Justin Leighton[1], Ephrem Chin[1], Yang Wang[1,3], Shruti Sureshkumar[2], Dhruv Bhatia[2], Alice Tanner[1], Christin Collins[1], Babi Nallamilli[1,2], Akanchha Kesari[1,2], and Madhuri Hegde[1,2,3]

1. PerkinElmer Genomics Inc., Pittsburgh, USA 2. PerkinElmer Health Sciences Pvt.Ltd., Chennai, India,
3. Suzhou PerkinElmer Medical Laboratory Co.,Ltd.

The era of Genomic Medicine is upon us as we charge towards the $1000 clinical genome. As we look towards the future of personalized medicine, it is helpful and interesting to look at how genomics is being leveraged in countries and markets beyond the US. One way to accomplish this is by looking at lower and upper middle-income economies such as India and China as compared to the US. In the US, individuals have access to a wide range of testing options from more recreational SNP-based testing to full clinical genome sequencing. Testing in the US is rapidly moving away from a single gene/gene panels towards whole exome and genome sequencing due largely to our changed understanding of disease pathology including genetic heterogeneity and pleiotropy. There has also been a rapid expansion of genetic testing options that have become available in China and India over the last four years. However, the areas of testing expansion and breadth of available testing options have been somewhat different than in the US. Testing growth in China has focused primarily on sequencing-based assays and microarrays, while the testing expansion in India has concentrated on more low-cost and mature technologies such as karyotyping and targeted mutation testing via Sanger sequencing largely due to socioeconomic pressures. However, as the cost of sequencing and new genomic technologies continue to decrease, the growth in availability and utilization of new testing technologies in these markets continue to evolve rapidly. Payment methods are another important metric to asses when evaluating genetic testing markets. In the US, payment methods have evolved over time, moving from a self-pay model to more of commercial insurance model as insurers began offering coverage for medically necessary tests. Meanwhile, elective genetic tests continue to utilize a self-pay model, and are rapidly increasing in popularity as individuals view these tests as a way to be proactive about their health. Contrarily, strict self-pay models remain the norm in China and India for all types of genetic testing. This singular payment model has led to immense downward pricing pressures in these markets to keep testing as accessible as possible since the majority of the population in both China and India would not be able to afford the current costs of clinical genetic testing in the US. In China, immense competition between various genetic testing providers has driven down the cost of testing to a point that now affords many Chinese patients access cutting edge testing. Similar to China, genetic testing in India is also facing immense downward pricing pressures due to a strict self-pay reimbursement model and lower income levels in several market segments. However, unlike in China, pricing pressures have served to reduce access to the newest technologies instead relying on more mature and lower-cost testing methodologies. While cutting edge assays utilizing next-generation sequencing are available in India, many of the tests remain unaffordable to patients due to the continued higher costs. Last but not the least is quality of the assays and variant data interpretation available in the market places. Though many new freely available databases such as GnomAD and ClinVar have facilitated interpretation, large proportions of variants identified in these populations are classified as variant of uncertain significance, or in many cases misinterpreted due to lack of large sequencing datasets from these regions. In many cases, it is impossible to follow up on variants due to local cultural complexities despite family structures spanning generations. Although it is clear that the use of genomics and genetic testing has both evolved and accelerated both in the United States and around the world, the breadth availability of genomic technologies varies greatly, and has been shaped by a number of factors including costs and acceptance of genetic testing into mainstream healthcare. We will present data comparing the trends in the number of genetic tests ordered, pricing pressures, current market trends and quality of data interpretation in these three markets.

