

Mountain States Report
Natural History of Recombinant 8 Syndrome

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Project Awarded: 1-1-2006

Project Started: 1-17-2006

IRB approval: 4-22-2006

First patient/family enrolled: 6-23-2006

Objectives and Progress:

1. To outline the morbidity and mortality associated with Recombinant 8 syndrome given modern acute and chronic medical management.
2. To measure adaptive functioning skills of children/adolescents/adults with Recombinant 8 syndrome.
3. To assess whether children/adolescents/adults with Recombinant 8 syndrome have access to appropriate medical care in the context of a medical home.
4. To develop a patient management tool for physicians involved in primary care for children/adolescents/adults with Recombinant 8 syndrome.

Objectives 1-4 are complete on time and within the budget parameters. We were able to enroll the anticipated number of patients (12) without difficulty. Families were satisfied with their assessments. The project results were shared in the following venues:

- Colorado Genetics Laboratory April 2007
- Department of Clinical Genetics and Metabolism Grand Rounds June 2007
- Mountain States Regional Collaborative Center Annual meeting July 2007
- Abstract at David Smith's conference July 2007
- Clinical Rare Diseases Research Network meeting September 2007
- United Physicians Incorporated February 2008
- American College of Clinical Genetics meeting March 2008

Publication: None at this time. Manuscript is in draft form.
Problems/Roadblocks: None.