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NEWSLETTER SUMMER 2014

Editor's Note

Welcome to this years summer edition! I hope you will find it informative with the responses to the Q & A from our experts featured on page 2 and 3.

Additionally, I would like to feature this beautiful thank you card that we received at the Family Day. Handmade by Wing Yi Cheung, it was a lovely reflection of how the day was received and appreciated. It was another huge success and wonderful to meet up with new and existing members.

Nicki Turley

In this issue...

Don't forget to send your articles or any news that you would like to see in future issues to: **secretary@wdssg.org.uk**

Please let me know if you have anything that you would like to share with other members.

Genetics:

- **In a study 6/47 families had a family history of WDS – what indications is there that having more than one incident could lead to another?**
- **When you have a child with WDS, what is the likelihood of having another child with WDS?**
- **Are there any patterns that suggest a family could have a much greater risk of carrying the gene that causes WDS**

There appear to be several causes of WDS which probably act on brain development in early pregnancy and in some cases this will be due to a genetic mistake which may have been inherited from parents, or may have occurred for the first time in this pregnancy. In around 15% of cases, several family members are affected suggesting a genetic cause that is passed through the family and this is true for cases with or without scan abnormalities. The recurrence rate after one affected child is about 1 in 10. However this will be a mixture of high risk and low risk families, and at the moment, there is no specific test to determine if a particular child has a genetic form of WDS, and whether the genetic abnormality has occurred as a genetic mistake for the first time (in which case there will be no increased risk for further siblings) or whether it has been inherited from the parents which would mean a higher risk for further affected children. In general, the risk for siblings of a child with WDS, of having affected children themselves, is lower than the above risk and for many will be no increased risk. There is some risk of the young person with WDS passing it onto their own children and they should be encouraged to discuss this with a geneticist at the appropriate time.

- **What tests can be done if the chromosome is passed down the line of a family?**
- **Are there any genetic screening tests available yet for pregnant women to see if they are at risk of giving birth to a child with WDS?**

At present we have no genetic screening tests.

- **Is there any evidence that WDS is more likely in IVF pregnancy?**

There is a slightly increased risk of congenital malformation after IVF, but this is not specific to WDS. There is a more specific risk of WDS in twin pregnancy where babies share a placenta and twin pregnancies are more common in IVF. However overall this increased risk is very small.

- **What are the characteristics of the gene that is understood to cause WDS is it a gene that could cause other illnesses or disabilities too neurologically or in the autistic spectrum?**

We are currently looking for 2 candidate genes and it is probably too early to give specific details. One gene is on Chromosome 2 and would be compatible with autosomal dominant inheritance. The other gene is on the X chromosome and therefore is most likely to affect boys.

Profiles in WDS:

- **Given the casual implications of Cerebral Palsy as one condition and genetic connections of Autism as another condition, could WDS be the product of a child who should have had say Asperger's but then suffers from cerebral palsy i.e. could WDS be a mixture of the two conditions?**

Children often present as an apparent mixture of conditions and it is very important to understand how these are related. The difficulty is often that we like to label things and create definitions before we truly understand how a particular clinical picture comes about, so a particular person may appear to fit several labels but of course these are often related.

For example, cerebral palsy simply means motor difficulties which are due to abnormal development of or injury to the young brain (either before birth or certainly within the first 2 years of life). But of course if there is such an injury or problem with development, it is likely to affect many aspects of brain function, not just control of movement, so affected children also often have difficulties with their learning and behaviour and even seizures. In the case of WDS, as well as the difficulties with movement around the throat and mouth, we know that learning difficulties, ADHD, autism, seizures etc are more common than in unaffected children and probably this is because of a general problem with how the brain functions. Our current project is trying to find out whether there is an underlying genetic cause that may explain the difficulties with brain function (and in some children also brain structural development) in WDS.

