NEWS IN BRIEF

Every Breath We Take

The deadliest enemies are invisible. Pollution is the elephant in the room, we choose to ignore. An Air Quality Index (AQI) upto 50 is considered healthy and upto 100 is satisfactory. As of 11th November 2017, AQI in New Delhi was touching 600, Lucknow 300, Mumbai 224 and Bangalore 191.

The Lancet Commission on Pollution and Health, published recently, has developed the concept of the 'Pollutome' to classify pollutants that affect health. This is a pyramid with the tip (Zone I) housing pollutants with well-established associations. Zone 2 has emerging but unquantified pollutants associated with common disorders like diabetes, prematurity, autism and dementia. Zone 3 has pollutants with still unknown health hazards such as neurotoxicants, endocrine disruptors, new pesticides and nanoparticles. The report quantifies disease burden, economic impacts and possible solutions to this deeply pervasive predicament. Pollution was responsible for 9 million deaths worldwide in 2015. Pollution is thus responsible for more deaths than a high-sodium diet (4·1 million), obesity (4·0 million), alcohol (2·3 million), road accidents (1·4 million), or child and maternal malnutrition (1·4 million). Pollution was also responsible for three times as many deaths as AIDS, tuberculosis, and malaria combined, and for nearly 15 times as many deaths due to war and all forms of violence. The bulk of global deaths due to pollution were in India and China. Deaths show a peak in children less than 5 years and elderly above 60 years.

In 1952, a combination of windless conditions, airborne pollutants and cold weather caused a severe fog to envelope the city of London. The Great Fog resulted in more than 4000 deaths and 100,000 became seriously ill. But since then strict legislation has seen much improvement in the air quality over London. Delhi and all large cities in India need to learn how to clean up India's air. (*Lancet 19 October 2017*)

Transgenic Stem Cells to Cure Epidermolysis Bullosa

A 7-year-old Syrian refugee suffering from severe junctional epidermolysis bullosa is now cured of his disease – thanks to German doctors and Italian researchers. Eighty percent of the boy's skin was involved in the blisters and his situation was critical. German doctors contacted Michele De Luca, an Italian researcher who had once used gene therapy in a single patient with the same illness. His work had been side-railed due to bureaucratic hiccups for several years. The German physicians sent a small sample of healthy skin to Italy. De Luca's team cultured the cells and using a phage inserted the normal copy of the *LAMB3* gene into the cell. They then grew sheets of the cells in the laboratory. This was then transplanted onto his skin in Germany. There was complete regeneration of the boy's epidermis. The experiment has proved that relatively few stem cells are adequate to regenerate the entire epidermis. The child will be monitored for any skin cancer, which is one of the major risks of gene therapy. (*www.nature.com/articles/nature24487; Scientific American 8 November 2017*)

CFTR Modulators for Cystic Fibrosis

Cystic fibrosis is due to mutations in a gene which results in dysfunctional or inadequate production of a protein – cystic fibrosis transmenbrane conductance regulator (CFTR). This results in abnormal chloride secretion, thick mucus, progressive lung disease and pancreatic insufficiency. CFTR modulators are a new class of drugs. They include potentiators, which increase function of the CFTR channels; correctors, which improve the processing and delivery of the functional CFTR protein to the cell surface; and production correctors, which promote the read through of premature stop codons in the mRNA.

The NEJM has recently published two studies evaluating the efficacy and safety of the oral CFTR modulators, tezacaftor–ivacaftor. The studies have shown statistically lower pulmonary exacerbations and significant improvements in lung functions in patients on therapy. (*NEJM November 10 2017*)

Gene Therapy for Spinal Muscular Atrophy

In Spinal Muscular atrophy, mutations of the *SMN* gene results in defective protein synthesis resulting in progressive motor regression and finally death in early childhood. Fifteen patients with Spinal Muscular Atrophy type 1 were infused with the adeno-associated virus, which had been modified to carry the DNA for the SMN protein. All patients were alive at 20 months compared to a historical control of 8%. Two were walking and 11 were sitting unassisted, which is remarkable for children with SMA type I.

The study opens new doors for a group of children whose outcome has been largely bleak. (*N Engl J Med. 2017;377:1713-22*)

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