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Coming to you –
Your Genomic Health Record

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Summary

- A **new genomic health record** will become a reality as genomic information gets combined with relevant data extracted from the traditional EMR

- **Rapid, precision oncology decision support** is expected to expand on a larger scale by performing sophisticated genome/proteome/RNA analysis coupled with cognitive computing.

- The capabilities and technologies associated with **cognitive computing** are critical to the ongoing genomic medicine evolution and will enable:
  - more sophisticated decision support
  - innovative clinical trials
  - new targeted therapies
  - a focus on health outcomes to prevent and alleviate disease symptoms.

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Agenda

- Summary

- Genomic Medicine is a reality

- Major forces impacting Genomic Medicine

- Three significant impacts of Genomic Medicine (focus on Genomic Health Record)

- Helping your organization develop genomic medicine strategies

- Conclusions

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Genetic Medicine vs. Genomic Medicine

Genetic Medicine

- Low frequency of usually "high penetrance genes"

- Uncommon diseases e.g. Huntington disease

- Focused on single genes

Genomic Medicine

- The genetic risk for common diseases will often be due to disease-producing alleles with relatively high frequencies (>1%).

- All genes may be disease causing.

- Common disorders

- Interactions of multiple genes and environmental factors

- Access to the entire genome

Slide 1

Slide 2

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In the Next year….

- It is now 2016 and whole-genome sequencing coupled with other genomic modalities like RNA-Sequencing (RNA-Seq) and oncology-specific multiplex panels of genes are **routinely** offered to cancer patients.

- The woman, now 47 years old, gets her tumor block analyzed by three such genomic modalities upon expert advice provided **by a cognitive system** that has been trained in medicine, genomics and the oncology specialty.

- The cognitive system continues to learn and provides the oncologist with key confidence intervals regarding options.

- Upon choosing the option with the highest confidence interval, another mutation is found in the primary tumor which, fortunately, also has a targeted treatment.

- **The woman is now taking two targeted medications** and leading a reasonably good quality of life.

- Her cancer can be treated as a "complex chronic condition" with care that is personalized for her own genetic makeup – targeted care that was not yet available to her in 2013.

Slide 3

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Genomic Medicine is a reality

➢It is 2014 and a 45-year-old woman has been diagnosed with adenocarcinoma of the lung.

➢ A PET-CT scan reveals that there is metastasis to the adrenal gland, brain and bone. She meets with her oncologist, who does a lung biopsy of the tumor.
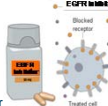
➢Immunohistochemistry confirms that she has Stage IV non-small cell lung cancer.

➢Her oncologist recommends that she submit her tumor biopsy for molecular testing for EGFR and ALK mutations, which confirms that she has an EGFR3 mutation.

➢This mutation is seen in 30 to 40 percent of women who are non-smokers and of Asian origin, as she is.

➢Her oncologist offers her first-line treatment with a particular EGFR inhibitor.

➢Eight weeks later, a PET-CT scan shows shrinkage of her tumors. She does not go through chemotherapy and continues taking a daily pill as part of her treatment regimen

EGFR Inhibitor
Blocked receptor
Treated cell

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Genomic medicine is at a unique inflection point in medicine's history due to three major forces:

1. **Innovation in Biology and Technology**
2. **Market Demand**
3. **Consumerism**

Slide 1

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1. Innovation in biology and technology

- Highly specific gene expression panels

- An increased understanding of complex pathways and corresponding development of targeted drugs.

- Decreasing cost and increasing capability of high-performance computing (HPC)

- A new era of computing – cognitive computing

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2. Market demand

- The healthcare and life sciences industries are at a crossroads:
  - The patent cliff
  - Reimbursement pressures
  - A thirst for new ways of impacting health outcomes through innovative therapies.
  - The increased expectation and market demand for targeted products (such as drugs and tests)

- Genomics converging with social media
  - This convergence has led to near-instant dissemination of new discoveries, as well as new forms of social and scientific collaboration.

- Increased computing power, sophisticated imaging capability

- The use of wireless sensors and the plethora of accessible digital health information are also part of the technological mix that is helping to push genomics forward.

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Plumetting costs and increased efficiency

Source: National Human Genome Research Institute" http://www.genome.gov/ images/content/cost_per_genome.jpg; Adapted by IBM Research.

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3. Consumerism

- Increased uptake of direct-to- consumer genomic testing, and in more recent regulatory concerns and recommendations about such technologies.

- Regulatory and legislative precedents in the U.S. –
  - 2008 Genetic Non-Discrimination Act (GINA)
  - 2013 Supreme Court decision regarding gene patenting with a biotechnology company to protect consumers of genomic data

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Exploring major changes from genomic medicine

Genomic medicine is a "game-changer" for important stakeholders that include patients, providers, researchers, payors, diagnostic companies, policy makers, life sciences and governments.

