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Treacher Collins Syndrome

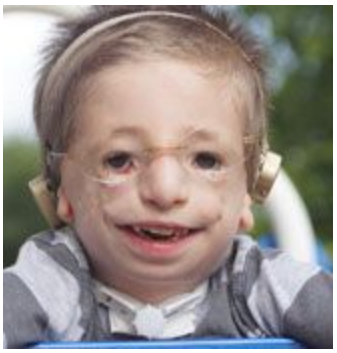
Treacher Collins Syndrome (TCS) is a congenital disease, which means that it cannot be acquired and is either present or absent at birth. TCS is sometimes so mild that individuals can go undiagnosed. There are also those who are very strongly affected to a life-threatening degree. TCS typically shows up as a visible face abnormality. These abnormalities are often symmetric. As this is a congenital disease, the abnormalities in the face can compromise speech and language development through hearing loss, cleft palate, and/or jaw and airway problems. Typically, infants with TCS have underdeveloped or absent cheekbones, making their face look sunken in. The outer parts of their ears can be almost completely missing and you often see a very small jaw coupled with a very large mouth. Symptoms also include a defect in the lower eyelid and scalp hair that reaches to the cheeks. It is estimated that TCS occurs in 1 in 50,000 live births, so the disease is not common at all. Even parents who have the disease have a 50-50 chance of their children having the disease as well.

In rare cases, TCS can be diagnosed before birth if facial abnormalities are seen during an ultrasound. Otherwise, a child can only be diagnosed after its birth. A doctor may take X-rays or CT scans to confirm the diagnosis. Typical traits are an abnormal eye shape, flat cheekbones, clefts in the face, a small jaw, low-set, abnormally formed ears (specifically the ear canal), hearing loss, coloboma, and decreased eyelashes on the lower eyelid. There is no “cure” for TCS, but children with the facial abnormalities can be helped with plastic surgery. Any hearing problems are also treated to better school performance. Research into the disease is slow and not entirely fruitful, but it is happening and it is improving the lives of all who have this disease.

By the disease itself, many aspects of the body are very affected. The respiratory system can be heavily affected. The lungs may not get enough oxygen due to this disease blocking any way for the afflicted person to breathe. This results in the person relying on some method of artificial respiration. It may, however, become impossible for this person to speak at all.

The organelle in the cell most affected by this disease is the ribosome. Ribosomes are found free in the cytoplasm or bound to the rough endoplasmic reticulum. They put together proteins using mRNA from the nucleus. TCS is due to a mutation that occurs in a gene responsible for the creation of rRNA, which is over half of what ribosomes are made out of. This mutation affects the formation of the proteins that make up the bones of the face. It is the TCOF1 gene that is involved in approximately 93% of all people affected. If the ribosomes don’t have

enough rRNA, then they simply cannot function, thus the disease. Treacher Collins Syndrome, in short, describes craniofacial deformities that an infant can be born with.



Works Cited

ScienceDaily. ScienceDaily. Web. 19 Dec. 2016.

"Craniofacial Abnormalities: MedlinePlus." *MedlinePlus*. Web. 19 Dec. 2016.

"Ribosome." *British Society for Cell Biology*. Web. 19 Dec. 2016.

"The Roles of RNA Polymerase I and III Subunits Polr1c and Polr1d in Craniofacial Development and in Zebrafish Models of Treacher Collins Syndrome." *PLoS Genetics.* U.S. National Library of Medicine. Web. 19 Dec. 2016.

"Treacher Collins Syndrome." *Treacher Collins Syndrome | Seattle Children's Hospital*. Web. 19 Dec. 2016.

"Treacher Collins Syndrome - NORD (National Organization for Rare Disorders)." *NORD (National Organization for Rare Disorders)*. Web. 19 Dec. 2016.

"Treacher Collins Syndrome: Facts About Life Expectancy." *MedicineNet*. Web. 19 Dec. 2016.

"Treacher Collins Syndrome." *Treacher Collins Syndrome*. Web. 19 Dec. 2016.

"Treacher Collins Syndrome: MedlinePlus Medical Encyclopedia." *MedlinePlus Medical Encyclopedia*. Web. 19 Dec. 2016.

"Treacher-Collins Syndrome - Mandibulofacial Dysostosis (MFD1)." *News-Medical.net*. 19 Apr. 2015. Web. 19 Dec. 2016.

Vanderburg, Bruce B. "Treacher Collins Syndrome: Three Genetic Mutations Identified As Cause." *ReliaWire*. 24 July 2016. Web. 19 Dec. 2016.