

to: csodraftresearchstrategy@scotland.gsi.gov.uk  
Response to the consultation on the CSO Research Strategy 2014  
From: Chromosome 18 Registry and Research Society (Europe)

Dear Sir

Two members of the Chromosome 18 (Europe) attended the Genetic Alliance focus group meeting on the CSO research strategy on September 17th. We found it very interesting and are grateful for the opportunity to participate. We understand that Natalie Frankish will be sending in a response from the meeting but we felt our members would be interested in the consultation and in supporting our recommendations to selected questions. We have sent this to all our members in Scotland.

Yours sincerely

Judy Wilkinson and Bonnie McKerracher  
on behalf of Chromosome 18 Registry and Research Society (Europe)

## SUMMARY OF QUESTIONS

### Chapter 2 – Partnership with Scottish Patients and Public

Question 8: Would a trial register be of benefit to patients seeking trials? Would it be an effective way to partner patients with researchers? Is there a danger that expectations of taking part could be unfairly raised?

*This could also be sent through the patient groups such as Chromosome 18 to disseminate to members. Results could be reported through Conferences to inform affected people and their carers. For example Dr Cody and Dr Hales from the Chromosome 18 Clinical Research Centre at the University of Texas report to the European Chromosome 18 conferences. In the same way relevant Scottish and European research could also be reported.*

Question 9: Would using electronic NHS patient records to alert GPs to research studies for which their patients may be eligible a service the NHS should offer? If so, would a process where NHS records are only accessed by identified NHS staff working in secure facilities, and only passing potential participant names to their GPs or hospital consultants for consideration, be a suitable way to proceed?

*GP's meet so few patients with rare diseases that the channel may be through the members associations as well.*

### Chapter 3 – Targeted Deployment of Resources and Infrastructure

Question 10: What proportion of CSO funding should be available for deployment in new research initiatives relevant to the NHS? In what areas should CSO seek to disinvest to free up resources?

*We do not have enough expertise to suggest where NHS should disinvest but we feel that new research is needed into physical support for patients (e.g better hoists, wheelchairs, accessing technology); therapies (such as music, communications, motor control) and into the effects of such therapies on development. Such research would be cross-disciplinary and complement the research into drugs and molecular biology etc. It would also probably be cheaper because it would not involve such large clinical trials but the results could make significant contribution to the quality of life of affected persons.*

Question 11: Is the focus of the CSO response mode grant schemes adequately defined and understood by the research community? Should there be a narrower focus to complement and avoid

overlap with other funding streams Scottish researchers have access to? What is a realistic upper level for CSO grants to allow worthwhile projects to progress?

*See our response to question 10. More cross-disciplinary research involving Engineering, Arts and Humanities is needed. Small pilot grants in these areas, could open up larger research collaborations and also economic benefits through the development of new hardware and technologies.*

Question 12: What should determine the creation and continued funding of a CSO unit? Should any new unit have a plan for CSO funding to be time limited?

*A cross disciplinary unit that is established for sufficient time to enable it to grow organically but has a reporting and evaluating structure to ensure the results of the research are relevant and sustainable.*

#### **Chapter 4 – Working in Collaboration**

Question 13: Are there other key areas of partnership CSO should be seeking to build?

*There is research on rare diseases being carried out in centres across Europe, the USA and other parts of the world. Eurodis is a dissemination for this research and perhaps linking to such international bodies could be part of the remit for the new NRS-GMS .*

Question 14: Would the creation of a CSO International Advisory Board be a positive step in raising Scotland's research profile and supporting our ambition? What should be the make-up of such a Board

*Inviting leading researchers in the different fields to present at Scottish conferences and at the same time request that they also evaluate the current research in Scotland in their area of expertise and discuss future strategies would be a positive step.*

Question 15: Are there other areas where CSO funded research could better support the Health Directorates Quality agenda?

*see our response to Q11 and 12. The quality of health and care will be increased by the interventions we have suggested.*

#### **Chapter 5 – Investing in the Future**

Question 16: Is the Primary Care Research Career Award scheme suitably focused to attract suitable high quality applicants? If not, what would a revised focus be?

*The discussion highlighted that perhaps GPs and health care nurses are more interested in their contact with patients than in research opportunities. A different kind of engagement may be more profitable in terms of research outcomes. A discussion with the Wellcome Trust about their Arts and Science initiatives suggest a new focus.*

Question 17: Do the current CSO personal award schemes targeted to meet our future needs? If not, should CSO conduct a wider review of its capacity building schemes?

*Exchanges with other countries enabling research staff to spend sabbaticals abroad (and discover the grass is not always greener) and for exchanges at all levels to take place would help to create a vibrant international centre. We trust this is part of the visions for the Scottish Science Improvement Centre at Dundee.*