PKS Kids Family Weekend

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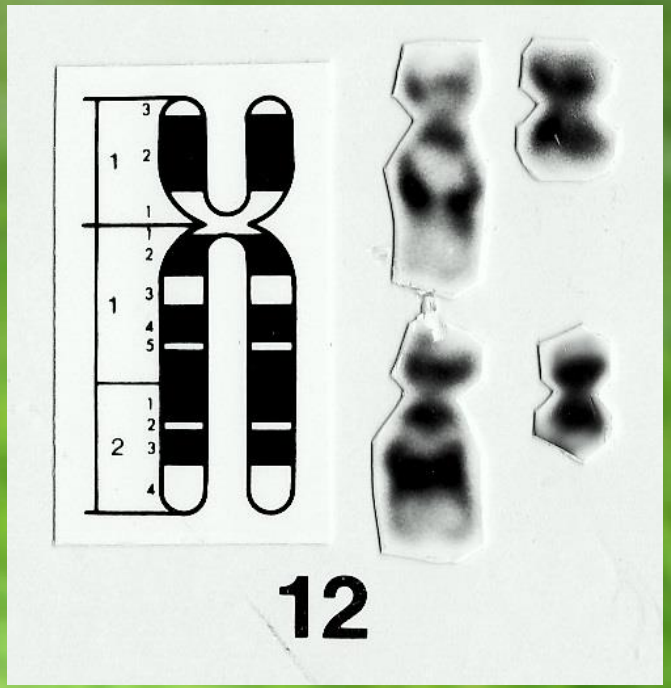
Talking Points

- Introduction
- Background on PKS
- Current Research Project
- Findings Thus Far
- Looking Ahead

Background

- What we know
  - Diagnostic Criteria
    - i12p tetrasomy
    - Craniofacial findings
    - Learning impairment
    - Seizures (40% of 67 cases)
  - Challenges
  - Desire to know more



Previous seizure reports/studies

- Limited information
  - Often incomplete, lacking EEG information
  - Collected differently by different groups
- Long term follow-up often not available
- Tendency to report more severely affected (or unusual) children
  - Maybe this makes the information look more negative?

Recent summary. 2010 Cerminara and colleagues

- Reported two children with PKS and “late-onset” spasms
- Provided detail on these two children
  - Case histories
  - EEG information
- Summarized previous reports from the literature that described seizures in PKS specifically

Summary of previous reports

- 14 children with sufficient information to analyze
- Average age of onset: 3 year of age
  - Range from “neonatal” to 9.5 yoa.
- Varied seizure types
  - Spasms>myoclonic=focal
- Very variable treatment--most effective not described
- “Outcome” not described in any detail.
- No mention of sleep-specific issues

Cerminara et al. J Child Neurol 25:238, 2010

Summary of previous reports- seizure medications used

- No information about which were most helpful

Chemical name	Brand name	No. children using
Valproic acid	Depakote	6
Topiramate	Topamax	5
Carbamazepine	Tegretol, carbatrol	3
Vigabatrin	Sabril	3

One each used: oxcarbazepine, vitamin B6, lamotrigine, clobazam, clonazepam, ethosuximide.

Cerminara et al. J Child Neurol 25:238, 2010

Current Research Objectives

- Characterizing Seizures in PKS
- Goals
  - Earlier recognition of seizures/seizure-like spells
  - Better parent education & anticipatory guidance
  - Improved understanding of cause(s), effect(s) & treatment(s)

