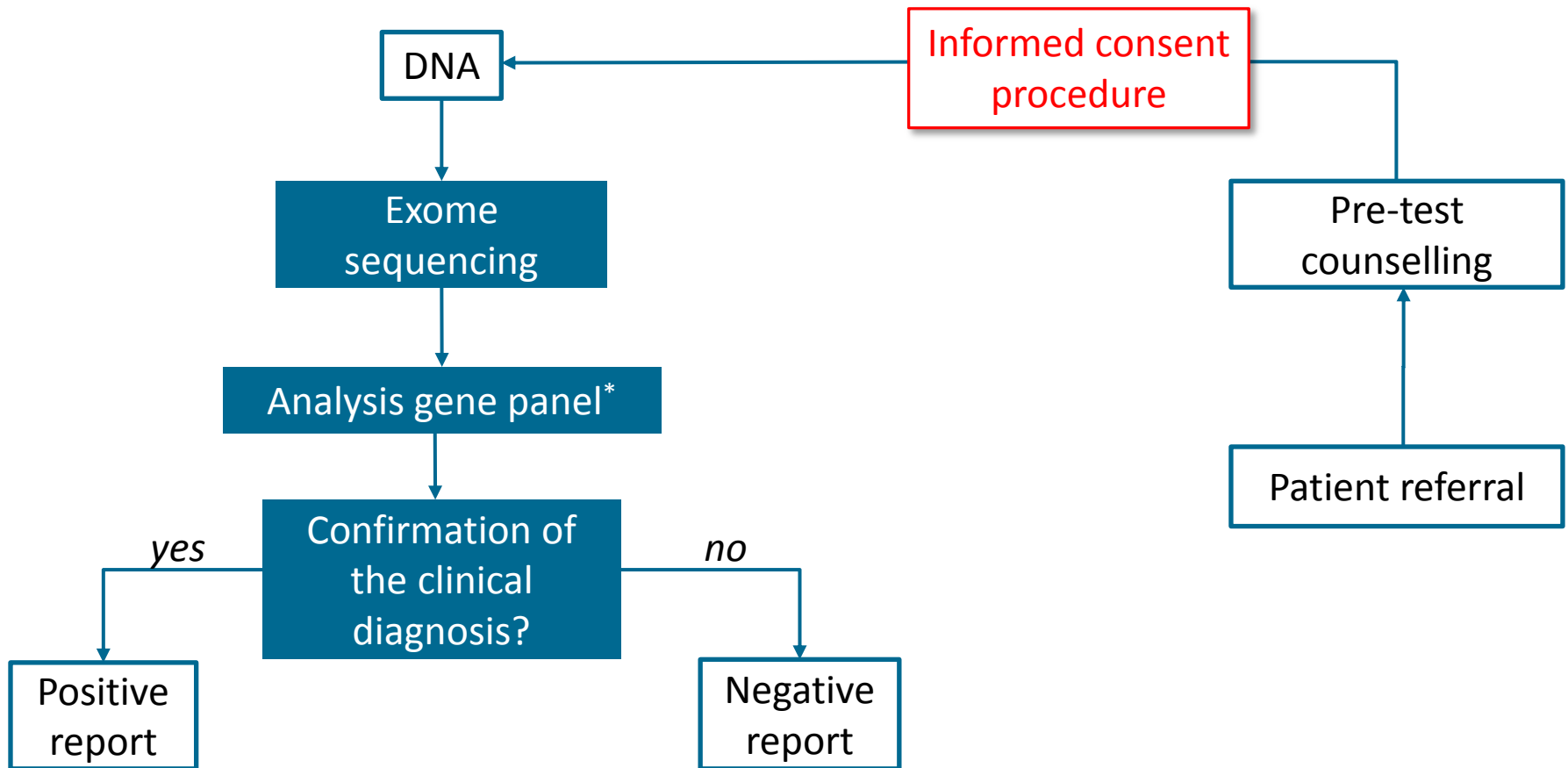
Radboud Biobank and Dept. of Human Genetics,
Nijmegen, The Netherlands

Whole exome sequencing – diagnostics

- Higher chance of finding the underlying genetic cause
→ **'All'** coding sequences of a human genome (>200,000 exons),
sequenced and analyzed in **one** experiment
- Number of diagnostic whole exome analyses ↑↑
- Chance for unsolicited findings;
reduced by a first step of filtering

