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USING MACHINE LEARNING ALGORITHMS TO ANALYZE GENETIC DATA FOR DISEASE DIAGNOSIS

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Today artificial intelligence (AI) plays an important role in many aspects, especially in genetics and reproductive science, including analysis of genetic data, prediction of genetic diseases, development of new methods of diagnosis and treatment. The integration of AI in medicine holds great promise for improving healthcare outcomes. With the ability to process vast amounts of data and recognize complex patterns, AI has the potential to enhance medical decision-making, accelerate diagnoses, optimize treatment protocols, and reduce medical errors. By augmenting the capabilities of healthcare professionals, AI can contribute to more precise and personalized healthcare delivery, leading to improved patient outcomes and a more efficient healthcare system.

The purpose of the article is to study the importance of using AI to prevent genetic diseases and increase the world demographic level with the help of biological and mathematical knowledge.

Neural networks are increasingly seeping into our lives. AI systems can help researches in interpreting images, identify early signs of diseases, and recommend appropriate treatment options. Furthermore, AI-powered predictive analytics can aid in forecasting disease progression, identifying high-risk patients, and optimizing resource allocation in healthcare settings. One of the most terrible diseases of modern civilization is cancer. Cancer is a disease caused by uncontrolled division. It is caused by mutations - changes in DNA that directly affect genes, which cause uncontrollable growth of cells [1].

The largest number of men die from cancer of the lungs, stomach, liver, colon and rectum, and esophagus. Most women around the world die from the following five types of cancer: breast, lung, stomach, rectum and colon, and cervical cancer. However, early diagnosis of cancer gives a chance for effective treatment, experts emphasize [2].

The InnerEye project at Addenbrooke's Hospital in Cambridge is a good example of using AI to diagnose cancer. Thanks to Kinect sensors and a smart image recognition system, InnerEye accelerates work with images of internal organs and tissues of patients. Instead of the many hours it takes doctors to process a series of hundreds of CT scans, InnerEye does it automatically in no more than 30 seconds. This significantly reduces the time for identifying malignant tumors in a person and

their subsequent treatment. In addition, InnerEye will help monitor the treatment process and adapt it according to the patient's reaction to the procedures. As a result, chemotherapy can become more targeted and effective, with fewer side effects for patients [3].

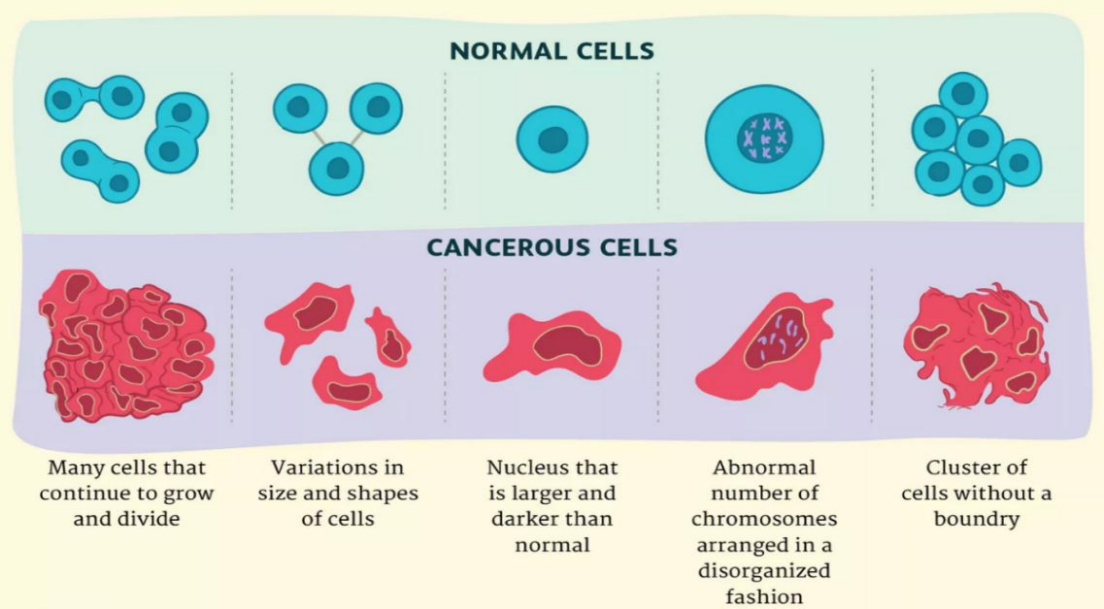

Fig. 1. **Illustrative example of the difference between normal cells and cancerous cells**

