

# Clinical Case Reports as a Tool for Advancing Personalized Medicine: Is this real?

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**Abstract:** Personalized medicine recognizes that diseases manifest and respond to treatment differently in each patient. While large trials provide general evidence, they often miss real-world variability. Case reports, long considered low-level evidence, are vital for documenting rare diseases, atypical presentations, and unique therapeutic responses that inform individualized care. Historically, case reports have led to major discoveries, embracing variability to reveal new mechanisms. In rare diseases, each report can serve as an “N-of-1” study, while in common conditions they help identify unusual patterns, reduce misdiagnosis, and define subgroups needing tailored management. They also record exceptional responders, non-responders, and rare adverse effects, advancing pharmacogenomics. Despite their value, challenges include patient privacy, publication bias, and inconsistent quality, which require robust consent and adherence to CARE guidelines. Future integration into searchable databases and AI analysis could amplify their impact, confirming that case reports remain central to the precision medicine era.

**Keywords:** Case Reports; Personalized Medicine; Precision Medicine; Rare Diseases; Therapeutic Response.

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## 1. Introduction

In recent decades, the paradigm of healthcare has shifted from a one-size-fits-all approach to a model that emphasizes the uniqueness of each patient. This transformation commonly referred to as personalized medicine or precision medicine, recognizes that disease manifestations, treatment responses, and prognoses can vary significantly among individuals, even those with the same diagnosis. Advances in genomics, molecular biology, imaging, and bioinformatics have accelerated this shift, enabling the development of diagnostic and therapeutic strategies tailored to the genetic, environmental, and lifestyle characteristics of each patient.

While large-scale clinical trials and population-based studies have been instrumental in providing generalizable evidence, they often fail to capture the nuanced variability seen in real-world patients. In this context, the humble case report, long considered the lowest rung in the hierarchy of evidence, emerges as an indispensable tool for advancing personalized medicine. Detailed documentation of rare cases, atypical disease presentations, unexpected therapeutic responses, and novel diagnostic approaches can provide critical insights that inform individualized patient care and spark new lines of research.

## 2. The Historical Role of Case Reports in Shaping Medicine

Case reports are among the oldest forms of medical literature. From Hippocrates' meticulous accounts of patient symptoms to the early 20th century documentation of rare

pathologies, they have served as the foundation upon which many medical breakthroughs were built. Seminal discoveries, such as the first description of Parkinson's disease, the identification of AIDS, or the recognition of novel drug toxicities, originated not from randomized controlled trials (RCTs), but from careful observation and reporting of individual cases.

In the era of personalized medicine, this role becomes even more relevant. Where RCTs often seek to minimize variability to achieve statistical significance, case reports embrace variability as a potential source of innovation. They highlight the "outliers" whose experiences challenge standard algorithms and whose unique biology or clinical circumstances can reveal new mechanisms of disease or therapeutic action.

Personalized medicine thrives on granularity. The more detailed the clinical, laboratory, imaging, and follow-up data, the better the ability to recognize patterns that might otherwise remain hidden. A well-written case report offers precisely this level of detail, including: comprehensive patient history capturing comorbidities, environmental exposures, lifestyle, and family background; genetic and molecular profiling when available, linking disease manifestations to underlying biological pathways; treatment course and rationale, describing not only what was done but why it was chosen over alternatives; outcome tracking over meaningful follow-up periods, documenting both objective measures and patient-reported experiences, and such richness of data allows clinicians and researchers to generate hypotheses about why a particular patient responded, or failed to respond, to an intervention. In some cases, it may reveal predictive biomarkers or contraindications that can later be validated in larger studies.

### 3. Rare Diseases and the Power of the *N-of-1*

The intersection between case reports and personalized medicine is most obvious in rare diseases. For conditions that affect only a handful of individuals worldwide, it is impractical, and often impossible to conduct large-scale trials. In these situations, each case becomes an "N-of-1" experiment, and cumulative knowledge is built one patient at a time. For example, in certain rare genetic syndromes, case reports have documented unexpected benefits from repurposed drugs based on shared molecular pathways with more common conditions. Similarly, in oncology, individual reports of patients with unique tumor mutational profiles responding dramatically to targeted therapies have led to the development of new treatment protocols and clinical trials. Without the careful documentation provided by case reports, these breakthroughs would likely remain anecdotal, unshared, and lost to the broader medical community.

