Institution: University of Oxford		
Unit of Assessment: 30 - Philosophy		
Title of case study: Demonstrating the value of embedding consideration of ethical issues into the day to day practice of genomic and genetic medicine		
Period when the underpinning research was undertaken: 2001-2020		
Details of staff conducting the underpinning research from the submitting unit:		
Name(s): Michael Parker	**Role(s) (e.g. job title):** Professor, Director of Ethox	**Period(s) employed by submitting HEI:** 1999-present
Period when the claimed impact occurred: September 2013 – November 2020		
Is this case study continued from a case study submitted in 2014? N		

1. Summary of the impact (indicative maximum 100 words)

Professor Parker's research on ethical issues arising in the uses of genetics has transformed policy and practice in three ways. Firstly, it has shaped the successful development and implementation of the UK's 100,000 genomes project and the establishment of the NHS Genomic Medicine Service (the world's first health system to provide whole genome sequencing at scale). Secondly, through his research, Professor Parker established the national ethics forum supporting UK genetics professionals facing ethically complex decisions to identify and address ethical issues and make better decisions: over 300 difficult clinical cases have since been discussed and resolved. Thirdly, his research has been incorporated into the Royal College of Physicians, Royal College of Pathologists and British Society for Genetic Medicine guidelines on Consent and Confidentiality in Genomic Medicine.

2. Underpinning research (indicative maximum 500 words)

Parker's research identifies and analyses ethical issues arising in the clinical and research uses of genetics. It combines qualitative social scientific research - interviews and focus groups with health professionals – with rigorous ethical analysis of cases. Several of his papers have been co-authored with Professor Anneke Lucassen (Clinical Geneticist at Wessex Genetics Service and Professor of Genetics at Southampton University), reflecting Parker's commitment to engagement with ethical questions arising in 'real world' settings, and to working closely with clinicians. In these papers, and in their subsequent work together, he has taken lead responsibility for the ethical analysis and Professor Lucassen leads on the medical aspects.[1]

Inspired by this case-based research, in 2001 Parker obtained Wellcome funding for a short pilot research project to explore clinicians' experience of addressing practical ethical issues. This identified a need for timely ethics support embedded in clinical practice, combined with ethics research on emerging issues. This led him and Professor Lucassen – later the same year - to establish the Genethics Club, a tri-annual national ethics forum for health professionals working in genetics. This forum has been very successful. As of 2020, it has met 57 times in locations across the United Kingdom. During this time, more than 684 clinically challenging cases have been presented and discussed (322 of the cases were discussed during this assessment period). In addition to its impact on practice and policy - outlined in sections below – the forum has always had an important research dimension. This reflects his interest in embedded ethics research. In 2012 he published a sole-authored book, *Ethical Problems and Genetics Practice*, which – through an analysis of cases presented at the Genethics forum - offers a critical case-based analysis of core ethical concepts, arguments, and assumptions shaping practice and policy in genetics and genomics. The book provides a rich description of the moral world of the genetics professional and the 'moral craft' central to good practice. [2] It is important to note that the relationship between research and impact works in both directions in this work. His research has been informed by engagement with the ethical issues arising in day-to-day practice of genetics. It has also had a significant impact on practice. A good example of this is *Ethical problems and Genetics Practice*, which reports research conducted on ethical aspects of practice arising in the Genethics forum but has also subsequently itself had an important influence on practice and policy – outlined in sections below – and, indeed, on the ongoing development of the Genethics forum itself.

The main results of his insights can be summarised as follows:

- He has demonstrated the value of embedding consideration of ethical issues into the day-to-day practice of genomic and genetic medicine. [2]
- He has developed arguments capable of recognizing both the importance of high standards of patient confidentiality and the existence of obligations to the family members of patients affected by inherited conditions. [2], [4], [5]
- He has made a convincing case for sharing familial information, where appropriate (after deliberation of benefits and risks of doing so). [3], [4]
- He has articulated important new ethical positions regarding the feedback of unexpected findings such as misattributed paternity. [2], [6]
- He has characterised the ethical issues arising in the use of new genomic technologies in reproductive medicine [2]
- He has described the emergence of a new hybrid form of clinical-research practice in data-driven genomic medicine and articulated the case for the development of a new social contract for contemporary medicine and medical research. [2]

