



**Early diagnostics:  
shaping healthcare and  
society through new  
technologies**





## Executive summary

New healthcare technologies are constantly developed at a rapid pace and allow for the detection and diagnosis of diseases at an earlier stage. This trend, also known as early diagnostics, is expected to lead to significant change within healthcare systems as the way that diseases are found and treated are set to be reshaped. It has the potential to improve the quality of care, lower costs and provide treatment at an earlier time.

There is a strong push on a global level to identify diseases and detect them at earlier rates to optimise the potential for healthy living. This trend is observed both in patients, with an increasing demand for healthy living and self-care, and in governments, where a growing focus on prevention can be observed.

Application of early diagnostics can be divided into three types of analyses: increased data (quantity), improved analysis (quality) and a new world (both quantity and quality). Within increased data, examples are the introduction of digital biomarkers. For improved analysis, the main changes could be found in the application of algorithms and Artificial Intelligence (AI). For a new world, examples are full genome sequencing and the introduction of new (digital) devices.

EIT Health supports a number of solutions in early diagnostics via its innovation projects, business creation programmes for entrepreneurs, and its educational

programmes for learners. The focus is on technology that enables prevention and monitoring so that diseases may be detected at an earlier stage. EIT Health facilitates the use of data to improve the quality of measurement using new algorithms and AI.

Early diagnostics shows a promising future if potential pitfalls could be avoided. It is important to strike a balance in cost effectiveness and to prevent potential overuse or misuse of diagnostics testing. Besides, it is important to take into account the potential economics and emotional costs of false positives.

Developments in early diagnostics go hand in hand with several other global health trends. Early diagnostics will allow healthcare to centre the delivery of care around the patient and is therefore expected to see a rise in point-of-care testing and a potential decentralisation of healthcare. In addition, a growing demand for personalised medicine could be strengthened by the effects of early diagnostics.

# 1.

## The emergence of early diagnostics

### Benefits of early diagnostics

- Reducing downstream treatment costs for payers and society
- Lowering hospitalisation rates and pressure on hospitals and clinicians
- Prescribing the right treatment for a patient, avoiding unnecessary intervention
- Initiating treatments earlier,<sup>6</sup> enabling minimally invasive procedures that reduce operating-room time, length of stay and rehabilitation.

Around 75% of clinical decisions are based on medical diagnostic tests,<sup>1</sup> with the goal of identifying a disease or confirming its presence.<sup>2</sup> There is a large range of different diagnostic tests, such as radiology (techniques used to create visual representations of the body for clinical analysis and treatment purposes),<sup>3</sup> in vitro (outside the body e.g. blood, biopsies) and in vivo tests (inside the body). Today, over 4,000 different diagnostic tests are available.<sup>4</sup> Early diagnostics is the result of applying new and advancing technologies to detect diseases at an earlier time than current common practice.<sup>5</sup>

Early diagnostics has gained increasing attention within the healthcare space. It is already altering healthcare by improving quality of care, delivering more timely treatment, and lowering costs, and the outcomes are expected to grow in the future. Yet there are also potential pitfalls to be considered. In this publication we will outline the dynamics of early diagnostics and analyse the implications of this trend on the healthcare sector.

For EIT Health,<sup>6</sup> early diagnostics presents a clear opportunity to enable longer, healthier lives for Europeans. Gains to be seen from the introduction and adoption of technology in this space will serve to aid healthcare services with improved tools that can speed up processes, drive efficiency, and improve patient care and therefore quality of life.



## 2.

# Trends that shape early diagnostics

New technologies are constantly developed at a rapid pace and allow the detection and diagnosis of diseases at an earlier stage, moving diagnosis from disease state towards healthy living.

In addition, there is a strong push on a global level towards the adoption of healthier lifestyles amongst patients and citizens to improve quality of life and reduce burden on health systems. This trend is observed on an individual level; in patients with an increasing demand for healthy consumption and self-care, and on a governmental level, where there is a push for a stronger focus on prevention.

