CHARGE Accounts

Fall 2004 | A Quarterly Newsletter for Families and Friends | Vol. 14 No. 3

CHARGE Syndrome:
CHARGE has four major features -
Coloboma, Choanal atresia, Cranial nerve abnormalities, and Characteristic ears
More information on website

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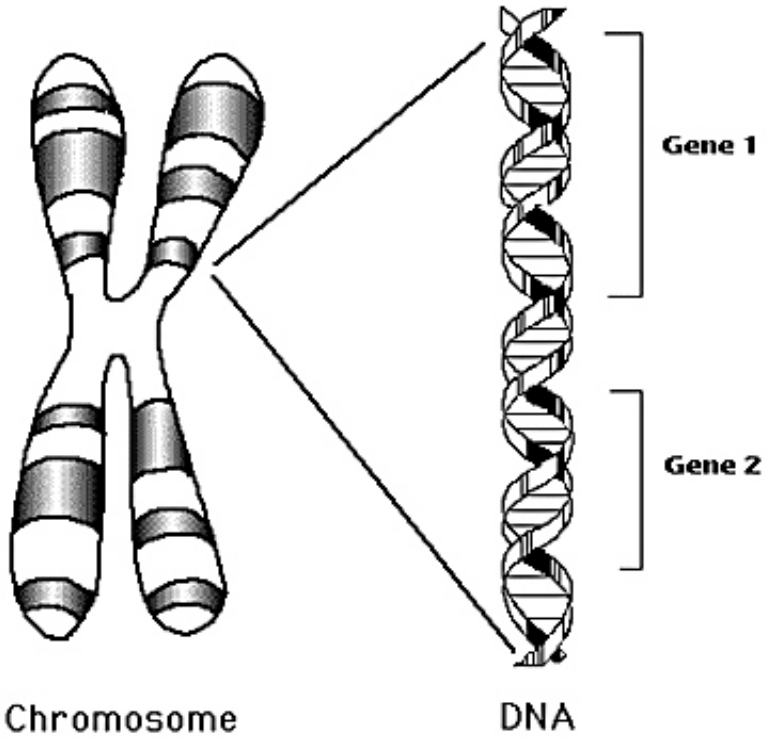
A CHARGE GENE DISCOVERED!



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YOUR HELP IS NEEDED!

Please share your stories, parent tips, questions, book reviews and suggestions for other features you would like to see included.
Remember this is your newsletter!

PRESIDENT'S NOTE
Bruce Appell

FOUNDATION ACCOUNT

Hi Everyone:

It is hard to believe fall is here already and the children are back in school. It also hard to believe that we will be meeting in Miami in just approximately nine months for the 7th International CHARGE Syndrome Conference. Plans are well under way for the conference and it should be an exciting time. The hotel is right on the beach with a beautiful pool and wonderful meeting rooms. Please start planning now to attend the conference next year. [Ed note: general information on the conference is in this issue of *CHARGE* Accounts]

The past couple of months have been exciting for the Foundation. We have opened our office in Columbia and have given Marion her house back. Thank you, Marion, for sharing your house for all of those years. We really appreciated your kind gesture. Please don't worry, Marion will still be answering the phones when you call and working hard for the Foundation and all of the families as the Executive Director of the CHARGE Syndrome Foundation. She has hired a part-time employee to help with some of the work load.

As many of you have heard, a gene has been found that is a cause of CHARGE. One of the authors of this study has written an account of the research for this issue. Hopefully this information will help Baylor University and Seema continue their research to confirm the findings using a larger sample of patients. [Ed note: Seema and the others at Baylor are thrilled with this news and will continue to work on CHARGE. Seema is currently on maternity leave with a new daughter]

I would like to welcome Dennis O'Toole back to the Board of Directors. Dennis will be finishing out Wendi Wood's term. I would also like to thank Jim Thelin for agreeing to become our new Vice-President. Thank you both for stepping forward and giving your time to the Foundation.

In closing, my family and I send our prayers to all who have been affected by the numerous hurricanes, tornadoes, and floods that the U. S. has experienced over the past month or two.

Sincerely,
Bruce Appell
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EXECUTIVE DIRECTOR'S NOTE
Marion Norbury

It has been a exciting summer here at the Foundation office with some changes and the news of a CHARGE gene discovery!

We have moved to an office in a *real* office building. This means that we now have everything in one place (and not in the back room in my house or my garage or a storage locker) and I have my house back.

The other big change is we now have a very well qualified assistant, Kyna Byerly. Kyna is a genetic counselor who has been a stay at home mom for a few years. She has experience and knowledge and has already proven to be a great asset to the Foundation. I am looking forward to completing several projects that we have been wanting to do with Kyna's help.

I hope you find this issue of *Accounts* to be helpful to you as we all learn about the gene discovery and what comes next.

Remember your suugestions and contributions for YOUR newsletter are always welcome.

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2005 Conference

The 7th International CHARGE Syndrome Conference will be at the Wyndham Miami Beach Resort July 22-24, 2005. Are you interested in attending the International CHARGE Syndrome Conference next July? Be sure to save the Conference Information insert. It will help you in your planning.

