Life's Blueprint

Within DNA lies a literal map of the human body, including potential diseases and abnormalities. The better doctors get at reading the map, the better these diseases can be treated and possibly avoided.

By Nicolè A. Staab

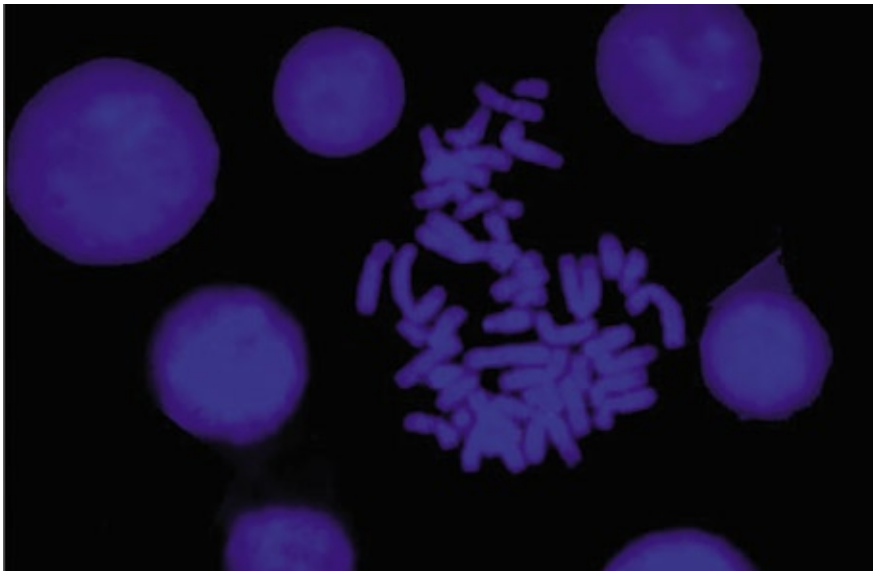
Humans come in all models. We're all unique on the outside, but probe beneath the skin and you'll find more similarities than differences. Our bodies are composed of cells and within each cell is deoxyribonucleic acid, or DNA, the source of heredity and the foundation of genetics. DNA, commonly compared to a blueprint, contains the coded instructions used in the development, replication and functioning of all living organisms.

Centuries before being able to clone Dolly the Sheep, humans recognized that living things inherit traits from their parents, with common-sense observation long applied in the selective cultivation of plants and the breeding of animals. Although it took some time to firm up the details of how traits were actually transferred, now it is clear that it is the genes that carry characteristics from one generation to the next.

The spiral staircase-shaped double helix contains the secrets of how genetic information is stored, transferred and copied. What's vital is that DNA can replicate, or make copies of itself. Each strand of DNA in the double helix can serve as a pattern for duplication. This is crucial when cells divide because each new cell needs to have an exact copy of the DNA present in the old cell.

In many developed countries, the scientific and medical communities are abuzz with talk of genetic engineering and its applications, but the field hasn't been receiving the same amount of attention in Egypt.

"It's not a new field," says Dr. Ezzat Elsobky, medical director and CEO of Mohandiseen's Medical Genetics Center and a graduate of Ain Shams Medical School. Today, he is one of the nation's top clinical geneticists with an international reputation in the field. "My [master's] certificate is from 1976, 30 years ago, but [the field] seems not very attractive to postgraduate students. In the field of genetics, you can count 50 doctors, or if you are very optimistic you can say 100, all over Egypt."



Those rice-shaped chromosomes can reveal almost everything about you. *(Courtesy Dr. Ezzat Elsobky)*

Genetics — the scientific study of heredity, or how particular qualities or traits are transmitted from parents to offspring — isn't just in the spotlight because of cloning; it is the science of the twenty-first century, with applications extending beyond the laboratory to a range of professions. Investigators use forensic tests based on DNA evidence to establish paternity and help convict criminals. Molecular biologists develop crops better suited for specific environments and pharmaceutical companies improve upon medicines.

And, of course, doctors are able to search for hereditary diseases and provide better medical care through genetic testing.

Genetics is gradually easing its way into doctors' offices and mainstream discourse. Elsobky views the success of a recent international conference he hosted in Cairo as evidence of its increasing popularity and importance. Close to 250 doctors and medical practitioners attended the Medical Genetics in Clinical Practice conference held in May.

Each year, says Elsobky, the conference focuses on a different topic of interest and "this time the core was molecular genetics and the role of regular genetics: offering better health services through better understanding of the role of molecular genetics."

Sometimes, mistakes are made during the

copying of DNA. This mistake is called a mutation. A mutation can be harmless, but in many cases it can lead to diseases such as cancer and genetic disorders, which can be passed on to children.