### Detecting diseases earlier

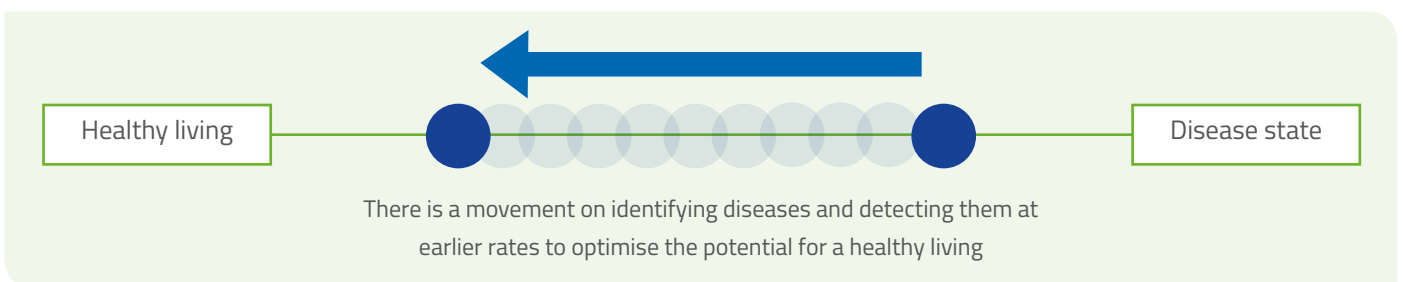
Digital tools can support by filtering, sorting and organising the large amounts of information being collected. They are able to provide clinicians with the ability to improve and speed up their diagnostic capabilities by managing information flow. This allows the identification of a patient's health state more quickly and leads to an earlier diagnosis.<sup>7</sup>

Biomarkers are often used to detect and diagnose diseases. They designate any indicator present in the body or excreted by the body as a biological response to a disease. A biomarker can therefore make it possible to identify the rapid or early detection of a disease, even before the first symptoms appear.<sup>8</sup> The use of biomarkers in research and diagnostics is expected to enable more precise, predictive and preventive clinical care.<sup>9</sup>

### Strong focus on healthy living

The introduction of new technologies and the growth of digital in healthcare have enabled a continuous monitoring of indicators and the collection of valuable insights about an individuals' health. Digital biomarkers, for example, provide these insights through digital tools that collect and / or track data, e.g. on exercise, sleep routines, and blood pressure. This allows individuals to monitor their own health and proactively focus on a healthier way of living, rather than reactively responding to diseases once they occur.

These developments, altogether, show a holistic approach to diagnostics that is focussed around the individual's health. Both earlier detection of diseases and a healthier way of living are in line with several global health trends in society, including prevention and healthy ageing.



# 3.

## Application of early diagnostics in practice

The (earlier) detection and diagnosis of diseases is the result of new technologies that generate new and improved ways to measure indicators of the human body. The application of this is two fold: on one hand, there is an increased capability to measure larger amounts and different types of indicators. On the other hand, new advancements allow for improved and more reliable ways of measuring these indicators.

### Using new data

With increased data, a previously known type of technique is used to either measure a new indicator or a larger amount of indicators. This could lead to more accurate outcomes and lower error margins. It could also provide earlier diagnosis for a disease that was previously diagnosed in a different way using more challenging or time consuming tests.

### New biomarkers

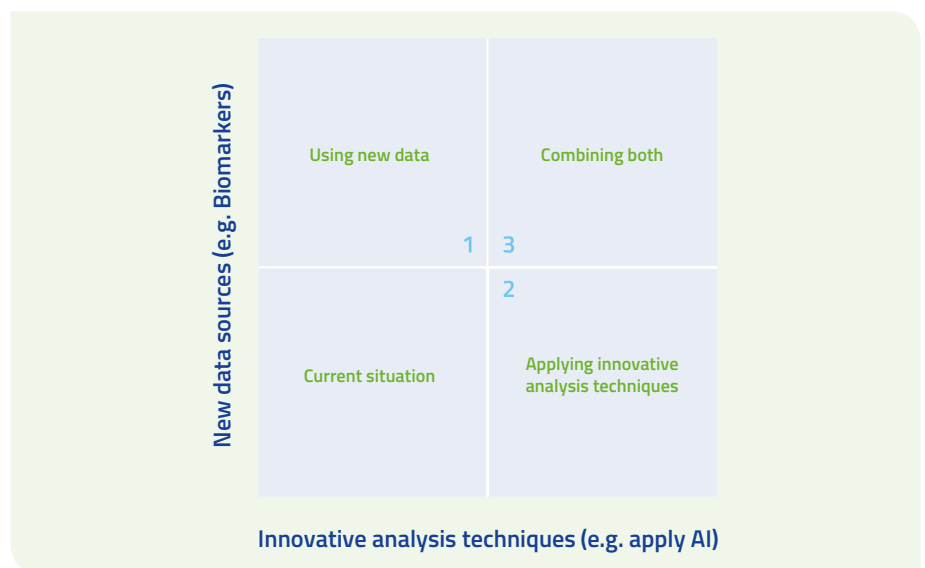
Biomarkers have been used as diagnostic testing for decades. The amount of indicators that one can test for has rapidly increased. Recent examples are biomarkers that can be detected using blood testing that measure an increased amount of indicators to test for different types of cancer cells, including lung- and breast cancer.<sup>10</sup>

### Digital biomarkers

In the healthy-living space, numerous currently existing applications are starting to gain new features that are able to measure additional functions in the human body. Examples are Apple Watch's fall detection and additional features added to existing fitness trackers and digital health systems such as Samsung Health.

