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Kartagener syndrome occurring simultaneously in a Filipino child with 5p- (Cri du chat) syndrome

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Kartagener syndrome (KS) is a genetic disease caused by defects of the structure and function of the cilia that leads to abnormal mucociliary clearance causing disease of the sinus and pulmonary regions. Kartagener syndrome is characterized by the triad of bronchiectasis, paranasal sinusitis and situs inversus totalis. The most common gene affected is DNAH5 which encodes ciliary dynein axonemal heavy chain. DNAH5 is linked to chromosome 5p which is the primary chromosome affected in Cri du chat syndrome. Here, we report a 7 month old Filipino female presenting with the common features of Cri du chat syndrome as well as situs inversus totalis, recurrent respiratory infections and bronchiectasis which point to a concomitant Kartagener syndrome.

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Late diagnosis of central giant cell granuloma in a patient with Noonan syndrome: Case report

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Noonan syndrome is an autosomal dominant condition with a variable phenotypic expression. It is characterized by short stature, distinct craniofacial features, congenital heart anomalies and developmental delay. The main craniofacial features include hypertelorism with a downward slanting palpebral fissures ptosis, low set posteriorly rotated ears, deeply grooved philtrum and a high arched palate. Furthermore, Noonan syndrome has been found to be closely linked to tumor development, such as central giant cell granuloma (CGCG). As in this report we present a case of an 11-year-old male diagnosed with Noonan syndrome and during his dental screening, a multilocular lesion in the right side of his mandible was discovered coincidentally in an orthopantomogram and was diagnosed to be central giant cell granulomas after the histopathological assessment following surgery.

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