Rachel's Rule: Protecting Today, For Tomorrow

Annual Hereditary Risk Reviews (AHRR)

Written by Stuart Ball, Founder of Rachel's Rule

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(Revised Policy Briefing 2025)

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Rachel's Rule – Revised Full Policy Briefing 2025

Turning Eligibility into Action – Making Genetic Testing Work in Reality

Prepared by Stuart Ball | Founder, Rachel's Rule Campaign
(Protecting Today, For Tomorrow)

Executive Summary

The NHS now possesses the tools to detect hereditary risk, yet thousands of families are still missed each year because guidance stops at eligibility and fails to ensure systematic follow-up.
Rachel Carole Ball's experience – three primary cancers between 2005 and 2019 plus benign hereditary indicators – shows that gap vividly.

Of those cancers, **only one** – her 2012 ovarian adenocarcinoma – would meet today's NHS Genomic Test Directory criteria.
Her 2005 non-epithelial dermoid squamous carcinoma and 2019 oestrogen-positive breast cancer (at age 41) still would not.
Yet even the qualifying ovarian tumour went untested because the universal-testing rule did not exist then.
Genetic referral finally occurred in 2019 – after three primary malignancies spanning fourteen years.

Rachel's Rule introduces an **Annual Hereditary Risk Review (AHRR)** so that every patient with cancer or hereditary indicators is re-evaluated each year and any clinician can trigger reassessment between reviews.
It creates a continuous safety net – an auditable process rather than a single missed opportunity.

Key facts (2025)

- 5 – 10 % of all cancers are hereditary (Genomics England 2024).
- ≈ 50 % of mutation carriers have no family history (Rahman 2021).
- ≈ 35 % of eligible breast-cancer and ≈ 60 % of ovarian-cancer patients are tested (Genomics England Audit 2024).
- Testing rates vary three-fold between regions (Ovarian DEMO 2024).
- £1 billion per year spent on avoidable late-stage treatment (NHS Cancer Data 2023).
- Diagnostic error claims = £13 billion liability (NHS Resolution 2023/24).

Headline Recommendations

1. Mandate an Annual Hereditary Risk Review across all NHS Genomic Medicine Service regions.
2. Launch a national recall audit for patients diagnosed before 2021.
3. Embed digital AHRR alerts within GP and oncology EHRs.
4. Require Cancer Alliances to report AHRR completion and referral rates annually.
5. Include AHRR compliance within the Quality and Outcomes Framework (QOF).

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1 Background and Context

1.1 Progress on Paper

Year	Guideline / Programme	Key Development
2013	NICE CG164 – *Familial Breast Cancer*	Introduced family-history thresholds for BRCA testing

Year	Guideline / Programme	Key Development
2021	NICE NG12 – *Suspected Cancer*	Recognised hereditary patterns as referral triggers
2021	NHS Genomic Test Directory v5	Created national testing codes
2023	CG164 update	Added < 40 yrs and triple-negative < 60 criteria
2024	NICE NG241 – *Ovarian Cancer*	Universal testing for *all invasive epithelial* tumours
2024	Genomic Test Directory v7 (R208/R213)	Re-confirmed eligibility rules for breast & ovarian testing
2024	GMS Transformation Programme	Acknowledged "equitable access remains inconsistent."

1.2 The Implementation Gap

- **Eligibility ≠ Access.** Guidelines specify who *should* be tested but not how to ensure it.
- **Historic Exclusion.** Pre-2021 patients rarely re-evaluated.
- **Regional Variation.** Testing rates differ three-fold between trusts.
- **Benign Indicators Ignored.** Non-cancerous findings (e.g. hamartomas, thyroid disease, fibroids) are not flagged for future review.

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2 Rachel's Case – A Timeline of Missed Opportunity

Year	Age	Diagnosis	Histology	Eligibility Then	Eligibility Now	Outcome
Dec 2005	28	Ovarian squamous carcinoma within dermoid cyst	Non-epithelial germ-cell	❌ No	❌ No	No referral
Mid-2012	34	Ovarian adenocarcinoma Grade 1C	Invasive epithelial (low grade)	❌ No (high-grade only then)	✅ Yes (universal epithelial rule now)	No referral
Jun 2019	41	Breast cancer ER+, HER2–, G2	Common luminal A	❌ No (> 40 yrs, not TNBC / bilateral)	❌ No	**First genetic referral after three primaries**

In 2012 Rachel was also found to have hepatic hamartomas – a benign feature linked to PTEN Hamartoma Tumour Syndrome (Cowden syndrome).
As stand-alone findings they did not meet referral criteria then and would not today; no mechanism flagged them for future review.
Rachel's Rule ensures such findings are logged and re-evaluated annually so that hereditary patterns are recognised before further disease develops.