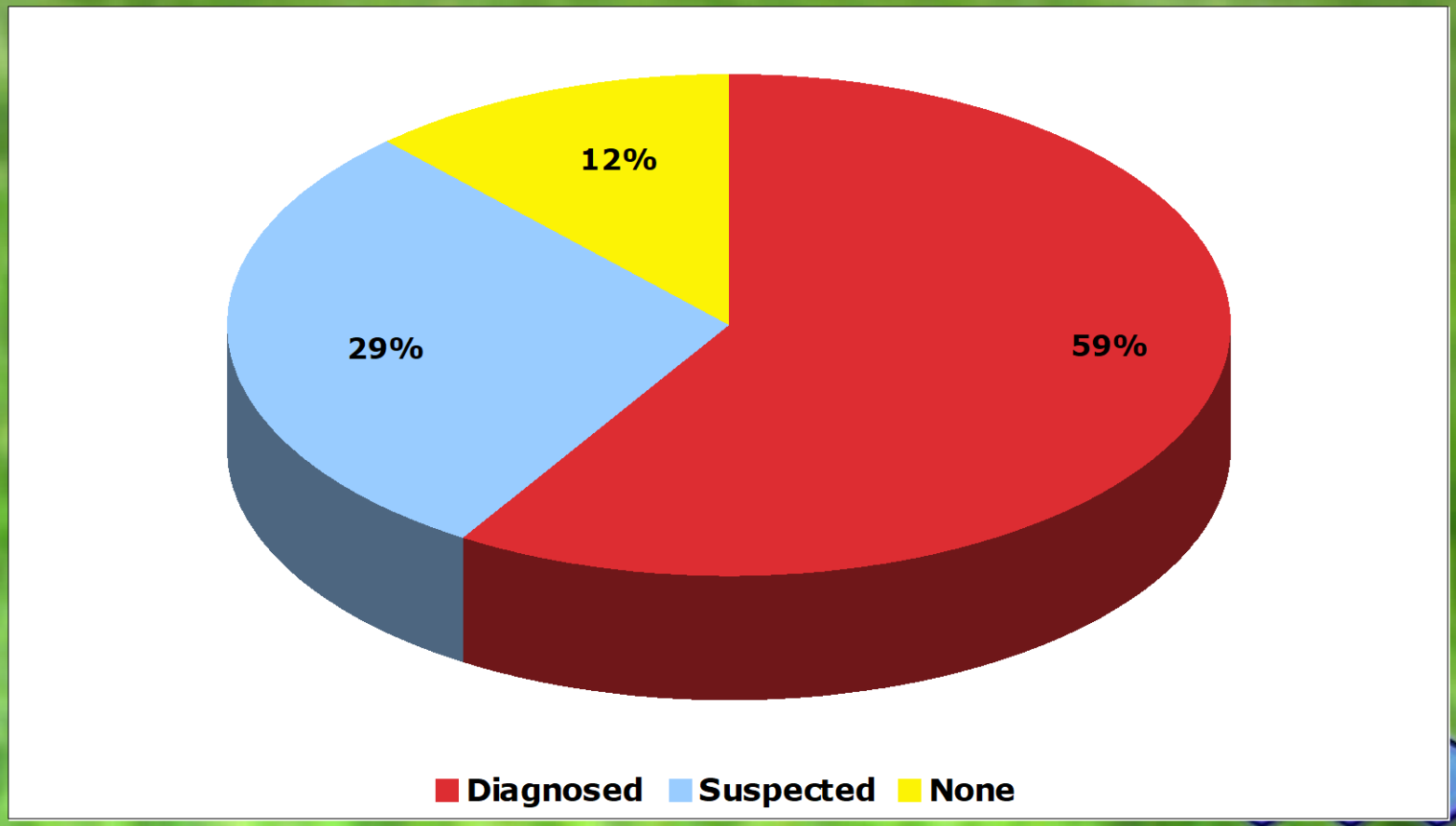
Current Research--Methods

- Patient selection/exclusion
- Consents
- Surveys
  - Onset, timing, frequency, triggers, therapies
- Medical record release forms
  - Neurology reports
  - EEGs
  - MRIs