Three significant impacts are underway:

(1) The growing use of a **new genomic health record**;

(2) Greater benefits for stakeholders in the **three genomic medicine "cornerstones"** of sequencing, translational medicine and personalized healthcare; and

(3) Opportunities for **radical industry transformation**.

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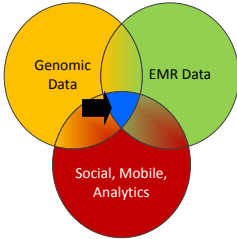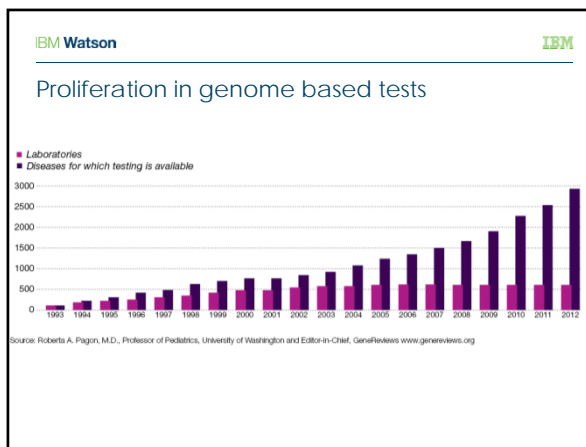## This prediction of widespread use stems from:

- The decreased costs of sequencing the human genome

- The proliferation and availability of genome-based tests in the past five years

- The rising adoption of electronic medical records

- The increased use of genome data to recommend targeted treatments using companion molecular diagnostics

- A growing willingness of payors to reimburse payments on some genetic tests today

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1. Growing use of a new genomic health record

In the next three to five years, it is likely that an individual who interacts with the healthcare system will have a genome-based electronic record or "genomic health record."

➢It will be used in tandem with other electronic healthcare tools for decision support, prevention, and customized testing and treatments

➢At first, the record may not contain the whole genome sequence, but instead perhaps the results from testing specific genes

➢This genomic health record will converge with mobile, social, cloud and analytics

Genomic Data

EMR Data

Social, Mobile, Analytics

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Proliferation in genome based tests

■ Laboratories
■ Diseases for which testing is available

Source: Roberta A. Pagon, M.D., Professor of Pediatrics, University of Washington and Editor-in-Chief, GeneReviews www.genereviews.org

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Genomic Health Record

- A genomic health record would extract and integrate relevant electronic health data **with a person's genome data**. To use such data effectively, **new types of decision support will be required to personalize risk, prevention and follow-up treatment**. Owing to the complex uses of and needs for such data, the genomic health record is likely to be distinct from the traditional EMR often generated from an EMR system.

- The healthcare and life sciences industries are poised for greater use of clinical decision support. This is supported by recent EMR adoption data.

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The role for Cognitive Computing and Genomic Data

- With the rising use of EMRs, healthcare integration and interoperability is finally maturing.

- Advances in cognitive computing – machine learning and natural language processing – will build on this trend to accelerate the adoption of genomic medicine and its integration with electronic health data into this new genomic health record.

- Adoption of genomic health records can provide a completely different level of decision support to users of genomic data through the application of cognitive computing.

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Greater Adoption of EMR's

➤ The bulk of the hospitals are in levels 3 -5
➤ A new demand for clinical decision support

US EMR Adoption Model℠

Stage	Cumulative Capabilities	2013 Q4	2014 Q1
Stage 7	Complete EMR, CCD transactions to share data; Data warehousing; Data continuity with ED, ambulatory, OP	2.9%	3.1%
Stage 6	Physician documentation (structured templates), full CDSS (variance & compliance), full R-PACS	12.5%	13.3%
Stage 5	Closed loop medication administration	22.0%	24.2%
Stage 4	CPOE, Clinical Decision Support (clinical protocols)	15.5%	15.7%
Stage 3	Nursing/clinical documentation (flow sheets), CDSS (error checking), PACS available outside Radiology	30.3%	27.7%
Stage 2	CDR, Controlled Medical Vocabulary, CDS, may have Document Imaging, HIE capable	7.6%	7.2%
Stage 1	Ancillary – Lab, Rad, Pharmacy – All Installed	3.3%	3.2%
Stage 0	All Three Ancillaries Not Installed	5.8%	5.6%

37.5%   53.2%

Source: HIMSS Analytics® EMR Adoption Model℠; 2013 N = 5458; 2014 N = 5445.

