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Syndromic Autism and Distinct Craniofacial Dysmorphism: Evidence for a Novel Autism Facial Dysmorphism Syndrome

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ABSTRACT

Background: Autism disorders are a very complicated and multifarious group of chronic disorders that are characterized by early impairment in social interaction and communication. The diagnosis of autism is clinical and is based on the presence of characteristic diagnostic manifestations resulting from impaired social interaction and communication, which cause the two major diagnostic features of autism: the lack of appropriate responsiveness to one's own name and the lack of eye contact. Poor speech development with difficulties in using and understanding language, and repetitive body movements including hand flapping, and spinning, or behavior patterns are important associated features of autism disorders. Case report: A six-year-old boy with autism disorder and cognitive impairment who displays a distinct constellation of craniofacial dysmorphic features. These include low-set, posteriorly rotated ears, mild hypertelorism, down-slanting palpebral fissures, a broad nasal bridge with a bulbous tip, full cheeks, flat midface, short indistinct philtrum, retrognathia, thin upper lip, and a short neck. There is no parental consanguinity, and no similar cases in the family. The phenotypic presentation does not align with any currently known genetic syndromes associated with autism, suggesting the possibility of a novel syndromic autism phenotype. We propose that this case represents a previously uncharacterized autism facial dysmorphism syndrome and highlight the need for further genetic investigations and phenotypic comparisons. Conclusion: This paper highlights a unique clinical entity "Syndromic Autism with Distinct Craniofacial Dysmorphism".

Keywords: Syndromic Autism, Craniofacial Dysmorphism, New Clinical Entity.

INTRODUCTION

Autism disorders are a very complicated and multifarious group of chronic disorders that are characterized by early impairment in social interaction and communication. The diagnosis of autism is clinical and is based on the presence of characteristic diagnostic manifestations resulting from

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impaired social interaction and communication, which cause the two major diagnostic features of autism: the lack of appropriate responsiveness to one's own name and the lack of eye contact. Poor speech development with difficulties in using and understanding language, and repetitive body movements including hand flapping, and spinning, or behavior patterns are important associated features of autism disorders. The variation in speech and cognitive development results in the subtypes of autism [1-18].

Traditional treatments for autism have predominantly focused on associated symptom management, including behavioral interventions and pharmacotherapy. However, the quest for a cure has remained elusive until recent breakthroughs, which have shed light on promising avenues for intervention. Neuroleptics and other medications such as baclofen and ondansetron have been used with some benefit to control behavioral abnormalities and hyperactivity [1,2,5,7,10,11,15-17,19].

For decades, autism disorders have been considered lifelong disorders without curative therapies, despite a variety of medications that have been tried, including pyridoxine, magnesium, thiamine, biotin, folic acid, and omega-three. However, during the previous decade, at least 19 patients have been reported in the literature including 18 patients from Iraq and one patient from Cuba have achieved cure of the major autistic diagnostic features with the use of a new therapeutic approach using individualized courses of intramuscular cerebrolysin as the curative agents [11,20,21].

Cerebrolysin has been used safely with benefit in a variety of neuro-psychiatric disorders including idiopathic mental retardation, cerebral palsy, brain atrophy, myelomeningocele, pediatric juvenile spinal muscular atrophy, pediatric Charcot Marie Tooth disease, kernicterus, and agenesis of corpus callosum with colpocephaly [22-38].

We have previously reported cases of syndromic autism such as Williams syndrome and Coffin Siris syndrome from Iraq and other countries [24,39]. We present a six-year-old boy with autism, cognitive delay, and a distinctive pattern of craniofacial dysmorphism not consistent with any recognized syndrome. This case may represent a novel syndromic form of autism.

CASE REPORT

A six-year-old boy was evaluated for developmental delay and autistic behaviors. Milestones were delayed across cognitive, language, and adaptive domains. The boy exhibits typical major diagnostic features of autism including poor social interaction, and stereotyped behaviors especially hand flapping. At the clinic he was hyperactive, not responding to name and had poor eye contact.

Developmental Profile:

- Speech: Delayed expressive and receptive language
- Motor: Fine motor skills are mildly delayed.
- Adaptive Behavior: Notably impaired; requires assistance with feeding (unable to eat using a spoon effectively) and other daily living tasks.

Family History:

The parents are not consanguineous. No family history of autism, developmental delay, or congenital anomalies was reported.

Physical Examination:

Weight, height, and head circumference were within normal limits. However, the child displayed multiple dysmorphic features (Figure-1), including:

- Low-set, posteriorly rotated ears
- Mild hypertelorism
- Down-slanting palpebral fissures
- Broad nasal bridge and bulbous nasal tip
- Full cheeks with flat malar areas
- Retrognathia (mild micrognathia)
- Short, indistinct philtrum
- Thin upper lip
- Short neck

Figure 1. The boy displayed multiple dysmorphic features.

