

## Healthcare industry BW

# Disease prevention through better diagnostics

**“Classical treatment approaches are not the only way to ensure good health, rehabilitation and care. In fact, it is envisaged that more effective prevention will contribute to halting the development of diseases and maintaining good health. Prevention is designed to prevent health risks and diseases, make them less likely and delay their onset.” (Declaration of the German government; Official Records of Parliament 17/845, 26th February 2010)**

“Prevention is better than treatment.” This proverbial saying is frequently repeated by representatives of medical science, practical medicine, government and funding authorities. In many cases prevention relates to advice such as eating a healthy diet, stopping smoking, doing physical exercise, etc. – sound advice that is little more than standard common sense. Reliable diagnostic measures are what are actually needed in order to establish specific prevention procedures to reduce the risk of developing severe diseases.

Due to the broad range of diagnostic methods for disease prevention used in Baden-Württemberg, it is not possible to give a complete overview of what is currently possible in this dossier. However, it is possible to give some insights into the topic and present state-of-the-art developments and notable projects, highlight future potential and perspectives and discuss problems. Diagnostic methods are used in a range of areas comprising conventional laboratory medicine, histological and cytological screening, high-throughput procedures involving molecular biology and genetic engineering methods, sequencing as well as high-tech medicine. Modern preventive medicine is characterised by the combination of such a broad range of different methods.

## Prevention and early diagnosis of cancer

The discovery of the intestinal bacterium *Helicobacter pylori* by Barry Marshall and Robin Warren is a convincing example of the importance of diagnostics in the prevention of disease. The two researchers, who were awarded the Nobel Prize in Physiology or Medicine in 2005, showed in experiments that they carried out on themselves that *H. pylori* was the cause of both gastritis and gastric ulcer. In addition, they showed that the diseases could be treated with antibiotics and hence prevented. In the time since the experiments were carried out, the bacterium has been identified as the major cause of gastric cancer. Patients diagnosed with *H. pylori* infections are treated with specific medicines that enable the permanent eradication of the pathogen.

Although coloscopy, the endoscopic examination of the colon that provides a visual diagnosis of polyps and ulceration, is not painful, it does usually cause some discomfort. Very few people in

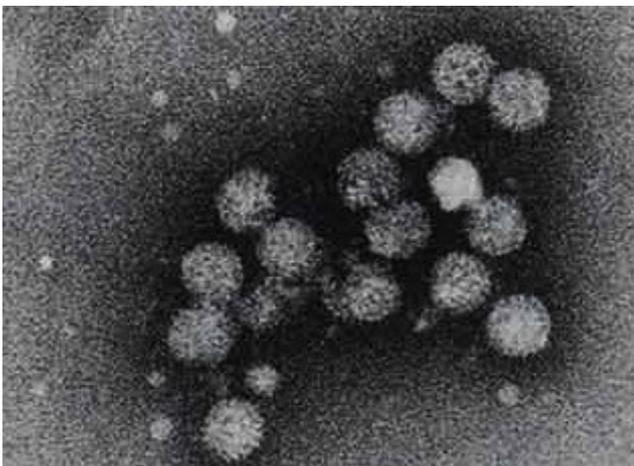


Coloscopy  
© German Cancer Society

Germany ever undergo a coloscopy, despite the fact that colon cancer is the second most frequent cause of cancer deaths amongst Germans and despite the fact that the compulsory health insurance pays for the endoscopic examination. Polyps discovered in the colon mucosa are regarded as potential precancerous stages and are removed. If scientists were able to agree amongst themselves that these polyps are in fact precancerous stages, the procedure could be called cancer prevention rather than just simply the early detection of cancer. The boundaries between the two often overlap, and as important as the differentiation is for oncologists, it is a question of mere semantics for patients, as they are mainly interested in measures that help to prevent this malignant, potentially terminal, cancer.

The most successful cancer screening test of all times, the Pap test, detects the abnormal growth of cervical cells at an early stage so that treatment can be started before the cells become cancerous and invasive. Since the simple Pap test is not 100% sensitive and can miss cervical abnormalities, the company mtm laboratories has since developed and launched molecular diagnostics tests that enable the reliable detection of precancerous stages of cervical cancer.