3 Evidence – Eligibility Versus Reality

3.1 Current Testing Performance Across the NHS

Indicator	Latest Finding	Source
Breast-cancer patients actually tested	≈ 35 % of those meeting NICE criteria	Genomics England Audit (2024)

Indicator	Latest Finding	Source
Ovarian-cancer patients tested	22–66 % (depending on region)	Ovarian Cancer DEMO Project (2024)
Carriers without family history	≈ 50 % of pathogenic-variant carriers	Rahman *Nat Rev Cancer* (2021)
Pathogenic variants missed under current eligibility rules	4.6 % of all variants	*Nature Sci Rep* (2022)
Patients offered re-testing when criteria expand	< 10 %	NHS GMS Evaluation (2023)
Ethnic and regional inequity in access	3–4 × variation	*BMJ Open* (2023)

Summary: Even in 2025, roughly half of hereditary cancers in the UK remain undetected or untested.

3.2 Why the Gap Persists

Tumour type bias – Testing is still focused on breast and ovarian cancers, leaving patients with other tumour types or non-cancerous syndromic signs unassessed.

Historic exclusion – Individuals diagnosed before 2021 rarely receive a recall for retrospective testing under new guidelines.

Funding and capacity limits – Some trusts cap the number of tests or rely on local commissioning policies that differ from national guidance.

Data fragmentation – Cancer, genetic, and primary-care records are not inter-operable; patterns spanning years or specialties remain invisible.

Cognitive overload – Front-line clinicians cannot be expected to cross-reference decades of records during time-limited consultations.

Benign signs overlooked – Features such as hamartomas, thyroid nodules, fibroids, or autoimmune manifestations are deemed non-actionable and are seldom logged as potential hereditary indicators.

3.3 The Statistical Reality of "Missed Hereditary Cancers"

Tumour type	Estimated hereditary proportion	Proportion meeting NHS criteria	Hereditary cases still missed
Breast	5–10 %	≈ 40 % eligible	≈ 60 % missed
Ovarian (all types)	15–20 %	≈ 90 % eligible (epithelial only)	≈ 10 % missed (non-epithelial)
Colorectal	5–7 %	≈ 75–85 % screened	15–25 % missed
Endometrial	3–5 %	≈ 70 % screened	25–30 % missed
Other tumours (pancreatic, prostate, thyroid, renal, brain)	5–10 % combined	< 30 % routinely tested	> 70 % missed

Tumour type	Estimated hereditary proportion	Proportion meeting NHS criteria	Hereditary cases still missed
Overall across NHS	≈ 8 % hereditary burden	≈ 3–4 % tested	**≈ 50 % of carriers unidentified**

3.4 Policy Interpretation

Even after two decades of policy progress, the system remains reactive.
Guidelines can only work if they are applied and re-applied as knowledge evolves.
Rachel's case illustrates that eligibility alone does not prevent loss of opportunity; without an annual structured review, each episode is treated in isolation and the hereditary thread is missed.

3.5 Economic Implications of Missed Testing (Overview)

- Late-stage treatment costs 6–8 × more than early intervention.
- Every 10 hereditary cases detected early can save > £1 million per Integrated Care System per year.
- Litigation arising from missed diagnosis is the costliest category of clinical-negligence claims (£13 billion provision in 2023/24).
- AHRR implementation would cost < £160 per patient per year and yield a return of £4–£6 for every £1 invested.

In summary: Policy frameworks define eligibility; Rachel's Rule creates accountability.
It ensures that eligibility criteria translate into consistent, year-on-year practice.

4 Economic and Legal Case

4.1 The True Cost of Late Detection

Late-stage and metastatic disease remains one of the NHS's most expensive areas of care.
Across major tumour types, delayed hereditary identification leads to repeated treatments, complex drug regimens and prolonged palliative support.

Cost Category	Examples of Components	Typical NHS Cost (2024 values)
Acute & Surgical Treatment	Major resection, reconstruction, multiple admissions	£40 000 – £70 000
Adjuvant & Systemic Therapy	Chemotherapy, radiotherapy, immunotherapy	£30 000 – £60 000
Targeted Drugs	PARP / CDK4/6 inhibitors, biologics	£3 000 – £6 000 per month
Diagnostics & Emergency Care	Imaging, endoscopy, emergency surgery	£5 000 – £10 000
Community / Palliative Care	Hospice, district nursing, medication	£800 – £1 200 per week
Average Total Late-Stage Cost per Case	—	**£120 000 – £150 000**

NHS England attributes **~£1.1 billion per year** to escalation and crisis oncology care (NHS Cancer Data 2023).

