Saved by screening ... Thuli Mthethwa and her daughter Thembelihle share a light moment while waiting for treatment at the Johannesburg Hospital's paediatric department. The little girl has a rare metabolic disease which fortunately was detected early, thanks to a special screening method that detects metabolic problems. Thembelihle can look forward to a healthier future if she is put on a special diet that excludes milk.

Early diagnosis puts brakes on suffering

Thembelihle Mthetwa sits happily on her mom's lap in Johannesburg Hospital's paediatric ward.

Her smiling eyes and playful gurgles mask her illness. In fact the only indication that she is sick is her swollen abdomen – a sign of her enlarged liver.

She suffers from Galactosemia. Thembelihle's disorder was picked up at Chris Hani Baragwanath Hospital when she was three months old. She later developed the complications of galactosemia because she was not tested at birth.

"She had a severe case of jaundice and had already developed cataracts. She was quite ill," said Dr Jennifer Cartwright, a paediatrician specialising in metabolic disorders.

Now diagnosed, her condition is treated with a special milk supplement.

However, if Thembelihle is fed normal milk, it is more than likely she could suffer mental retardation later in life.

The cost of a baby screening test is about R20 in public hospitals and about R130 in the private sector.

If the test is not done and a child has a metabolic disorder, there is a possibility that the child could end up in an intensive care unit, the cost of which runs into thousands of rands.