For the application of diagnostics we are therefore distinguishing three different types of analyses:

1. Using new data
2. Applying innovative analysis
3. Combining both



## Applying innovative analysis

Improved analysis is characterised by a new type of measurement that improves current diagnostic techniques without measuring new indicators of the human body. These improved techniques could result in higher detection rates, earlier detection or lower error margins for diseases.

There are a few examples of new advancements that improve current diagnostic testing:

### Algorithms

In healthcare, algorithms are used to improve, speed up and standardise the decisions made. They are often used in the process of diagnostic testing and have the potential to contribute to an earlier diagnosis. Aside from in-depth diagnostic testing, many mobile health solutions adopt algorithms to look for patterns and predict future outcomes. The insights that follow are provided to the patient and / or healthcare provider through a feedback loop. This empowers patients to make well informed decisions and introduce small adjustments to their lives that could improve their health.

### Artificial Intelligence (AI)

AI refers to the simulation of human intelligence in machines that are programmed to think like humans and mimic their actions.<sup>11</sup> The key difference between algorithms and AI is the computer's ability to change, adapt and grow based on new data, which is therefore described as "intelligence".<sup>12</sup> AI tools have the ability to reduce human errors and can be used for different applications, such as informing diagnosis and assisting in treatments.<sup>13</sup> An important way in which AI is used is as visual pattern recognition software, where preliminary results show similar accuracy to clinicians.<sup>14</sup>

### Examples

#### Improved analysis

Google DeepMind Health: an AI technology platform that can read 3D retinal OCT (Optical Coherence Tomography) scans, diagnose and prioritise patients most in need of urgent care.<sup>15</sup>

#### Increased data

Cancer detecting blood test: A blood test has been developed to detect over 50 different types of cancer before symptoms appear. These cancer types include pancreatic cancer, usually diagnosed at a very late stage. The approach used is feeding a system with data on methylation patterns in DNA. The machine learning system then sorts the samples into groups, which reflect different types of cancer.<sup>16</sup>

## Combining both

When we apply an innovative analysis technique to a new data set, we can uncover insights that we have never had access to before. These types of analyses can bring completely new insights to healthcare. When applied in Early Diagnostics, these are often focused on prevention by monitoring health functions of the human body or foreseeing potential diseases in the future by understanding individual genes. Some examples of newly developed analyses are DNA testing and the introduction of new (digital) devices.

### A new world

Withings: a physical sleep tracking mat connected to your smartphone. It performs sleep cycle analyses and introduces sleep coaching programmes.<sup>17</sup>

### Improved analysis

My Sugr: a diabetes solution platform combining physical testing with a digital application. A personalised patient interface with data analysis and recommendations for the patient to improve and monitor the disease.<sup>18</sup>

## DNA testing

Testing DNA has become one of the most obvious examples of newly developed analysis, and is rapidly growing in popularity as well as in the sectors within which it can be applied (e.g. health, ancestry, beauty).<sup>19</sup> Since the 2017 FDA approval of 23andMe's direct-to-consumer DNA test, access to DNA testing has become possible for the consumer.<sup>20</sup> This has led to a large growth in the amount of people having their DNA tested. However, due to ethical and data privacy concerns the growth has largely disappeared from this market. Nonetheless, this has still opened up a new way of Early Diagnosis for consumers

## The introduction of new (digital) devices

In recent years, numerous new digital devices have been introduced in the healthcare and consumer health space. These devices often use new techniques and sensors to measure new indicators of the human body.

## EIT Health examples

### Improved analysis

OncoWatch: aims to provide faster, better and cheaper image analysis of biopsies, using Big Data, AI and cloud-based technologies.<sup>21</sup> It also brings new analytical tools for precision medicine. This improves diagnostics of prostate cancer, reduces time to treatment and lowers healthcare costs.

### A new world

Alzheimer's Disease Prediction Service – ADPS: a smartphone test to predict whether someone is likely to develop Alzheimer's in the next six years, using data that has shown 87-94% accuracy.<sup>22</sup> It is one of the first pre-symptomatic digital biomarkers that will be able to predict the risk of Alzheimer's.

