

CASE REPORT:

**CORNELIA DE LANGE SYNDROME AND
PSYCHIATRIC'S ASSISTANCE**

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ABSTRACT

Cornelia de Lange's syndrome (CdLS) is a genetic disorder characterized by developmental disorders in several organ systems, including the brain, bones, digestion, immunity, endocrine, and others. This syndrome is mainly caused by mutations in the NIPBL, SMC3, and SMC1A genes. CdLS are generally comorbid with developmental or intellectual-level disorders, autism spectrum disorders, self-injury behavior, difficulty speaking, anxiety, hyperactivity, and sleep problems. This CdLS has a significant impact on the quality of life and maladaptive function in patients, as well as causing psychological disorders for families. Therefore, the need for psychiatric assistance for family psychoeducation, psycho-social interventions and cognitive-behavioral education.

Keywords: Cornelia de Lange Syndrome, Comorbidity, Autism, Psychiatric's Assistance

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INTRODUCTION

Cornelia de Lange's syndrome (CdLS) is a genetic disorder characterized by specific facial features, limb abnormalities, and intellectual disabilities. Somatic phenotypes also include upper limb abnormalities, gastroesophageal, cardiac dysfunction, ophthalmological and genitourinary anomalies, and hirsutism. Phenotype characteristic of individuals with CdLS has autistic features that characterized by repetitive behavior, self-injuring behavior, and deficits in communication abilities, with a smaller impact on social deficits. Common psychiatric manifestations associated with individuals with CdLS include hyperactivity, anxiety symptoms, and sleep problems. Older people with CdLS can manifest neuropsychiatric deficits as concomitant symptoms. Doctors who treat CdLS individuals must have a high index of suspicion in autistic features, and preparing integrated psychiatrists evaluation for psycho-social interventions, cognitive-behavioral education, and work together with medical rehabilitation for speech therapy with the aim of improving receptive and expressive communication. CdLS treatment needs of multidisciplinary involvement.

CASE REPORTED



A baby boy, 9 months old, small thin body, body length 55 cm, body weight 3,3 kg, face younger than age, lanugo hair appeared in most areas of the face, unformed hands, and his two-fingers at right toes have joined together. Patient referred from pediatric outpatient clinic for psychiatric's assistance in determining the sex and less of eyes contact. The patient had a small penile shape and testicles that are not clearly visible so the patient is diagnosed with suspected Cornelia de Lange Syndrome + testicular DSD + micropenis chordae + undescended testis + severe stunted + severe underweight + microcephaly + syndactily region of

pedis dextra et sinistra. The patient was the only child, born prematurely at 7 months gestation due to antepartum bleeding. At birth, patient had chest retraction and used oxygen support in an incubator for 1 month. He had various physical limitations and developmental disorders. Patients had been consuming Formula milk from birth until now because his mother's milk did not come out, due to her stressed saw patient's abnormality condition. The patient got complete basic immunization. He had growth retardation, began on prone position at 6 months, head raised at 6 months, "bubbling" at 8-9 months. From Psychiatric's assistance and follow up, patient began to have eye contact at least around the age of 8-9 months.

DISCUSSION

From the physical examination, it was found from ultrasonography that the left testicle was in the left inguinal region (distal high scrotum). Clinical diagnose : Disorders of Sex Development (DSD). Endocrinology examination resulted of children with Undescended testicle. From metabolic nutrition consultation resulted children with Retarded development following protein-energy malnutrition. From Andrological examination resulted with Indetermined sex, unspecified. Planning examination will be chromosome test, parental and maternal. Heart examination resulted with Tetralogy of Fallot. From psychiatric observations, there was non-specific symptoms of autism. Psychiatric observation still continue, because CdLS is a severe genetic disorder [1,2], often has another impact besides physical development disorders, and on the developmental / intellectual level disorders, autism spectrum disorders, self-injury behavior, physical conditions, and medications as well as speech difficulties, anxiety, hyperactivity, and sleep problems [3–11]. Psychiatric diagnose based on DSM-5 and psychometric, including: *The Checklist for Autism in Toddlers (CHAT)* at 18 months old and *Childhood Autism Rating Scale (CARS)*, also consultation needed to psychological test at preschool-age (4-6 years old) for intellectual ability.

Autism spectrum disorders are shown to be part of the behavioral phenotype of CdLS syndrome and higher compulsive behavior [4]. The study classified 17% as having no autism, 41% mild autism, and another 41% severe autism [5]. ASD is found in 27% - 82% of total CdLS patients [3,12,13]. Intellectual disability is a hallmark of CdLS, with 33% -74% of CdLS patients categorized as low intellectually, with IQ in the range of 55-70 [5]. For present, Psychological assistance is

needed for the families of CdLS child, especially related to his sex determination, and psychiatric disorders of children that potentially appear in the future, which mostly become severe autism. Psychiatric's assistance also needed for parents in parenting their child [14,15].

CONCLUSIONS

Psychiatric's assistance is important in the case of Cornelia de Lange Syndrome, especially related to the determination of the sex and potential psychiatric disorders of children who often appear later in life such as intellectual disorders, autism, self-injury behavior, anxiety, hyperactivity, and difficulty speaking

Acknowledgments

The authors would like to thank subject and his families.

Conflicts of Interest

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

This case report was funded by author's personal funding.

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