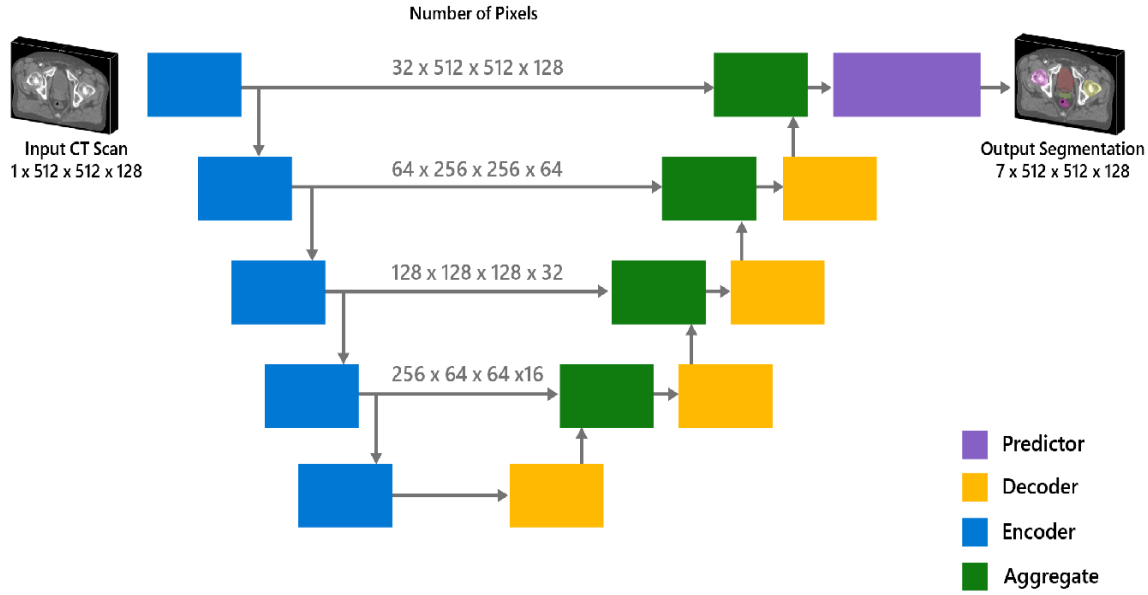

Fig. 2. **General logic of Inner eye project**

In Fig. 3 the algorithm predictions are shown in darker colors in comparison to the ground-truth OAR annotations. The corresponding expert reader annotations are shown in lighter colors. From left to right (columns displaying mid-axial and mid-coronal slices): for comparison purposes the first scan on the left is retrieved from the main dataset and the remaining two are from the external dataset. Here we see differences between the datasets in terms of patient anatomy and through-plane scan resolution [4].

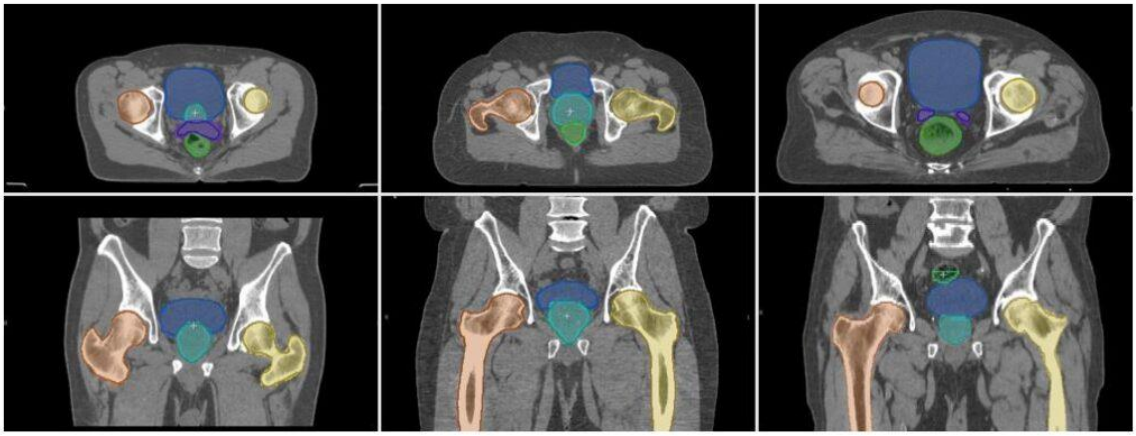

Fig. 3. **Datasets in terms of patient anatomy and through-plane scan resolution**

A group of scientists led by the Institute of Cancer Research in London and the University of Edinburgh has used artificial intelligence to predict the development of cancers and developed a new technique known as a "revolver". The point of this technique: helps to identify patterns of mutation in DNA during cancer and use this information to predict future genetic changes. The team also found an association between certain sequences of recurrent tumor mutations and survival outcomes. This suggests that patterns of DNA mutations can be used as a prognostic indicator to help shape future treatment. During the study, scientists used 768 tumor samples from 178 patients with lung, breast, kidney, and colon cancer. This powerful tool predicts the development of tumors based on certain types of mutations that have so far remained hidden in complex datasets. With the help of this tool, the team hopes to eliminate the main trump card of cancer - its unpredictability [5].

