

## Laurence-Moon\_Bardet-Biedl Syndrome (LMBBS)

### WHAT IS LMBBS?

LMBBS is a rare genetic condition. The word 'Syndrome' is used to show that a diagnosis is made on the basis of a collection of characteristics (some of which are described below).

### HOW IS LMBBS PASSED ON?

The pattern of inheritance for LMBBS is autosomal recessive.

A person who inherits one faulty gene for LMBBS will be a carrier. Carriers are usually unaffected but can pass the faulty gene onto any children they may have. If one or both parents are a carrier, there is a 50% (1 in 2) chance that each child of theirs will also be a carrier.

A child who inherits two copies of the faulty gene (one from each parent) will have LMBBS. If both parents are carriers there is a 25% (1 in 4) chance of this happening.

### A PERSON WITH LMBBS MAY BE AFFECTED BY SOME, OR ALL, OF THE FOLLOWING:

- Eye problems leading to night blindness and poor vision (most young people with LMBBS will be registered blind by the age of 16).
- Extra fingers and/or toes at birth, ranging from a single skin tag to a fully-formed extra finger on each hand and an extra toe on each foot. Short, stubby toes and fingers can make balance and some manual tasks (such as using a keyboard or reading braille) difficult.
- It is difficult for people with LMBBS to use up calories efficiently so they are usually overweight.
- Mild to moderate learning difficulties and poor short-term memory (although long-term memory may be excellent).
- Sitting, standing, talking and walking are usually delayed in children with LMBBS, and there may be some behavioural problems.
- Clumsiness and poor coordination.
- The voice may be breathy and a high-pitched nasal tone.
- Problems with the kidneys and sometimes the liver.
- High blood pressure.
- Abnormally small genitals
- Pain in weight bearing joints.

### OTHER INFORMATION

LMBBS is relatively common in certain regions of the world (1 in 13,500 of the Bedouin population in Kuwait, and 1 in 17,500 in Newfoundland). This may be partly because interfamily marriage has increased the chance of both partners sharing a greater number of genes. In Britain, it is estimated that 1 in 125,000 of the population have LMBBS.

LMBBS is named after the doctors who first described the condition –Laurence and Moon in the 19th century, Bardet (pronounced Bar-day) in France in 1920 and Biedl (pronounced Bee-die) in Austria in 1922. LMBBS cannot be cured, but extra fingers/toes can be removed in childhood and obesity controlled with careful eating and regular exercise. People with LMBBS can also be helped to prepare for life with poor vision.

If you are interested in finding out more about LMBBS, you can write (enclosing an A5 stamped addressed envelope) to: 1 Blackthorn Avenue Southborough Tunbridge Wells Kent TN4 9YA