- **Is there any evidence to show if people with WDS have a shortened life pan. Are there figures for average life expectancy?**

People with WDS do not generally have life-limiting conditions, although for some, the difficulties with swallowing may lead to serious problems with breathing and chest infections. However some children with WDS have epilepsy and we also know that in general for those with epilepsy, there is an increased risk of dying (1.6 to 3 times higher than for the general population) and this includes the risk of Sudden Unexpected Death in Epilepsy (SUDEP). We also know that some people with WDS have a learning disability and that people with learning disabilities can have a shortened life span and that some of this is due to the underlying condition but some is due to the difficulty they have in accessing suitable medical care.

In general, WDS is not seen as a condition that leads to a shortened life span but we have no figures for average life expectancy.

- **Although WDS comes in different severity is there an understanding how the syndrome will affect the diagnosed as he/she reaches adulthood?**

This will depend on level of any learning difficulty, severity of epilepsy and presence of other difficulties such as autism. There has never been a study of adults with WDS for us to have good information about this.

- **Any practical advice for improving Literacy skills and educational methodologies for a teenager with WDS, Microcephaly and ASD?**

It is important that the teenager's profile is understood so that if there are particular weaknesses, then a learning programme can be set up to support these and perhaps use their stronger skills to help with this and to compensate for their weaker areas. Hence it will be important to understand issues around any learning disability, attention difficulties and autism etc. The fact that spoken language is often so difficult in WDS, means that learning to read, particularly where there is a lot of emphasis on phonetics, can be tricky, and they may need particular strategies to address this.

Epilepsy and Brain Scans:

- **Difference in symptoms between WDS patients where brain malformation is visible and those where nothing can be found on MRI's?**

The main difference between WDS patients where there is an abnormality on brain scan and those where there is not, is that epilepsy is more common where there is a visible brain abnormality. In general, epilepsy is more common than in the general population and occurs in around a third of children with WDS who have normal brain scans, and more frequently in those with imaging changes on brain scan. In our experience, the average age at first seizure was around 3years of age and most of those who develop epilepsy do so in early childhood.

- **Has there been any documented cases that epilepsy can return once cleared?**

Yes, particularly if there is a visible abnormality on the brain scan.

- **Regards to MRI scans of children who have WDS, I have heard that the MRI scan may not been interpreted correctly. What part of the brain shows that the person may have WDS and how could this be misinterpreted?**

In around 15% of people with WDS, there is an abnormality (polymicrogyria) visible on a brain scan, usually on both sides of the brain the in perisylvian regions (congenital bilateral perisylvian polymicrogyria).

The brain undergoes a lot of maturation in infancy and early childhood, and so sometimes an abnormality may not be visible on an early scan, but becomes visible as the brain matures and the tissues take up the adult form. There is also improvement in detection of brain abnormalities as our scanning techniques become more sophisticated so more up-to-date scans may show abnormalities that could not be detected with older scanners.

Speech and Language and Feeding Issues:

- **My son does not know how to chew his food. Speech therapists all try to desensitize his mouth in the hope he will chew. Is it possible to train him to chew instead of swallowing food whole?**

In general in WDS, the problem with feeding is not to do with hypersensitivity but is rather a motor problem. This means that desensitisation and chew sticks are unlikely to be successful. Direct oral motor intervention is not usually very helpful in WDS as it is neurological in nature. However a careful oromotor assessment would be very helpful in identifying the specific difficulty and perhaps developing appropriate goals. For example, if there is no lateral tongue movement, then it would be very difficult for the child to move food from the molar teeth into the main oral cavity without using their finger. Often it is helpful to explore functional solutions such as cutting food up into smaller pieces or giving softer foods that can be squashed using the tongue against the hard palate so that the person with WDS can manage a reasonably 'normal ' diet.

Occasionally a person with WDS will also have autism and then there can be issues around oral sensitivity and unusual food preferences.