Will it be the first conference you have attended? Did you know that the CHARGE Syndrome Foundation provides scholarships for families attending their first International CHARGE Syndrome Conference? The scholarship covers a family's hotel and registration fees. An application is included with this newsletter. The deadline for applying is February 1, 2005.

See you on the beach in Miami in 2005

Committee Chairs

Conference – Neal Stanger

Research – Meg Hefner

Fundraising – Brownie Shott

Education/Outreach – Jim Thelin

All board members have a special email address:
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SPECIAL RESEARCH ACCOUNT

Editor's note: They Found a Gene for CHARGE!!!

As many of you have heard through the grapevine, researchers in the Netherlands have identified a gene for CHARGE syndrome. This issue of CHARGE Accounts includes an account, by Dr. Conny Ravenswaaij, one of those researchers, of the successful hunt for the gene. There are also commentaries on the news by Dr. Sandra Davenport and the Baylor group (Dr. John Belmont and Dr. Seema Lalani) in accompanying articles.

A Major CHARGE Gene Has Been Found

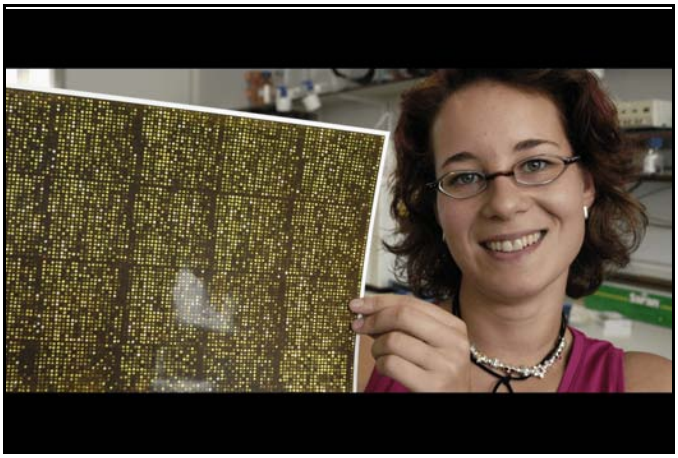
Dr. Conny van Ravenswaaij

"Mutations in a new member of the chromodomain gene family cause CHARGE syndrome."

This is the title of a paper that summarizes the research of a Dutch group looking at possible genes for CHARGE syndrome. The paper appeared on the Nature Genetics web site on August 8, 2004 and confirmed the rumors that were circulating among the CHARGE communities. Many of you may have seen the publication on the Internet and may have asked yourself what language it was written in. Well, it is written in science weirdo and I shall try to translate it into normal everyday English.

Who am I?

Let me start with a short introduction of our research group and myself. My name is Conny van Ravenswaaij, clinical geneticist/cytogeneticist at the University Medical Centre of Nijmegen. The first author of the paper is Lisenka Vissers, a junior researcher at our institute. She did most of the laboratory work. Ronald Admiraal, the third author, is an ENT specialist -- he knows many Dutch children with CHARGE syndrome and studied the semicircular canal abnormalities in CHARGE. Joris Veltman is head of our microarray lab facility and supervises Lisenka. All other authors contributed more or less to the research project.



How did our project get started?

Ronald Admiraal has been involved with CHARGE for many years. He has collected clinical information and blood samples of children with CHARGE syndrome and is involved in the Dutch parent support group. I always thought that CHARGE was caused by a microdeletion and have been looking for possible sites of such a microdeletion (I will come back to that later). One day Han Brunner, the head of our department, showed me a paper about a girl with a small deletion of chromosome 14 who looked very much like she had CHARGE syndrome. We studied blood we had collected from children with CHARGE to look for a possible deletion on chromosome 14. However, none of the children we tested had a chromosome 14 deletion and the samples were stored for later research.

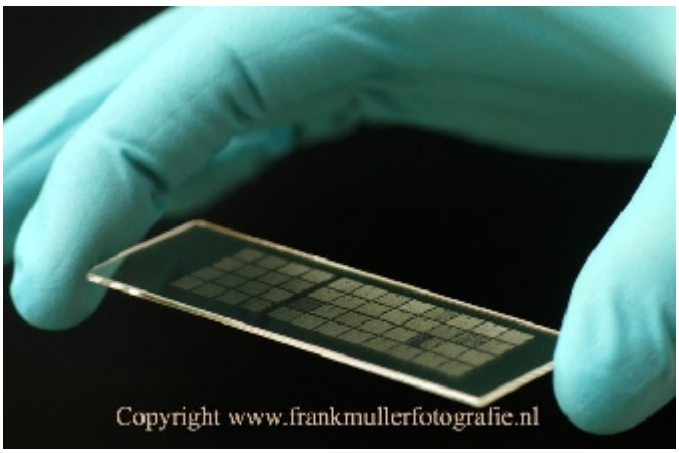
What causes CHARGE syndrome, a microdeletion or a single gene defect?

