Learning & Self-Awareness Pack (Version 1.0)

For patients, families, clinicians, and decision-makers

A patient-safety awareness resource about hereditary risk when family history is absent

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Disclaimer (Please Read)

This learning resource is provided for **general awareness and patient-safety learning** only.

It is **not medical advice**, does not provide diagnosis, and must not be used as a substitute for professional clinical judgement.

If you have concerns about your health or cancer risk, you should speak to your GP, specialist team, or a qualified healthcare professional.

This resource reflects **lived experience** and the patient-safety learning drawn from Rachel's case. It is intended to support awareness, reflection, and earlier recognition of risk patterns — especially when those patterns may be missed.

Why This Resource Exists

Rachel's Rule exists because hereditary risk can be missed when:

- care is fragmented across specialties
- health issues are treated one at a time
- no one is responsible for reviewing the whole picture
- the question of hereditary risk is delayed or never raised
- **family history is absent**, so risk is assumed to be low

Rachel died aged 47. Her death was not inevitable. It followed years of missed opportunities — signs that were there in plain sight but never joined together.

The Most Important Message

Hereditary risk does not require a family history.

Many people assume hereditary risk only applies when:

- "everyone in the family has cancer"
- there is a dramatic cluster of deaths
- there is an obvious inherited pattern

That assumption is one of the biggest reasons people fall through gaps.

Hereditary cancer syndromes can be missed because:

- a condition is **de novo** (a new genetic change not inherited from parents)

- family size is small
- relatives are mostly male (so risk patterns are less visible)
- relatives died young of other causes
- family medical history is unknown or incomplete
- previous cancers were never discussed openly

Rachel's Rule exists to ensure the absence of family history does not lead to false reassurance.

What "Hereditary Risk" Means — and What It Does Not

Hereditary / genetic risk does not require:

- a known family history
- multiple relatives with cancer
- a dramatic "textbook" presentation

Hereditary risk can still exist when:

- there are multiple health issues over time
- there is a pattern of benign growths or lesions
- there are multiple cancers, especially at a young age
- there are unusual combinations of conditions across different organs
- there are repeated "rare" findings that are never linked together

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The Core Patient-Safety Problem: Siloed Care

Healthcare is often excellent within each specialty.
The danger arises when:

- one team focuses on one organ
- another team focuses on another
- no one is tasked with joining the dots
- risk is assumed to be "someone else's responsibility"

Rachel's Rule is a safeguard against that system blind spot.

When One Is Enough (Key Principle)

Rachel's Rule is built on two principles:

1) One significant event can be enough to trigger review.

This is especially true when that event is unusual, early, or complex.

2) Surveillance must follow the person — not just organs.

A genetic label alone is not enough if follow-up remains fragmented.

Triggers That Should Prompt Hereditary Risk Consideration

(Even when family history is absent)

A hereditary risk question should be considered when someone has:

A) Cancer-related triggers

- A first cancer diagnosis at a younger age (for example under **50**)
- Multiple primary cancers (more than one separate cancer diagnosis)
- Rare cancers, or unusual combinations
- Cancer plus multiple "non-cancer" red flags over time

B) Multi-system / long-term triggers

- Longstanding benign lesions or recurrent growths
- Multiple unexplained symptoms affecting different body systems
- A history of hamartomas, polyps, or repeated "benign" findings
- Thyroid issues that persist alongside other red flags
- Patterns of health issues from childhood onward

Important:
These do not prove hereditary risk.
They simply justify a **whole-person review**.

Rachel's Case: The Patient-Safety Learning (Non-Blame)

Rachel experienced multiple health challenges across her life.

From childhood and early adulthood, there were red flags, including:

- recurrent skin issues and lesions
- benign lesions
- thyroid problems (including goitre)
- hamartomas discovered later
- multiple cancers across different organs

The crucial patient-safety issue was not that any one clinician missed everything.

It was that **no system safeguard existed** to ensure the overall pattern was reviewed.

The Critical Delay (What Must Not Happen Again)

In Rachel's case, the hereditary risk question was not raised until **after her third primary cancer diagnosis**.

By the time hereditary risk is raised at that stage:

- opportunities for earlier surveillance have already been lost
- risk has already had time to progress
- the person may already be living with advanced disease

Rachel's Rule exists to ensure the hereditary risk question is raised **earlier**, and revisited regularly.

Why a Genetic Diagnosis Alone Is Not Enough

Rachel was later diagnosed with **Cowden Syndrome (PTEN Hamartoma Tumour Syndrome)**.

However, even after diagnosis, the surveillance that followed was largely:

- organ-based
- siloed
- disconnected from her overall risk
- not clearly owned by any one accountable process

In Rachel's case:

- breast surveillance ceased after preventative mastectomy
- follow-up did not sufficiently reflect recurrence risk
- surveillance did not feel whole-person, joined-up, or reassessed over time

Key learning:
A genetic diagnosis can become a label — but labels do not automatically create safe, joined-up care.

What Rachel's Rule Calls For (Conceptually)

Rachel's Rule calls for a system safeguard that ensures:

- hereditary risk is not missed
- patterns are reviewed, not just isolated events
- the absence of family history does not stop the question being asked
- the person's overall risk is reassessed regularly
- surveillance is coordinated around the person, not individual organs

This learning pack is not a policy document.
It is an awareness resource.

Rachel's Rule and Rachel's Pathway proposals are set out separately.

For Patients and Families: What You Can Do

If this resource resonates, you can consider:

- Writing down your health history in one timeline
- Listing cancers, benign findings, lesions, and long-term conditions
- Noting ages at diagnosis (even approximate)
- Asking whether anyone has reviewed your history as a whole
- Asking whether hereditary risk has ever been considered

You are not being difficult by asking.

You are asking for the system to do what it is meant to do:
join the dots.

For Professionals: A Reflection Prompt

Rachel's Rule is not an accusation.
It is a patient-safety observation:

When care is fragmented, cumulative risk can become invisible.

Consider:

- Where whole-person review currently sits
- Whether it has an accountable "home"
- Whether family history absence creates false reassurance
- Whether a patient's multi-system history is being actively integrated
- Whether surveillance after genetic diagnosis remains siloed

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The Point of Annual Review (Plain English)

An annual hereditary risk review is not about diagnosing everyone.

It is about:

- pausing once a year
- reviewing the whole picture
- asking "does anything now add up differently?"

- ensuring risk is not lost between specialties
- ensuring the patient has a plan that follows them

This is a safety check — not a judgement.

Closing Statement

Rachel's Rule exists because hereditary risk can hide in plain sight.

Not because the signs are invisible —
but because the system often lacks a dedicated mechanism to connect them.

Rachel's story shows why the hereditary question must be asked:

- **earlier**
- **more consistently**
- **without relying on family history**
- **with surveillance that follows the person, not just organs**

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