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Current Research in Down Syndrome

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Editor's note: In the last year, I've seen several newspaper articles with headlines that suggest a "cure" for Down syndrome is within reach. If you read beyond the headlines, you understand that it's really not happening tomorrow or even next year. But, not everyone reads beyond the headlines and sometimes well-intentioned friends, family members and co-workers want to pass along what they think is great news. It can be difficult to know what is hope and what is hype. Laura is a pediatrician and parent of a 12-year old son with DS.

Many exciting areas are currently being investigated in relation to Down syndrome. The genome of human chromosome 21 was elucidated three years ago and right now about 360 genes have been identified. Most of these genes will, when entering the cell plasma, produce specific proteins. Proteins play important roles in various chemical reactions that occur within the body. There is ongoing research to identify the structure and the function of these proteins and how they affect the phenotype. In mice that have segmental trisomies similar to human chromosome 21, studies are underway to investigate various bodily functions such as learning and behavior and fetal brain development. There continue to be studies on conditions associated with DS such as celiac disease. These studies attempt to better define the progress of these conditions, and evaluate how to improve screening and treatments.

Alzheimer disease is a very active and important area of research. About 20 to 25 percent of older people with DS may develop Alzheimer disease when they are 50 to 60 years old. The beta-app gene is coded for on chromosome 21 and is involved in the senile plaque formation in DS and Alzheimer disease. Much of Alzheimer research could be helpful to people with Down syndrome even if not directly done with people with DS. An example of this is an article published by investigators from Johns Hopkins that documented a decreased prevalence of dementia in people who took supplements of vitamin C and vitamin E, both of which are antioxidants.

Antioxidant therapies are another important area of research. Antioxidants help to prevent tissue damage. The gene for one of the key enzymes, superoxide dismutase is located on chromosome 21. Having an extra copy of the gene that produces superoxide dismutase results in hydroxyl radicals that will cause damage to cells. Scientists theorize this damage may lead to some of the abnormalities associated with DS. Further research could lead to ways to adjust the imbalance caused by having an extra copy of the gene that produces superoxide dismutase and hopefully limit or prevent the cellular damage.

The biology underlying DS is very complex for several reasons. Chromosome 21 codes for about 360 proteins. The effects of the abnormal number of chromosomes (three copies of chromosome 21) begin in utero when the baby is developing. Major organs are formed during the first trimester of gestation. These factors make it unlikely that a single intervention could ameliorate or cure all the abnormalities that are associated with DS.

When evaluating treatments or therapies it is important to look at the respective research. Different types of research take various amounts of time to accomplish. Studies of potential drug treatment can take many years because FDA approval is not given until safety and efficacy has been shown in very large studies. Research dealing with gene function is very promising but must be viewed in the long term. First, the genes need to be identified and their function to be determined. It will also be important to investigate how they interact with other genes. Once the functions and interactions are understood, then ways to ameliorate the effects must be investigated. It will likely take many years until effective therapies based on these types of research are found.

It also is important to understand where and how the research was done. Was it done in a reputable laboratory that followed standard scientific methods? It is important that these studies include a large number of people, that the investigators are blinded to the study groups, and that other variables that could affect the outcome are controlled for and adverse affects of the intervention documented. One should know if a pharmaceutical company or other group that will make money if the therapy becomes popular funded the study.

It is a very exciting time in research on DS. With the advent of the completion of the human genome project, more is known now than ever before. It is assumed that, in the future, there will be interventions to help people with DS be healthier, function better and have a better quality of life.