Supplementary Material

Policy-making in newborn screening needs a transparent and structured approach

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**Supplementary Table 1. Articles included in the review (n = 26) with a short summary of each article, as well as the main stakeholder groups discussed, articles are sorted based on publication date to illustrate the shift in the technologies discussed.**

| **Author** **(year of publication)** | **Main technology discussed (MS/MS; genetic technologies)** | **Key words** | **Type of article, region** |
| --- | --- | --- | --- |
| Hiller *et al*. (1997)(1)  | PKU and CH | N/A | Research, USA |
| Atkinson *et al*. (2001)(2) | MS/MS | N/A | Practice article, USA |
| Therrell (2001)(3)  | MS/MS and genetic technologies | newborn screening; policy; tandem mass spectrometry; DNA; public health. | Review, USA |
| Elliman *et al*. (2002)(4)  | MS/MS | N/A | Review, UK |
| Comeau *et al*. (2005)(5)  | Genetic technologies  | N/A | Review, USA |
| Bailey *et al*. (2006)(6) | MS/MS and genetic technologies | Consumer advocacy; early intervention; family; infant; newborn; newborn screening | Review, USA |
| Grosse *et al*. (2006)(7) | MS/MS and genetic technologies | N/A | Review, USA |
| Arn (2007)(8) | MS/MS and genetic technologies | N/A | Review, USA |
| Borowski *et al*. (2007)(9) | None | Health policy, Healthcare funding, Health technology assessment, Decision making, Organizational | Research, USA |
| Bailey *et al*. (2008)(10)  | Genetic technologies | Newborn screening | Review, USA |
| Little *et al*. (2008)(11)  | Genetic technologies | Biobanking; Birth defects surveillance; Canada; Genetic counseling; Genetic testing; Health technology assessment; Newborn screening; Prenatal diagnosis | Review, Canada |
| Potter *et al*. (2008)(12) | MS/MS and genetic technologies | neonatal screening, policy development, stakeholder involvement | Review, UK,USA, Australia, Canada |
| Therrell (2008)(13)  | MS/MS and genetic technologies | Newborn screening, Paediatrics, Screening | Report, USA |
| Bailey (2009)(14)  | MS/MS and genetic technologies | N/A | Review, USA |
| Plass *et al*. (2009)(15) | Genetic technologies | neonatal screening, extension, parents’ opinion, untreatable diseases, childhood onset diseases | Research, NL |
| Potter *et al*. (2009)(12)  | Genetic technologies | Ethical, legal, and social issues (ELSIs); Genetic screening; Health technology assessment; Neonatal screening; Prenatal screening; Public health ethics | Workshop report, Canada |
| Simopoulous (2009)(16) | Genetic technologies | Ethical, legal, social, and economic aspects; Genetic counseling; Genetics education; Genetic screening; Public acceptance | Review, NA |
| Benkendorf *et al*. (2010)(17)  | MS/MS and genetic technologies | newborn screening; dried blood spots | Meeting report, USA |
| Bombard *et al*. (2010)(18)  | Genetic technologies | newborn screening; reproductive risk information; policy; ethics; informed decision making | Syst. review, global |
| Burke *et al*. (2010)(19)  | Genetic technologies | Clinical utility; Evidence-based practice; Genetic testing | Review, NA |
| Cornejo *et al*. (2010)(20) | MS/MS | N/A | Review, Chile |
| Dhondt (2010)(21)  | MS/MS and genetic technologies | N/A | Review, NA |
| Kasper *et al*. (2010)(22)  | MS/MS | National Austrian Newborn Screening Program, tandem mass spectrometry, inherited metabolic disorders, inborn errors of metabolism. | Research, Austria |
| Fisher *et al*. (2011)(23) | MS/MS | Neonatal screening, Decision-making processes, Reimbursement, Health technology assessment, Fourth hurdle | Review, Europe |
| Hayeems *et al*. (2013)(24)  | None | expanded newborn screening, mixed methods, public engagement, public expectations | Research, Canada |
| Bombard *et al*. (2014)(25)  | Genetic technologies | N/A | Research, Canada |
| Botkin *et al*. (2014)(26)  | None | newborn screening, research, research ethics, informed consent, parental permission | Meeting report, USA |

**Organizational abbreviations**: AAP: American Association of Pediatrics; ACMG: American College of Medical Geneticists; CADTH: Canadian Agency for Drugs and Technologies in Health; CCASS: Canadian Congenital Anomalies Surveillance System; CCMG: Canadian College of Medical Geneticists; CDCs: US Centers for Disease Control and Prevention; CORD: Canadian Organization for Rare Disorders; CPS: Canadian Paediatric Society; HRSA/MCHB: Health Resources and Services Administration/Maternal and Child Health Bureau; INTA: Institute of Nutrition and Food Technology; IOM: Institute of Medicine; NAS: National Academy of Sciences; NSC: UK National Screening Committee; PHAC: Public Health Agency of Canada; SACHDNC: Secretary’s Advisory Committee on Heritable Diseases in Newborns and Children; SOGC: Society of Obstetricians and Gynaecologists of Canada; WHO: World Health Organization.

**Other abbreviations**: CF: cystic fibrosis; DTC: direct-to-consumer; ELSIs: ethical, legal, and societal issues; HTA: health technology assessment; IDM: informed decision making; IEM: inborn errors of metabolism; MS/MS: tandem mass spectrometry; NBS: newborn bloodspot screening; NL: Netherlands UK: United Kingdom; USA: United States of America; WG/ES: whole genome/exome sequencing.

a For example covering the following disciplines: expert on metabolic diseases, representative from the Newborn Screening Program, parent/consumer representative, researcher, representative of a Hospital Association, official from a Department of Public Health, medical ethicist, representative from public health laboratories; screening program administration; health care providers

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