3. References to the research (indicative maximum of six references)

1. [Journal article] Revealing false paternity: some ethical considerations. Lucassen, A. and **Parker, M.** *The Lancet* (2001) Vol 357(9261) pp.1033-1035. DOI: https://doi.org/10.1016/S0140-6736(00)04240-9
2. [Authored book] Ethical problems and genetics practice (2012) **Parker, M.** Cambridge: Cambridge University Press https://doi.org/10.1017/CBO9781139107792
3. [Journal article] Using a genetic test result in the care of family members: How does the duty of confidentiality apply?" (2018) **Parker, M**., and Lucassen, A., *European Journal of Human Genetics* (2018) Vol 26, pp 955-959. https://doi.org/10.1038/s41431-018-0138-y
4. [Journal article] Confidentiality and sharing genetic information with relatives. Lucassen, A. and **Parker, M.** *The Lancet* (2010) Vol 375(9725) pp.1507-1509. DOI: https://doi.org/10.1016/S0140-6736(10)60173-0
5. [Journal article] Genetic Information: a joint account? **Parker, M** and Lucassen, A., *British Medical Journal* (2004) Vol 329 pp.165-167. DOI: https://doi.org/10.1136/bmj.329.7458.165
6. [Journal article] When genomic medicine reveals misattributed genetic relationships – the debate about disclosure revisited. Wright, C., **Parker, M.,** and Lucassen, A. *Genetics in Medicine* (2018) Vol 21 pp. 97-101. DOI: https://doi.org/10.1038/s41436-018-0023-7

Key Research Awards

M. Parker (PI) with Dr Jeremy Howick, Professor Hazel Everitt, Dr Felicity Bishop, Professor Christian Mallen, Professor Paul Little, Wellcome Trust Centre Grant, 'Wellcome Centre for Ethics and Humanities' (2017-2021). Award of GBP2,991,157; Grant no. 203132/Z/16/Z.

M. Parker (PI), Health Innovation Challenge Fund from Department of Health and Wellcome Trust, 'Prenatal Assessment of Genomes (PAGE)' (2014-2018). Award of GBP328,297; Grant reference HICF-R7-396; WT Reference 101177.

M. Hurles, Wellcome Trust Sanger Institute (PI), with M. Parker (Co-I) et al. Health Innovation Challenge Fund from Department of Health and Wellcome Trust 'Deciphering Developmental Disorders' (2010-2017). Award of GBP4,405,588; Grant reference HICF-1009-003.

4. Details of the impact (indicative maximum 750 words)

Enabled the successful completion of the UK's 100,000 Genomes Project and the establishment of the NHS Genomic Medicine Service. In December 2012, the Prime Minister, David Cameron announced an aspiration to sequence 100,000 genomes. In December 2012, having read *Ethical Problems in Genetics Practice*, the Chief Medical Officer, Professor Dame

Sally Davies invited Parker to chair a high-level advisory group to make recommendations on the ethical requirements to inform the establishment of the 100,000 Genomes Project.[A] Parker's published recommendations [B] were accepted by the Chief Medical Officer and the Department of Health in full and informed the subsequent implementation of shape of the 100,000 Genomes Project. [C, especially chapter 16] In 2013, the Secretary of State established Genomics England - a company wholly owned by the UK Government - as the mechanism for the implementation of the 100,000 Genomes Project. [D] Parker was appointed as a non-executive director (with particular responsibility for embedding ethics in the 100,000 Genomes Project). The inaugural board meeting took place on the 13th August 2013. Parker established and chairs an Ethics Advisory Committee and an in-house ethics advisory team. He led the development of the 100,000 Genomes Project's approaches to consent, data sharing, feedback of findings, recontact, diversity and equity, and consent for children and young people. The 100,000 Genomes Project – the world's first genomics initiative at this scale and in the context of a national health service - was successfully completed in 2019. On its completion, the Chief Medical Officer for England, wrote to Parker for his significant level of leadership on relevant ethical issues played by Parker.

