Chimerism in humans



Chimera according to the genome glossary is an organism which possesses cells or tissues with a different genotype. This can be caused due to mutated cells of the host organism or cells from a different organism or species.

According to Greek mythology, the original " Chimera" (pronounced as kih-MEE-ra, and the adjective form " chimerical" pronounced as kih-MER-ih-cal) was a fire-breathing female monster with the head of a lion, the body of a goat and the hindquarters of a dragon. The terrifying Chimera may have merely been a product of a substandardfamilyenvironment-- her father was the giant Typhon, her mother the half-serpent Echidna. Her siblings were the Cerebrus (the three-headed hound who eventually found work guarding the gates of Hell), Hydra (a nine-headed aquatic monster) and Orthrus (the runt of the litter, a prosaic two-headed dog). As per the mythology, Chimera pounced around making everyone wretched until one day a man named Bellerophon, riding on the winged horse Pegasus slays her. [Author unknown, Chimera, 2004]

Though believed to be a mythological epic, it was found that cases of Chimera were actually prevalent in humans when certain intriguing criminal cases erupted. One such case was when a woman has been raped by a man and the women claimed a particular person to be the culprit. However, the crime justice authorities had found it difficult to convict the suspect. It so happened that the DNA taken from his blood sample did not match the DNA of the suspect. However, extensive investigation and the victim's statements revealed it to be same person being responsible for the crime. When another series of DNA tests were taken from the suspect, this time from his hair sample it was found that the DNA sample perfectly matched. This revealed a

breathtaking phenomenon among human genome which has resulted in various problems in the criminal justice system.

Those people who have two types of DNA in their body are called chimeras after the mythological creature with a head of lion, body of a goat and a tail of a serpent. These people are sometimes also referred to as mosaics. It has found that these people have two different types of DNA in various parts of their body. Though a rare occurrence, this could have been due to various reasons. [Vladar, 2004]

One of the common reasons is that chimeras are formed if developing fraternal twin embryos join together to become one embryo. This is something which is completely opposite of identical twins who are formed when a single embryo splits into two. This occurrence takes place in the very early stages of the embryo development i. e. when it is still in the form of unspecialized cells, so when it develops the baby that is born is healthy but with two types of DNAs. It is also found that fraternal twins don't have same set of DNA, however, they the two fraternal twin embryos fuse together it results in chimera.

Another reason for chimeras to form is when developing fraternal twin share the same blood supply. This usually happens when the twins with different DNA share a placenta in the mother's womb and cells from their blood mix together. This results in the twins being chimeras based on only their blood as other cells are not affected by the blood supply. Chimeras are also formed through some mistakes or mutations taking place in the division of the cells in the development embryo. This mainly takes place when the cells split into

two so that they are able to be formed into more of themselves for the growth of the embryo. It also results in the the doubling up of the DNA by the cells which gets divided between the two new halves. During this process, if something goes wrong, then some new cells result in having different DNA. If this happens in the early stages of the development, then the tissues which are developed from the cells will have different genotype.

It is difficult to gauge the exact number of chimeras in the world. We are able to find out whether a person is a chimera or not only when his / her DNA is analysed. This causes varied problems with regards to solving of crime cases and also results in complication with regards to donation of organs. In recent years, exciting results have been found out that a section of genetically incompatible cells may contribute to conditions like infertility, autism and Alzheimer's disease. [Migliore, L et al, 1999]

Human chimeraism came into being with advent of blood typing and it was found that some people had more than two types of blood group. Most of the cases of chimeraism are the blood chimeraism i. e. non-identical twins sharing a blood supply in the womb. Twin embryos often share a blood supply in the placenta, allowing blood stem cells to pass from one embryo and settle in the bone marrow of the other, seeding a lasting source of blood. As a result, almost 8% of non-identical twin pairs have chimeric blood. [van Dijk, B A; Boomsma, D I and de Man, A J M, 1996]

Apart from those who have chimera due to different blood types, there are even larger numbers of people who have this phenomenon due to carrying smaller numbers of different blood cells that may have passed through the mother and the fetus across the placenta or persist from a blood transfusion. It has also been analysed that the presence of some foreign white blood cells might be the cause for autoimmune diseases wherein the immune system affects the body's own tissues. [Nelson, J L, 2002]

True chimeras are usually identified when they possess male and female cells resulting in the formation of hermaphrodites or any related problem in the person's sexual organs and their chromosomal sex is identified by their blood test. So the real number of chimeras and mosaics are much more than those that have been identified till date. Unless and until a chimera comes forward to get his blood sampled or has been convicted for a crime, it wouldn't be possible to know whether he actually is a chimera. This has resulted in further complication in solving some crime justice cases.

It has been also analyzed that with the increase in in-vitro fertilization (IVF), there has an increase in the cases of chimeras. To avoid infertility among women, IVF has been quite common in the present day scenario. In this process, two or more embryos are placed in a woman's uterus so there is an increased success rate. This results in 25% more cases of twin pregnancies than expected. With the increased number of twins, there is likelihood of more cases of chimeras. This was proved by Bonthron who found out that the British hermaphrodite boy who was a chimera was formed by IVF. Strain, L; Dean, J C S; Hamilton, M P R and Bonthron, D T – 1998]

According to various tests undertaken along with ultrasound examinations performed on the mother who was carrying the single fetus, the baby that was born had a normal right testis and a semi-developed left testis. An

operation was conducted when the child was 15 months old and the surgeons removed " an abnormal gonad and vas deferens." Pathology studies later exposed that these were actually an ovary with a fallopian tube attached.

Studies revealed that the child's chromosomes had both female (XX) and male (XY) cell lines. This was explained to be that the child was an exceptional case of a true chimera which was a mixture of two embryos from separately fertilized eggs. The other likelihood was that two different sperm had unusually developed into a single mature egg before cell division had taken place. To distinguish between these possibilities, the researchers analyzed DNA near the centromeres of the child's chromosomes, which are involved in cell division. [Author unknown, DNA puzzle of mother 'made of two women', 2004]

Apart from this, the study conducted also revealed that chimerism can occur naturally, resulting in children with vague genitalia, however published data on the phenomenon is very restricted and many XX/XY children may go unnoticed. Even so, the fact that chimerism occurred in a child born following in vitro fertilization should be taken seriously. [Author unknown, 2004 In Vitro Fertilization Results In Chimera, 2004]

Considering these above points, it is becoming more and more difficult for the crime justice system to analyze the crime due to the increase in the cases of chimeraism and mosaicism which in turn is increasing with the increase in in-vitro fertilization. The criminals are able to move about freely as their blood samples usually differ from their other DNA samples taken

from different parts of the body. It is also becoming difficult for the crime justice system as well as the medical system to analyze the exact number of chimeras as it is very difficult with the increase in mutations.