According to Elsobky, genetic disorders are "diseases that are not acquired or caused from infection or trauma," but rather, children inherit the gene from their parents. He adds, "It could be congenital, if you can see the disease at birth immediately — but sometimes, if they have the gene, the disease will develop later on. It's still genetics and still transmitted from the parents."

Familial Mediterranean Fever (FMF) is one such genetic disorder passed on by parents that specifically affects populations living in the Mediterranean region. One in five people in Egypt are a carrier for the disease.

Impact

Genetic diseases represent a major public health problem in the Arab world. Many of the genetic defects seen in this region are nowhere else in the world. Elsobky says that of 1,500,000 births per year, 30-50 cases will have some sort of congenital anomaly, with genetic background to blame for half of these cases.

Among the factors at play here is the high rate of consanguinity (close-family marriages). The fact that people of the Mediterranean region are prone to inherited diseases such as thalassemia and glucose-6-phosphate dehydrogenase has also contributed to this. The lack of public awareness about the early recognition of inherited disease has not helped either.

The custom of consanguineous marriage has its roots in a variety of cultural and historical factors rather than religious ones. Contrary to the belief of many, Islam does not advocate or encourage the practice, as evidenced by the Prophet Muhammad's (PBUH) hadith: "Ightaribu" (Go beyond your relatives). When asked by his companions what he meant by the advice, the Prophet Muhammad (PBUH) replied that close marriages, specifically those between first cousins, should be avoided as they could result in weak offspring.

The geographical concentration of many population groups in small isolated areas and the fact that many families view it as a way to maintain family assets promoted the practice of consanguineous marriages.

It is a custom that Elsobky doesn't think very highly of. "The cause underlying [the high number of genetic disorders] is the very high rate of consanguineous marriages and a very limited level of awareness," he says.

The genetic dangers associated with consanguineous marriage are often overestimated, but scientific results link it to many abnormalities including increased frequencies of recessive disorders, disability, learning disorders, psychiatric morbidity, central nervous system anomalies and cancer. The reason behind this is that the more closely two people are related, the more genes they share. A child from a marriage between first cousins is two-and-a-half times more likely to develop a genetic disorder, since their parents share one-eighth of their genes.

Genetic Testing

Genetic tests use a variety of laboratory techniques to determine if a person has a genetic disorder or is likely to develop a disease. The tests look for abnormalities in the genes and include gene tests (DNA testing) and biochemical tests (protein testing).

Gene testing involves examining DNA — taken from cells in a sample of blood or, occasionally, from other body fluids or tissues.

Genetic testing, says Elsobky, "means that you look at the genetic content of a human cell. Any source where you can find cells, you can do a genetic test. Genetic testing can help in confirming the diagnosis, and also it gives us an idea for the need for treatment — [whether] for life, or only during the attack."

Nowadays, genetic testing is widely available, and more affordable. The most common type is newborn screening. Each year in the United States, four million newborn infants have blood samples tested for genetic disorders and diseases, including Phenylketonuria (PKU). The Medical Genetics Center in Egypt is continually expanding its screening program, recently adding PKU tests to its battery of exams as the risk for this disease increases with consanguineous marriages.

Carrier testing is also very popular, and is used by couples to determine if they carry a recessive allele (specific DNA sequence) for inherited disorders such as Cystic Fibrosis, Sickle-Cell Anemia or Tay-Sachs disease. A person who has only one abnormal copy of a gene for a recessive condition is known as a carrier — carriers won't get the disease, but they can pass on the faulty gene to their children.

Dr. Ezzat Elsobky says social customs have contributed to the prevalence of certain genetic defects. *(Khaled Habib/Egypt Today)*

Prenatal screening is available to parents who are at risk of having children with a chromosomal abnormality or an inherited genetic condition. Often referred to as the Triple Test, this test screens for Down Syndrome, Trisomy 18 and neural tube defects such as Spina Bifida. Although it does not have the same predictive power of amniocentesis (sampling amniotic fluid from the womb) or chorionic villus sampling (tissue sample from outside the sac where the fetus develops) it poses a much lower risk to the fetus.

Much of the current excitement in the field, however, centers on predictive gene tests. These tests can be comforting to individuals in families that are at high risk for a disease or cancer, because some disorders can be identified before any symptoms appear.

Genetic tests help doctors provide better medical care, because if symptoms are present they can confirm a diagnosis. A negative test can provide a sense of relief while a positive test can at least relieve uncertainty and enables the person to take steps to reduce risk and make informed decisions about their future. et