Although CHARGE syndrome is usually sporadic (i.e. only one person in a family is affected), many scientists already were convinced that it is caused by a genetic factor. What is the evidence for this? First, there is no pattern of prenatal exposures that would suggest anything done during pregnancy causes CHARGE. Second, twin studies of children with

CHARGE showed that identical twins always have CHARGE but fraternal (non-identical) twins never both have CHARGE. Third, there are rare familial cases, where two siblings have CHARGE or a parent and child are affected. And finally, many groups have confirmed advanced paternal age: that is, fathers of children with CHARGE are, on average, slightly older than fathers of children with non-genetic conditions such as isolated cleft lip. It is well known that new changes in DNA (new mutations) happen slightly more often with advanced paternal age.

A "new mutation" causing CHARGE could be either a microdeletion or a change in a single gene. "Microdeletion" refers to a very tiny missing piece of chromosome. Microdeletions are too small to be seen by routine chromosome studies in which the chromosomes are visualized under a microscope with a magnitude of 1000x. However even such a tiny piece of chromosome can contain tens to hundreds of genes. The FISH study many of your children have had was to rule out a microdeletion on chromosome 22 which causes VCFS (velocardiofacial syndrome), a syndrome with many features similar to CHARGE.

The fact that CHARGE syndrome affects many different organ systems (ears, eyes, heart, etc.) fit with a microdeletion involving a number of missing genes. At least, that is what I always thought. If, on the other hand, CHARGE is due to a single gene defect, that gene must play a key role in early embryonic development – such a gene could have an effect on many different organs. As you know, many researchers around the world have been using a number of different laboratory techniques to try to find a gene or microdeletion that could cause CHARGE.

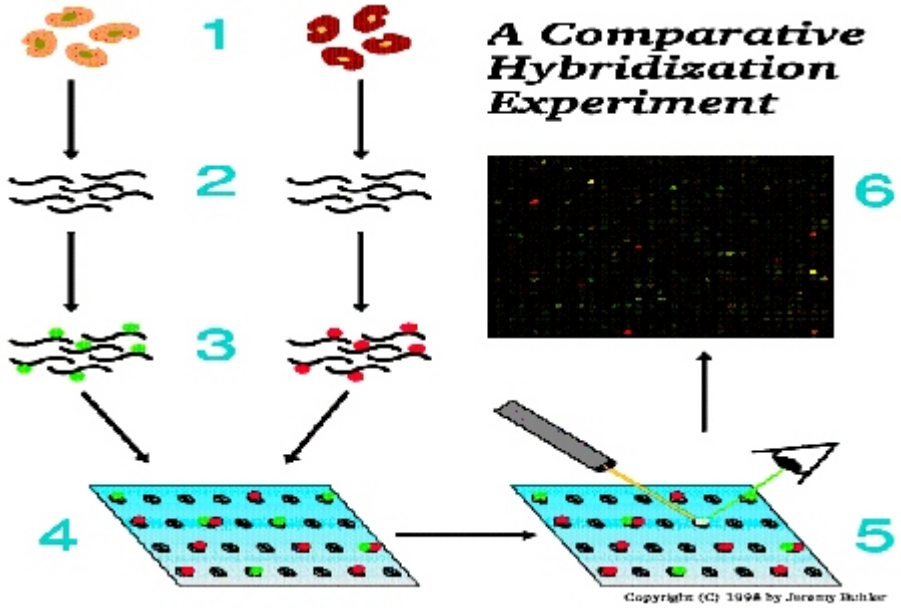


Micro array

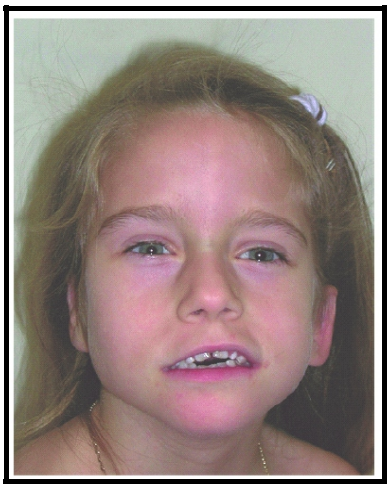
Array CGH, a new method to search for microdeletions.

Over the last two years, our cytogenetics research laboratory has worked very hard to develop and improve a method to identify microdeletions. This method is called "array CGH." "Array" refers to a glass slide with many little spots of DNA ordered in rows (see figure). This is a very efficient way to study many parts of DNA at the same time.

"CGH" is an abbreviation for Comparative Genomic Hybridization. "Comparative," because DNA from a person without the disorder in question (control) is compared to DNA from a person with the syndrome or disease (patient). "Genomic" means that we are looking at all of the DNA at once, not just the DNA of one chromosome or only DNA that is active in a cell at a particular time. "Hybridization" refers to the tendency of DNA to stick to (hybridize with) its counterpart. If a spot on the array contains a piece of DNA that originates from a specific point on, let us say, chromosome 1, the DNA from exactly the same location on chromosome 1 of the control person and of the patient will both stick to this spot.



