Fragile X syndrome

Fragile X syndrome (FXS) is a rare neurodevelopmental condition and is the leading genetic cause of learning difficulties in the world.

It affects roughly

1/4,000 males

1/8,000 females

It is caused by a fault in the **FMR1 gene**

which is responsible for making a protein (FMRP) that is needed for normal brain development. With fragile X, the protein isn't made and the brain doesn't develop as it should.

1943
James Purdon Martin and Julia Bell first described fragile X syndrome.

1991
The FMR1 gene was discovered by Drs Ben Oostra, David Nelson and Stephen Warren. They named this mutation FRAXA.

Most common symptoms

- Mild to severe intellectual disabilities
- Attention deficit and hyperactivity
- Anxiety
- Speech delays
- Sensory integration problems, such as: hypersensitivity to loud noises or bright lights
- Epilepsy in 25% of cases

Treatment

There is currently no cure for fragile X, but treatments are available

- Speech therapy
- Behavioural therapy
- Occupational therapy
- Special education

Working closely with the **FRAXA Research Foundation**

We're combining
- Patient insight
- Our AI drug discovery expertise

To identify novel therapies and fast-track them towards patients in need.

References

FRAXA - www.fraxa.org/fragile-x-syndrome | Centers for Disease Control and Prevention - www.cdc.gov/ncbddd/fxs
NORD - www.rarediseases.org/rare-diseases/fragile-x-syndrome