4.2 The Cost of Prevention and Annual Review

Cost Element	Assumption	Annual Cost per Patient
Administrative time (10 min GP + 5 min nurse @ £3.20/min)	15 minutes	£48
Digital infrastructure (EHR query, data storage & audit)	Fixed per-patient share	£10
Germline panel test (≈ 20 % patients flagged annually)	£400 × 0.2	£80
Follow-up surveillance (≈ 5 % patients under monitoring)	£300 × 0.05	£15
Total Average Annual Cost	—	**≈ £150 – £160 per patient**

4.3 Per-1 000-Patient Cohort Example

Item	Volume / Assumption	Annual Cost
Administrative review	£48 × 1 000	£48 000
Digital infrastructure	£10 × 1 000	£10 000
Genetic testing	200 tests × £400	£80 000
Surveillance imaging	50 patients × £300	£15 000
Total Implementation Cost	—	**≈ £153 000 per 1 000 patients**

4.4 Savings and Net Value

Avoiding just **10 advanced hereditary cases per 1 000 patients** (1 %) saves ≈ £1.2 – £1.5 million.
After implementation costs, **net saving ≈ £1.0 – £1.3 million per 1 000 patients**.

Scenario	Detection Improvement	Net Annual Saving / 1 000 Patients
Conservative	0.5 % (5 cases prevented)	≈ £450 k
Base Case	1 % (10 cases prevented)	≈ £1.0 m
Ambitious	2 % (20 cases prevented)	≈ £2.0 m

4.5 Macro-Economic Benefits

- **Workforce productivity:** Cancer-related absence ≈ £3 billion per year (ONS 2024).
  Early detection typically halves absence duration.
- **Public finances:** Each working-age person kept well preserves ≈ £14 000 in annual net tax benefit (£9 000 tax + £5 000 welfare avoided).
- **Household impact:** Average family spends £500 – £1 000 per month on travel and care during advanced treatment; these costs are largely preventable.
- **Health inequality:** Early risk review benefits areas of high deprivation most, advancing the NHS Long Term Plan goal of equitable outcomes.

4.6 Legal and Professional Protection

Missed hereditary diagnoses sit among the highest-value negligence claims.
NHS Resolution (2023/24) reports £13 billion in outstanding provisions, with diagnostic error a major component.

AHRR provides:

- A recorded annual safety check for every eligible patient.
- An auditable trail demonstrating reasonable preventive practice.
- Alignment with the patient-safety culture exemplified by *Martha's Rule*.

Overall return: for each £1 invested in AHRR and testing, the NHS saves £4 – £6 through reduced treatment and litigation costs.

The Annual Hereditary Risk Review (AHRR) Framework

5.1 Purpose and Principles

The AHRR transforms hereditary-risk management from a one-off eligibility check into an ongoing, auditable process. Every patient with a personal or family history of cancer, a relevant benign condition, or other hereditary indicator will receive a documented annual review.
Any clinician may trigger a review mid-cycle if new information emerges.

Objectives

- Detect hereditary patterns earlier and more consistently.
- Ensure that evolving guidelines automatically reach existing patients.
- Create an electronic audit trail proving proactive prevention.
- Reduce regional inequality in access to genetic testing.
- Protect clinicians and trusts through transparent documentation.

5.2 Operational Process

Step	Action	Responsible Professional	Outcome
1 Automated Case-Finding	EHR algorithms identify patients with multiple primaries, early-onset cancers (< 40 yrs), or benign hereditary indicators.	ICS digital-genomics team	List of patients for review
2 Annual Review	Structured 15-minute template used to confirm diagnoses, update family history, and screen for new red-flags.	GP / Genetic liaison nurse	Risk score updated
3 Trigger Thresholds	(a) Cancer < 40 yrs (b) Multiple or bilateral cancers (c) Rare histology (d) Benign hereditary indicators (e) Change in guidelines	Automatic via template	Referral to local Genomic Medicine Centre
4 Inter-Year Escalation	Any clinician can manually flag new concerns between annual cycles.	Any registered staff member	Immediate review

Step	Action	Responsible Professional	Outcome
5 Audit & Reporting	Quarterly compliance and outcome reporting to Cancer Alliance board.	Trust genomics lead	Public-domain metrics

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5.3 Integration with Existing NHS Systems

Existing Structure	Added Value of AHRR
NHS Genomic Medicine Service	Turns national policy into operational assurance.
Federated Data Platform	Enables cross-trust pattern recognition and recall.
Cancer Alliance Audits	Adds measurable hereditary-review compliance.
Martha's Rule Framework	Parallel right of escalation for patients and staff.
Primary-Care QOF Metrics	Incorporates AHRR completion as a quality indicator.

