

WOLF HIRSCHHORN SYNDROME 4p- BOOKLET

The diagnosis of Wolf Hirschhorn Syndrome is made when blood has been taken from a newborn baby to try to discover why he or she has a number of unusual physical features. If a baby is found to be lighter than expected or to have a shape of face very different from the parents, a close examination may show a number of different variations. There may be unusually shaped ears, hands or genitalia and there may be evidence of defects in the heart or palate. The name `Syndrome` is used when there is a cluster of these abnormalities. Before it was possible to examine the chromosomes, a number of syndromes were described and then the proof depended upon whether there were a reasonable number of the described features in any individual. Since the 1960's it has been possible to count the chromosomes and later developments have allowed individual chromosomes to be examined very carefully. In Wolf Hirschhorn Syndrome there is a small piece of the short arm of chromosome 4 missing.

It is very difficult to understand this unless we know more about chromosomes. Let's start from the beginning. We are made up of millions and millions of tiny cells. These *bricks* which build up our body can only be seen under a microscope. Different parts of the body have different types of cells, but even if the cell is a tireless heart muscle cell or an inspired brain cell, the centre of the cell is very similar. This centre is called the nucleus. The nucleus is made up of an amazingly neat chemical called desoxyribose nucleic acid (DNA). This DNA is a long spiral shaped molecule which contains all our genes. Genes are the bits of information which tell our developing bodies exactly what shape to be. Put scientifically, the genes which are coded in the DNA sequence control the types of chemicals which build up the shape and size of every part of the body. Try to imagine that the DNA in every nucleus is like a tape bound in a cassette. The words on the tape could be commands which tell people what to do. If the instructions coming from the tape were telling someone how to construct a car or computer, just imagine what would happen if a piece of the tape were damaged or missing. This is roughly what happens in conditions like Wolf Hirschhorn Syndrome. Here some of the DNA is missing. So what are chromosomes?

When cells divide the DNA gathers up into convenient rolls like skeins of wool. The tangles of DNA are called chromosomes and they are of different lengths. They gather in 23 pairs and are numbered from 1 to 22 with 1 being the largest. The 23rd Pair are the sex chromosomes and are called XX (girls) and XY (boys). Wolf Hirschhorn Syndrome affects chromosome 4. This is a large chromosome but it is only the short arm which is affected (Chromosomes have a lump near the middle and the pieces either side are called arms, since the lump is not exactly in the centre there is a short arm either side of it). This diagram might make that clearer:

Occasionally this missing piece of chromosome is stuck onto another chromosome (This is called translocation). A few parents have a balanced translocation meaning that they have the correct amount of DNA and therefore haven't lost any genes. However, when the cells divide up their chromosomes to form their eggs or sperms the resulting offspring may have that short chromosome and not have the piece which is stuck onto the other chromosome. Now this is very complicated so perhaps this diagram will help:

Most parents have normal chromosomes and the chances of having another baby with the same condition is rare, but if a parent has a balanced translocation of the short arm of chromosome 4 then there is a much higher chance of another baby being affected. Your Genetic Counsellor will give you more information about this. Some parents feel very upset when they find that it was their chromosome which led to this condition. It is most unfair to blame themselves for this since we have no idea ourselves about how our chromosomes are arranged. There is no way in which we can change this nor was there any way in which we could have caused it to happen.

This might be a good point to look at a philosophical point. When the difficult news is broken to parents that their little baby has such a major problem which will severely affect his or her future, a number of parents feel a great bitterness about the whole design of the universe as well as the sadness and bereavement for the baby they were expecting to have. They may feel that it is totally unfair that chromosomes should be so weak that they can break or get damaged in this way. They may feel that the whole of life has ganged up against them to produce such bad luck. This is very difficult and can distance them from supportive family or friends especially if linked to religious groups. It seems that we could not have developed into such complicated but remarkable beings if it hadn't been for occasional breaks in the chromosomes way back at the dawn of life. Spontaneous changes in the chromosomes which led to alterations in the shape or activity of life forms were probably the way we developed from the original one cell creatures. Perhaps it is at least some consolation that the problem is caused by the very activity which allowed us to evolve to where we are today.

Every child with Wolf Hirsch horn Syndrome is different but they share a number of characteristic. It will be most helpful to consider these in the order of importance to the child at various ages. If we summarise these in this table we can put in a brief check on what treatment or other activity might be needed.

