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Early detection of defects saves lives

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The early detection of congenital defects in babies can save lives, Hong Kong University's faculty of medicine revealed.

It said Monday primary immunodeficiency, or PID, can be detected and cured with early diagnosis through DNA testing.

PID often presents itself in the form of ordinary infections. Physicians sometimes treat the infections while missing the underlying cause, allowing the illnesses to recur, and leaving the patient vulnerable to vital organ damage, physical disability, and even death.

"It is a matter of life or death," said Lau Yu-lung, chair professor and head of the department of pediatrics and adolescent medicine.

Forms of PID, which are caused by mutations of the X-chromosome, are carried by mothers and female members of the family, who then pass it down to their newborns - especially boys.

In fact, 72 of the 117 cases studied by Queen Mary Hospital last year were boys.

Lau insists that early detection and diagnosis is crucial in determining the baby's ability to live. With a death rate of 100 percent, babies born with PID die within six months of birth.

The most common clinical problem faced by PID patients is the susceptibility to recurrent infections, especially since babies with PID are prohibited from taking live vaccines.

"Early treatment can increase their chances for a brighter future," Lau said.

After a series of immune function tests and precise diagnosis, patients can then undergo one of two treatments: Immunotherapy or Immuno- reconstitution. Immunotherapy supplements deficient components, alleviating symptoms, while Immuno-reconstitution requires stem-cell transplantation.

Both treatments, available at Queen Mary Hospital, have revealed remarkable results with a success rate of 75 percent for transplantations.

If not treated, forms of PID can develop into even more severe diseases such as cancer.

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Lau and his colleagues recently created a Web-based chatroom for use by doctors.

The innovative research of genetic analysis has contributed to the detection of rare cases of PID, such as IPEX syndrome, the first diagnosed case in Hong Kong and China.

Three-year-old Ho Wai-hang was diagnosed with IPEX at 21 months. Chronic diarrhea and bruising were just some of the symptoms he experienced.

Ho, who is currently awaiting a bone marrow transplant, has also been experiencing side effects to medication with swollen cheeks and thinning hair.

He was often affected by the sneezing of others around him. His mother was forced to quit her job in order to take care of him.

"One of the best options for treatment is umbilical cord transplant," said Dr Ho Hok-lung of HKU. He said cell doses from umbilical cords are adequate for transplants in babies, who can receive small doses according to weight.

Other treatments, such as antibody injections, are usually taken every four weeks and can cost parents up to HK$2,500 a month.

Mothers are encouraged to watch out for certain symptoms including eight or more ear infections a year; two or more bouts of pneumonia a year; and recurrent deep skin or organ abscesses caused by the inability of white blood cells to function normally.

Bone marrow transplant, the most effective form of treatment, is also one of the most difficult because the chances of locating suitable donors for 100 percent compatibility is just one in 200,000. Worldwide registries revealed that the occurrence of PID is approximately one per 5,000 births.