Clinical Genetics

AB108. Oral findings of Apert Syndrome case found in Cirebon

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Background: Apert syndrome is characterized by structural abnormalities of skull, face, eyes, hands and feet. This autosomal dominant disorder is caused by mutations in *FGFR2*, a gene encoding for fibroblast growth factor receptor protein. Despite its rare frequency in general population, several cases have been identified in Indonesia.
Methods: Physical and radiological examination was conducted to confirm the patient's deformities. Analysis using POSSUM software was done to diagnose the patient.
Results: We report on a 26-year-old man with striking dysmorphisms. He presented with acrocephaly, asymmetrical flat facies, hypertelorism, down slanting palpebral fissure, strabismus, ptosis, depressed nasal bridge, nasal deformity, short philtrum, low set ears and prominent jaw. The oral deformities showed maxilla hypoplasia with high arch palate. The V-shaped maxillary arch is filled with double rows of teeth. Besides, there is dental fusion between maxillary premolar and first molar. Panoramic radiographs were obtained for the confirmation. Other abnormalities found are mild scoliosis, mild pectus excavatum, type II symmetrical syndactyly of 4 fingers of both hand and of 5 toes of both feet. POSSUM software used confirmed the diagnosis as Apert syndrome with cut-off point of 14. He is as a slow learner and attended a school for children with special needs in Cirebon, Indonesia. The dysmorphisms found are very characteristic, thus molecular confirmation for *FGFR2* mutation is complementary for the diagnosis. Since the patient is the only affected person in his family, the mutation is possibly *de novo*.
Conclusions: An Indonesian man with Apert syndrome was reported with typical deformities of the skull and face, including maxillary hypoplasia and dental anomalies. Therefore, counseling for the patient should involve referral to dental specialist in order to provide an adequate treatment plan for his needs.

Keywords: Apert syndrome; dental fusion; oral deformities

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