

Diagnosis of Angelman Syndrome

The syndrome can be diagnosed clinically by observing distinctive facial and other physical and neurological characteristics that are common in Angelman syndrome.

Often a child can develop [seizures](#) which may alert professionals to consider a diagnosis of Angelman syndrome. For others, [delayed development](#) may initially alert carers to seek advice from a professional.

The first developmental changes are noted from 6 months of age. A professional will look at the developmental history, [physical](#) and [behavioural characteristics](#) of a child and from this information determine whether the child has a clinical diagnosis of Angelman syndrome. For example, persistent smiling and happy demeanour around 3-6 months of age may be an indication of Angelman syndrome.

A clinical diagnosis can be given by paediatrician, GP or a clinical geneticist. However, a clinical diagnosis will only be made if a genetic diagnosis cannot be established.

Diagnosis is mainly made on the basis of genetic testing. Someone with a genetic diagnosis will have had a genetic test. This involves taking a sample of blood or saliva and sending it to be tested for one of the [genetic changes](#) known to cause Angelman syndrome on chromosome 15.

In the short film above Prof Jill Clayton-Smith describes the process of diagnosis and the range of emotions families may experience when the first receive a diagnosis. For families who have recently received a diagnosis they may find some useful information and links to support groups on our [welcome section](#).

In this clip, Andrea describes her experience of getting a diagnosis of Angelman syndrome for her son Woody.

*In the following short films two families describe their experiences of receiving a diagnosis of a rare genetic syndrome. In the first film Nathaniel **does not** have Angelman syndrome; however, many families may still relate to his father's experiences.*

For more information about diagnostic characteristics for Angelman Syndrome, please visit [Angelman UK](#) - the syndrome support website.

To read more about some of the genetic tests that may be used for diagnosis [click here](#).