

calories be delivered daily as fats. If pancreatic insufficiency is present, the patient should take pancreatic enzyme supplements with each meal and snack in doses adequate to maintain weight and prevent fatty, greasy stools. Fat soluble vitamins should be administered daily, including vitamins A, D, E, and K.

Men with CF who desire to have children should be evaluated for candidacy for techniques to harvest spermatozoa directly from the testes. These sperm samples can be used for fertilization via artificial insemination. Women with CF who desire to have children are frequently able to conceive without much difficulty. Obviously, if a woman with CF anticipates becoming pregnant, it would be in her best interest to work with a physician to develop a plan to optimize her physical state and pulmonary function; doing so would allow for the best possible chance for a successful pregnancy.

Regular exercise and physical conditioning is extremely important in CF patients. Exercise has been shown to improve physical fitness, possibly increase secretion clearance, and improve quality of life. The most common exercises recommended include aerobic forms of exercise such as swimming, walking, jogging, and cycling.

ADDITIONAL THERAPIES

Lung transplantation has been performed in some individuals with CF who have developed end-stage pulmonary involvement. The long-term survival rates in CF patients undergoing lung transplantation are estimated as 60 percent at five years post-transplantation. Generally accepted indications for lung transplantation in patients with CF include FEV₁ (lung function test) less than 30 percent of predicted value; low blood oxygen levels or high blood carbon dioxide levels; increased hospitalization frequency; frequently coughing up blood; or extreme malnutrition.

Difficulties with lung transplantation in CF patients are similar to those encountered with patients requiring other organ transplants; these include paucity of donor organs, deterioration of the CF patient's condition prior to the time that a donor is located, and so forth. Survival of CF patients post-transplant is estimated to be 80 percent at one year, and less than 50 percent at four years. Several ongoing research trials are underway that should assist practitioners and CF patients with new therapies to

extend their life span. Examples of new therapies include gene therapy to correct the basic genetic defect. A good resource for patients with CF and their families is the Cystic Fibrosis Foundation.

SEE ALSO: Bronchitis; Genetic Disorders; Genetic Testing/Counseling; Lung Transplantation; Pneumonia.

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Cytogenetics

Cytogenetics deals with chromosomal structures and their inheritance as applied to clinical medical genetics. Chromosomes are microscopic structures found in cells and malformations associated with them lead to numerous genetic diseases. Over the years, chromosomal analysis has improved in precision and resolution, and has led to improved diagnosis of various genetic diseases in all areas of medicine.

Genetic diseases begin with a mutation in chromosomal structure. The hundreds of types of genetic diseases include congenital abnormalities, reproductive wastage, and retarded mental growth. Statistics show that approximately 1 percent of all live births have congenital abnormalities (2 percent to women over 35) and they are present in 50 percent of all spontaneous abortions. Some common genetic disorders are Down syndrome, Turner's syndrome, cystic fibrosis, and Huntington's disease.

The study of chromosomes begins with the extraction of intact chromosomes from live body cells. The most common cells used for chromosomal studies

are white blood cells (T lymphocytes) because they grow rapidly in cell cultures. These cells are arrested in metaphase, and treated with hypotonic solutions to release their chromosomes. The only disadvantage is that these cell cultures live only three to four days. Chromosomes may also be extracted from skin fibroblasts, bone marrow cells, lymphoblastoid cells, and fetal cells (from amniotic fluid and chorionic vil-lus biopsy).

After cells are extracted, they have to be viewed so that chromosomes can be identified for abnormalities. The 48 pairs of chromosomes can be identified by using various staining techniques such as Q banding, R banding, C banding, G banding, and fluorescence in situ hybridization (FISH). G banding is the most used chromosomal staining method. Chromosomes are first treated with trypsin and then with Giemsa stain. All chromosomes can be individually identified using this technique. With Q banding, chromosomes are stained with quinacrine mustard or related compound, and examined by fluorescence microscopy, which is useful for detecting heteromorphisms—occasional chromosomal structural and staining variants. Before chemical staining using the R banding method, chromosomes are treated with heat, resulting in patterns that are easier to analyze. In C banding, regions of chromosomes containing heterochromatin are stained.

FISH is used to examine the presence or absence of a particular DNA sequence, and the number or organization of a chromosome or chromosomal region. With FISH, technicians use DNA probes to detect individual DNA sequences and copies of a particular chromosome. These probes can also be developed to scale entire lengths, and “paint” target chromosomes in metaphase and anaphase.

CHROMOSOMAL ABNORMALITIES

There are two main types of chromosomal abnormalities: aneuploidy and translocations. Aneuploidy is an abnormal chromosomal number due to extra or missing chromosomes, for example, Down syndrome. This is the most common type of chromosomal abnormality, and persists in 3 to 4 percent of all clinically recognized pregnancies. Aneuploidy is further divided into trisomy or monosomy. Trisomy is usually caused by meiotic nondisjunction. Nondisjunction is most common at meiosis I, but can occur during meiosis II, and even less frequently during mitosis.

Translocations are due to abnormal rearrangements in chromosomal material. Material may be exchanged between different chromosomes, causing disease. There are two types of translocations: balanced and unbalanced. Balanced translocations occur if the cell ended up with a normal number of chromosomes; no symptoms are presented. Unbalanced translocations occur if there is additional or missing material and abnormal phenotype may be visible. They are more commonly found in couples who have had two or more spontaneous abortions.

SEE ALSO: Chromosome; Genetic Disorders; Genetics; Genetic Testing/Counseling.

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Cytokine

Cytokines are small secreted protein messengers that are involved in a range of physiological processes, including development and wound healing. They have a particularly pivotal role in the immune system. Similar to hormones, cytokines affect the behavior of their target cells—causing effects such as growth, proliferation, and migration. Yet, unlike hormones, cytokines are produced by a variety of cells, rather than specialized glands.

The roles of different cytokines are not completely understood, and they are the subject of intense research efforts. As a result of the important role of cytokines in the immune response, these molecules may have medical applications that have only begun to be explored.