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Survey of Epilepsy Presentations in 8p-Related Disorders

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BACKGROUND

- 8p-related disorders are genetic conditions associated with chromosomal rearrangements on chromosome 8p
- Characterized by structural brain and cardiac abnormalities, intellectual disability, autism, and epilepsy
- There is currently no in-depth survey of epilepsy in 8p-related disorders

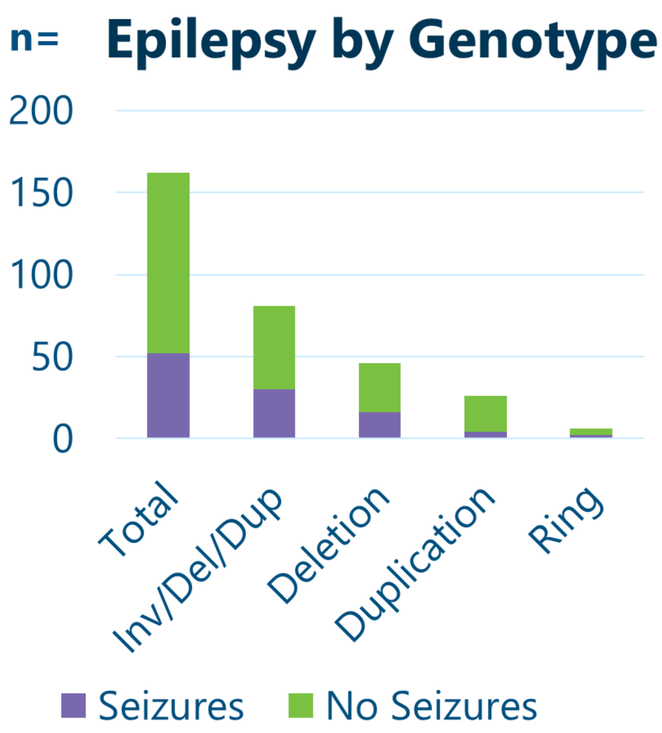
METHODS

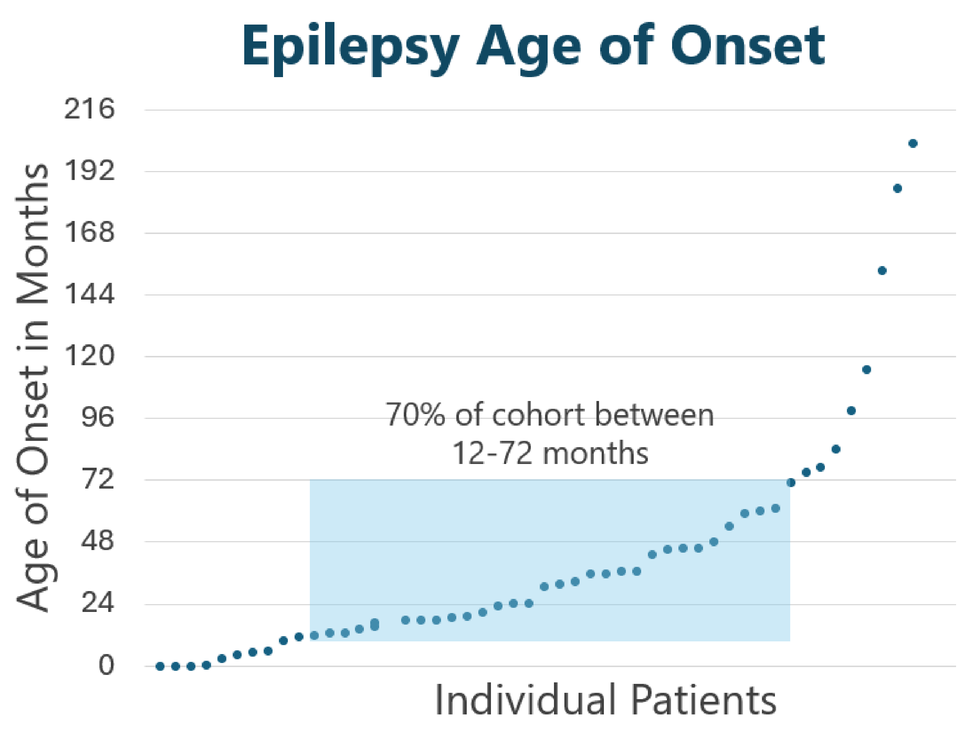
- Data obtained from patients with 8p-related disorders seen at Children's Hospital Colorado (CHCO) and/or recorded in the Project 8p Foundation Natural History database (genotype, presence of epilepsy, & age of seizure onset were able to be combined)
- Retrospective chart review conducted for patients seen at CHCO from 2020 to 2024
- Recorded epilepsy prevalence, age of seizure onset, treatment response, and EEG features
- Assessed differences in epilepsy presentation and severity among patients with Invdupdel(8p), isolated deletions, and isolated duplications.

