A hereditary form of anaemia called Sickle Cell Anaemia or Sickle Cell Disease (SCD) often afflicts tribal communities in the deepest parts of central, western and southern India. This form of anaemia is unlike other forms of anaemia as it may lead to lifelong chronic pain, fatigue, increased chances of infections, stroke, organ failure, and even premature death. Around five million people worldwide suffer from this disease.

The Red Blood Cells (RBCs) in our body contain haemoglobin, a protein that carries oxygen in our blood. It is round in shape and flexible. But in case of sickle cell disease or sickle cell anaemia the RBCs acquire a crescent shape or sickle shape or the characteristic ‘C’ shape and they clump together and stick to walls of blood vessels thereby slowing down or even blocking blood flow along with oxygen from parts of the body. In SCD, the RBCs also break apart easily and live only for 10-12 days in place of the normal 120-day-period.

Persons with SCD have abnormal haemoglobin, called haemoglobin S or sickle haemoglobin, in their red blood cells. A person inherits two haemoglobin genes, one from each parent. SCD is an inherited disease so when the haemoglobin S gene is inherited from only one parent and a normal haemoglobin gene is inherited from the other parent, a person will not have the disease but the person will have sickle cell trait. Generally, people with sickle cell trait live a comparatively healthier life and face fewer complications as faced by persons with SCD. Since people with sickle cell trait are carriers of the abnormal haemoglobin S, they can pass it to their offspring and if the person with whom they reproduce also carries an SCD trait or any other defective haemoglobin gene then the child has a chance of getting SCD.

A person has SCD from birth but the problems associated with the disease do not appear till the baby is about 5 or 6 months of age. Some may have early onset symptoms and some may have delayed appearance of symptoms. The early symptoms of SCD usually may be one or all of them which are fatigue from anaemia, painful swelling of the hands and the feet which is known as dactyilitis, jaundice and icterus which means the yellowing of the whites of the eyes.

The disease can be diagnosed through blood tests. A diagnosis for SCD is also possible before a baby is born – it is done by using a sample of amniotic fluid or tissue taken from the placenta as early as 8-10 weeks into the pregnancy.

Disease Scenario in India
The first description of sickle cell haemoglobin in India was made by Lehman and Cutbush in 1952 in the tribal populations in the Nilgiri hills in South India. In the same year, Dunlop and Majumdar also reported the presence of sickle haemoglobin in the tea garden workers of Upper Assam who were migrant labourers from tribal groups in Bihar and Odisha.

Chhattisgarh is home to the largest number of sickle cell anaemic people in India. Ten lakh tribals of this state are affected with it and according to a study conducted by the Jharkhand State Health Department the state is fast emerging as home to an alarming number of people afflicted with sickle cell anaemia with numbers pegged at over nine lakh tribals, with 60% of it being women.

Treatment of SCD
Most SCD complications are treated as they occur. These treatments include antibiotics, pain relieving tablets along with pain management skills, Vitamins, blood transfusions, and surgery, for example, removal of spleen (splenectomy), removal of the gall bladder (cholecystectomy), hip replacement if the tissue of the hip breaks down because it does not get enough blood and dies (osteonecrosis).

Hydroxyurea is an oral medicine that has been shown to reduce or prevent several SCD complications. This medicine is known to increase the amount of foetal haemoglobin (Haemoglobin F) in the blood. Increased haemoglobin F has been found to provide some protection against the effects of haemoglobin S. Hydroxyurea can cause the blood’s white cell count to drop. In rare cases, it can worsen anaemia.

Doctors also use acute and chronic red blood cell transfusions to treat and prevent certain SCD complications. It is done in case of acute stroke, in cases of acute chest crises or pain, and in multi-organ failure. Chronic transfusions are recommended for people who have had an acute stroke. Chronic transfusions are also used where complications of SCD are not controlled by hydroxyurea and where a person has many side effects due to the use of the medicine.

At present Haematopoietic Stem Cell Transplantation (HSCT) is the only cure for SCD. In HSCT, stem cells are taken from the bone marrow or blood of a person who does not have SCD (the donor); however the donor may have sickle cell trait. The donor is often the person’s brother or sister. This is because the safest and most successful transplants use stem cells that are matched for special proteins called HLA antigens. Since these antigens are inherited from parents, a brother or sister is the most likely person to have the same antigens as the person with SCD.

At present most SCD transplants are performed in children. But since only about 1 in 10 children with SCD has a matched donor without SCD in their families, the number of people...
with SCD who get transplant is low. HSCT is more risky in adults because with increasing age the organs are damaged by the sickled cells.

In the field of HSCT, a new promising treatment for adults with SCD has been developed by the National Institutes of Health, U.S. and validated by the University of Illinois at Chicago that does not require the use of chemotherapy. With the new procedure, patients receive immunosuppressive drugs, along with a small dose of body irradiation prior to stem cell transplantation. A recent study of 13 patients that received this treatment yielded promising results.

According to Dr Santosh Saraf, a doctor of Indian origin and one of the co-authors of the study, “As soon as one month after the transplant, we’ve been administering surveys for the patients, and in that one month we’ve seen improvements recorded for that patient’s quality of life and that reported quality of life continues to improve one year out from the transplant.”

In 2017, a team from the AP-HP University Hospital Group in Paris, the Imagine Institute of Genetic Diseases, and gene therapy company Bluebird Bio claimed that they got a boy suffering from SCD off from transfusions. The boy was the first person to be treated in Paris in October 2014 for SCD in a clinical trial with gene therapy.

The team collected hematopoietic stem cells from the bone marrow of the youngster, who was then aged 13. The immature cells were treated with a therapeutic gene, carried in a deactivated virus, which re-coded their DNA to correct blood cell production. The treated cells were then re-injected into the boy’s body.

Awareness about SCD is increasing worldwide and happily also in our country. With successful adoption of screening programmes what more is desired is a nationwide reporting system or registries that will help to gauge the real picture of SCD in India and which in turn will help in understanding the natural history of the Sickle Cell Disease in India.

The Indian Council of Medical Research and National Rural Health Mission in different states are undertaking outreach programmes which in turn will aid in the better management and control of the disease. Moreover, the establishment of centres for the diagnosis of patients and comprehensive care as well as new and cost effective therapy in curing the disease is the need of the hour.

Mr Shakunt Pandey is a popular science writer, freelance journalist and author. Address: Lake Utsav, P-331 Parnasree Pally, Flat-3A, Kolkata-700060, West Bengal. Email: shakuntpandey33@gmail.com