"*Thank you for your tremendous contribution to this work. With the successful completion of the 100,000 Genomes Project and the launch of the NHS Genomic Medicine Service the UK is firmly established as the world leader in genomic medicine. It would not be an overstatement to say that this initiative would not have been the success it is and might perhaps not even have been possible without your crucial leadership on ethics."* Chief Medical Officer for England. [A]

The NHS Genetic Medicine Service, which launched in 2018, builds upon on existing NHS clinical genetic services and the 100,000 Genomes Project; it includes a national genomic test directory and the national provision of whole-genome sequencing.[E i] In 2018 the Health Secretary announced that from 2019, all seriously ill children will be offered whole genome sequencing as part of their care, and that adults with certain rare diseases or hard-to-treat cancers will also be offered the same option from 2019.[E ii]

Embedding the analysis of ethical issues into genetics practice to enable high ethical standards in the care of patients and families. Between September 2013 and November 2020, the Genethics forum took place 23 times. Genethics is a national forum for health professionals, patients, and ethicists to discuss practical ethical problems encountered in the working lives of clinical genetics departments in the United Kingdom. The outputs of the Genethics meetings include recommendations to improve ethical decision-making in practice. During this period, the Forum was attended by approximately 1,000 genetics professionals and over 322 cases were presented and discussed. The **most important impact** of the Forum during the assessment period is that health professionals facing complex ethical challenges in their clinical work have been able to obtain (otherwise unavailable) ethics support and advice in their decision-making involving complex ethical judgements. A Consultant Genetic Counsellor in Manchester highlights: *["… it has been a particularly invaluable resource for trainees in genetic medicine (both doctors and genetic counsellors). The fact that it has remained active for all these years is testament to how useful we have found it."* During the assessment period, versions of Genethics have been established in the Netherlands, [F] Italy, [G] and Israel [H]. *"Forum OncoGenEtica was established in 2014 […]. The idea for the format directly drew from the Genethics Forum, […] Attendees have included cancer geneticists, oncologists and pediatricians and almost 70 cases have been discussed."* [G].

Led and informed public and policy debate on the future ethical shape of the social contract for genomic research. In 2017, together with Professors Anneke Lucassen (Professor of Clinical Genetics at Southampton University) and Jonathan Montgomery (Professor of Medical Law at UCL), Parker contributed the ethics chapter to the Chief Medical Officer's Annual Report (2017) 'Generation Genome' reflecting on the ethical requirements for a future data-driven, genomic medicine service. The CMO's Annual Report is her most important formal policy statement. [C, especially chapter 16] Drawing upon the 100,000 Genomes Project experience, and looking forward to the future uses of genomics, the chapter introduced the concept of the

'social contract' and argued that there is a need for the development – through a national process of deliberation - of a new 'social contract' as the basis for well-founded public trust and confidence in data science and genomics. The concept of 'social contract' struck a chord and was picked up by the CMO in her introduction as one of the main recommendations of the report. The importance she placed on this was clear in her evidence (in April 2018) to The Science and Technology Committee on Genomics and Genome Editing in the NHS,

"[In her evidence to us] The CMO […] cited the importance of routine large-scale data sharing as a reason "to rethink—or at the very least reinforce—elements of the current 'social contract' as set out in the NHS Constitution, to take account of the advances in genomic medicine". [At this point a footnote in the Committee's report includes a direct quotation from our chapter *"The report refers to this 'social contract' as a "common set of principles and values that bind together patients, the public and [NHS] staff in order to ensure that [the NHS] can be effective and equitable", as laid out in the NHS Constitution, with each party having rights and responsibilities." para 68 on page 33* [I]

As a consequence, the concept of the 'social contract' became central to policy debate about genomics in the UK. A good example of this influence is that, in 2018-19, Ipsos MORI conducted a national consultation on public and patient views about the 'social contract' between medical research and society. This was directly inspired by our chapter and the work on social contract. This is reflected in the fact that when the report was published in April 2019 it included an invited foreword co-authored by the CMO and Parker. [J]

Led changes to policy, practice, and the law regarding the familial uses of genetic information. Parker's research has identified important ethical considerations in favour of a more familial approach to genetics practice. In his published work – both sole authored and with Professor Anneke Lucassen – he argued such information should be more available for care of family members. [2], [3], [5], [6]. Their recommendations were incorporated into the Royal College of Physicians, Royal College of Pathologists and British Society for Genetic Medicine guidelines on Consent and confidentiality in genomic medicine: Guidance on the use of genetic and genomic information in the clinic (2019), which are the primary source of guidance on these issue for all genetics professionals in the UK. [K] A key paragraph in the guidance states,

