Assessing auditory processing endophenotypes associated with schizophrenia in individuals with **22q11.2 Deletion Syndrome**

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BACKGROUND

- 22q11.2DS is the most common chromosomal microdeletion disorder, with a prevalence ranging from 1:1000 to 1:4000 live births[1,2]
- It is characterized by a highly variable phenotypic expression, including: multi-organ dysfunction such as cardiac and palatal abnormalities, variable developmental delays, cognitive deficits, neuropsychiatric conditions[3]

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30% of individuals with 22q11.2DS go on to develop schizophrenia[4]; about 50% experience schizotypical traits and transient psychotic experiences[5]

AIMS

- To characterize basic auditory processing and sensory memory using EEG in a group of adolescents and adults with 22q11.2DS, with and without psychotic symptomatology.
- To relate these measures to cognitive function.
- To assess the potential informativeness of these measures with regard to vulnerability for psychosis.

PARTICIPANTS

- 11 individuals with 22q11.2DS without psychotic symptoms **(22q-)**
  (age: M = 23.26; SD = 7.75; 5 males, IQ: M=73.91; SD=10.97
- 15 individuals diagnosed with 22q11.2DS with 1+ psychotic symptoms **(22q+)**
  (age: M = 20.87; SD = 6.25; 4 males, IQ: M=69.43; SD=13.79)
- 26 neurotypical controls **(NT)**
  (age: M = 21.88; SD = 6.86; 10 males, IQ:M=112.17; SD=14.76)

EEG PARADIGM

passive duration oddball paradigm
standard tone: 100ms (85%); deviant: 180ms (15%)
SOAs: 450, 900, 1800 ms

EEG DATA COLLECTION

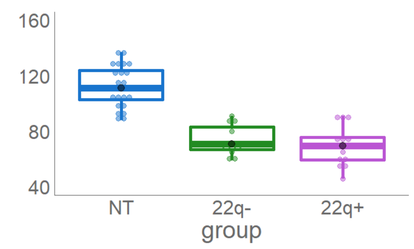
BiosemiActiveTwo 64-channel electrode cap
Data recorded at 512 Hz, filtered between 1 & 45Hz, re-referenced TP8; artifact cutoff at 120uV.

CLINICAL ASSESSMENT

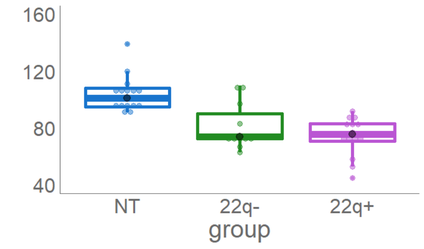
WAIS/WISC (IQ, working memory)
SCID-V/Kid-SCID (presence of psychotic symptoms)

RESULTS

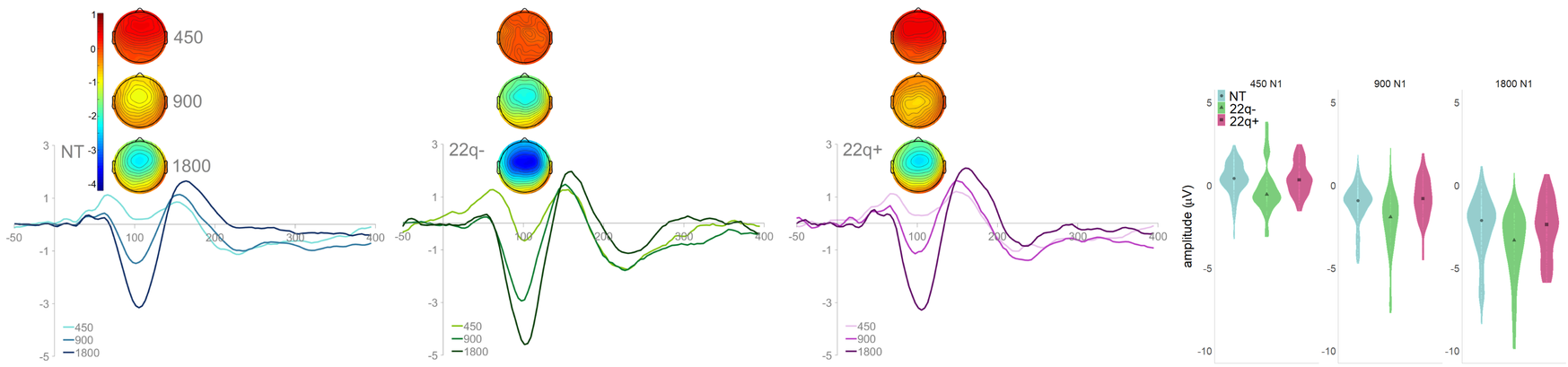
IQ



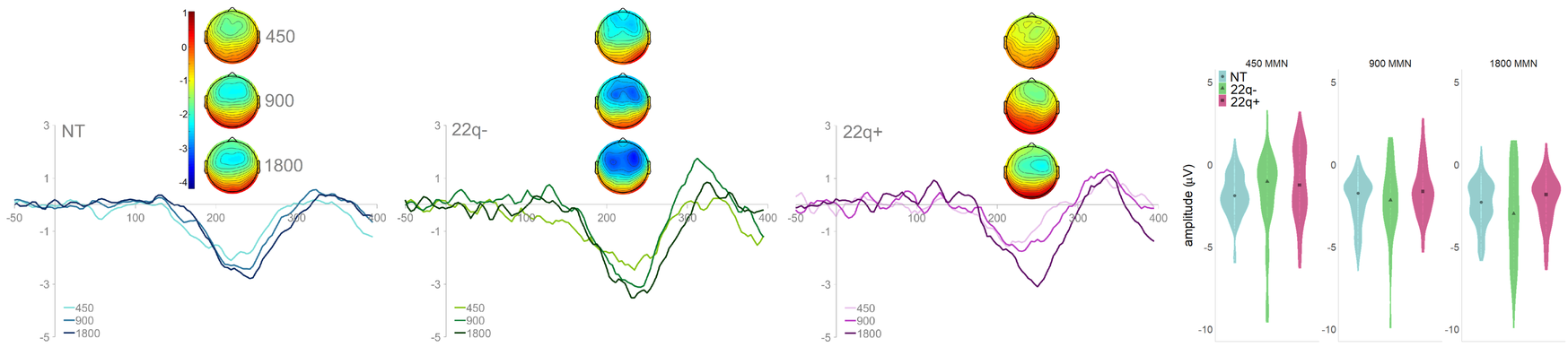
WORKING MEMORY



N1: standards (basic auditory processing)



MMN: deviants-standards (sensory memory)



DISCUSSION

- All groups presented typical N1 modulation as a function of SOA (all adapt).
- All groups processed change in duration (MMN).
- N1 adaptation effects interacted with psychotic symptomatology: When compared to the NT group, the 22q- group presented larger adaptation effects, whereas the 22q+ presented smaller effects
- In contrast, individuals with 22q11.2DS showed increased effects of presentation rate on MMN amplitude, regardless of the presence of psychotic symptoms.
- While IQ and working memory were lower in 22q11.2DS, these measures did not correlate with the electrophysiological data.
- These findings suggest the presence of two distinct mechanisms: One intrinsic to 22q11.2DS resulting in increased N1 and MMN responses; another related to psychosis leading to a decreased N1 response.

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