Published in final edited form as:

Pharmacogenomics. 2013 October ; 14(13): 1529–1531. doi:10.2217/pgs.13.121.

Let's make data on essential pharmacogenes available for every patient everywhere: The Medicine Safety Code initiative

Matthias Samwald^{1,*} and Robert R. Freimuth²

¹Section for Medical Expert and Knowledge-Based Systems, Center for Medical Statistics, Informatics, and Intelligent Systems Medical University of Vienna Spitalgasse 23, A-1090 Vienna, Austria

²Division of Biomedical Statistics and Informatics, Department of Health Sciences Research Mayo Clinic Rochester, Minnesota, U.S.A. freimuth.robert@mayo.edu

Keywords

pharmacogenetics; health care systems; biomedical informatics; clinical decision support systems

Pharmacogenetic data can significantly alter physicians' prescribing behaviour and lead to reduced hospitalization rates [1]. If pharmacogenetic data were widely used to inform medical treatment, patients could profit from reduced incidence of adverse drug events and an improvement of effectiveness of treatment. Similar to the *essential medicines* that the World Health Organization lists as indispensable for health care systems [2], one could devise a list of *essential pharmacogenes* – genes that contain variants that significantly influence an individual's response to medications – with the goal of having a patient's genotype data readily accessible at the point of care.

Barriers to the global implementation of clinical pharmacogenetics

Even though pharmacogenetics holds great potential for improving the quality of pharmacotherapy, there are several major barriers to the integration of pharmacogenetics data into routine clinical practice. Major obstacles include the financial cost and feasibility of pre-emptive genetic testing, as well as the availability and turnaround time for indication-specific tests. The storage, exchange, and interpretation of pharmacogenetic data also represent barriers to widespread adoption. In particular, interoperable electronic health record systems that are capable of storing structured genetic test results and transmitting those results to other care providers are needed; unfortunately, this type of infrastructure is not available in most parts of the world. There is also a lack of easily accessible computer-based clinical decision support systems that can assist clinicians with the interpretation of pharmacogenetic data – a vital component to the use of these data at the point of care. Finally, few resources exist to help patients understand the results of a pharmacogenetic test, which is necessary to enable patients to be more proactive with their health care.

* Author for correspondence: matthias.samwald@meduniwien.ac.at.

A possible solution: the Medicine Safety Code

Many of these barriers are due to limitations in our ability to share and interpret pharmacogenetic test results. To make patient genotype data on essential pharmacogenes globally available for routine medical care, technologically-simple and intuitive systems are required in order to minimize the need for specialized infrastructure, software, and knowledge.

We have recently started the non-profit Medicine Safety Code initiative (<http://safety-code.org/>) in an effort to improve the ability of clinicians and patients to share pharmacogenetic data and make it available at the point of care. This system consists of two major components:

1. The Medicine Safety Code (MSC), a standardized two-dimensional (2D) barcode that captures data about genetic variants in essential pharmacogenes for an individual patient. The 2D barcode is based on the Quick Response (QR) code standard [3], which has become widely popular in recent years because it can be decoded quickly and reliably, has relatively high information density and can encode hyperlinks to pages on the World Wide Web.
2. A centralized knowledge base that contains information about essential pharmacogenes, including haplotype definitions, phenotype translations and clinical guidelines. This knowledge base is used to automatically interpret the genetic data that is encoded within the 2D barcodes. The knowledge base leverages semantic technologies and ontologies to ensure a high degree of interoperability with other systems [4].

A proof-of-concept prototype of the MSC system has been developed and which has performed well in preliminary tests. For the prototype, data on 385 pharmacogenetic markers were encoded as a 2D barcode, which could quickly be decoded and interpreted using a standard smartphone device [5].

Benefits and applications of the Medicine Safety Code

The MSC could act as an enabling technology for the widespread dissemination and clinical implementation of pharmacogenetic data for several reasons:

- **Immediately implementable.** The MSC is based on technology that is low-cost and widely available, and implementations would require minimal integration with existing clinical infrastructure.
- **No specialized hardware or software is required to use the codes.** The 2D barcodes used by the MSC can be printed on personalized cards that patients can carry in their wallets or incorporated in paper-based lab reports. The image can be saved electronically and shared via e-mail. The 2D barcodes can be quickly decoded using common smartphone devices to URLs, which link to pages that describe the genetic results that are encoded in the barcodes as well as interpretive clinical decision support messages that are pertinent to the individual's genetic profile.