Breast cancer diagnostics has become more effective thanks to state-of-the-art ultrasound methods. Numerous studies have shown that ultrasound is able to detect breast cancer much earlier than inspection and palpation or X-ray examinations. Novel technological developments such as three-dimensional sonography and colour duplex sonography, which do not expose the patient to irradiation, considerably extend the capabilities of ultrasound diagnostics, especially in the field of prenatal diagnostics.



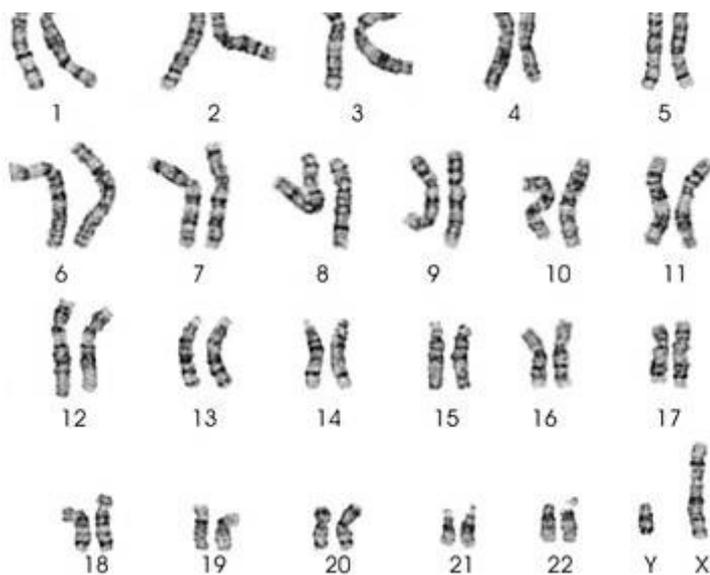
Hepatitis C viruses. Electron microscope image, negative staining.  
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The early detection of malignant diseases is of course an important objective in the endeavour to prevent health problems. In the case of cancer, the early detection of malignant cells, i.e. before the tumour spreads in the body through metastases, might be decisive in determining whether patients live or die. The lack of reliable recognition features is a major reason for the high mortality rates in some cancers, such as lung or liver cancer. The goal of the LUSI (lung cancer screening intervention study), which was started by the German Cancer Research Center in 2007, is to improve the diagnosis of lung cancer through the use of multilayer computed tomography.

Liver cell cancer (hepatocellular carcinoma) is often fatal. However, the only development of more effective diagnosis and prevention measures at present is for cancer types that are associated with hepatitis C virus (HCV) infections. Reliable diagnostic tests, and medication given in the early phase of infection are available against HCV. HCV is often asymptomatic; however, once chronic infection is established it can progress to scarring of the liver (cirrhosis) and eventually trigger the development of liver cancer.

## Genetic tests for the identification of risk factors

In order to enable the successful treatment of HCV infections, it is necessary to genotype the virus, as responses to treatment can vary by genotype. Genetic testing has become indispensable for the diagnosis of some diseases, in particular in oncology. Personalised medicine, which seems set to become the medicine of the future, heavily relies on gene or genome testing. Biotech companies that specialise in sequencing and array technologies such as the Tübingen-based company CeGaT GmbH offer genetic tests for the diagnosis of complex diseases. While such genetic testing services are strictly regulated by the Gene Diagnostics Law in Germany, numerous companies offering direct-consumer genetic testing have emerged in the USA and Great Britain. Many of these tests are misleading and their marketing borders on the fraudulent and deceptive. The USA and Great Britain are also now putting regulatory and control measures in place to combat the misuse of such tests.



Human karyotype with 46 chromosomes  
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In many cases, in particular for the diagnosis of common diseases such as cardiovascular diseases,

diabetes, metabolic diseases and osteoporosis, traditional diagnostic methods are much better than the new gene tests. This is particularly true in cases where different parameters, for example laboratory blood values, molecular markers, imaging methods, etc. are critically assessed and included in the diagnosis. University hospitals, private institutes and companies such as IPM in Freiburg or BioLabs GmbH in Ulm all apply such approaches. Under the coordination of the German Cancer Research Center and Helmholtz Zentrum München, the largest ever medical study has been initiated in Germany. The study, known as the Helmholtz Cohort study, is investigating a broad range of such parameters over long periods of time in order to identify risk factors for common diseases and new ways to prevent them.

Dr. Ernst Jarasch - 30.08.2010  
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## Dossier

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