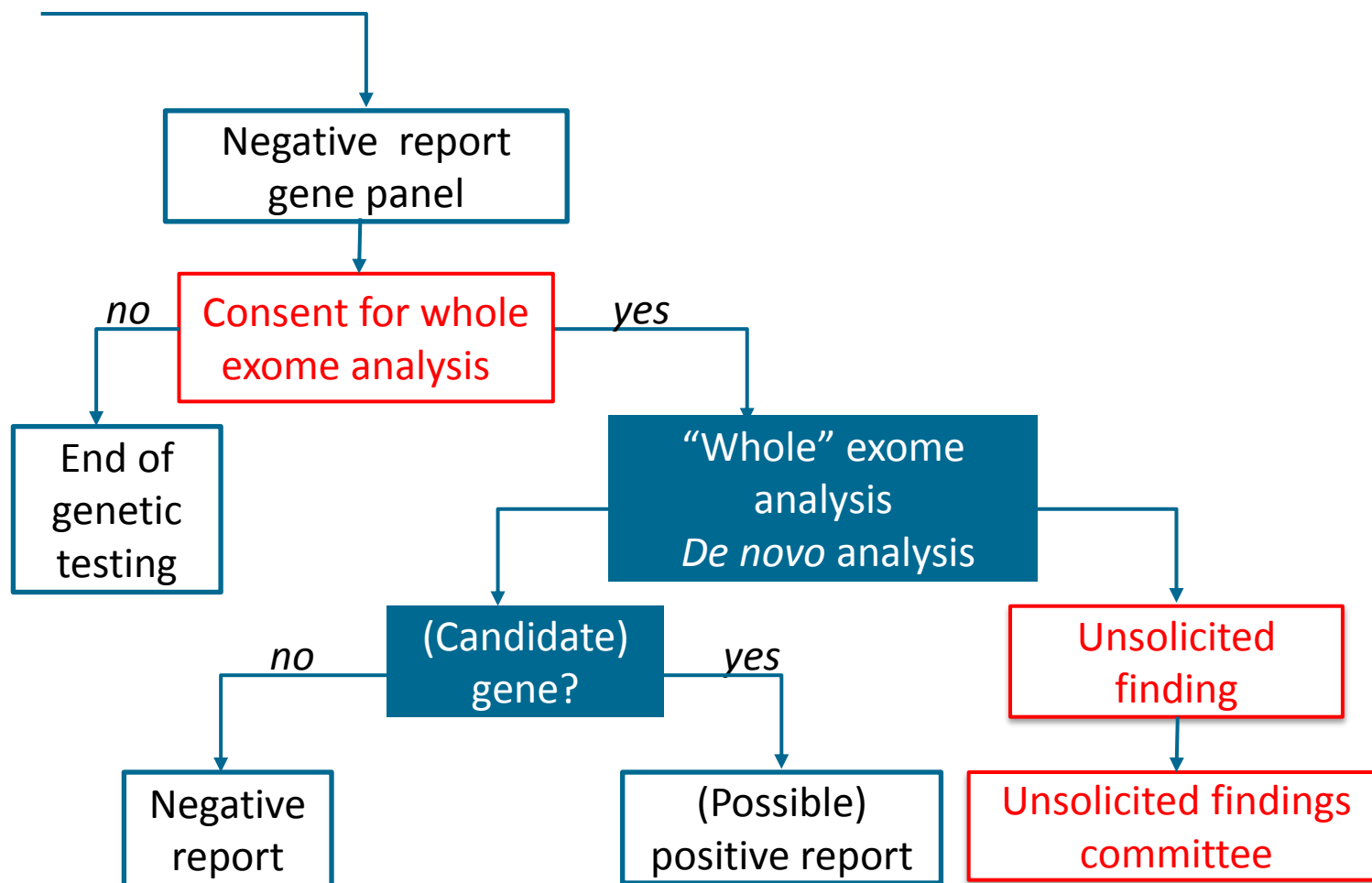


Workflow diagnostics – 1



* Available gene panels - www.genomediagnosticsnijmegen.nl

Workflow diagnostics – 2



Unsolicited findings committee

- Different disciplines, e.g. clinical geneticist, molecular geneticist, social worker, lawyer, ethicist
- Return of results
 - Which gene, which mutation
 - Classification of the mutation
(3 – unknown, 4 – probably pathogenic, 5 – pathogenic)
 - Patient and family characteristics
 - Type of disease, risk, screening available, penetrance



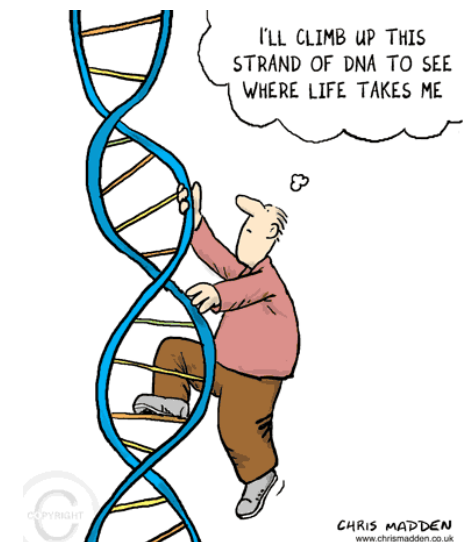
Statistics exome diagnostics

- ~4,000 gene panel analysis
- ~1,500 whole exome analysis
- 35 potential unsolicited findings reported (~2.3%)
 - 21 unsolicited findings reported to referring clinical geneticist (~1.4%)
 - 14 findings not reported (~0.9%)

