
Revisiting Recombinant 8 Syndrome

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Historical Phenotype Description

- Characteristic facial appearance
- Other minor anomalies on physical exam
- Cardiovascular malformations
- Genitourinary tract malformations
- Clefting of the lip/palate or both
- Seizures possibly related to brain anomalies
- Cognitive impairment – often significant
- Early mortality

Sujansky et al. AJMG 47:512-525, 1993.

2007 Rec 8 Goals

- **Hypothesis** – Early intervention and modern surgical cardiac repair techniques result in improved outcomes in affected individuals.
- **Goals** –
 - To characterize the natural history of Rec 8
 - To measure adaptive functioning skills
 - To assess whether patients have access to a medical home
 - To develop a management tool for physicians to help guide primary care decisions

Methods

- Clinical Interview and Physical Examination
- Review of any available medical or school records
- Formal developmental testing including cognitive and adaptive assessment
- Medical Home Family Index survey

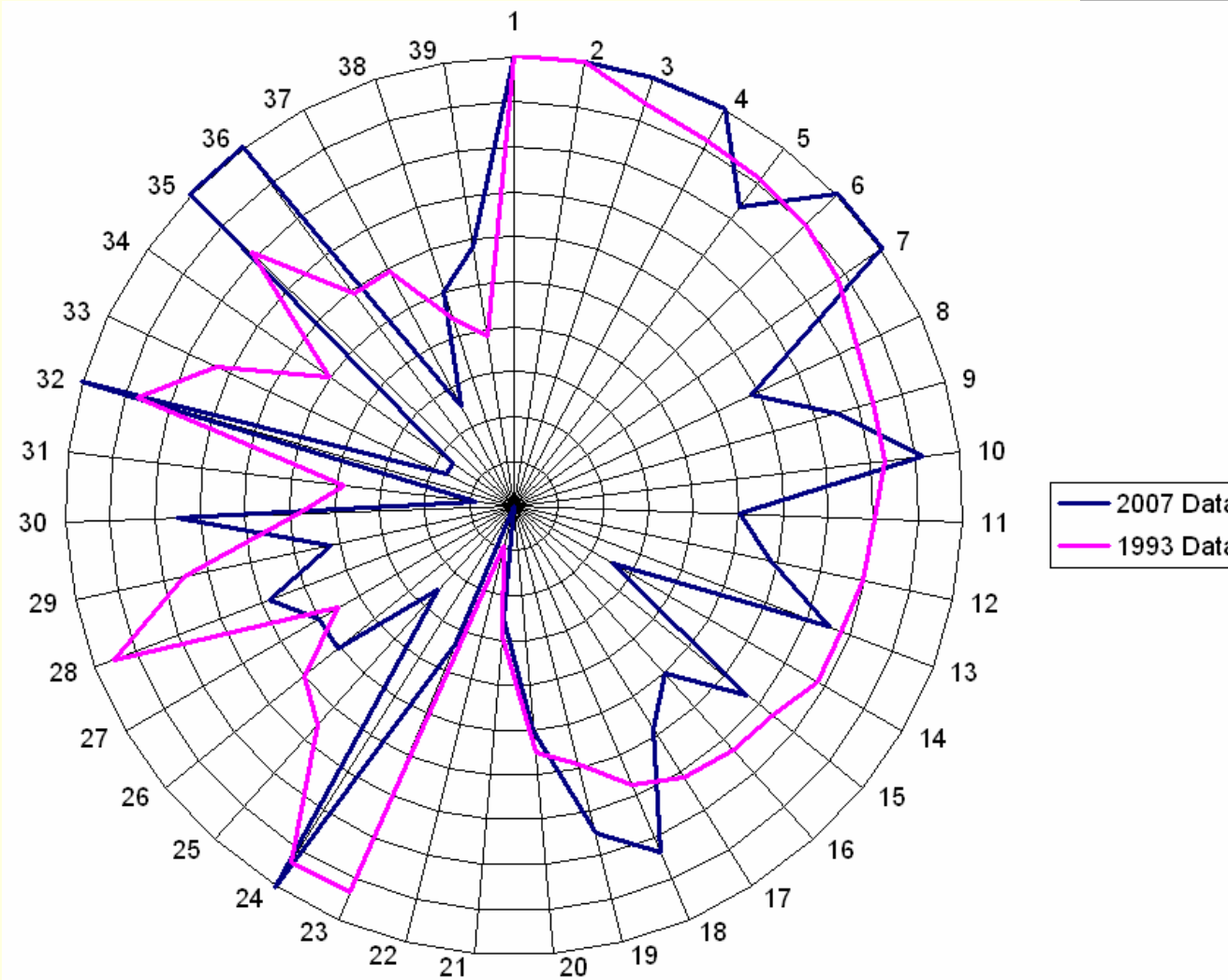
Results - Demographics

- Age Range: 2 – 21 years
- 7 boys, 5 girls
- Urban: 5, Suburban/Rural: 7
- Represent families in 4 States
- Private Insurance: 8
- Medicaid only: 4
- Premature birth in 3/12 (25%)
- Delivery by LTCS 7/12 (58%)
- Lives with adoptive family 2/12 (16%)