Beyond rare diseases, case reports play a critical role in capturing atypical presentations of common conditions. Personalized medicine depends on recognizing that not every patient will fit the textbook description. Misdiagnosis or delayed diagnosis often occurs when clinicians are unaware of the full spectrum of a disease's manifestations. By describing unusual symptom clusters, unexpected disease trajectories, or rare complications, case reports expand the diagnostic horizon. This can lead to earlier recognition, more accurate diagnosis, and appropriately tailored interventions for future patients who present in similar ways. Moreover, these atypical cases can highlight subgroups within a broader disease category who may require distinct management strategies. For instance, patients with diabetes who exhibit unique glycemic patterns or those with autoimmune disorders who respond paradoxically to standard immunosuppressive therapies may represent biologically distinct subsets that warrant further investigation.

One of the most compelling contributions of case reports to personalized medicine is their ability to document unusual therapeutic responses. These can take several forms: 1. exceptional responders: patients who achieve remarkable benefits from a treatment typically considered ineffective in the general population; 2. primary non-responders: patients who fail to benefit from therapies that are highly effective for most, prompting exploration of alternative mechanisms; 3. adverse drug reactions: unique toxicities or hypersensitivities that suggest genetic predispositions or drug-drug interactions requiring

individualized caution. By recording and publishing these experiences, case reports can catalyze pharmacogenomic research, support the development of predictive tests, and inform personalized prescribing guidelines.

Another often-overlooked strength of case reports is their ability to bridge the gap between cutting-edge research and everyday clinical practice. Personalized medicine technologies, such as next-generation sequencing, advanced imaging modalities, and AI-driven diagnostics, are often developed in highly specialized centers. Case reports allow clinicians in diverse settings to see how these tools can be applied to real patients, including those with comorbidities, limited resources, or cultural contexts not represented in major trials. In turn, this democratizes access to personalized care by providing relatable examples that can be adapted to different healthcare environments.

Despite their value, case reports must navigate several challenges to fulfill their potential in personalized medicine. The most pressing include concerns about patient privacy, as personalized medicine often involves detailed genetic and phenotypic data, increasing the risk of patient identification even when names are removed, thus making robust consent processes essential. Another challenge is publication bias, given the tendency to report only “successful” or dramatic cases, which can distort the understanding of true variability; therefore, publishing negative or inconclusive outcomes is equally important. Finally, quality and reproducibility remain critical issues, as inconsistent reporting standards can limit the ability of others to learn from or replicate findings, underscoring the need for adherence to guidelines such as CARE (CAse REport).

#### 4. The Future: Integrating Case Reports into Data Ecosystems

Looking ahead, the role of case reports in personalized medicine could be further amplified by integrating them into structured, searchable databases linked to genomic, proteomic, and metabolomic repositories. Such integration would enable researchers to rapidly identify patterns across dispersed individual cases, accelerating the validation of hypotheses generated from singular observations. Moreover, artificial intelligence could be harnessed to mine published and unpublished case reports for trends in therapeutic responses, adverse effects, and disease evolution, ultimately guiding more precise treatment recommendations.

#### 5. Conclusion

In the era of personalized medicine, the value of case reports cannot be overstated. They provide the level of detail, the recognition of uniqueness, and the clinical context that are essential for truly individualized care. By capturing the stories of patients who fall outside the average, case reports illuminate pathways for innovation, challenge established paradigms, and remind us that medicine is, at its core, about the care of individuals, not populations. Far from being an outdated form of medical literature, case reports are poised to become a cornerstone of the precision medicine revolution. As such, journals dedicated to their publication, including the *Brazilian Journal of Case Reports*, have a unique opportunity and responsibility: to preserve, disseminate, and elevate these singular narratives so they can shape the future of healthcare, one patient at a time.

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