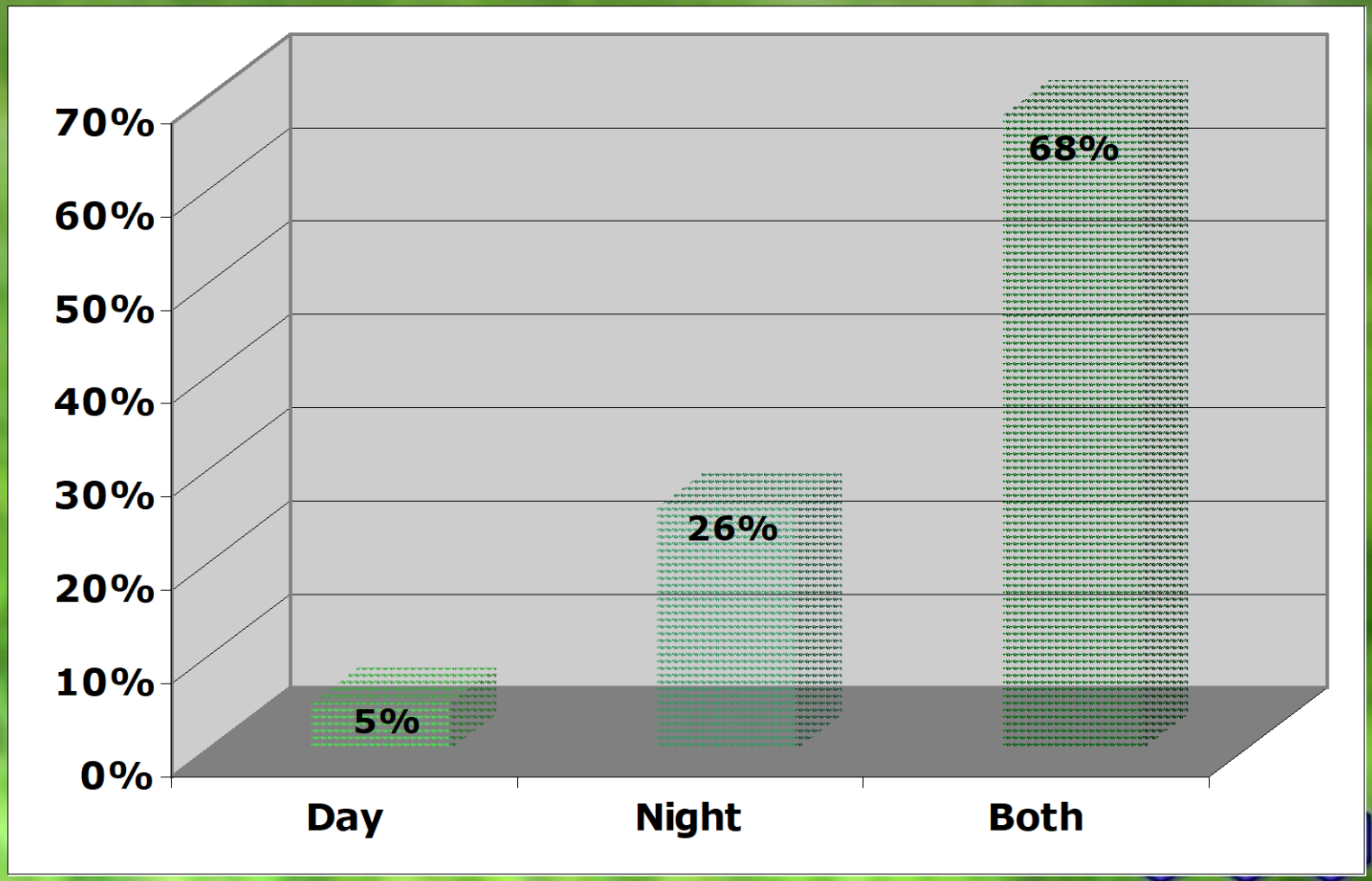
Preliminary Findings--
Selected Patient Characteristics

Total number of subjects	34
Practice	4 (12%)
Interview	10 (29%)
Questionnaire	20 (59%)
Mean Age at time of study	6 years 4 months
Median age at time of study	4 years 6 months
Total number diagnosed with seizures	20
Mean Age at Seizure Onset	2 years 8 months
Median Age at Seizure Onset	2 years 3 months
Percent with history of status epilepticus	7 (35%)
Percent taking anti-epileptic medications	9 (45%)
Percent taking >1 anti-epileptic medications	2 (10%)

