

Paper Summary

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Title: Reconciling evidence-based medicine and precision medicine in the era of big data: challenges and

Authors: Jacques S. Beckmann, Daniel Lew

DOI: 10.1186/s13073-016-0388-7

Year: 2016

Publication Type: Journal Article

Discipline/Domain: Medicine / Clinical Bioinformatics

Subdomain/Topic: Precision Medicine, Evidence-Based Medicine, Big Data Integration

Eligibility: Eligible

Overall Relevance Score: 85

Operationalization Score: 70

Contains Definition of Actionability: Yes (implicit, in terms of “clinically actionable knowledge”)

Contains Systematic Features/Dimensions: Yes

Contains Explainability: Yes

Contains Interpretability: Partial

Contains Framework/Model: Partial (conceptual integration model)

Operationalization Present: Yes (data integration, bioinformatics workflow)

Primary Methodology: Conceptual / Review

Study Context: Integration of precision medicine and evidence-based medicine in big data healthcare

Geographic/Institutional Context: Switzerland (SIB Swiss Institute of Bioinformatics), global implications

Target Users/Stakeholders: Clinicians, bioinformaticians, healthcare policymakers, patients/citizens

Primary Contribution Type: Conceptual framework and challenges/opportunities analysis

CL: Yes

CR: Yes

FE: Yes

TI: Partial

EX: Yes

GA: Yes

Reason if Not Eligible: N/A

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****Title:**** Reconciling evidence-based medicine and precision medicine in the era of big data: challenges

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****Year:**** 2016

****Publication Type:**** Journal Article

****Discipline/Domain:**** Medicine / Clinical Bioinformatics

****Subdomain/Topic:**** Precision Medicine, Evidence-Based Medicine, Big Data Integration

****Contextual Background:**** The paper addresses the tension and complementarity between precision medicine and big data

****Geographic/Institutional Context:**** Switzerland; global healthcare systems

****Target Users/Stakeholders:**** Clinicians, bioinformaticians, policymakers, healthcare IT developers, patients

****Primary Methodology:**** Conceptual / Review

****Primary Contribution Type:**** Conceptual framework and challenge–opportunity mapping for integration

General Summary of the Paper

This paper outlines how high-resolution, high-throughput biomedical technologies and big data can drive precision medicine

Eligibility

Eligible for inclusion: ****Yes****

How Actionability is Understood

The authors frame actionability as the ****translation of big, heterogeneous biomedical datasets into “clinically actionable knowledge”**

> “Proper data mining and translation of the vast datasets into clinically actionable knowledge will require a paradigm shift”

> “The real challenge... will be to curate, store, federate, integrate, share, mine, interpret, and transform the data into actionable knowledge”

What Makes Something Actionable

- Standardization and interoperability of clinical and laboratory datasets
- Integration of multi-layered data (genomics, microbiome, lifestyle, environmental)
- Statistical robustness from large cohorts combined with individual-level granularity
- Ethical, legal, and privacy safeguards for trust in data sharing
- Explainable outputs that can guide clinical decisions at the point of care

How Actionability is Achieved / Operationalized

- ****Framework/Approach Name(s):**** Evidence-Based Precision Medicine
- ****Methods/Levers:**** Big data integration, bioinformatics analysis, interoperability standards, meta-analysis
- ****Operational Steps / Workflow:**** Data collection (EHR, wearables, genomics, etc.) → Standardization → Data integration → Analysis → Actionable knowledge
- ****Data & Measures:**** Omics, imaging, clinical measures, behavioral and lifestyle data, environmental exposure
- ****Implementation Context:**** Multi-institutional, transnational data-sharing systems

> “...collation and meta-analyses of big data from cross-institutional and transnational large-scale registries and biobanks”

> “...create sustainable federated, safe data commons or warehouses...” (p. 6)

Dimensions and Attributes of Actionability (Authors' Perspective)

- **CL (Clarity):** Yes — data must be interpretable for clinicians and patients.
- **CR (Contextual Relevance):** Yes — personalized care requires context-specific relevance.
- **FE (Feasibility):** Yes — operational constraints considered in prevention focus.
- **TI (Timeliness):** Partial — early detection emphasized, but not deeply operationalized.
- **EX (Explainability):** Yes — clinical bioinformatics bridges data complexity and clinical understanding
- **GA (Goal Alignment):** Yes — alignment to prevention, wellness, and patient-centered care.
- **Other Dimensions Named by Authors:** Interoperability, standardization, patient empowerment.

Theoretical or Conceptual Foundations

- Systems medicine (P4: predictive, preventive, personalized, participatory)
- Holistic integration of biological and environmental determinants of health
- N-of-one to N-of-many cohort aggregation

Indicators or Metrics for Actionability

- Early detection of symptoms
- Identification of pre-symptomatic individuals
- Delay or prevention of disease onset
- Integration and usability of diverse data streams in decision-making

Barriers and Enablers to Actionability

- **Barriers:** Heterogeneous EHR systems, lack of interoperability, semantic complexity, data silos, privacy
- **Enablers:** Federated safe data commons, standardized vocabularies/ontologies, citizen participation

Relation to Existing Literature

Positions precision and evidence-based medicine as complementary paradigms; builds on P4 medicine, etc.

Summary

Beckmann and Lew (2016) argue for integrating the population-level rigor of evidence-based medicine with

Scores

- **Overall Relevance Score:** 85 — Strong conceptual framing of actionability in a healthcare big data context
- **Operationalization Score:** 70 — Provides a clear conceptual workflow and enabling infrastructure, but

Supporting Quotes from the Paper

- “Proper data mining and translation of the vast datasets into clinically actionable knowledge...” (p. 1)
- “The real challenge... will be to... transform these extensive heterogeneous data into scalable, medically
- “...collation and meta-analyses of big data from cross-institutional and transnational large-scale registers

- “Shifting emphasis of medicine more from therapy to prevention, and from disease to wellness” (Table 1)

Actionability References to Other Papers

- Auffray et al. (2009, 2015) on systems medicine
- Hood & Price (2014) on prevention-focused precision medicine
- Schwaederle & Kurzrock (2015) on actionability in oncology
- Collins (2015) on NIH precision medicine perspective