5.4 Non-Malignant and Sub-Clinical Indicators

Current NHS guidance largely confines germline testing to malignant diagnoses.
Benign or functional features that may signal hereditary syndromes—hamartomas, thyroid nodules, fibroids, macrocephaly, recurrent urticaria, or autoimmune manifestations—are rarely recorded for future review.
Even today these standalone findings would not prompt referral or a note to watch for future conditions.
Rachel's Rule bridges this gap.

AHRR Inclusion Criteria for Benign Indicators

Indicator / Early Feature	Associated Hereditary Syndrome (examples)	Later Life Risks if Unrecognised
Multiple hamartomas (skin, hepatic, mucosal)	PTEN Hamartoma Tumour Syndrome / Cowden	↑ Breast, thyroid, endometrial cancer
Thyroid nodules / multinodular goitre	PTEN / DICER1	Thyroid or ovarian tumours
Benign uterine fibroids / heavy periods / endometrial polyps	PTEN / Lynch	Endometrial and ovarian cancer
Benign breast fibroadenomas or fibrocystic change	PTEN / CHEK2 / ATM	Breast cancer later onset
Recurrent urticaria / autoimmune-type inflammation	PTEN / PIK3CA	Immune dysregulation, neoplasia
Skin tags, lipomas, oral papillomas	PTEN / NF1 / TSC1-2	Multisystem tumour risk
Renal cysts / angiomyolipomas	VHL / Birt-Hogg-Dubé	Renal carcinoma, lung cysts
Macrocephaly ± developmental traits	PTEN / NF1 / TSC	Neuro-oncologic risk
Benign thyroid or parathyroid lesions	MEN 1 / MEN 2 / PTEN	Endocrine tumours

AHRR Action:
Each benign or early finding must be recorded, coded, and re-evaluated annually against updated genomic guidance. Where patterns accumulate—e.g., hamartomas + thyroid nodules + ovarian tumour—the patient moves automatically to genetics referral.

Supporting Evidence Base

- **ERN GENTURIS PTEN Guidelines (2020):** multiple hamartomas → PTEN testing recommended.
- **NICE NG241 (2024)** and **CG164 (2023)** acknowledge hereditary predisposition beyond family history.
- **Genomics England PTEN Learning Hub (2024)** emphasises cross-organ pattern recognition.

The AHRR extends prevention upstream—from reacting to cancer to recognising the silent precursors of hereditary disease years earlier.

5.5 Governance and Data Security

- AHRR records stored within NHS EHR under existing GDPR lawful-basis provisions (public task / patient interest).
- Data visible only to authorised clinicians within the patient's care circle.
- Annual pseudonymised audit submitted to NHS Genomics for national benchmarking.
- Patients retain full rights to access and opt-out under NHS Genomic Data Policy (2024).

6 Implementation and Evaluation

6.1 National Implementation Model

The AHRR can be deployed using the NHS's existing genomic and data-infrastructure, minimising cost and disruption. It requires only modest software modifications, annual professional-development sessions, and clear accountability through Cancer Alliances.

Implementation Phase	Key Actions	Lead Bodies	Outputs / Deliverables
Phase 1 – Pilot (Year 1)	• Select 3–5 Integrated Care Systems (ICS) representing urban and rural areas • Baseline audit of hereditary testing rates and time to referral • Train multidisciplinary teams on AHRR template use • Link FDP query to Genomic Medicine Centres	NHS Genomics / Cancer Alliances / ICS Boards	Proof of concept and cost model validated
Phase 2 – National Roll-Out (Years 2–3)	• Embed AHRR digital prompts in EHR systems (EMIS, SystmOne, Cerner etc.) • Publish national AHRR manual & training modules via Genomics Education Programme • Set annual compliance targets (> 90 % completion within 24 months)	NHSE Genomic Medicine Service / HSSIB / Trusts	Nationwide coverage with standardised reporting
Phase 3 – Sustainability (Years 4–5)	• Annual review of guidelines and data integration • Embed AHRR within CQC inspection framework	NHSE / CQC / Cancer Alliances	Continuous improvement cycle and public accountability

Implementation Phase	Key Actions	Lead Bodies	Outputs / Deliverables
	• Publish national league table of AHRR performance • Link AHRR metrics to Quality & Outcomes Framework		

6.2 Digital and Data Requirements

- **Template integration** – short structured fields in GP and hospital EHRs; flag appears automatically when criteria met.
- **Federated Data Platform (FDP)** – hosts annual query search across Trusts to identify eligible patients.
- **Audit dashboard** – aggregated regional data on AHRR completion, referrals made, variants found.
- **Inter-operability** – FHIR-standard APIs allow cross-system communication without extra licences.