- **Providing access to decision support, anywhere.** The MSC enables patients and care providers to access pharmacogenetic data and relevant clinical decision support algorithms in all health care settings, including outpatient and emergency situations, regardless of whether the care facility has implemented a genome-enabled electronic medical record and local clinical decision support algorithms.
- **Patients can opt-in and opt-out at any time.** Patients can choose whether or not to carry their anonymized pharmacogenetic data in their pockets and make their data available to a care provider, putting the issue of privacy in the hands of the patient.
- **Independent of the genotyping platform.** The MSC captures the result of genetic testing independently from the platform that was used to produce the results. Therefore, a wide variety of genotyping or sequencing platforms can be used to generate standardized representations of the most important test results on essential pharmacogenes.
- **Local implementations can be deployed.** The MSC system aims to be independent from local technological platforms, since no specialized equipment is needed for creating, transporting and interpreting the pharmacogenetic information. No investment in a dedicated infrastructure is required to start using the system, but if a customized implementation is desired the MSC can be deployed locally. The system specifications and source code will be made openly available.
- **Lowering costs through data re-use.** The MSC provides a simple, effective means for sharing genetic test results at the point of care. Costs for pharmacogenetic testing could be reduced by facilitating the re-use of genetic test results and a reduction in redundant testing.
- **Barcodes are customizable.** Custom 2D barcodes can be created for specific gene panels, if needed.
- **Facilitating return of results.** The MSC could be used as a simple, low-cost method for returning genetic test results to patients.
- **Facilitating the development of tailored therapeutics.** A broad adoption of the MSC could also provide benefits for drug development. When data on a minimal set of essential pharmacogenes can be made cheaply available for large patient populations, these pharmacogenes could be used for patient stratification during clinical trials ('tailored therapeutics'). This could help to bring new therapies to market faster and with fewer losses caused by concerns about safety and efficacy, which are often encountered in the development of non-tailored therapeutics.

The MSC is intended to complement local pharmacogenetics initiatives by providing a simple method for making pharmacogenetic data more portable across geographic regions and health care networks. The MSC could be utilized in both clinical and research settings and can be tailored to custom gene arrays, such as the *Personalized Medicine Program Custom Array*, which was developed by the University of Florida and Stanford University to genotype 252 pharmacogenetic variants [6]. We are also exploring methods to encode next-

generation sequencing data to support platforms such as PGRN-Seq, an 84 gene array that is being developed by the Pharmacogenomics Research Network (PGRN) [7] and adopted by the eMERGE PGx project [8].

Limitations

The MSC was designed to improve the portability of pharmacogenetic data and provide access to a basic level of interpretive information when specialized systems and knowledge are not available. It is not meant to replace robust, certified clinical infrastructure nor does it substitute for the clinical judgement of a physician or genetic counselor. Nonetheless, while there may be ethical and legal considerations to its use, the MSC does enable patients to be more proactive with their health care.

The amount of information that can be encoded by the MSC is limited by the physical resolution of the QR code. The current implementation of the MSC can support genotypes for up to 500 variants, but improvements in data compression could increase this number. As a result, the MSC can represent the clinically actionable variants that are known today but currently it is not capable of encoding results from whole exomes or genomes.

Patients may decide whether or not to carry their MSC-encoded genetic data with them, but since the code itself is not encrypted it is no more or less secure than any other personal information that might be present within their wallet. If privacy is a concern, additional protections may be necessary.

Future directions and call for participation

The MSC is a technologically-simple and intuitive system that could address many of the barriers that limit the ability to share and utilize pharmacogenetic test results in clinical practice, but the true potential of this project will not be realized until the MSC is tested in different scenarios. Partnerships with clinical and research centers are needed. A call for participation has been posted at: <http://safety-code.org/support.html>.

Acknowledgments

The MSC system is being developed in the context of the World Wide Web Consortium (W3C) Health Care and Life Science Interest Group. The research leading to these results received funding from the Austrian Science Fund (FWF): [PP 25608-N15]. The authors have no other relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript apart from those disclosed. No writing assistance was utilized in the production of this manuscript.

References

1. Epstein RS, Moyer TP, Aubert RE, et al. Warfarin Genotyping Reduces Hospitalization Rates: Results From the MM-WES (Medco-Mayo Warfarin Effectiveness Study). *J. Am. Coll. Cardiol.* 2010; 55(25):2804–2812.
2. WHO Model Lists of Essential Medicines [Internet]. Available from: <http://www.who.int/medicines/publications/essentialmedicines/en/>
3. QR Code.com [Internet]. Available from: <http://www.qrcode.com/en/aboutqr.html>
4. Samwald M, Coulet A, Huerga I, et al. Semantically enabling pharmacogenomic data for the realization of personalized medicine. *Pharmacogenomics*. 2012; 13(2):201–212.

5. Samwald, M.; Adlassnig, K-P. Pharmacogenomics in the pocket of every patient? A prototype based on quick response codes. J. Am. Med. Inform. Assoc. 2013. [Internet]Available from: <http://jamia.bmj.com/content/early/2013/01/22/amiajnl-2012-001275>
6. Johnson JA, Burkley BM, Langaee TY, Clare-Salzler MJ, Klein TE, Altman RB. Implementing personalized medicine: development of a cost-effective customized pharmacogenetics genotyping array. Clin. Pharmacol. Ther. 2012; 92(4):437–439. [PubMed: 22910441]
7. Network-wide Projects - PGRN Web Site - Confluence [Internet]. Available from: <http://pgrn.org/display/pgrnwebsite/Network-wide+Projects>
8. Workgroups | eMERGE [Internet]. Available from: <http://emerge.mc.vanderbilt.edu/workgroups>