- **In some literature it states that normal Speech and Language Therapy (SALT) will stop being a benefit to someone with WDS as their speech will not improve. This has not been the case in our daughter and we would like to know why this has been written when there appears to be limited research on this. We feel that it does not help our children get the right amount of SALT with their local authority post 16, unless parents fight for this provision.**

SLT can always be of benefit but may not always be helpful in improving clarity as neurological speech disorders may not always be amenable to therapy (Lindsay Pennington's research in CP speech disorders). For all people with WDS, SLT is useful in developing vocabulary, social communication and introducing AAC devices if appropriate. There is a significant number of people with WDS in whom speech will never be their main way of communicating but SLT will be important in support multimodal communication. However there also appears to be number of people with WDS who develop spoken language much later than is usually thought possible (e.g. after 9 years) and who appear to benefit from focussed direct speech work. The benefit of any such work, must be balanced with the cost of how arduous and difficult it is to work on an area where improvements are slow and not guaranteed. Often a compromise is best, with some work being built into a daily routine and intense periods of SLT being trialled at intermittent times over the years and being extended when there is evidence of benefit.

Diagnosis:

- **My sons paediatrician is indicating that his speech is too good to diagnose him with WDS but he has all the other symptoms, is it acceptable to say that the speech will prevent him being diagnosed with WDS?**

As with all conditions, there is a spectrum of severity and speech clarity will depend on whether the main movement problem is anterior (lips and tongue) or more posterior (throat and palate). It is important that any diagnosis takes into account all aspects and in particular there needs to be a history of difficulty with more than one of the mouth/throat functions (feeding, speaking, drooling etc) with examination showing difficulty with movements of the lips, tongue, palate, and to be secure, the diagnosis is best made after the age of 4 years.

- **My sons MRI scan was inconclusive as was his sleep EEG is there a definitive test that can be done and how can I access the test?**

There is no definitive test. WDS is a clinical diagnosis.

- **Can I self refer my son to a specialist or pay privately for him to be seen to help get a diagnosis then access a plan for the future?**

A GP or Paediatrician's referral to a specialist tertiary service that can offer a multidisciplinary assessment with input from a geneticist would be the best solution.

- **It seems that WDS is still a fairly rare disorder is that down to wrong diagnosis or lack of cases nationally?**

There is lack of awareness of this diagnosis and in our experience it is not that uncommon. However it is most important that the children is properly understood and their needs met, this does not necessarily need a diagnosis though many families and services find it helpful.

FABULOUS FUNDRAISING..

WDS parent brings Porsche Carrera 993RST to show and committee member - Callum Turley gets to ride round the Top Gear track in it ... twice!

MARK SQUARES UP TO THE STIG AT THE

THE SUPERCAR EVENT

Some of our members met up the day before the Family Day and had a fantastic time!

From the Chair

Wow, what a fantastic time we had at the Top Gear track this year and I got to meet the Stig. I challenged him to a race but he just looked at me and walked off! Maybe he knew I would win? Ha, Ha!

The Family Day was also really good. I am so glad to have met so many people this year, at the Thames Valley Adventure Playground. The talks from Maria Clark and Professor Neville were very interesting and informative. I am also delighted that I have been able to represent the group for another year as your Chairman, where does the time go? I am also really happy to now have new members of the committee and would like to thank them for their contributions to the team.

I am looking forward to the next 12 months, both supporting you all and campaigning for more awareness of Worster-Drought. I am lucky as now I have more time available to do this since becoming unemployed on the 4th July.

I feel there is something ironic, as I have now got my independence as I am not having to worry about work.

Mark Mayer

and greatful giving...

We would also like to say a huge thank you to everyone that helps us at our family days. We appreciate your continued support.

It's also a real treat when we are able to personally give out birthday gifts at Family Days. Two of our members, Vanessa and Anthony, were celebrating birthdays that week.

We would also like to thank Sarah Sugden, James Marygold and Robin Gruchy Wilson for volunteering to give some of their time to the group as new committee members. We look forward to receiving their fresh input and ideas for the future of the group.