Problem	Effect on child	Treatment or Activity
Chromosome 4	Wolf Hirschhorn Syndrome	Genetic counselling
Small size before Birth	Probably easier birth	Early sign of the Syndrome
Small jaw, downturned mouth, cleft lip/ palate in some	Feeding and breathing difficulties	Help with feeding/sleeping position
Wide spaced eyes with squint or iris defect	Possible vision problems	Detailed eye examination. Vision checks when older
Small & unusually developed Brain	Major learning problem	Special education
	Tendency to convulsion	Anticonvulsant drugs
Floppy muscles	Movement difficulty Poor cough therefore Chest infection risk	Physiotherapy Antibiotics sometimes
Heart defects in Some	Depends on severity	Difficult decisions about possible operations

This list is rather daunting but it may help in making sure that your child has had all these areas checked. The severe learning difficulties and the muscle weakness are the most difficult problems. There is no medical treatment which can improve these and it is these which are going to restrict his or her opportunities. It is also these which increase the risk of chest infections and ultimately reduce the life expectancy.

There is no information about average lifespan but if the child has no severe heart defects and does not have serious chest infections or uncontrollable fits there is no reason why they cannot survive into adulthood. If there are no major medical problems with the heart, lungs or with controlling fits then decisions have to be made on just how intensive treatment should be. We have to consider philosophy (ethics) again. If a treatment is necessary to sustain life but the treatment itself may be very burdensome to the patient then a decision must be made on whether it is in the patient's best interest to be treated. Usually it depends on the quality of the life which has been extended by this treatment. Quality of life is difficult to measure but is usually defined as the sort of life which the average person would like to have. Usually this is one which has the minimum of pain or distress and the maximum of opportunities to enjoy pleasure in its widest sense. Difficult decisions about when to stop active treatment in those who are very elderly, those with advanced cancers, those with major brain damage after accidents and also those born with major congenital malformations are based on this type of thinking.

Doctors will try to explain the nature of any major treatment or operation and how that might either extend or enrich the life of their patient. They will also give details of how burdensome or unpleasant the procedure might be. They will usually go through the balancing of the factors for and against an active treatment with the parents. Most doctors will state what they would prefer to do in the particular circumstance. In Wolf Hirschhorn Syndrome, the child will never be able to understand enough to make a decision like this so parents have the right and responsibility to follow the doctor's advice or not. In most situations these decisions are made with the parents, the child's doctors and nurses in a team. Usually there is plenty of time to discuss it with the family, friends and religious advisers where available.

It would be most inappropriate to finish at this sad point although it is essential to consider these problems long before they arise even if this is painful. It also helps to have a realistic view of the child with Wolf Hirschhorn Syndrome rather than try to avoid or deny it. If parents build up a fantasy idea of their baby then they may miss the reality of the character of their child which in spite of all the problems will shine through.

Many of us who work with children with major disabilities and their families are inspired by what we have the privilege to share. We see the way that the families cope with their original sorrow and worries and the gradual realisation of the strong human qualities which show through the child's many limitations. However, this can only happen if the families have enough support from both their local community of family and friends as well as contacts with professionals. These professionals may be very daunted by coming across a child with such a rare syndrome and may be very embarrassed by their lack of knowledge. We do not expect doctors even working exclusively with children to have details of Wolf Hirschhorn Syndrome in their heads. Parents should not be surprised by this nor put off by the professional's reaction to their lack of knowledge. Maybe some of the details in this paper might help them before they have had a chance to look up Wolf Hirschhorn Syndrome 4p- in the textbooks.

As this is so rare, it is very helpful for families to keep in touch with others who have similar children. Please remember that, even though they have many features in common, every child has a different combination and severity of problems.

"Having read the medical side of Wolf Hirschhorn Syndrome, please look at this smile and the smiles on the faces of the children on the front cover.

This is Steven, my son. He is a very loveable, happy young man who tries hard to achieve the skills that we take for granted. My family and I love Steven very much and we are proud of him.

There can be difficult times, but many happy ones too - you can see that from the smiles."

*Mrs Christine Hilder
Founder
Wolf Hirschhorn Syndrome Support Group*

WOLF HIRSCHHORN SYNDROME SUPPORT GROUP

This is a small self-help group; it offers:

- Contact with other parents with affected children
- Information about the condition

Activities

- Liaison with doctors and other professionals
- Compilation of information about the Syndrome
- Bi-annual National Meeting

Publications

- Newsletter (three times per annum)
- Website
- Booklets about Wolf Hirschhorn Syndrome.

Subscriptions

- £20.00 per annum (voluntary)