

FRAGILE X SYNDROME

Speech Pathology casenotes by Louise Tyquin

In the last edition of Woman and Child we presented a case about an eight and a half year old girl called Jan who had difficulties with her auditory skills particularly in the area of auditory short term memory i.e. remembering spoken information that has just been said.

Today we are discussing a two and a half-year-old boy called Paul. Paul's mother recalled that he was born at term with no complications.

Mum's concern:

Paul's mum reported that he was a 'floppy' baby and had great difficulty learning to sit by himself. Paul saw a physiotherapist to help with his gross motor skills like crawling and walking. The physiotherapist noted that Paul had hypotonia (low muscle tone), flat feet and some loose joints.

At two years of age, Paul's mother took him to an Ear, Nose & Throat (ENT) Specialist because he had suffered numerous recurring ear infections since he was 6 months old. The ENT inserted grommets in Paul's ear to avoid the build-up of fluid, which was resulting in the ear infections. The ENT told mum to expect an increase in Paul's language. However, the increase in language was only minimal so Paul's mum sought a Speech Pathology assessment.

Assessment:

Assessment revealed that both Paul's comprehension and expression of language was significantly delayed. At two and a half, Paul was just starting to understand some very simple instructions enhanced by a lot of gesturing. Expressively, Paul was grunting and pointing to communicate his needs.

Behaviour:

Throughout the assessment it was noted that Paul's attention span was very short. He would flit from toy to toy and not spend more than one minute on any particular activity. Paul did not give any adults in the room any eye contact and would often flap his hands or a piece of paper rather than play with the toys.

Follow up:

I recommended that Paul see a Paediatrician to assess his overall development. The Paediatrician noted that Paul had an unusually large head with large ears. He also noted that Paul had a squint and observed some difficulties with Paul's overall co-ordination.

The Paediatrician diagnosed significant attentional difficulties and noted Paul's fixation on objects, hand flapping and poor eye contact. Paul's mother also spoke about his difficulty adjusting to change.

After a number of different tests, Paul was diagnosed with Fragile X Syndrome.

Fragile X Syndrome (FXS) is the most common genetically inherited cause of intellectual impairment ranging from mild learning difficulties through to profound intellectual impairment.

FXS is one syndrome presenting with speech and language delays that Speech Pathologists come into contact with.

In most instances however, children with speech and/or language delays have at least average intelligence.