"*Health professionals can find it difficult to know how to preserve the confidentiality of one patient and at the same time alert a family member of their risk of a particular condition. One way of doing this … relatives can be alerted that they might be at risk of developing a condition because of a family history (they perhaps already know about), or because of other information the clinician does not need to specify. This provides a way of alerting relatives that does not breach the confidence* (a footnote here links to his research output [3]). [K, page 10])

In 2018, the Appeal Court decided to allow a genetics case (ABC v St Georges Healthcare Trust) concerning the appropriate limits to patient confidentiality to be heard at the High Court. The case was heard in November 2019 and the High Court's decision announced in April 2020. Although the decision went against ABC, the case established for the first time that health professionals have a legal duty to balance the interests of genetic relatives with those of a patient who has refused permission for the disclosing of confidential information. This case will significantly change the use of genetic (and possibly other) healthcare information in the United Kingdom. Because of his work in this area, Parker was invited to provide expert ethics input into this case to support the claimant. The contents of this advice are confidential, but his input took the form of meetings with the legal team and the claimant and preparation of a substantial report for her legal team. [L]

5. Sources to corroborate the impact (indicative maximum of 10 references)

A. Letter of thanks from the Chief Medical Officer, Professor Dame Sally Davies FRS FMedSci, 30 July 2019. This letter outlines Parker's impact on the 100,000 Genomes Project and the development of genomics in the UK during the reporting period.

B. Letter from Professor Parker, as chair of the Chief Medical Officer's Advisory Committee to CMO about how to make the 100,000 Genomes Project ethical (2013).
C. Selections from the Chief Medical Officer Annual Report 2016– Genome Generation, which includes a chapter by Professor Michael Parker, Professor Anneke Lucassen, and Professor Jonathan Montgomery. The CMO's annual reports are the main/most influential statement produced by CMO each year. In her introduction to the report, the CMO draws explicitly on Professor Parker's concept of the social contract. Full report available at: https://www.gov.uk/government/publications/chief-medical-officer-annual-report-2016-generation-genome
D. POST Note (No. 504 Sept 2015) – publication for MPs about Genomics England highlighting the role of ethics and citing Professor Parker's letter [C] to Chief Medical Officer. Available at: https://post.parliament.uk/research-briefings/post-pn-0504/
E. i. Details of the NHS Genomic Medicine Service https://www.england.nhs.uk/genomics/nhs-genomic-med-service/
ii. Announcement by the Health Secretary, October 2018 https://www.gov.uk/government/news/matt-hancock-announces-ambition-to-map-5-million-genomes
F. Letter from a Dutch Clinical Geneticist at the University Medical Center Groningen in The Netherlands about the role of Genethics Forum in influencing a similar initiative in the Netherlands, 10 January 2020.
G. Email statement from OncoGenEtica Steering Committee member about the role of Genethics Forum in influencing a similar initiative in Italy, 16 December 2019.
H. Letter from Vice Dean - School of Law at Netanya Academic College in Israel about the role of Genethics Forum in influencing a similar initiative in Israel, 10 January 2020.
I. Report of the Science and Technology Committee on Genomics and genome editing in the NHS. Published April 2018, citing the importance of Professor Parker's contribution directly. Available at: https://publications.parliament.uk/pa/cm201719/cmselect/cmsctech/349/34902.htm
J. Foreword from IPSOS MORI report (April 2019), 'A public dialogue on genomic medicine: time for a new social contract?' This report, with a foreword co-authored by the Chief Medical Officer of England and Professor Parker, was prompted by his research on the 'social contract'. Full report available at: https://www.ipsos.com/ipsos-mori/en-uk/public-dialogue-genomic-medicine-time-new-social-contract-report
K. Selected pages from Royal College of Physicians, Royal College of Pathologists and British Society for Genetic Medicine. *Consent and confidentiality in genomic medicine: Guidance on the use of genetic and genomic information in the clinic*. 3rd edition. Report of the Joint Committee on Genomics in Medicine. London: RCP, RCPath and BSGM, 2019. The guidance cites Professor Parker's influence on the guidelines. Full guidance available at: https://www.rcplondon.ac.uk/projects/outputs/consent-and-confidentiality-genomic-medicine
L. Emailed letter of thanks from lead barrister for the claimant in the ABC court case, 15 April 2020. The letter acknowledges Professor Parker's role in helping a QC prepare for a court case which was eventually won.