Data protection follows the NHS Data Security and Protection Toolkit; patients' rights of access and opt-out are preserved.

6.3 Workforce and Training

- **Primary Care Teams:** annual CPD module (1 hour online) covering AHRR criteria, data entry and referral workflow.
- **Secondary Care:** integration into Trust mandatory training for oncology, gynaecology and pathology staff.
- **Genetic Counsellors:** capacity planning through Genomics Education Programme expansion.
- **Administrative Support:** utilise existing Cancer Alliance data teams for quarterly reporting.

Estimated workforce impact: +0.2 FTE per 10 000 patients at ICS level – absorbed within current staffing variability.

6.4 Evaluation Metrics

Domain	Indicator / Measure	Target (3 Years)	Data Source
Safety	AHRR completion rate	≥ 90 %	Cancer Alliance audit
	Number of missed hereditary cases identified post-AHRR	Year-on-year reduction ≥ 30 %	Genomic Medicine Centres
Equity	Regional variation in testing rates	≤ 10 % difference between regions	NHSE Genomics Dashboard
Efficiency	Average cost per detected carrier vs cost per advanced case	≥ 5× return on investment	NHS Finance Model
Experience	Patient and clinician satisfaction surveys	≥ 85 % positive response	PROMs / PREMs

Quarterly progress reports feed into Cancer Alliance governance and the NHSE Genomics Board.

6.5 Alignment with National Strategies

- **NHS Long Term Plan (2019):** supports 75 % early-stage diagnosis target by 2028.
- **Genomic Medicine Service Strategy (2024–27):** commits to equitable access to testing.
- **Patient Safety Strategy (2023):** emphasises learning from diagnostic error.
- **Women's Health Strategy (2022):** highlights early identification of hereditary gynaecological conditions.

AHRR directly delivers on all four.

7 Policy Recommendations

The following actions translate Rachel's Rule into deliverable national policy.
Each is aligned to existing NHS structures and requires minimal new legislation—relying instead on directive letters from NHSE Genomics and accountability through Cancer Alliances.

No.	Recommendation	Implementation Route	Expected Outcome
1	**Mandate the Annual Hereditary Risk Review (AHRR)** within all NHS Genomic Medicine Service regions.	NHSE Genomics circular; amendment to GMS operating manual.	Annual review becomes standard of care for hereditary-risk patients.
2	**Retrospective recall audit** of patients diagnosed before 2021 to ensure they are re-assessed under current testing rules.	One-off data query across FDP and NCRAS datasets.	Thousands of previously excluded patients offered updated testing.
3	**Digital integration funding** to embed AHRR prompts within primary- and secondary-care electronic records.	Targeted Digital Investment Fund; supplier collaboration.	Automatic identification of eligible patients and annual reminders.
4	**Annual Cancer Alliance reporting** on AHRR compliance, referral volumes and variant yield.	Add to existing Quality Surveillance indicators.	Transparent measurement and reduction in regional inequality.
5	**Quality & Outcomes Framework (QOF)** inclusion.	NHS England Primary Care contract update.	Incentivises routine completion of hereditary reviews.
6	**Professional recognition & indemnity support.**	Work with NHS Resolution and GMC.	Clinicians protected by demonstrating adherence to proactive safety process.
7	**Public and patient awareness campaign.**	Collaboration with NHS Genomics and charities.	Patients request hereditary reviews proactively, driving bottom-up safety culture.

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8 Conclusion – Protecting Today, For Tomorrow

Rachel's life and story highlight a truth that policy documents rarely expose:
guidelines, no matter how sophisticated, cannot save lives unless they are re-applied as knowledge evolves.

Only one of Rachel's cancers—the 2012 ovarian adenocarcinoma—would meet today's testing rules.
Her first (2005 dermoid squamous carcinoma) and third (2019 ER-positive breast cancer at 41) would still fall outside eligibility even now.
She was referred for genetic testing only after three primaries.
Earlier benign clues—hamartomas, thyroid and gynaecological abnormalities—were recorded but never connected.

Rachel's Rule converts those missed fragments into an annual, system-wide safety check:
a low-cost, high-value review ensuring that no hereditary risk remains hidden in plain sight.
It turns written eligibility into living accountability, protecting patients, clinicians, and the NHS alike.

Guidelines are promises on paper; Rachel's Rule makes those promises real.

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End of Rachel's Rule – Revised Full Policy Briefing 2025