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Short Communication

Xeroderma Pigmentosum

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Xeroderma pigmentosum, which is known as XP, is a rare inherited disease characterized the maximum sensitivity of the sun and ultraviolet rays, resulting in pigment changes, sunburn and increased rates of mucus membrane cancers. It is an autosomal recessive disorder with the possibility of starting a 10,000-fold growth in skin cancer in patients having XP below the age of 20 years. It was in the late 19th century when dermatologist Moritz Kaposi led the road to understanding XP. During 1874, Kaposi used the term "xeroderma" to describe four patients with this disease. Dr. Albert Neisser reported the first XP case with neurological indications and later in the second decade, DeSanctis and Cacchione outlined XP with acute neurological deficiency known as "DeSanctis-Cacchione syndrome" [1].

Xeroderma pigmentosum usually affects the eyes and parts of the skin that has been exposed to the sun. People who are affected also have problems implying the nervous system. XP signs have been noticed in early childhood. Children who have been affected develop an acute sunburn just after a few minutes in the sun, causing blisters and redness that can last for weeks, while other children tan normally after less sun exposure. Most children by the age of 2 with XP grow freckling of the skin in areas like the face, lips and arms. Affected individuals have dry skin (xeroderma) when often exposed to sunlight and skin color changes (pigmentation). Hence this condition is called xeroderma pigmentosum. There is a higher risk of skin cancer developing in people with XP. These cancers happen on the lips, eyelids, tip of the tongue, eyes and scalp. XP is also related with noncancerous growths on the eye. This can lead to vision impairments. Malformations like difficulty in walking, hearing loss, problems in moving, poor coordination, loss of mental function and seizures occur in 30 percent of people who develop neurological abnormalities after having XP [3,4].

Genes that are associated to XP are part of the DNA-repair procedure called nucleotide excision repair (NER), mutations in more than one NER XP gene caused by molecular faults in cellular DNA repair system would lead to extreme physical sensitivity to UV radiation. This would cause a build-up of unrepaired UV-induced DNA damage which can cause cell death leading to an increase in skin ageing or can promote cellular modification in cancer development. Studies have also indicated a high incidence of XP seen in people where marriage of blood relation (blood relatives) in countries like Libya, Nigeria and Pakistan is common. Other than UV radiation, DNA-damaging agents have been known to produce excessively sensitive responses with XP cell [2].

Usually, diagnosis is carried out on clinical findings and family history. Display of hypersensitivity to UV in a person at an early age can be diagnosed. Measuring post-UV unscheduled DNA synthesis (UDS) from DNA repair tests and UV survival by colony development can be also the diagnosis of XP. Unscheduled DNA synthesis in refined skin fibroblasts can be measured as well as the complementation assay for NER defects. There is molecular diagnosis of XP, these include using PCR to diagnosis XPA, plasmid host cell reactivation (HCR) and prenatal tests [2].

To date, there is no cure for XP. To prevent skin cancers in people having XP and prolong their life expectation, early diagnosis and protection from the sun is vital. The damage of the DNA increases and is irreversible. It is important to take protective measures, medical care and therapy.

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