Diagnosis of Williams Syndrome

The first stage in diagnosing Williams syndrome oftens begins with recognition of the physical characteristics, which may include; puffiness around the eyes, a short nose, a broad forehead, cardiovascular problems and also feeding disturbance in early childhood.

This will be followed by a confirmatory genetic test. The most common genetic test is the Fluorescence In-Situ Hybrid (FISH) test.

The test involves taking a sample of blood and sending it to be tested for the deletion of elastin in chromosome 7.

The test maps the genetic material of a person and can determine if there are genes missing.

There is now a newer genetic test called a micro-array analysis. This test provides additional information, such as the size of the deletion on chromosome 7.