Equally important is the use of artificial intelligence in reproductive science, which helps analyze sperm, eggs, embryos and other factors related to reproduction, helping to identify the most viable elements for better reproductive outcomes. In 2019, a group of American scientists led by M. VerMilia published data on their new AI development for selecting and predicting embryos with the highest implantation potential. The development is called The Life Whisperer [6].

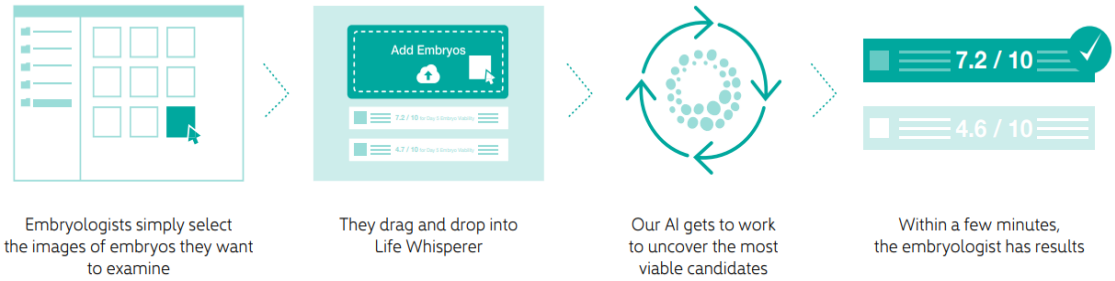

Fig. 4. **How the Life Whisperer supports embryologist to enhance your chances of pregnancy and reduce IVF cycles**

The main work of AI is the analysis of photographs of embryos and the selection of those with the most promising appearance. The algorithm examined numerous embryo images obtained from real IVF cycles, encompassing both successful and unsuccessful cases. This data was retrieved from Presagen's globally connected clinical data platform hosted on the AWS cloud, ensuring secure storage of information collected from clinics worldwide. Critical to a global undertaking, the AWS cloud gave

the opportunity to use this global medical data without crossing any patient data privacy laws. The AWS cloud provides a distributed cloud solution that guarantees a secure, dependable, and easily accessible platform capable of delivering the required computing power for practical implementation. By utilizing a distributed cloud system, data remains securely stored at its original source, while federated learning, a machine learning technique that trains algorithms across multiple decentralized servers, effectively addresses the crucial concerns of data privacy and security [7].

Since 2019, the Ukrainian clinic of reproductive medicine "Nadia" together with the Israeli company Embryonics has taken part in the development of AI for the selection of embryos with the greatest ability for implantation. Currently, the Nadia clinic is conducting a clinical study on the benefits of using AI, so every patient has the opportunity to participate in this study and additionally analyze the development of their embryos with the help of AI. After several days of embryo development in a time-lapse incubator, embryologists have a video recording of the development of each of them. Such videos are uploaded to a program (AI), which further analyzes the video data and gives each embryo a score. The higher the embryo's score, the greater the probability that it will implant in the uterus [6].

The use of AI and sperm analysis was not missed. In 2019, a group of American scientists led by Irene Dimitriadis and Charles Borman published interesting results regarding the use of a portable ejaculate analyzer. A portable analyzer is a smartphone with an application installed on it, a video adapter and a special chip where the ejaculate sample is placed. In this case, the smartphone camera functions as a microscope. With the help of such an analyzer, scientists determined the level of viable spermatozoa in ejaculate samples and compared the results with a similar analysis in the laboratory. If we deviate a little from the purely scientific direction and wonder what additional "goodies" patients can get, we can mention the Spanish software Fenomatch. It is currently the only tool developed specifically for centers of reproductive medicine, the task of which is to select an egg donor that is as close as possible to the patient's facial features [6].

AI has the potential to revolutionize the field of medicine by improving diagnostic accuracy, enhancing treatment planning, and enabling personalized healthcare. Its ability to analyze complex data, identify patterns, and provide insights can empower healthcare professionals and contribute to more efficient and effective healthcare delivery.

Summarizing all the material, we can conclude that the application of AI is extremely important in genetics and reproductive medicine for several reasons. Firstly, it helps to accelerate the analysis of genetic data. Secondly, AI also helps to improve reproductive medicine by analyzing clinical data and genetic information to improve the selection of the most viable sperm, eggs and embryos. Thirdly, it promotes to the rapid prediction of the risks of disease development. Thus, the use of AI in genetics and reproductive medicine provides faster, more accurate and more efficient study of genetic features and improves the quality of medical services in these fields.

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