**Diseases of Cellular Organelles Case Studies:**

**Case 1:** Cystic fibrosis (CF) was first described in medical journals in 1938 as a defect in the channels leading from certain glands, resulting in a variety of problems--chokingly thick mucus in the lungs and frequent infection there; a clogged pancreas, preventing digestive juices from reaching the intestines; and salty sweat. A child with CF is often small and sickly, and until the recent availability of biochemical tests, was often initially diagnosed simply as having failure to thrive.

Researchers identified the cellular defect behind cystic fibrosis in 1989 as abnormal channels in lung and pancreas cells that trap salt within cells. The salty cellular interiors draw moisture in from surrounding tissue, drying out the mucus until it is so sticky that it clogs organs. Several new treatments, including a healthy gene introduced into the lungs in a nasal spray, target the illness at the cellular source.

Which organelle is not functioning properly? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Case 2:** Michael was a pleasant, happy infant who seemed to be developing normally until about six months of age. Able to roll over and sit for a few seconds, suddenly he seemed to lose those abilities. Soon, he no longer turned and smiled at his mother's voice, as he had before, and he did not seem as interested in his mobile as he once was. Concerned about Michael's reversals in development, his anxious parents took him to the doctor. It took exams by several specialists to diagnose Michael's Tay-Sachs disease, because, thanks to screening programs in the population groups known to have this inherited illness, fewer than ten new cases appear each year. Michael's parents were not among those ethnic groups and previously had no idea that they both were carriers of the gene that causes this very rare illness.

A neurologist clinched her suspicion of Tay-Sachs by looking into Michael's eyes, where she saw the telltale cherry red spot indicating the illness. Biochemical tissue analysis indicated that Michael Lacked one of the forty types of enzymes used to break down fatty material in his nerve cells. The result is that the organelles that should have broken down this fatty tissue had begun to store it and swell. His nervous system would continue to fail, and he would be paralyzed and unable to see or hear by the time he died, before the age of four.

The cellular and molecular signs of Tay-Sachs disease had been present long before Michael began to lag developmentally. The next time his parents expected a child, they had her tested before birth for the enzyme deficiency. They learned, happily, that she would be a carrier like themselves, but not ill.

Which organelle is not functioning properly? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Case 3:** Emily was a happy, active 12 year old, until she began to have unexplained seizures. Over the next 12 months, she began to have problems with her vision, lose muscle control, and was eventually hospitalized for stroke-like symptoms following her seizures and headaches.

A muscle biopsy revealed that Emily’s cells were missing an enzyme involved in the production of ATP in her cells. The result was that her muscles and nerves lacked the energy required to function.

Which organelle is not functioning properly? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_