

Dear friends and colleagues,

Thank you for the opportunity to update you on some of our research activities.

With this newsletter I want to highlight a new publication led by my colleague Dr Renuka Dias, and Prof Julia Rohayem in Germany, investigating testicular function in young men with Wolfram syndrome in Europe. They did this study because there is very little information on how boys go through puberty in Wolfram syndrome. The aim of this study was to assess testicular function and pubertal progression in a cohort of adolescent and young adult men with Wolfram syndrome. They included 21 young people aged 16–30 years. All were treated in two tertiary European health care centres: in Birmingham, UK and Münster, Germany. They looked up details of hormone levels, and who was being treated with hormone supplements. In addition, the presence or absence of erectile dysfunction was explored. In a subset of men, semen data were analysed. They found that 57% of young people with Wolfram had delayed puberty and needed testosterone supplements to complete their pubertal development. The remaining 43% of young men went through puberty normally, and one, aged 30 years, had normal sperm count in semen.

I think this study is important as it tells us we should check that all young men with Wolfram get through puberty 'under their own steam', and if not, should be offered testosterone supplements to help their development.

The team are now looking at pubertal development in females, to complete the picture and inform our clinical services. Dr Dias is hoping to complete the data collection and analyse the results this Summer.

Three other updates: Dr Sovan Sarkar has 2 PhD students and 2 visiting fellows from Brazil working to find new candidate treatments for Wolfram and will be presenting some of his results at the Wolfram international research meeting in June. And relating to the TREATWOLFRAM trial, Tori Homer is currently hard at work analysing the results of the trial again, this time with data from Prof Tammy Hershey's St Louis Wolfram Natural History Study. She hopes some of this will be ready in time for the Wolfram international meeting. Finally, Dr Josie Elliot has been working hard with myself, Fumi Urano, the St Louis team, Patrick Yu Wai Man, Tracy Lynch, and others, to complete the International Wolfram Syndrome consensus guidelines on clinical management. These will be presented in June before publication, and we hope will make a real difference to how affected people are

offered care, in whatever centre they visit. We have had over 50 experts from across the world contribute to these, so we hope the guidelines will lead to a more consistent standard of care for all Wolfram patients.