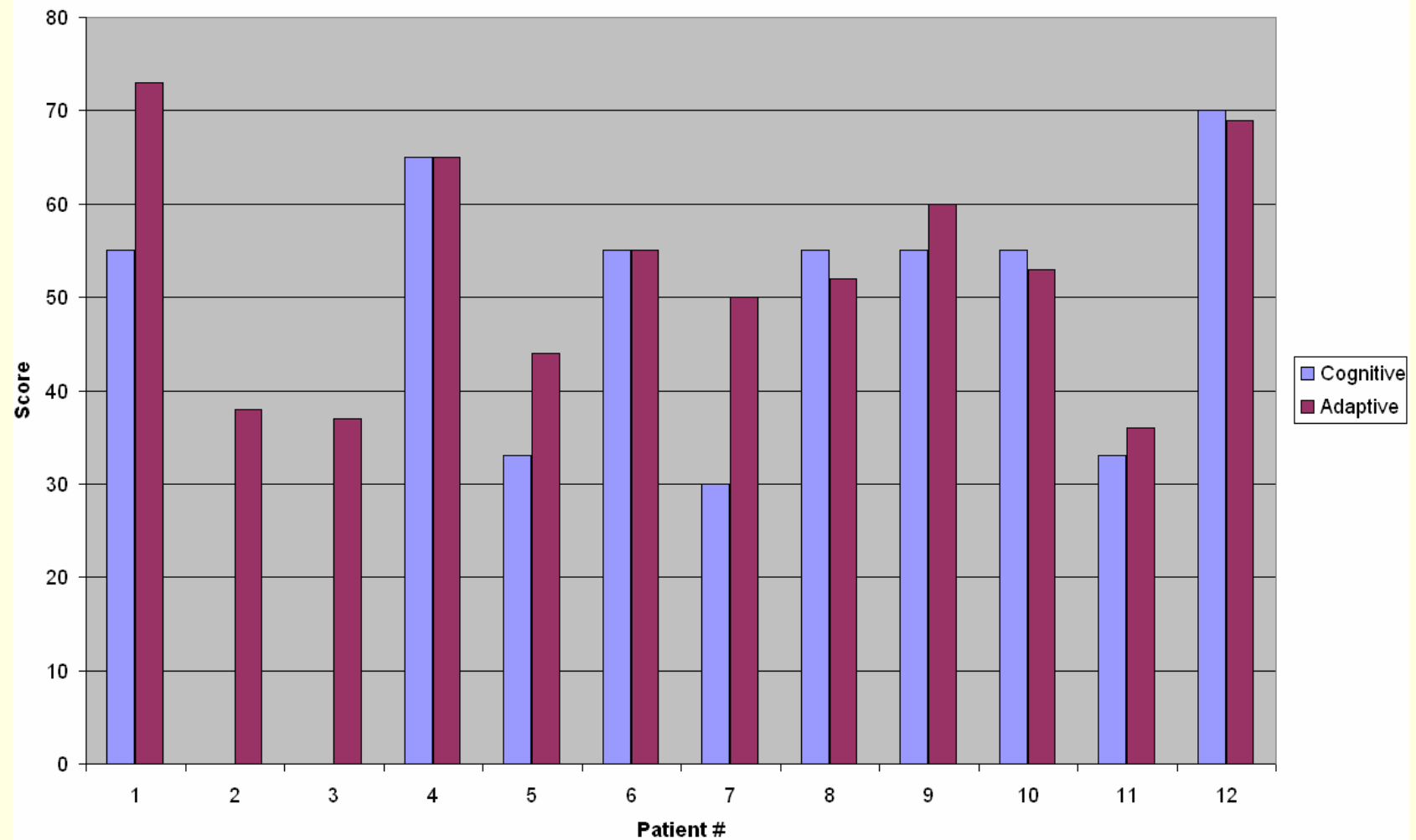
Results - Demographics

- Mean number of surgeries/child = 3 (range 0-7)
- Mean number of medications/child = 2 (range 0-7)
- 83% had a known parent who was a carrier
- 58% had other known relatives with Rec 8
- 91% were of Hispanic ethnicity (1 family with no known link to Hispanic heritage)
- 100% receive age appropriate therapies for developmental delays at least weekly
- 66% of families reported some degree of ongoing financial or family planning stress

Results - Physical Characteristics



Results – Cognitive Assessment



Results – Functional Abilities

Activity	Number	Percent
Diet	6/12	50
Sleep	3/12	25
Constipation	6/12	50
Play	5/12	42
Communication	8/12	67
Toilets	4/12	33
Dresses	1/12	8
Feeds	9/12	75
Ambulates	6/12	50
Chores	3/12	25

Results – Social Skills

Skill	Number	Percent
Joint Attention	10/12	83
Reciprocity	10/12	83
Social Smile	12/12	100
Eye Contact	12/12	100
Point	9/12	75
Nonverbal Gestures	9/12	75
Shared Enjoyment	12/12	100

Results – Social Skills

Skill	Number	Percent
Restricted Interests	5/12	42
Aggression	2/12	17
Skin Picking/Digging	0/12	0
Self Injurious Behavior	0/12	0
Spitting	0/12	0
Sensory Integration	0/12	0
ADHD	0/12	0
Anxiety	0/12	0
Smearing Feces	0/12	0
Sexually Inappropriate Behavior	0/12	0

Results – Medical Home Family Index

- All responses by our families were compared to national survey data (N=227) and to each other broken down by insurance type and demographic setting.
- Individuals with private insurance were generally more satisfied with the care they received due to perceived greater choice.
- Urban residents reported greater satisfaction for 3 items all pertaining to the degree of communication between medical providers (care provided in a team approach) and availability of resources. ($p < .05$)

Results – Medical Home Family Index

- Rural residents reported greater satisfaction with two items both pertaining to communication the family received from their medical care provider ($p < .05$).
- Compared to national data our respondents were more satisfied with face to face contacts with their doctors.
- Compared to national data our respondents were less satisfied with care coordination, parent advocacy/partnerships, and were dissatisfied with the lack of written care plans and anticipatory guidance.

Conclusions

- The Rec 8 phenotype is consistent across studies.
- Lethality is not necessarily a given and depends on surgical outcomes of cardiac repair.
- Learning does occur, although more slowly than in typically developing people.
- Quality of life is considered “good” by many families and their doctors.
- Social/adaptive skills are better than expected for cognitive ability.

Conclusions

- A team approach to the Medical Home concept was associated with improved medical outcomes and greater family satisfaction with care.
- Continued longitudinal studies with greater numbers of patients should be completed to help guide families making decisions surrounding long-term care.