# 4.

## Structuring the developments of early diagnostics

Based on the trends and application of early diagnostics in practice, we have developed a matrix in which early diagnostics projects could be positioned, depending on their trend (healthy living or disease state) and type of analysis (increased quantity, improved quality of measuring or both).

The matrix below shows examples of projects in the field, both EIT Health projects and non-EIT Health projects that can be positioned on the axes of the matrix.



Figure 1: Matrix of projects in the field

<p><b>Healthy living</b></p>	<ul style="list-style-type: none"> <li>▪ Apple Watch fall detection</li> </ul>	<ul style="list-style-type: none"> <li>▪ My Sugr</li> </ul>	<ul style="list-style-type: none"> <li>▪ Family Tree DNA</li> <li>▪ WHS@work (EIT Health)</li> </ul>
<p><b>Disease state</b></p>	<ul style="list-style-type: none"> <li>▪ Diagnosing cancer through blood tests</li> <li>▪ DeTecT2D (EIT Health)</li> </ul>	<ul style="list-style-type: none"> <li>▪ Google DeepMind: 3D retinal OCT scans</li> <li>▪ LUCINDA (EIT Health)</li> <li>▪ OncoWatch (EIT Health)</li> <li>▪ Smart Drain (EIT Health)</li> </ul>	<ul style="list-style-type: none"> <li>▪ ADAPT (EIT Health)</li> <li>▪ ADPS (EIT Health)</li> </ul>
<p><b>Using new data</b></p>		<p><b>Applying innovative analysis</b></p>	
<p>New tests / More biomarkers</p>		<p>AI / Algorithms</p>	
<p><b>Combining both</b></p>			<p>Digital devices / self testing</p>



# 5.

## The future of early diagnostics

### Upcoming trends in early diagnostics

The developments in early diagnostics are expected to go hand in hand with several global health trends observed. Early diagnostics allows healthcare to adjust care towards and around the patient to an increasing extent. This follows the growing demands of patients to personalise their medication and bring care to the place where they'd like to receive it. In addition, early diagnostics leads the way for further focus on healthy living and prevention. It is to be expected that this field will become even more prominent in the future.



#### Rise in point-of-care testing and decentralisation of healthcare

One of the major growths in the diagnostic market is point-of-care diagnostic testing. This entails testing at or near the point of care for the patient. This results in a faster diagnosis and a more rapid treatment.<sup>23</sup> Together with attempting to cater medical facilities remotely and curb costs, there is an upcoming trend that shows a decentralisation of healthcare, focussing healthcare around the patient.<sup>24</sup>



#### Growing demand for personalised medicine

The shift towards personalised medicine is likely to increase in-vitro diagnostic testing and service costs. Further innovation and demographic pressures will likely lead to demand accelerating over the coming years.<sup>25</sup>



#### Further focus on prevention

The last few years have marked a change in governmental policies to call for preventative healthcare through screenings, a healthy lifestyle and a higher awareness of health among citizens. Technological advancements allow for this trend to continue to grow rapidly with patients monitoring their own vital health functions through wearables and other technologies.

## The potential pitfalls of early diagnostics

Diagnostic testing is incredibly important in healthcare. A vast majority of the decision making is dependent on diagnostic testing. The development of early diagnostics has the potential to give diagnostic testing an even more important role in healthcare. However, there are some challenges that come with the developments in early diagnostics that must be kept in mind as the movement grows.



### Cost effectiveness

The advances in diagnostic testing have led to improved techniques, an expanding number of available tests and an increased demand for them.<sup>26</sup> However, the cost effectiveness of early diagnostics can be unclear at times. Although diagnosing diseases earlier – or even before they occur – could prevent unnecessary intervention, treatment and hospitalisations, the costs of diagnostic testing are high and rapidly rising.



### Misuse of diagnostic tests

Some diagnostic tests are misused or overused, with waste from diagnostic imaging alone estimated at more than US\$25 billion in the United States alone.<sup>27</sup> Generally speaking, it is challenging to determine whether diagnostic tests are being overused or underused and at what times ‘more’ diagnoses is not equal to ‘better’ diagnoses.<sup>28</sup>



### The economic and emotional costs of false positives

With a stronger focus on prevention, governments worldwide have introduced various screening programmes for their citizens in higher risk categories. Examples are screenings for cervical cancer, colon cancer and breast cancer. Although these screening programmes can be very effective in detecting diseases at an earlier stage, their psychological and economic impact should not be underestimated. Each screening programme is different and a careful assessment needs to be made in order to understand whether the benefits outweigh the economic and emotional costs.

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