Mind the gap: Differences in sensory memory throughout development in individuals with Cystinosis

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BACKGROUND

Cystinosis is a rare, **autosomal recessive disorder** characterized by a bi-allelic mutation in the 17p13.2-located CTNS gene (Town et al., 1998) that promotes **excessive storage and crystallization of cysteine**, causing damage to organs such as the kidney, thyroid, eye, and brain (Gahl & Kaiser-Kupfer, 1987).

Cystinosis appears to be associated with an **impairment in sensory memory in children and adolescents** (Francisco et al., 2020), but the developmental trajectory of this impairment is unknown.

Aim: to assess the developmental trajectory of basic auditory processing and sensory memory in individuals with Cystinosis.

PARTICIPANTS

15 adults diagnosed with Cystinosis (CYS adults)
(age: M = 27.46; SD = 6.49; 3 males)
17 adults neurotypical controls (NT adults)
(age: M = 26.94; SD = 6.47; 6 males)
22 children and adolescents diagnosed with Cystinosis (CYS children)
(age: M = 11.15; SD = 2.66; 9 males)
24 child and adolescent neurotypical controls (NT children)
(age: M = 11.71; SD = 3.38; 10 males)

TASKS

WAIS-IV and WASI-II (intelligence quotient measures)
EEG task: (passive) duration oddball paradigm
standard tone: 100ms; deviant: 180ms
SOAs: 450, 900, 1800 ms

EEG DATA COLLECTION

BiosemiActiveTwo 64-channel electrode cap
Data recorded at 512 Hz, filtered between 1 & 45Hz
Re-referenced to TP8; artifact cutoff at 120uV.

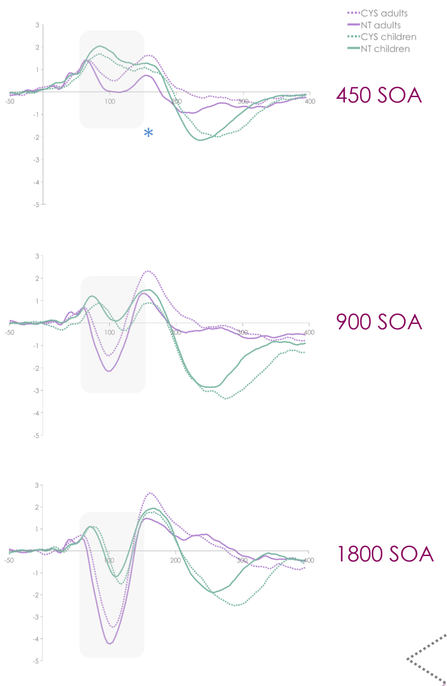
RESULTS

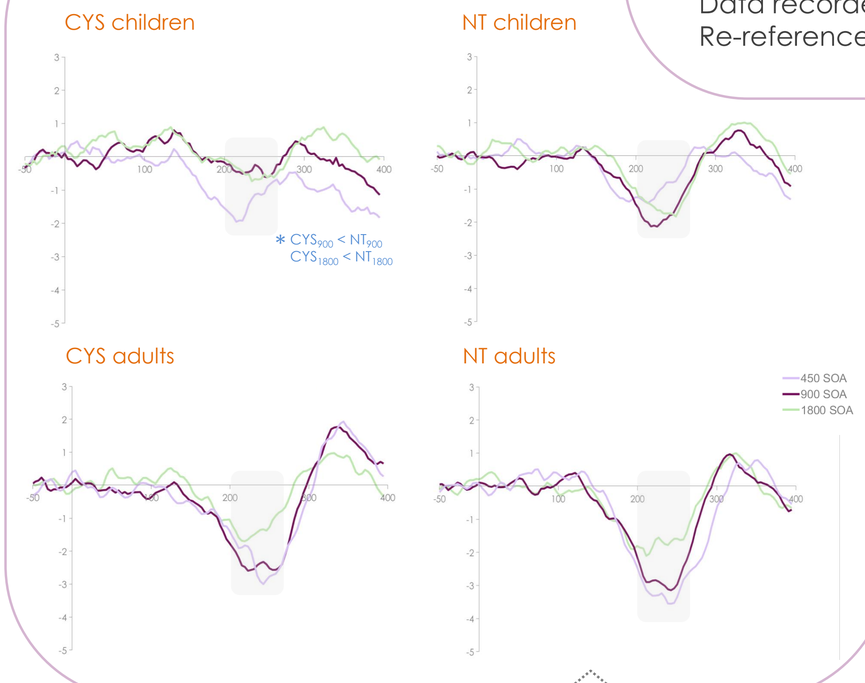
mismatch negativity (mmn); standards - deviants