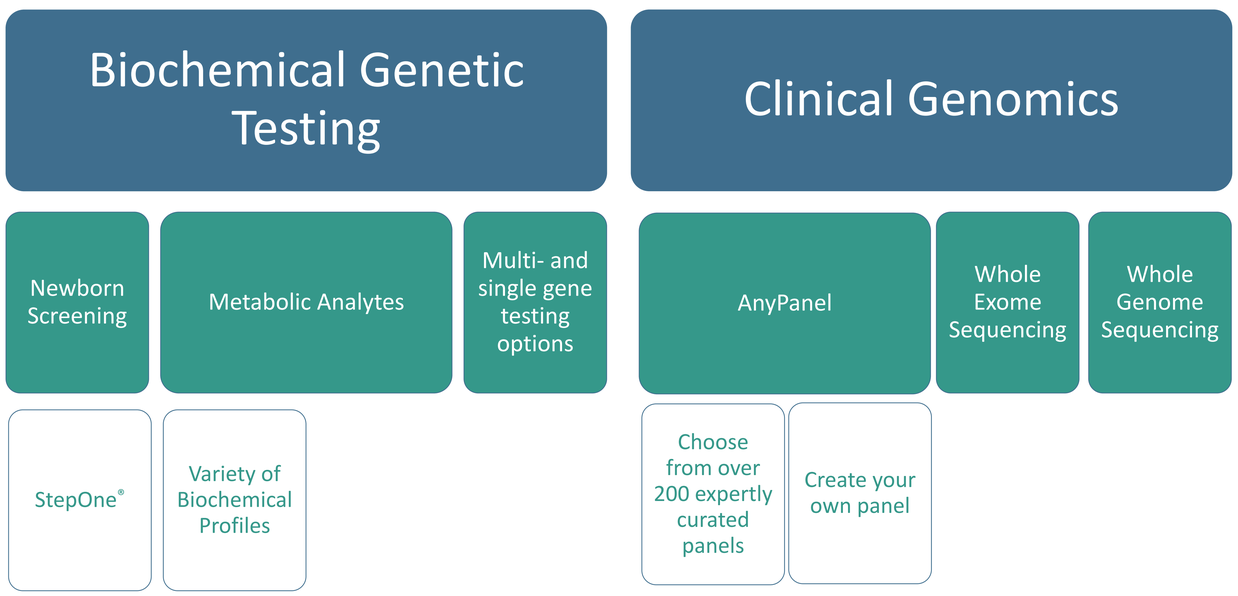
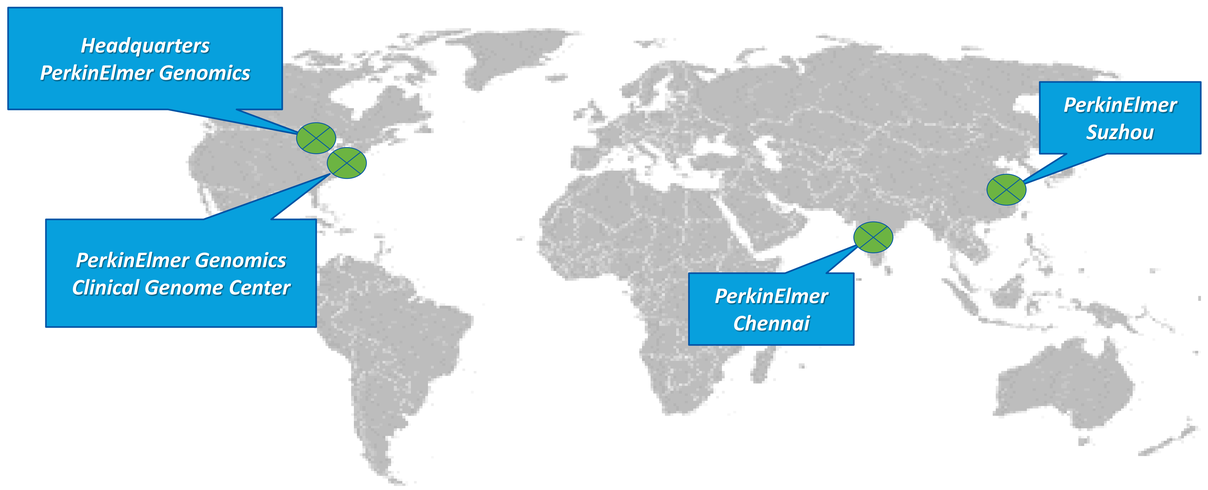



Figure 1: Global Test Offering at PerkinElmer Genomics

| Payment types | | | |
|---|---|---|---|
| | USA | India | China |
| Insurance | Very common | Just started coverage | No coverage |
| Institution | In use decreasing | No | Uncommon |
| Self Pay | Uncommon | Very common | Very common |

Table 1: Payment types in China, India and USA




**Pittsburgh, PA**
- Global leader in Newborn Screening utilizing DBS card assays
- Serving: Global

**Branford, CT**
- Next generation Sequencing Clinical Genome Center
- Serving: Global

**Chennai (India)**
- Prenatal & newborn Screening
- Cytogenetics (FISH, Karyotyping)
- Molecular Diagnostics (BoBs)
- Serving: India, Middle East & Asia

**Suzhou (China)**
- Molecular Cytogenetics
- Molecular Diagnostics (NGS)
- Newborn Screening
- Serving China

Figure 2: PerkinElmer Genomics lab locations market served

| Usage | Biochem | CMA | WGS | WES | Any Panel | Single Gene | Clinical Expertise | Variant reporting |
|---|---|---|---|---|---|---|---|---|
| High ↑ | | | | | | | Seq 1st due to clinical overlap & disease heterogeneity | Focus on clinically relevant variants but ask about VOUS rates |
| | | | | | | | Test driven by clinical expertise. Disease get named by HCP before DNA testing. Many times pathogenic variants not found | Wants all variants |
| ↓ Low | | | | | | | Test driven by clinical expertise. Disease get named by HCP before DNA testing. Many times pathogenic variants not found | Do not want >2-3 variants |

Table 2: Relative demand of tests in China, India and USA

| India Experience – Regional differences | |
|---|---|
| North region | ~50% of testing request are for exomes. Single gene testing request is low. |
| South region | ~30% of testing are for exomes. Majority of tests are AnyPanels and single gene testing. |
| West region | Majority of testing request >75% are for AnyPanel and single gene testing. |

Table 3: Regional difference in test orders. A focus on India.