Medical information on **Achromatopsia (also known as Rod Monochromatism or Stationary Cone Dystrophy)**

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 focusing 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. There are two types of photoreceptors named by their shape when looked at in fine detail. They are called ‘rods’ and ‘cones’.

**Rod and cone photoreceptors are good at seeing different things**

Rods are good at seeing:

- things that move
- in the dark
- but only in black and white
- and in less detail.

Cones are good at seeing:

- things that are still
- in daylight
- in colour
- and in fine detail.

The covering of rod and cone photoreceptors at the back of the eye makes a thin film called the retina. The central bit of the retina is made up of cones. They help us see the central bit of vision that we use for reading, looking at photographs and recognising faces. The area of the retina around the central bit is made up of rods. The rods see the surrounding bits of vision and help us to walk around and not bump into things especially in the dark. 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 Achromatopsia?
Some children are born with cone photoreceptors that do not work correctly. This usually leads to blurred vision, no colour vision and a dislike for bright light (photophobia). The medical word for colour is ‘chromat’. If ‘A’ is placed at the beginning of a word it changes the word to mean ‘without’ or ‘no’. Achromatopsia means to see ‘without colour’ or ‘no colour’. The name of the condition tries to describe how a child with Achromatopsia might see. It is also known as Rod Monochromatism. Mono means one. This other common name also tries to describe how a child with Achromatopsia might see and what with. It means seeing in only one colour using rod photoreceptors.

The condition does not get any worse as the child grows up. Because of this it is often described as ‘stationary’. A medical word for a condition which a child is born with is ‘dystrophy’. Another common name for Achromatopsia is ‘Stationary Cone Dystrophy’. This should not be confused with ‘Progressive’ Cone Dystrophies where the condition may become worse as the child gets older. Progressive Cone Dystrophy is a different eye condition from Achromatopsia, Rod Monochromatism and Stationary Cone Dystrophy. But Achromatopsia, Rod Monochromatism and Stationary Cone Dystrophy do all refer to the same eye condition. They mean the same thing in different words.

What is the cause of Achromatopsia?
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 small part of the body may not work correctly. A child with Achromatopsia has inherited a copy of the same gene from each parent with a misprint and the wrong plan for making cone photoreceptors. This means that when a child is born the cone photoreceptors do not work. Most cases of Achromatopsia occur by chance however occasionally 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’.

This is only a brief summary of the genetics of Achromatopsia. There are exceptions to these general rules. This is why it is important that families with a child affected by Achromatopsia receive counselling from a specialist in genetics.

How is the diagnosis made?
Young children with Achromatopsia may develop:

- Fast ‘to-and-fro’ movements of the eyes called Nystagmus.
- ‘Roving’ eye movements where the eyes appear to slowly wander around not fixing and staying still on any objects.
- Blurred vision that is worse in bright light and better in dim light.
- Children may dislike bright light and try to avoid daylight and well-lit rooms. This is known as photophobia.

Parents will often notice these signs. Sometimes parents also notice (by the way their child acts) that their child’s vision is reduced. If they discuss their concerns with their Family Doctor an assessment can be arranged.

An eye doctor can check the way the eyes behave to bright lights. If the pupils of a child move slowly to a bright light then Achromatopsia is more likely. Using a special instrument the eye doctor can look at the optic nerve and retina at the back of the eye. In children with
Achromatopsia sometimes these parts of the eye look different from normal. Often the central bit of the retina (the macula) can appear to have circular bands of different shades of pink and orange. Eye doctors often describe this appearance as ‘Bull’s Eye Maculopathy’.

There are also special tests that can be done to help the eye doctor decide what is wrong. These tests measure signals from the eyes when a child is shown a bright light. Sticky patches are placed around the eyes. The sticky patches are attached to wires that lead to a machine. The machine records the electrical signals made by the eyes. The record of the signals will help the doctors decide what the matter is. Achromatopsia is more likely if the signals are weak or absent. This test is called an Electroretinogram (ERG).

How does Achromatopsia affect the way a child sees? Achromatopsia can affect different children in different ways. Some children have an ‘incomplete’ or ‘partial’ kind of Achromatopsia. These children tend to have slightly better vision when they grow up.

All young children with any kind of Achromatopsia will however feel their vision to be ‘normal’. At first they assume that everyone else has vision the same as their own, as they have never known anything else but their own visual world. They do not realise that other people see things differently.

Cone Photoreceptors are found in the centre part of the retina. This bit ‘works’ the central part of the visual world. Cones are good at seeing:

- things that are still
- in daylight
- in colour
- and in fine detail.

Because children with Achromatopsia have Cone photoreceptors that do not work they will have problems with:

- Blurred vision, especially in the central bit of vision
- Poor colour vision
- Dislike of bright light (photophobia)
- Fast to-and-fro movements of the eyes (nystagmus)

Most children with Achromatopsia can still see well in the outside bit of vision, away from the centre. They usually have no problem getting around although reading, recognising faces and seeing small toys can be difficult. Children with reduced vision often develop fast to-and-fro movements of the eyes. This is called Nystagmus and usually gets better as a child grows up.

Long-sight (hypermetropia) is common in children with Achromatopsia. This means that their eyes find it easier to focus on things in the distance than for near. Spectacles will often help to improve vision for near and far in children with Achromatopsia and Hypermetropia.

Is there any treatment for Achromatopsia? There is no good way to fix the Cone photoreceptors that are not working. But many things can be done to help children maximize their vision and fulfil their potential.

What can be done to help? We use our vision to get around, learn new things and to meet other people and make friends.

It is important, if children have been prescribed spectacles, that they are encouraged to wear them. Often a dark tint to the spectacle lens will help the child see more clearly and reduce glare and photophobia. It will also help the vision parts of the brain to grow and develop
correctly. Some children find wrap-around glasses very helpful. When a child is older tinted
contact lenses can be worn to help in the same way.

Sun hats or baseball caps with a large sun visor can also help to reduce photophobia.

Most children with Achromatopsia have few problems getting around. The way they act can
often give the impression that their vision is normal. It is important however to be aware of some
of their own personal difficulties with vision.

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.

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.

Many children with Achromatopsia need early and specialist care from the local visual
impairment support team. Even if a child has very poor vision many useful and practical things
can be done to help. Ask the visual impairment teacher or habilitation specialist for advice.