

# DNA holds code to rare 'Ondine's Curse'

Until now, little has been known of what causes rare diseases. However, geneticists tell us that eighty percent of illness is either hereditary or a result of gene mutation during conception. Only 20 percent are of an infectious nature or caused by immune failure

By Lidia Zhelezovskaya

"In Europe, rare diseases are classed as those suffered by just one in 2,000 or more," explains Konstantin Mosse who heads the Laboratory for Cytogenetic, Molecular-Genetic and Morphological Research at the Republican Mother and Child Scientific-Practical Centre. "Even the most widespread genetic diseases are considered rare; many are severe chronic diseases accompanied by life-threatening symptoms. These may be seen from the earliest age but might just as easily become apparent only in later childhood or adulthood."

Four year old Polina, from Belarusian Novogrudok, has perplexed her parents and doctors from the first days of her life. Until recently, she was the only such case in the country, suffering from 'Ondine's Curse' (the unofficial name for sleep apnea). Polina lacks autonomic control of her breathing while sleeping, so stops breathing independently each time she falls unconscious. As a baby, doctors were obliged to use artificial pulmonary ventilation to keep her alive. Today, she can breathe on her own, courtesy of a Swedish clinic which gave her an implanted diaphragm pacemaker. Sadly, no other cure exists for the defective



Konstantin Mosse: 'Genes will reveal their defects in a couple of hours'

gene responsible for her disease. "About 400 people worldwide suffer from 'Ondine's Curse', with 90 percent featuring a new mutation. Others have inherited their illness," says Mr. Mosse.

The Centre's geneticists aim to detect the primary cause of each disease, so that they can determine whether someone is likely to be susceptible. Their equipment allows them locate all possible gene mutations, with special attention given to the most common pathologies (there are about 50 in Belarus). However, specialists are also ready to cope with others. "Often, we detect genetic defects responsible for neurological disease and

metabolic irregularities. However, doctors' prognoses do not always coincide with the real situation," notes Mr. Mosse, adding, "For example, none of those whom we suspected of having Friedrich's Ataxia had gene abnormalities (which lead to this pathology). Geneticists face a huge challenge in diagnosing rare diseases, since it's impossible to identify a neurological pathology purely by molecular means. It takes time and money."

Belarusian geneticists have already detected several hundred gene problems but still have much work to do to decipher the hidden secrets of our DNA. In fact, many people may not even realise that

genes are the cause of their illness. Moreover, there are no more than ten specialists in our country qualified to conduct such research into DNA.

No effective treatment exists to cure the rarest diseases, although some drugs can improve our quality and length of life. Sadly, the most suitable approach — gene therapy — is not yet widely affordable. Some successful attempts have been made but their practical implementation remains out of reach. Mr. Mosse is sure that the future will see each 'rare' patient receiving the chance to regain full health, but who knows when that day may come.

# Display can even endure battle

By Dmitry Toporkov

**Display Design Bureau, from Vitebsk, specialises in producing monitors suitable for operating under toughest conditions**

The Display Design Bureau produces monitors for the military, suitable for use on land, at sea and in the air, failsafe under any conditions. The shock resistant technology remains operational regardless of terrain, extreme temperatures, radio-electronic block-



Display's designers discuss production novelties

ing devices and, even, human clumsiness. Its monitors (which use the latest LCD and plasma panels) and portable computers are well known worldwide.

Among the company's innovations is an information display system using a 13 metre high-resolution screen, unique in the CIS. Staff are also very proud of their aviation LCD displays,

aimed at 4th-5th generation aircraft. These remain operational in the toughest conditions, displaying information under ambient illumination. The company also manufactures an automated, remote control, observation-firing ADUNOK complex; it has already been highly praised by military experts around the world.

# Border closed to potentially dangerous goods

**Radioactive products from Japan excluded from entering Belarus**

The whole world is attentively following the radioactive situation in Japan, with many states banning agricultural imports from Fukushima and neighbouring provinces. Belarus is also implementing enhanced control over imports of food from Japan. "We've not received any applications for permission to import animal derived materials from Japan. Before the disaster, we purchased only a narrow range of veterinary goods from Japan and we once imported Japanese dried broth," notes the Deputy Head of the Agriculture and Food Ministry's Main Veterinary Department, Alexander Kutsko.

All products arriving in Belarus from abroad undergo radiation control. "Border checkpoints have 'radiation gates' through which cars pass; an alarm sounds on detecting a vehicle bear-

ing a hazardous load. Such vehicles are then withdrawn and, if further measurements indicate raised radiation levels, they're denied entry," explains the Head of the Customs Control Organisation Department at Belarus' State Customs Committee, Vladimir Orlovsky. "Minsk National Airport also has such gates, ensuring that no contaminated products enter the Republic."

The press service of an importer of Japanese cars tells us that any vehicles affected by the catastrophe won't be allowed into Belarus. "Mazda plants, in particular, are situated a thousand kilometres away from the zone. Like other Japanese plants, they have stopped operations. However, orders have been placed and are to be completed on schedule, while new orders are still being made. Only the logistics have changed, with transport routes avoiding territories affected by the disaster," notes the company.

# Supercomputers and super-work

By Olga Bestuzheva

**Belarusian scientist expands participation in Large Hadron Collider experiments**

Domestic physicists have received a new order to develop electronic elements for the Large Hadron Collider, used to 'read' signals from its muon cham-

bers (high-precision devices, registering high-energy particles). Development has begun, with our Belarusian scientists joining in analysing collected data from the gigantic particle accelerator, using domestic supercomputers. Our physicists expect further orders relating to the project.

The Director of the Be-

larusian State University's National Centre for Particle and High Energy Physics, Nikolay Shumeiko, notes that Belarus' contribution to the Large Hadron Collider is highly appreciated abroad. Our BSU specialists are continuing to work within the European Organisation for Nuclear Research (CERN), which

is located on the border of Switzerland and France. "Research continues and we'd like to do much more in Belarus. As the application of wide communication channels remains expensive — despite some fall in costs — we're primarily working abroad, via other missions," notes Mr. Shumeiko.