No other systemic abnormalities were noted. No cardiac, renal, or skeletal anomalies were detected on routine clinical assessment. Neuroimaging and metabolic workup were not available at the time of reporting.

The phenotypic presentation does not align with any currently known genetic syndromes associated with autism, suggesting the possibility of a novel syndromic autism phenotype. We propose that this case represents a previously uncharacterized autism facial dysmorphism syndrome and highlight the need for further genetic investigations and phenotypic comparisons.

The patient was treated initially with:

Intramuscular cerebrolysin, 4 ml given every other day in the morning. Oral risperidone 0.5 mg daily at 9 pm. Oral piracetam 800 mg daily in the morning.

After this initial treatment, the parents reported improvement in his behaviors and at the clinic he showed a better eye contact and less hyperactivity.

Therefore, the decision was made to continue oral risperidone and oral piracetam, and give intramuscular cerebrolysin 4 ml every third day (Ten doses monthly) until cure of the two major diagnostic features of autism.

DISCUSSION

We have previously reported cases of syndromic autism such as Williams syndrome and Coffin Siris syndrome from Iraq and other countries [24,39].

Identification and reporting of such cases are crucial for the delineation of new syndromes and the advancement of personalized diagnostics and care.

This case represents a child with syndromic autism, developmental delay, and distinct facial dysmorphism not corresponding to known entities such as Fragile X syndrome, Smith-Lemli-Opitz syndrome, Niikawa-Kuroki syndrome, Mowat-Wilson syndrome, or other recognizable syndromic autism conditions.

James Purdon Martin and Julia Bell from the United Kingdom reported a pedigree of X-linked mental retardation, without mentioning macroorchidism (larger testicles) in 1943 [40]. Herbert Lubs reported the association of mental retardation with an unusual “Marker X chromosome” in 1969 [41]. In 1981, Brown and colleagues suggested screening for fragile X syndrome by testicular size measurement [42].

In 1982, Brown and colleagues reported the association of fragile X syndrome with autism [43].

Smith-Lemli-Opitz syndrome was first described in 1964 by David Weyhe Smith, an American pediatrician, Luc Lemli, a Belgian physician, and John Marius Opitz, a German-American physician [44].

In 2000, Tierney and colleagues reported the association of Smith-Lemli-Opitz syndrome with autism [45].

Niikawa-Kuroki syndrome was first reported independently by in 1981 Norio Niikawa and Yoshikazu Kuroki from Japan [46]. 1997, Ho and Eaves reported the association of Niikawa-Kuroki syndrome with autism [47].

Mowat Wilson syndrome was first described in 1998 by David R. Mowat Meredith J. Wilson and their colleagues [48]. In 2024, Oliveira emphasized the association of Mowat Wilson Syndrome with autism disorder [49].

In this patient, de novo variants or novel chromosomal anomalies cannot be excluded. The facial gestalt and neurodevelopmental profile suggest the possibility of a previously uncharacterized syndrome.

Similar cases have not been reported in the immediate or extended family, nor have we identified identical phenotypes in existing literature or dysmorphology databases.

This underscores the potential for a new genetic syndrome associated with autism and facial dysmorphism:

1. Dysmorphic Features

The boy presents with a distinct and consistent pattern of craniofacial dysmorphism, including:

- Low-set, posteriorly rotated ears
- Mild hypertelorism and down-slanting palpebral fissures
- Broad nasal bridge and bulbous tip
- Flattened malar area and full cheeks
- Short indistinct philtrum and thin upper lip
- Retrognathia (or mild micrognathia)
- Short neck

2. Neurodevelopmental Impairment

- Autism disorder diagnosis
- Cognitive delay evidenced by:

o Poor adaptive behavior including spoon-feeding difficulty at age of six.

3. Negative Family History and Parental Consanguinity

4. Distinct Facial Phenotype

These features do not correspond neatly to common known syndromes such as:

- Fragile X syndrome
- Smith-Lemli-Opitz syndrome
- Mowat-Wilson syndrome
- Niikawa-Kuroki syndrome
- CHARGE syndrome
- Sotos syndrome
- Angelman
- or others commonly linked with syndromic autism.

5. Novelty

- The facial gestalt does not clearly match existing syndromes.
- Syndromic autism with this pattern of dysmorphism is not well characterized in the literature.

CONCLUSION

This paper highlights a unique clinical entity "Syndromic Autism with Distinct Craniofacial Dysmorphism".

A six-year-old boy with autism, cognitive impairment, and consistent craniofacial dysmorphic features not aligning with any known syndrome.

The combination of findings in this patient raises the possibility of a novel autism facial dysmorphism syndrome.

Genetic testing, such as whole-exome sequencing, is warranted and may clarify the etiology. We recommend further documentation of similar cases to support syndrome delineation.

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CONFLICT OF INTEREST

None.

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