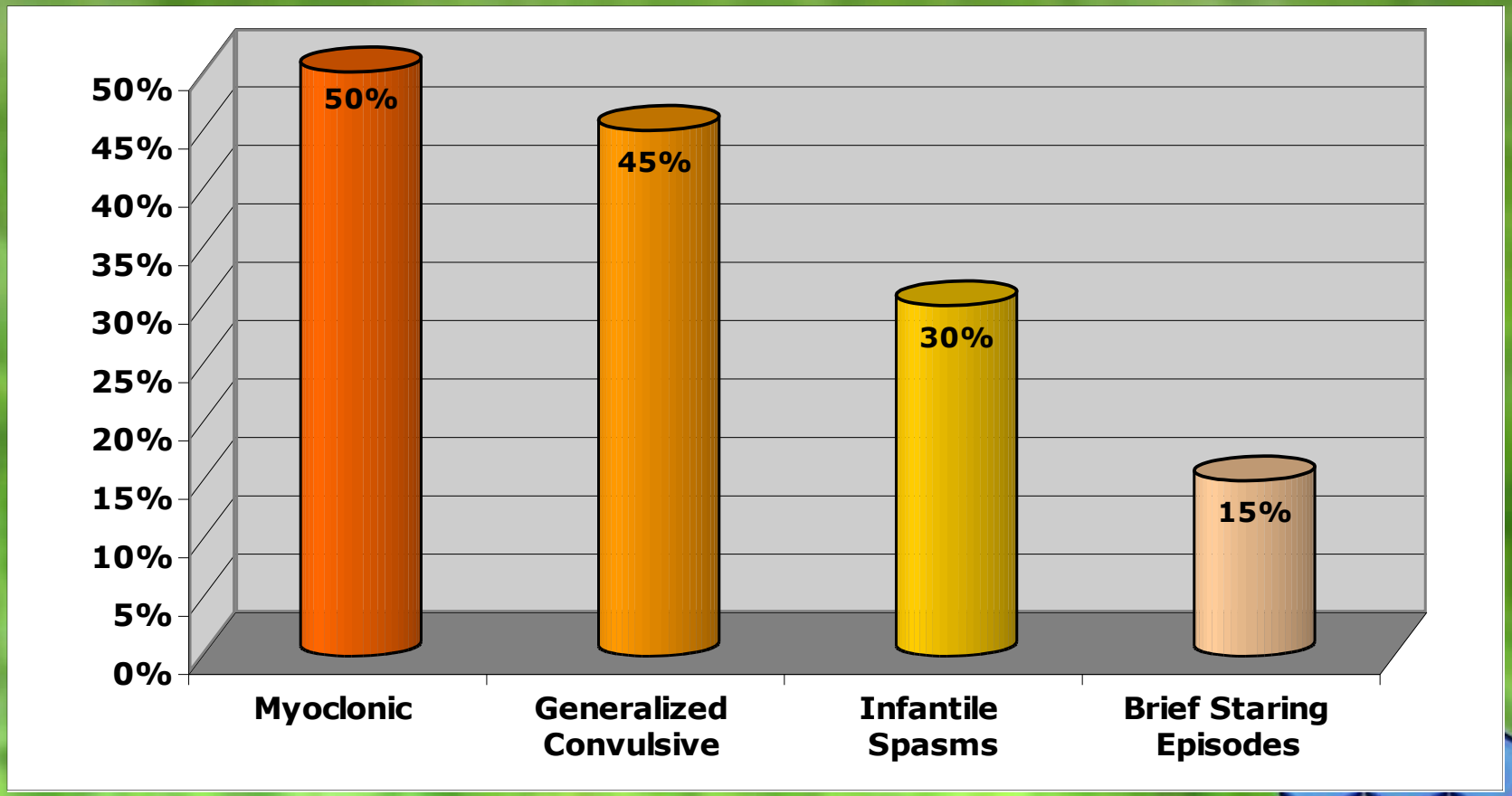
Preliminary Findings--
Seizure Prevalence



Preliminary Findings--Seizure Timing



Preliminary Findings--
Common Seizure Types



"Most helpful" antiepileptics

Most helpful medications (of those used by at least 3 children)

Chemical name	Brand Name	No. Children Used	No. of times "most helpful"	Percent successful
Levetiracetam	Keppra	6	3	50%
Zonisamide	Zonegran	3	1	30%
Clonazepam	Klonopin	3	1	30%
Vitamins (+)	--	3	1	30%
Lamotrigine	Lamictal	7	2	29%
Oxcarbazepine	Trileptal	4	1	25%
Valproate	Depakote	9	2	22%
Topiramate	Topamax	8	1	12.5%

Vigabatrin, Stiripentol each used by one child and deemed "most helpful." Diet (ketogenic) used by two children and deemed "most helpful" by one.

Antiepileptics which were never "most helpful"

"Never" "most helpful" medications

Chemical Name	Brand Name	No. Children Trying Medication
Phenytoin	Dilantin	2
Phenobarbital	--	2
Carbamazepine	Tegretol,carbatrol	2
Clobazam	Frisium	2
Gabapentin	Neurontin	1
Nitrazepam	--	1

Preliminary Results

- Most common seizure types:
  - myoclonic (10/20, 50%)
  - generalized convulsive (9/20, 45%)
  - infantile spasms (6/20, 30%)
  - brief staring episodes (3/20, 15%)
- Seven of the 20 patients with seizures (35%) had >1type.
- Six of the 20 patients diagnosed with seizures (30%) were seizure free on medication, while only 2 (10%) were in remission.
- Among interviewed subjects, 50% had paroxysmal events of uncertain etiology, most of which were associated with sleep disturbance

Preliminary Conclusions

- 88% of these PKS children exhibited seizure-like events. Definitive diagnosis was not always possible.
- Onset in early childhood was typical, but only one child presented with neonatal seizures.
- Paroxysmal disorders, often related to sleep, appear common.
- Relatively few of the children had experienced status epilepticus requiring emergency room visits or intractable epilepsy requiring hospitalization.
- Further study of this group of children may yield further insights into the seizure characteristics of PKS.

Looking Ahead

- Meeting with Families tomorrow
- Continued Data Collection
  - consents, surveys, med record release forms
  - By USPS or email

Upcoming Meetings/Events

- University of Utah Pediatrics Research Conference--June 28, 2010
- Child Neurology Society Meeting--October 2010
- Potential for additional research projects
- PKS Family Weekend 2011!

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- Kate Hettiger
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- Dr. John Carey
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