Schematic representation of array CGH procedure



We first made an array with 3600 little spots of DNA from all over the chromosomes. We purified DNA from controls and labeled this with a fluorescent color (green). We also purified an equal amount of DNA from a child with CHARGE syndrome and labeled this a different color (red). Spots in the array with equal amounts of control and CHARGE DNA will show up yellow (an equal mixture of fluorescent green and red will appear yellow). If a piece of DNA from the child with CHARGE is missing, then there will be more green (control) than red (CHARGE) labeled DNA and the corresponding spot will show up as green. On the other hand, if the child with CHARGE has a duplication of a tiny part of a chromosome, the corresponding spot on the array will show up as red, since there is more CHARGE DNA compared to control DNA. This is shown in the figure on page 4. The control DNA is on the left, the CHARGE DNA on the right.

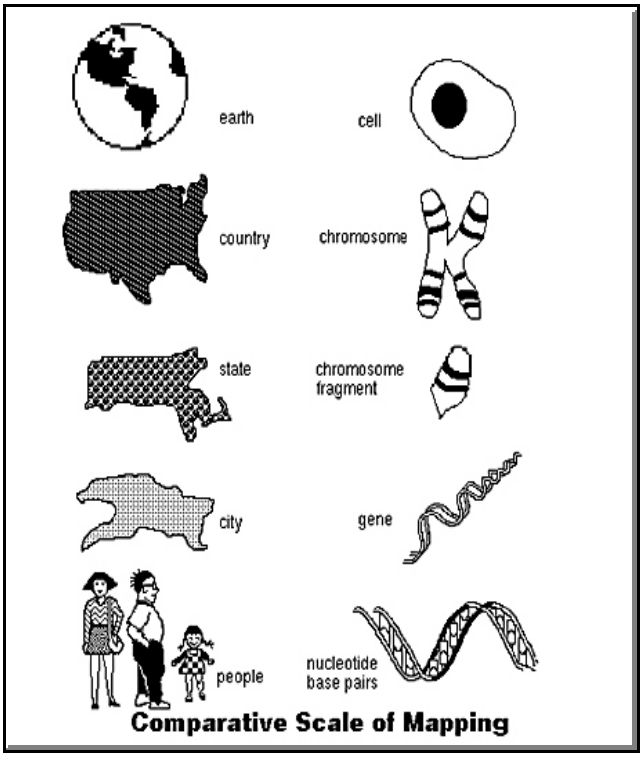
The first microdeletion found.

With our new technique (the array with 3600 tiny spots of DNA) we identified a deletion on chromosome 8 in S, a girl with CHARGE syndrome and severe mental retardation. We did not find the same deletion in any other patients with CHARGE, but it gave us a place to start looking for changes in genes that might be associated with CHARGE. We made a new array just for chromosome 8, with 918 very little pieces of chromosome 8 DNA. With this array, even smaller chromosome 8 deletions can be detected. However, still none of our patients, except S, showed a deletion with this very high-resolution array.

We did not give up and searched the literature for children with CHARGE with chromosome 8 aberrations and found a paper written by Jane Hurst in 1991. In this paper an English child with CHARGE syndrome and an apparently balanced translocation between chromosomes 6 and 8 is described. A translocation is an exchange of two pieces of chromosome. "Apparently balanced" means that by looking through the microscope it appeared that no part of either chromosome 6 or 8 was missing. We asked Jane Hurst to send us DNA from this patient. Our tests on this DNA showed nearly the same microdeletion of chromosome 8 as in our own patient with the high-resolution chromosome 8 array! This confirmed that there must be a very important role of this specific region of chromosome 8 in CHARGE syndrome. But what is wrong in the DNA of all the other patients with CHARGE without a chromosome 8 microdeletion?

The search for genes on chromosome 8.

By comparing the deletions of the two patients, we defined a stretch of DNA of 2.3 Mb (2,300,000 basepairs - a basepair is the minimal building block of DNA) that was missing in both patients. At the time, this stretch of DNA was known to contain eight genes. We sequenced the DNA of all 8 genes in this region in 17 patients with CHARGE who did not show a microdeletion of chromosome 8. Sequencing means that the make-up of the DNA in the entire gene is decoded. To our disappointment we did not find anything. None of the eight genes showed any changes in any of the patients.



However a few weeks later Lisenka studied an up-dated map of chromosome 8 gene information that became available on the Internet late 2003. She found that two additional genes were predicted to be in our chromosome region of interest. She compared these human genes to equivalent genes in the mouse and found that actually it was just one large gene, which in the mouse is expressed in the ear, heart and brain during early embryological development. Well, that was promising. It is a large gene and sequencing it in our 17 patients took a lot of time and effort. ***But was worthwhile because we found changes in this new gene in 10 out of the 17 patients!*** Since the paper was published, we have tested 28 individuals with CHARGE and found mutations in two–thirds of them (19).

In which persons did we look for CHD7 mutations?