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## Cognitive systems expand the problems we can address
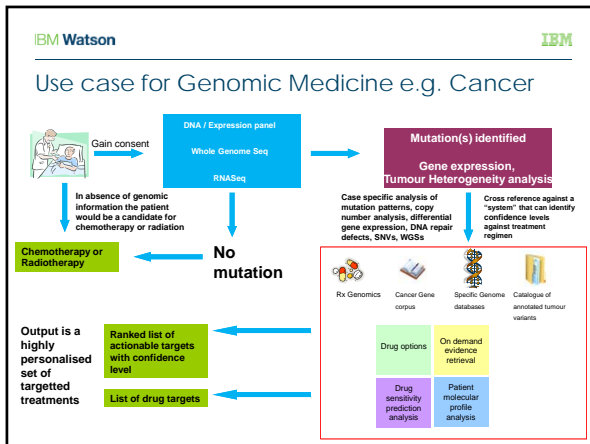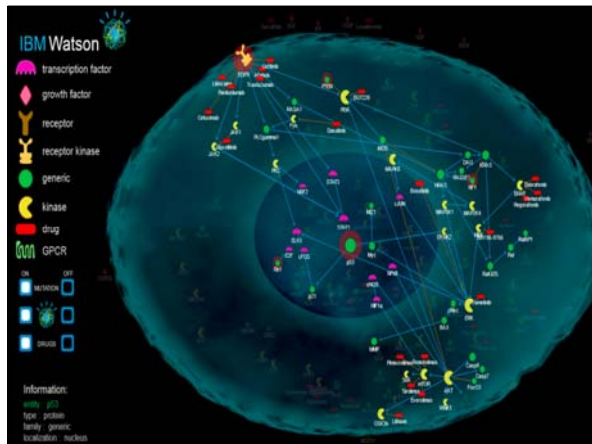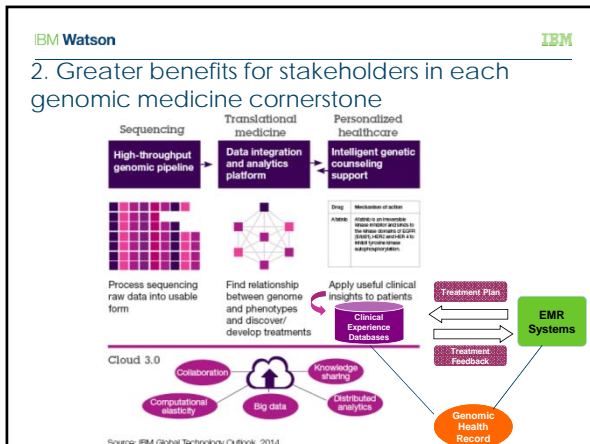

Programmatic Systems
- Leverage traditional data sources
- Follow pre-defined rules (programs)
- Provide the same output to all users

Cognitive Systems
- Are taught, not programmed.
- Learn and improve based on experience
- Interpret sensory and non-traditional data
- Relate to each of us as individuals
- Allow us to expand and scale our own thinking
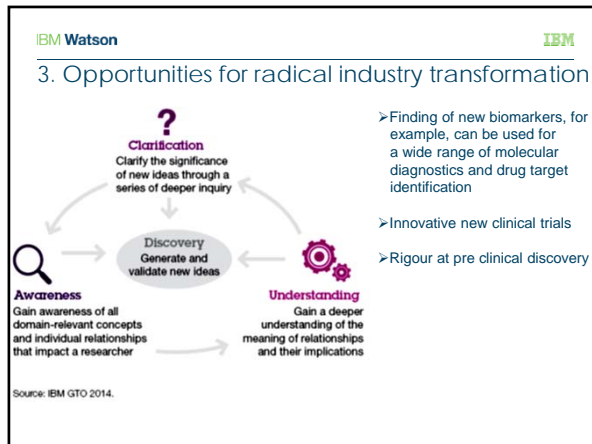
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Help your healthcare or life sciences organization develop its genomic medicine strategies

- How have you incorporated genomic medicine into your enterprise vision and strategy at the clinical, research and IT levels?

- Which types of capabilities, skills and personnel will your organization need so you can use genomic medicine? Which do you already possess and which do you currently lack? How will you develop a plan to fill any identified gaps?

- Understand the requirements for your organization to leverage advances that enable patient care to be of higher quality, more accurate and safer.

- Develop competencies to comply with evolving regulatory standards, privacy and security requirements (for example, HIPAA) will be crucial in an increasingly personalized world where data breaches can be very damaging.

- What is your approach to deciding who you should partner with to build complementary capabilities and skills in genomic medicine?

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What we don't envision happening…

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Conclusions

- A **new genomic health record will become a reality** as genomic information gets combined with relevant data extracted from the traditional EMR.

- Rapid, precision **oncology decision support** is expected to expand on a larger scale

- The capabilities and technologies associated with **cognitive computing** are critical to the ongoing genomic medicine evolution

- To benefit from the far-reaching industry transformation that has begun, forward-thinking healthcare leaders can:
  – verify that genomic medicine is part of their enterprise vision and strategy;
  – assess and plan to fill existing and future skill gaps;
  – look closely at how and when partnering will help their organizations succeed in meeting stakeholder needs.

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"Thank you!"

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New Thought Leadership report

Download at:
www.ibm.com/us/en

The evolving promise of genomic medicine
How advanced technologies are transforming healthcare and life sciences

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