What we see is made in the brain from signals given to it by the eyes.

What we see is in fact made in the brain. The brain makes sight from signals given to it by the eyes.

What is the normal structure of the eye?

The eye is made of three parts.

- A light focussing bit at the front (cornea and lens).
- A light sensitive film at the back of the eye (retina).
- A large collection of communication wires to the brain (optic nerve).

A curved window called the cornea first focuses the light. The light then passes through a hole called the pupil. A circle of muscle called the iris surrounds the pupil. The iris is the coloured part of the eye. The light is then focused onto the back of the eye by a lens. Tiny light sensitive patches (photoreceptors) cover the back of the eye. These photoreceptors collect information about the visual world. The covering of photoreceptors at the back of the eye forms a thin film known as the retina. Each photoreceptor sends its signals down very fine wires to the brain. The wires joining each eye to the brain are called the optic nerves. The information then travels to many different special ‘vision’ parts of the brain. All parts of the brain and eye need to be present and working for us to see normally.

What is Ritscher-Schinzel Syndrome?

Very rarely when a baby is growing in the womb not all of the parts of the body grow correctly. In Ritscher-Schinzel Syndrome a child is born with conditions of several different parts of the body. These include:
• Changes to the shape of bones of the face
• Poor or absent growth of some parts of the brain
• Problems with the heart
• Eye conditions

This syndrome is sometimes also known as the cranio-cerebello-cardiac, 3C or CCC syndrome. Cranio is a medical word for head and face. Cerebello is a medical word for a part of the brain that is often affected. Cardiac is the medical word for the heart.

What is the cause of Ritscher-Schinzel Syndrome?

The body has a “built-in” plan to make sure all the parts of the body work correctly. This plan is written in our genes. Genes are a chemical alphabet stored in the body. Every person has two copies of each gene. If both copies of a gene have a misprint in the chemical alphabet then a part of the body may not work correctly. A child with Ritscher-Schinzel Syndrome has both copies of an important gene misprinted. This leads to several different parts of the body not developing correctly while in the womb.

Most cases of Ritscher-Schinzel Syndrome occur by chance. Occasionally however the condition can occur as a result of the parents of a child being cousins or even a more distant relative. This is called ‘recessive inheritance’.

How does Ritscher-Schinzel Syndrome affect eyes and vision?

Ritscher-Schinzel Syndrome is a rare condition. There is very little information available on how it affects the eyes and vision. It is likely that not all children will be affected by all the eye conditions described in this document. It is also possible that other conditions that affect vision may occur in children with this syndrome.

Children with this syndrome may have poor vision for a number of different reasons. The conditions that have been described in children with this syndrome include:

• Coloboma
What is Coloboma?

Very rarely when a baby is growing in the womb not all of the parts of the eye grow. If a part of the eye does not grow then this is known as a coloboma. The parts that often do not grow include the iris, lens, retina or optic nerve.

How might coloboma affect a child's vision?

Coloboma can affect the eyes in different ways. It depends on which bit of the eye is affected and how much of it is missing. If only a small part of the iris is missing then the eye may work almost normally. Sometimes more important parts of the eye are affected. This may stop the eye giving the brain a clear picture of the visual world. This happens if a large part of the retina or optic nerve is missing.

What Is Glaucoma?

Eyes keep their shape by being pumped up with water. It is a bit like a football being pumped up and inflated with air. You can feel the ‘pressure’ in your own eyes by gently feeling them through your closed eyelids. The amount of pressure depends on the flow of water in and out of the eye. If the flow of water out of the eye becomes blocked the pressure goes up. High pressure in the eye can cause damage to the optic nerve. Glaucoma is when the optic nerve is damaged by high pressure in the eye. This can occur in children with Ritscher-Schinzel Syndrome.

How does glaucoma affect the eyes of children?

High pressure can slowly cause loss of communication wires (nerves) between the eye and brain. If the high pressure is not treated the optic nerve damage can become worse. As more communication wires are lost, less information about the visual world is given to the brain. This can cause visual impairment. Sometimes high pressure can make the eye painful and red. Treatment with drops and an operation can help lower the pressure. This makes the eye more comfortable and stops damage to the optic nerve.
What is Cerebral Visual Impairment?

Cerebral Visual Impairment (CVI) is a condition where some of the special ‘vision’ parts of the brain and its connections do not work normally. This causes visual impairment even though the eyes may be normal. Often children with CVI have clear vision but cannot ‘make sense’ of what they see. As the child grows older some of the visual difficulties may improve.

Often the best way to find out if a child has Cerebral Visual Impairment is by asking questions. A doctor can find out from the parents and teachers of the child what kind of problems they seem to be having. The questions are based on the visual difficulties that commonly occur in children with CVI.

What kind of visual difficulties is a child with CVI likely to have?

Most young children with Ritscher-Schinzel Syndrome and visual impairment feel their vision to be ‘normal’ as they have never known anything else but their own visual world. At first they assume that everyone else has vision the same as their own. They may not realise that other people see things differently.