For the purpose of this study, I asked Ronald Admiraal, the ENT specialist, to select children with an abnormality of the semicircular canals as part of their CHARGE diagnosis. This vestibular abnormality is very common in children with CHARGE syndrome, but is also seen in children with other syndromes. So at this moment we do not yet know if mutations can also be found in the people with CHARGE without a vestibular malformation. However we already know that the

spectrum of CHARGE features due to CHD7 mutations is very broad. For instance one of our children did not have coloboma or choanal atresia, but still had a CHD7 mutation. We have also found a CHD7 mutation in a 26 year old woman with normal intelligence, visual field defect, heart malformation, facial nerve palsy and subfertility. She told us that she had problems with her balance and she appeared to have coloboma on ophthalmologic investigation.

How do we know that this gene really is causing CHARGE syndrome?

We tested DNA from the parents of the children with the CHD7 mutations. If the changes we saw in this gene did not cause CHARGE, we would expect them in at least some of the parents. We did not find any changes in the DNA of this gene in any of the parents. That means the change in the DNA on chromosome 8 is de novo – new in the child with CHARGE. The change must have arisen during egg or sperm formation. The fact that all the changes were de novo and that the gene has a presumed function during early embryogenesis is convincing evidence that this gene is really involved in CHARGE syndrome.

Why is this CHARGE syndrome gene called CHD7?

I must admit that this sounds like an awkward or random name. However, scientific protocol insists that we not give the new gene a name. We had to ask an international committee what this gene should be called. CHD7 means that it is the seventh gene within the family of "chromodomain" genes that has been discovered. Thus there are already six other human genes that appear to be similar in function to the CHARGE gene.

What is the function of CHD7?

Now I need to be a bit speculative, since there still is a lot that has to be studied. We know that CHD (chromodomain) genes code for proteins that play an important role in the regulation of the expression of other genes. The chromosomes in each of our cells contain approximately 35,000 genes. Every cell does not use every gene. For example, some genes are needed in skin cells, while others are needed in the thyroid gland. Some genes are only needed during early embryonic development, others later in life. Some embryonic genes help orchestrate proper development of very sophisticated organs like the heart, eye and semicircular canals. The CHD family of genes play very important roles in regulating which genes need to be used at which time, especially in early embryonic development. It will be years before we fully understand the role of such regulator genes. But now we have a place to start.

Can CHARGE syndrome also be due to mutations in other genes?

So far, we have found changes in the CHD7 gene in about two-thirds of patients tested. To be certain that we did not miss anything we plan to repeat the analysis on new samples from these patients. The other third of patients definitely have CHARGE -- it is likely that there are more genes that can cause CHARGE syndrome and our search still continues. (Ed. Note: Many other researchers will continue to look for additional CHARGE genes.)

What does DNA diagnostics contribute to our knowledge of CHARGE syndrome?

At this moment we have only performed DNA testing in a small number of children with CHARGE. But I have already learned a lot. For example:

1. It confirms that normal intelligence is entirely consistent with a diagnosis of CHARGE syndrome.
2. CHARGE is extremely variable – even children who do not have colobomas or choanal atresia can still have CHARGE syndrome. At this point, we do not know if children without the vestibular anomalies have mutations in the CHD7 gene

We hope that finding this gene will help us learn more about the clinical spectrum in CHARGE syndrome with CHD7 mutations. It is also not clear yet whether a relationship exists between the type of mutation and the type or severity of the clinical symptoms. There are likely to be other genes that, if mutated, can also cause CHARGE syndrome.

Can CHARGE-syndrome be cured now the gene is known?

No. I realize this is a great disappointment for all of you. The gene has its main influence during the first 12 weeks of embryonic life. Any organ that is not formed properly at that time due to a mutation of the CHD7 protein can not be repaired afterwards.

Future research

A very big step has been made in CHARGE research. However, there is still a lot of research to be done. The Nijmegen research group and I assume many other research groups will collaborate in this, plans to study the following:

1. Is there a phenotype-genotype relationship? (Do the particular changes in the CHD7 gene predict particular medical problems?)
2. Is there a difference in children with CHARGE who do and do not have a CHD7 mutation?

3. How broad is the clinical spectrum of children with CHD7 mutations?
4. How is the puberty development in children with a CHD7 mutation?

And many other questions that undoubtedly will come onto our path.

Is a blood test already available? Can you test my child?

Yes. Immediately after we discovered the CHD7-gene we implemented the sequencing of this gene into our DNA diagnostics laboratory and told this to all Dutch clinical geneticists. So, in the Netherlands diagnostics is offered on the same conditions as for other genetic disorders in which the gene is known. For the countries that do not have the facilities (yet) the laboratory in Nijmegen is willing to offer the test. *If you and your doctor want this, please ask your doctor to contact me by e-mail. The costs are 650 euro (currently about $800 US, $1000 Canadian).* This may seem very high, but the CHD7 gene really is a large gene and since there are no so-called "mutation hot-spots" it needs to be sequenced completely.

What if a mutation is found in your child?