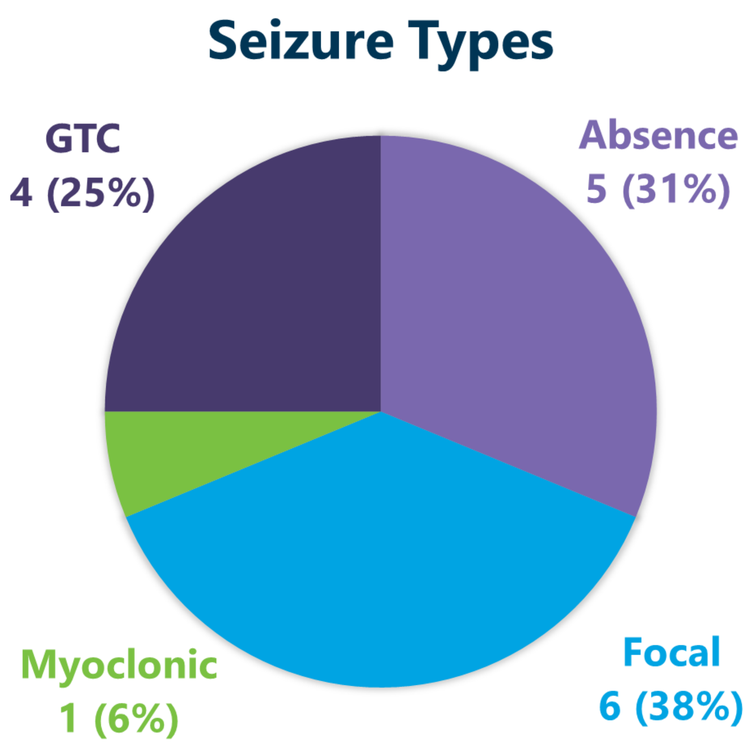
RESULTS

Characteristic	CHCO	Project 8p Database	Total
Average age (age range)	10.7 years (7 months – 42 years)	10.6 years (3 months – 52 years)	
Invdupdel(8p)	22/42 (52%)	59/120 (49%)	81/162 (50%)
Deletion Only	12/42 (28%)	34/120 (28%)	46/162 (28%)
Duplication Only	7/42 (17%)	18/120 (15%)	25/162 (15%)
Trisomy or Ring	1/42 (2%)	5/120 (4%)	6/162 (4%)

- Cohort included 162 patients with 8p-related disorders (42 at CHCO, 120 from the Project 8p database)
- 32% of all patients (53/162) experienced at least one lifetime seizure: 37% (30/81) with Invdupdel(8p), 35% (16/46) with deletions, and 15% (4/26) with duplications
- Among CHCO patients with epilepsy (11/42), only one had intractable epilepsy, while 9 were seizure-free (5 off AEDs)
- EEG abnormalities present in 43% of CHCO patients







IMPLICATIONS

- First detailed analysis of epilepsy in 8p-related disorders
- Epilepsy in 8p disorders is relatively common, but typically well controlled
- Genotype-specific patterns seen: Invdupdel(8p) was associated with the highest epilepsy prevalence and 8p duplication with the lowest epilepsy prevalence
- We will need to evaluate larger numbers of patients in order to verify relationships

DISCLOSURES

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DEMAREST LAB

PROJECT 8P