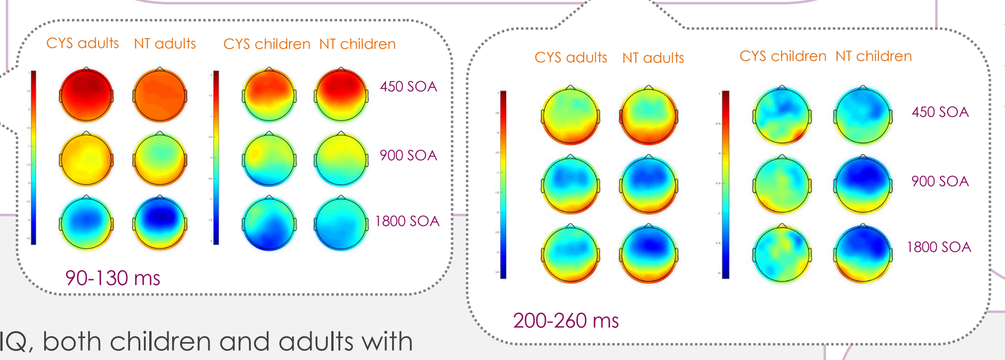
sensory memory

N1

basic auditory processing

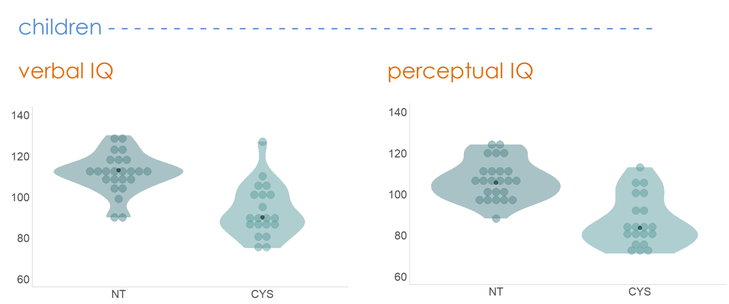


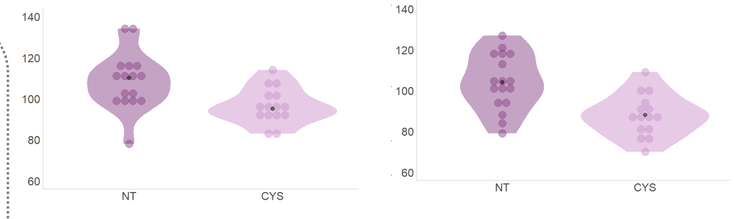




standardized cognitive assessments

verbal and perceptual IQ





DISCUSSION

With respect to verbal and perceptual IQ, both children and adults with Cystinosis had significantly lower scores than their neurotypical controls. Within both Cystinosis groups, perceptual IQ scores were consistently lower than verbal IQ scores, suggesting this finding is consistent across development in this population. Unexpectedly, this difference was also present in neurotypical adults.

Children with Cystinosis demonstrated typical auditory processing, with robust N1 responses of similar amplitude to the NT control group. Slight group differences were found in the adult group for the fastest presentation rate (450 SOA).

The MMN data suggested that children with Cystinosis have impaired sensory memory: In comparison to the control group, intact MMNs were observed for the fastest presentation rate, but these were greatly diminished for the slower presentation rates. In contrast, in our adult Cystinosis group, robust MMNs were evident for all conditions that were largely the same as the MMNs for the control group. Thus, deficits in Cystinosis in maintaining short-term auditory sensory memory in childhood seem to be resolved by adulthood.

Despite the limitation of our cross-sectional design, our findings suggest improvements in sensory memory and consistency in basic auditory processing and IQ throughout development in individuals with Cystinosis.

references

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