Role of cholesterol in craniofacial bone development

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Infants with cholesterol metabolic aberrations have malformations in the craniofacial region. For example, mutations in genes involved in cholesterol synthesis (DHCR7, SC5D, and DHCR24) have been found in Smith-Lemli-Opitz Syndrome [SLOS], lathosterolosis, and desmosterolosis. Patients with these syndromes display craniofacial bone abnormalities. In addition, high cholesterol diets during pregnancy are known to be a risk factor for birth defects including craniofacial bone abnormalities. However, it is still largely unknown how cholesterol metabolism contributes to bone formation. Taking advantage of our animal models, we are trying to determine new roles of cholesterol and its related molecules. I will discuss new mechanisms of how cholesterol is involved in the differentiation and function of osteoblasts during skull formation.