The next step we recommend is testing both parents (remember, the charge for each person tested is 650 euro). Although we do not expect to find mutations in parents, we don't know what the mild end of the CHARGE spectrum is like, so it is important to rule out a mutation in the parents. If your child has a CHD7 mutation that is de novo (not present in either parent), than we may conclude:

1. Your child definitely has CHARGE syndrome. As you know, in some children, the clinical diagnosis is uncertain. It is helpful for parents and professionals to have a definitive diagnosis.
2. Your unaffected children do not have an increased risk of having a child with CHARGE syndrome.
3. Your recurrence risk remains 1-2% for another child with CHARGE syndrome. This is due to what we call germ line, or gonadal mosaicism. In most cases, the CHD7 mutation occurred in the single egg or sperm that created your child with CHARGE. In a small number of cases, the mutation occurred in the cells that make eggs or sperm (gonadal cells). In those few cases, there could be other eggs or sperm carrying the same CHD7 mutation. The good news is that once a specific mutation is identified in a family, reliable prenatal diagnosis (of the presence or absence) of that CHD7 mutation is possible with chorionic villus sampling (CVS) or amniocentesis. Genetic counseling is recommended when considering these options.
4. If he or she is able to have children, your child with CHARGE will have a 50% chance of passing on the CHD7 mutation with each pregnancy. Again, prenatal diagnosis is possible. We are not at a point where we could predict the severity of CHARGE in a child who inherits the CHD7 gene.

Finally . . .

I want to thank all the children and parents who have helped us with our research. I also want to thank the Dutch CHARGE syndrome support group for their faith in us. And, of course, I want to thank all my collaborators with whom I have shared a very exciting time.

Please feel free to contact me in case you or your doctors have any questions. This story will undoubtedly be continued . . .

Warm greetings to all of you!

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We've found a gene for CHARGE -- So Now What??

As usually happens, the parents got the word out about the gene first. We are delighted that Dr. van Ravenswaaij has written such a comprehensive article on the search for and discovery of this first major gene for CHARGE syndrome. We think it is important to make a few things clear.

First, this is the FIRST major CHARGE gene to be isolated but it undoubtedly will not be the last. Let us illustrate by telling you about Sandra's experience with Usher Syndrome (another deafblind condition). The first gene was discovered in France. When other labs around the world started checking their own patient samples, they could not find the gene. Later, two more genes were discovered on a different chromosome. It turned out that the French gene really was a "French gene" for Usher syndrome. That means that the only people who had that gene came from or were descended from people who lived in a particular area in France. Now we know of 11 different genes for Usher Syndrome and more will undoubtedly be discovered. There are not significant clinical differences between people with these different genes.

CHARGE is a very complex syndrome, much more so than Usher. Many organs are affected and each one can be 0-100% affected. There is no one feature seen in every person with CHARGE. This means that lots of combinations exist. Dr. van Ravenswaaij works with Dr. Admiraal who is an ENT interested in the vestibular system of these children. All 17 of the children studied had vestibular abnormalities. There are many kids who meet the criteria of CHARGE (have a diagnosis of definite CHARGE syndrome) who do not have vestibular abnormalities. What we are getting at is that the Dutch group looked at a particular population of CHARGE. We don't know whether people with CHARGE with vestibular anomalies are different from those without vestibular anomalies. Of the patients they studied, two thirds had an abnormality of the CHD7 gene. This means that one third of the patients did not. Therefore, there will be more genes discovered later.

The Dutch discovery of a new gene for CHARGE is indeed exciting. It confirms what we, Bonnie Pagon and others have been saying for 20 years. We believed that CHARGE syndrome was genetic but it was just a matter of time until this was proved. This also puts to rest the issue of syndrome vs. association since the first gene has been discovered. Now other geneticists will come on board, too, using the term syndrome.

What does this mean to the families? In other words, so what? What will knowing that your child has the gene do for your child, your family and your child's future?

Confirm the diagnosis

If the diagnosis of CHARGE is uncertain in your child, finding a mutation would confirm the diagnosis. However, not finding a mutation does not rule out CHARGE because at least a third of children with CHARGE do not have a CHD7 mutation. Before considering testing, check with your medical insurance carrier. Many companies will not cover costs of tests at overseas labs, even if it is the only place in the world offering testing. Although 650 euro is relatively inexpensive for DNA sequencing, it's still a lot of money.

Prenatal diagnosis

The most immediate benefit could potentially be prenatal diagnosis for future pregnancies. IF your child has an identifiable mutation, prenatal diagnosis would be very accurate and in most cases would reassure families that the next child does not carry the same CHARGE gene. But what if the next baby does carry the mutation? CHARGE is so variable, that the presence of the mutation tells us nothing about the severity of CHARGE in a pregnancy. If the child with CHARGE does not have an identifiable mutation, then prenatal diagnosis would still not be available.

Medical and educational value – does finding a gene help treatment?

No, the discovery of a CHARGE gene and confirming a mutation in your child is not likely to lead to any changes in his or her medical or educational programs. We will need to look carefully at the medical and educational histories of the children who have the confirmed gene change and, particularly if there are older children or adults, we might eventually be able to say something about what to expect in the future given particular mutations. That kind of information will take many years to accumulate.

Can we now cure CHARGE?