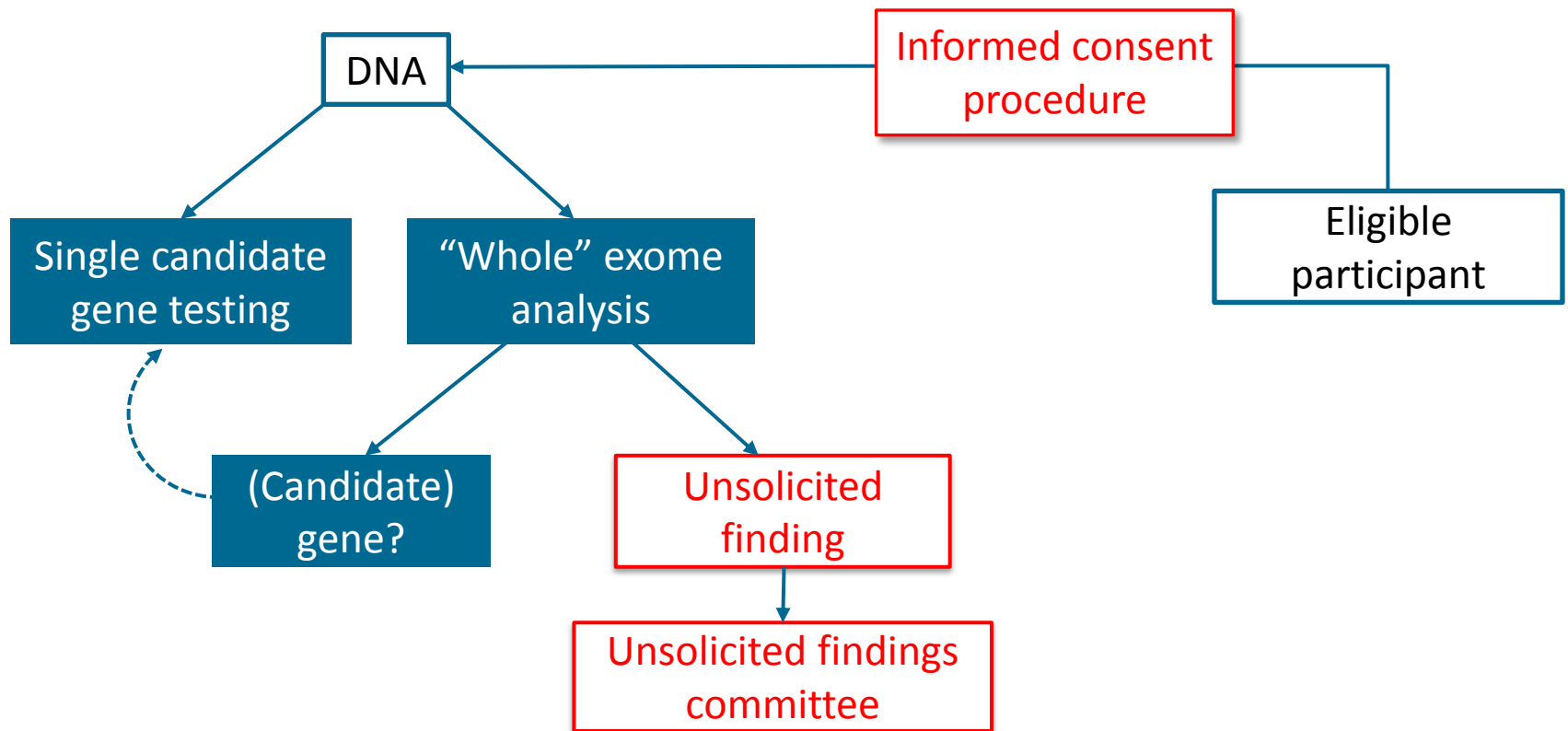


Whole exome sequencing – research

- Experiences gained in diagnostics → used for implementation of WES in research
- Broad informed consent for future research
 - Single candidate gene testing
 - Whole exome sequencing



Workflow research



Discussion

- Implementation of newly developed documents and procedures
- Find out what participants think of newly developed documents and procedure
- Assessment of all whole exome analyses performed in research plus the number of (potential) unsolicited findings

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peggy.manders@radboudumc.nl