Cerebral Visual Impairment can affect vision in many ways. All or only one of the special ‘vision’ parts of the brain may be damaged and to different degrees. If there has been lots of damage to many parts of the brain the vision can be very poor. However the child may still see movement and bright lights. Complete blindness due to brain damage is rare.

If there has been only a small amount of damage in only one part of the brain the visual problem can be mild.

Children with CVI may have difficulty with:

- Visual field loss
- Moving around safely
- Recognising objects and people
What is Visual Field Loss?

Visual field is the medical word for the full area that we can see: our visual world. If an area of our visual world is blurred or missing with the rest clear then visual field loss is present. It is due to damage to some of the special vision parts of the brain. The relationship between brain damage and visual field loss is all opposite to what you might think.

The right side of the brain makes vision for the left side of the visual world. If this side of the brain is damaged the opposite side of the visual world may not be seen. The left side of the brain makes vision for the right side of the visual world. If this side of the brain is damaged the opposite side of the visual world may not be seen. In the same way the upper part of the back of the brain is responsible for seeing the lower part of the visual world. A child with damage in this area will not see the ground when looking straight ahead. The child may then tend to trip over things.

Problems with getting around and recognising objects

The many different ‘vision’ parts of the brain combine together, to make two visual ‘systems’.

- The ‘dorsal stream’ helps children get around safely and quickly

One system helps the child to get around safely and quickly. It also helps the child pick objects up and avoid bumping into things and falling over. This visual system that tells the body how to get around is called the ‘dorsal stream’. It is called a ‘stream’ because it is a flow of information about the visual world from one place to another like water flowing in a stream. ‘Dorsal’ describes the part of the brain where the system is.

When the dorsal stream is damaged it is difficult to know precisely where things are in three dimensions. It can be difficult to:

- Use stairs without falling.
- Step onto pavements without tripping.
- Reach forward and grab a cup or handle.
Damage to the dorsal stream can also make it difficult to see a lot of different things at the same time. This means it can be difficult to find a toy on a patterned carpet or to see something that is pointed out in the distance amongst other things.

- **The ‘ventral stream’ helps a child recognise objects**

The other system, called the ‘ventral stream’ helps us to recognise faces, objects and places. Damage to this system leads to problems:

- Recognising familiar faces.
- Knowing what common everyday objects are.
- Losing the way in places that should be well known to the child.

There are a number of other problems that can occur in children with CVI who still appear to have good vision. These include:

- Difficulty remembering things they have seen.
- Difficulty imagining ‘seeing’ things in their minds.
- Some children’s vision can become ‘tired’ more quickly than others. This means that their ability to see can vary from one time to another.
- Difficulty reading. This can be due to lots of different reasons.

**What can be done to help?**

There are no medicines or surgical treatments that will fix or improve the special vision parts of the brain. There are lots of things that can be done to help children with Ritscher-Schinzel Syndrome and visual impairment see better.

We use our vision to get around, learn new things and to meet other people and make friends. It is important to consider what your child’s particular problems with vision might be.

If your child has been prescribed spectacles, contact lenses or a Low Visual Aid (LVA) it is important that they are encouraged to wear and use them. This will help your child see more clearly and ensure the vision parts of the brain grow and develop correctly.
Wearing a hat with a broad brim can also help reduce problems from bright light. If your child has been prescribed drops they should be used regularly as advised by the nurses and doctors at the eye clinic.

Problems at school may be due to some of the reading books being hard to see. This often means it takes longer and more effort to do the work. If the size of print is increased and words spaced out a bit more, many children will find schoolwork easier. Good bright lighting and crisp black print on a clean white background will also help. Sometimes placing reading books on a slope, which tilts the print towards the child, will improve reading speed as well.

All these ‘tactics’ can help to improve reading speed though not all children will benefit from them.

It is worth watching carefully to find out what the smallest toys are that a child can see and play with. Then try to only play with toys that are the same size or bigger. If your child has a part of the visual world missing it is important to know where this is. Then try to always place toys in the part of vision that is working. Placing one toy on a plain background will often help children see it more readily. Placing lots of toys of different size and colour close together on a patterned background can make them ‘invisible’ to some children with poor vision.

Recognising facial expressions can often be difficult. It is worth trying to find out at what distance facial expressions can be seen and responded to. Then always try to talk and smile from within this distance. This helps a child to learn what facial expressions mean and to copy them.

Both head movements and eye movements may be slow and difficult to control. Careful positioning of the head to prevent it falling to the side or falling forward can help a lot.

Infants and young children need to learn about the world around them. Visual impairment teachers, physiotherapists and occupational and speech therapists may all add to the child’s care and education. It is important to continue the programmes that they recommend. If the child is involved in family activities vision can improve and new skills can develop.

Even if a child has very poor vision many useful and practical things can be done to
improve the ability of the child to get around, interact with other children and learn. Advice can be given on ways to support your child by your VI teacher or habilitation specialist.