Does finding the gene also mean finding a cure? NO. This is a long process. First, researchers have to figure out what the gene does. Dr. van Ravenswaaij has said that it appears to be involved in early (prenatal) development. That has been what geneticists have postulated for two decades now, so that makes sense. If this is a gene that acts in early development, there is now, and for the foreseeable future, no way to correct the gene abnormality. Much more research needs to be done. This means years not weeks or months.

Should I send blood to the Netherlands?

If, after reading all of this information, you are interested in pursuing gene testing for your child, go ahead. The group in the Netherlands is offering a very good test at a very reasonable price. Your insurance may not pay for it, so be prepared for a big bill. You may want to discuss it with your child's pediatrician and/or geneticist. Please remember, it may not answer all of your questions.

What about the Baylor research?

As many of you know, Dr. Seem Lalani and Dr. John Belmont and their team at Baylor University in Houston, Texas, have been searching for a CHARGE gene for several years. They have DNA from blood samples on 96 triads (individuals with CHARGE and both parents), drawn at the 1999 Houston CHARGE conference and later. They are also very excited about the CHD7 discovery, even though it happened in another lab. They are already in the process of screening their samples for changes in the CHD7 gene. They are using a slightly different process from the Dutch group. It is a little faster, not quite as labor-intensive (read: expensive) and is usually nearly as accurate as sequencing the entire gene. So far, the positive rate at Baylor has not been as high as in the Netherlands. Those of you who have sent blood to Baylor can expect to hear from them in the next few months with results of their screening. They will tell you whether or not they were able to detect a CHD7 mutation in your family. The Baylor group will also continue looking for other CHARGE genes. Finding a major CHARGE gene opens up may areas of research. If an animal model can be found (e.g. a mouse with CHARGE-like features), even more research can be done.

Will Baylor be offering clinical CHD7 mutation testing?

Not any time soon. For Baylor to offer CHD7 testing as a clinical test (as opposed to a research test), they have to go through a very lengthy approval process through the University. With current rules on privacy and so on, Dr. Belmont expects the approval process could take a year or more. And if it turns out full DNA sequencing is necessary, the charges at Baylor would be likely to be about twice as high as those in the Netherlands. That's why we are calling an $800 test "very reasonable."

For those of you that would like more information about chromosomes and genes, check out
http://ghr.nlm.nih.gov/

and look under "Help Me Understand Genetics." CHARGE is not yet one of the diseases referenced in this new online service because no genes had been discovered until now.

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A RESEARCH STUDY UPDATE ON "Osteoporosis in Adolescents and Adults with CHARGE Syndrome"

As you may be aware, a number of families have expressed an interest in having Osteoporosis in Adolescents and Adults with CHARGE Syndrome updates. Karen has put together this little blurb and was hoping it could be posted on the websites.

It is becoming increasingly evident to parents and health care workers caring for individuals with CHARGE syndrome that the bone health of these individuals is often neglected. Adolescents and adults with CHARGE syndrome appear to be at increased risk for developing frail bones and low bone density which, if left untreated, may lead to the development of osteoporosis.

At the IWK Health Center in Halifax, Nova Scotia, Canada, we are currently examining the relationship between osteoporosis and CHARGE syndrome in adolescents and adults. If you know of an individual (age 13+) who would like to participate in this study, please contact Dr. Kim Blake (kblake@dal.ca) or her research assistant, Karen Forward (keforwar@dal.ca). The study is questionnaire based; however, we would be very interested in receiving bone scan results (DEXA scan results) and hormone reports if these have been completed. The study protocol and the questionnaire were approved by the Isaac Walton Killam (IWK) Health Center Ethics Review Board and by the CHARGE Foundation USA Advisory Board. Thank-you to all those who have completed the questionnaire thus far, your interest and participation in this research has been vital.

Karen Forward

California Deaf-Blind Services Fact Sheet #35

Suggestions for Creating Successful Transitions from School to Adulthood

by Maurice Belote, CDBS Project Coordinator

While this is not an exhaustive list of steps towards successful transitions, it represents a few of the things I've learned over the years.

Mind the gap. The subways in London remind you, as you step off the trains, to mind the gap—the space between the subway car and the platform. In the same way, mind the gap between the end of a school career and the beginning of adult services. We know that the longer the gap in services, the greater the likelihood that persons who are deaf-blind may not have meaningful employment, adequate housing services, and/or community access to recreational and social opportunities. For example, if you can find a permanent job placement for an individual a few months before that person would otherwise age-out of special education services, why not take it? If the IEP can be modified so that services can be provided in this new environment, all the better. But if it can't, don't regret the little bit of missed school. A seamless transition into adult services may be more important than those last few weeks of school. Of course, major transitions cannot be rushed but must be thoughtfully planned so the individual has time to prepare for the changes.

Plan early. The law states that at age 14 IEPs must include transition service needs and at age 16 IEPs must contain needed transition services. (Yes, even educators are confused by this wording.) Don't let this requirement be satisfied with the attachment to the IEP of a single sheet of paper with a few boxes checked. By this time in students' lives, educational programs should be leading to clearly defined outcomes. All components of educational programs should be preparing students for success beyond school—at home, at work, and in the community (see next paragraph).

