

Williams Syndrome Clinical Management Guideline Development Project

This project, which started in July 2008, is based at the Nowgen Centre in Manchester (www.nowgen.org.uk), and led by Dr Kay Metcalfe and Professor Dian Donnai.

It is generously supported by The Williams Syndrome Foundation UK (www.williams-syndrome.org.uk) and the post of 'Guideline and Care Tool Developer' was created in order to produce UK-specific clinical management guidelines and other care tools for people with Williams Syndrome (WS).

The first phase of the project was to review and update the 2001 recommendations for WS follow up, published by the American Academy of Pediatrics. A thorough search of the literature on managing WS, published

since the AAP guidelines came out in 2001, was undertaken, and the resulting publications were reviewed by WS experts from across Europe.

A consensus meeting, attended by the reviewers, patient representatives and other healthcare professionals with clinical experience of managing WS was held in May 2009, at which the findings from the literature review were discussed and recommendations for the management of WS in the UK were proposed and finalised.

The aim was to establish an expert consensus on the management of WS, based on the literature and clinical experience, that would inform the recommendations and guidance contained in the clinical management

guidelines.

The second phase was to draft the guideline document and other care tools, including a personal health record (PHR) and checklist for non-medical personnel involved in the care of people with Williams Syndrome. These drafts are now complete and the guidelines and personal health records are piloted nationally, by healthcare professionals and families affected by WS.

The third stage will involve an evaluation of the guidelines and care tools, and the subsequent drafting of the final versions of the documents. These will be completed by July 2010 and we hope that they will become helpful resources for people with or affected by WS, and the professionals involved in their care.

DYSCERNE's Dysmorphology diagnostic System (DDS) – Results from the pilot and details of the full launch.

DYSCERNE an EU funded, European Network of Centres of Expertise for Dysmorphology, successfully launched its electronic Dysmorphology Diagnostic System (DDS) in May 2009. 76 centres with known expertise in Dysmorphology were invited to become submitting nodes and at the end of December 2009, 77 centres from 27 European countries have access to the on-line dysmorphology diagnostic system DDS.

The aim of the DDS is to provide rapid and equitable access for clinicians to expert dysmorphologist's opinions through Europe. The DDS software links expert dysmorphologist's from 30 European Centres of Expertise forming a powerful web-based diagnostic resource. It allows clinicians to submit difficult to diagnose dysmorphic cases for review by this Expert Panel. A diagnostic report including suggestions for further investigation and clinical management of the case is prepared from the consensus of opinions received and sent to the submitting clinician.

Prior to the full launch of the DDS a pilot of the system took place in spring 2009. The pilot ran for 4 months and involved 7 centres (Istanbul, Leuven, Manchester, Marseille, Nijmegen, San Giovanni Rotondo and Warsaw) who were invited to submit cases for review by the DDS Expert Panel.

During the pilot 20 cases were reviewed by the expert panel's members. All the cases could be described as complex phenotypes with combination of dysmorphic features, varying congenital abnormalities affecting different body systems and a range of neurocognitive disabilities. Diagnoses were suggested for all the cases, with molecular or cytogenetic tests being suggested for 19 out of the 20 cases. Summary reports of the expert opinions on each case were prepared and sent to the referring clinician within six weeks of the case being accepted onto the system. The traditional route for these difficult to diagnose dysmorphic cases would be presentation at national and international

dysmorphology meetings, a process which can take many months and still not result in a diagnosis.

These promising early results, coupled with positive feedback from the pilot participants, indicate that the DDS provides a rapid and effective diagnostic service.

Since the full launch of the DDS in May 2009 case submissions have averaged over 5 per month with Summary reports of the expert opinions on the majority of cases being produced within 35 days of case acceptance.

The DYSCERNE Coordinating Centre, based at the University of Manchester is anticipating that case submissions will steadily increase over the next few months as the centres with DDS accounts families themselves with the system and incorporate it into their clinical practice.

If you would like more information on DYSCERNE Network and the DDS please visit our website at www.dyscerne.org