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Diagnosis of childhood TB could be improved by genetic discovery

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A distinctive genetic 'signature' found in the blood of children with tuberculosis (TB) offers new hope for improved diagnosis of the disease.

TB is very difficult to diagnose in children and is often recognised late when the child is already critically ill and the disease has spread from the lungs to the brain or other organs. Now an international team of researchers has shown that the disease can be identified in over 80 percent of cases by looking at 51 specific genes in the blood of affected children.

The researchers hope the findings – published on 30 April in the *New England Journal of Medicine* – could be used to develop a cheap, quick and effective diagnostic test.

Lead researcher, Professor Michael Levin, Director of the Wellcome Centre for Clinical Tropical Medicine at Imperial College London, explained: "We urgently need better methods to diagnose TB in children, so treatment can be started earlier and to avoid unnecessary treatment of children who are wrongly diagnosed. The symptoms of TB in children are common to many other childhood diseases, and the standard tests used on adults are not effective in children. Although the disease is treatable, thousands of children still die each year due to late diagnosis and many more are left with damage to their brain, bones and lungs."

The study – funded through the EU and carried out at Wellcome Trust-supported units in Africa –looked at over 2,800 children admitted to

hospitals in South Africa, Malawi and Kenya with symptoms of TB. The researchers identified those who had proven TB and those in whom TB was excluded as the cause of the child's illness.

Blood samples from the South African and Malawian children were examined to see which genes were activated or suppressed in those with the disease. The researchers found that TB could be distinguished from other diseases by looking at just 51 genes from over 30,000 in the human genome and seeing whether they were activated or suppressed. This information was used to give a single TB risk score for each child which, when tested in the Kenyan patients, accurately diagnosed over 80 percent of the children with TB.

Professor Levin said: "It has taken seven years and the combined efforts of clinicians and scientists in the UK, Africa and Singapore to identify this gene signature of childhood TB. What we now need is collaboration from biotechnology and industrial partners to turn these findings into a simple, rapid and affordable test for TB that can be used in hospitals worldwide."

According to World Health Organisation (WHO) statistics, TB is second only to HIV/AIDS as the greatest killer worldwide due to a single infectious agent. A significant proportion of TB cases worldwide are children. An estimated 530,000 children became ill with TB in 2012 and 74,000 HIV-negative children died of TB.

Professor Brian Eley from the University of Cape Town, who led the clinical study in South Africa, said: "Childhood TB is a major problem in African hospitals. An accurate test for childhood TB would be an enormous breakthrough, enabling earlier diagnosis, reducing long hospital admissions for investigation of TB suspects, and limiting the number of children treated inappropriately."

Dr Suzanne Anderson from Brighton and Sussex Medical School, who led recruitment in Malawi, said: "This study has highlighted the benefit of research institutions in Europe collaborating with hospitals in Africa to apply sophisticated technology to major public health problems."

More information: Anderson et al. 'Diagnosis of childhood tuberculosis and host RNA expression in Africa. The *New England Journal of Medicine*, 1 May 2014. DOI: 10.1056/NEJMoa1303657

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