Does every step lead towards the desired outcome? Ask yourself at IEP meetings: does each goal and objective move this child towards a concrete and functional outcome? If a student is 20 years old and hasn't mastered tying shoelaces after years and years of trying, let it go; the student will probably be just as relieved as you are. The same goes for writing a signature, spreading on bread, or any other skill that has been worked on for years with little or no success. There may be other things for the child to learn that are more important, such as personal hygiene skills. Employment and housing personnel report that this is one area they would really like the persons they serve to take care of themselves—if they can. And remember the importance of cleanliness when it comes to social interactions (see next paragraph).

The importance of social skills. Social skills are just as important—if not more important—than competence. People will put up with a lot of incompetence if you have good social skills. Think about your own experiences. Have you ever worked with someone who, although he or she wasn't the hardest worker at your place of employment, was friendly, brought fresh-baked cookies on Fridays, told good jokes, or pitched in for the office parties? Imagine that same person, who wasn't the hardest worker, if he or she hadn't contributed positively to the work environment. Stopping at the donut shop once a week on the way to work to bring a box of donuts to the office may contribute more to longevity and social relationships than performing flawless work tasks day after day.

Document everything. It is important to document everything that might someday be necessary to know. This includes tasks at which the person who is deaf-blind excels, their expressive and receptive communication systems, preferences and dislikes, favorite leisure time activities, etc. This documentation will be useful as video resumes and/or personal communication dictionaries are compiled. Consider the following example. A student paddles a

kayak across a lake at age 16, has a great time, is good at it, and then doesn't have the opportunity to do it again for years. By the time the student is 22 years old, will anyone remember this event and the fact that kayaking might be a great recreational activity for this person? They will if it has been documented. This can be accomplished with videotape, photographs, journal entries, or any other method that works for those involved.

The "readiness model" might impede success. There was once a belief that students had to prove they were ready for jobs, living situations, etc. by demonstrating readiness. Consider the following example. A student wants a work experience placement at a plant nursery watering plants. Under the readiness model, the student would have to prove his or her readiness by successfully watering plants in the classroom for a period of time, which would then be followed by a trial placement watering plants on the school grounds. If all of this goes well, the student would then graduate to watering plants at an actual nursery. The problem with the readiness model is that the student may never get past watering in the classroom for reasons that have nothing to do with the ability to water plants. Perhaps the student is bored with the classroom because he or she has spent too many years there. The student may be loud and unfocused while watering in the classroom, and the assumption is that the student will behave in a similar way out in the real world. But given the opportunity to do this job in a natural environment, the same student might very well succeed. The student's behavior might have been saying "I'm sick of the classroom", but in a real environment with natural motivators and consequences, the student may pleasantly surprise the doubters.

It's all about who you know. It's true that much of what we have in life, e.g., jobs, apartments, significant others, we got through someone we know, or through someone who knows someone we know. For example, when considering work experience placements for students, think about people you know who have small businesses such as restaurants, hair salons, offices or warehouses. When looking for apartments, think about people you know who live in desirable buildings and may know of unpublished vacancies, or people you know who work as property managers or real estate agents. Even if it's a friend who knows someone, have him or her make an initial call on your behalf. It will make your subsequent call much easier and will probably make the person more interested in what you have to say because they know you're a friend of a friend. This is something we need to learn from people in the private sector who practice this well: never underestimate the power of personal contacts and connections.

Get the relevant facts. Make sure you know everything there is to know about the individual who is deafblind: likes, dislikes, activities in which he or she excels, dreams, fears, social connections, and anything else that might impact future success. Gathering this information might be accomplished though processes such as personal futures planning, MAPS, person-centered planning, etc. Parents, siblings, extended family members, neighbors, and former teachers are all vital sources of useful information. These same people are also vital sources of information about interpreting the individual's wishes if the person has limited formal communication skills.

Fact sheets from California Deaf-Blind Services are to be used by both families and professionals serving individuals with dual sensory impairments. The information applies to students 0–22 years of age. The purpose of the fact sheet is to give general information on a specific topic. More specific information for an individual student can be provided through individualized technical assistance available from CDBS. The fact sheet is a starting point for further information.

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THE FOUNDATION OFFICE HAS MOVED!

Our new address is 409 Vandiver Dr Ste 5-104
Columbia MO 65202-1563

Telephone 573-499-4694 voice/fax
1-800-442-7604 for families

The office will be staffed Monday Wednesday Friday 9 am to 1 pm CT
The answering machine is always available

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Calendar

December 1, 2004	**Deadline for submitting items for Winter 2004 *CHARGE* Accounts**
Late December	**Winter Issue of *CHARGE* Accounts**
June 3-7, 2005	**The 8th Helen Keller World Conference and the World Federation of the Deafblind 2nd General Assembly Tampere, Finland (www.helenkeller2005.com)**
July 22 - 24, 2005	**7th International *CHARGE* Syndrome Conference Miami Beach, Florida**