Introduction

A 39-year-old healthy pregnant woman, G3P1, The ultrasonic examination at 23 weeks 1 day of age demonstrated that there were plenty of abnormalities, including flat face, disappeared ogee of jaw, inconspicuous lower lip, thick tissue layers of forehead skin together with, abnormal posture of both hands and feet. The male neonates was delivered at 23 weeks and 3 days, the postmortem examination of the fetus was characterized by demonstrated including micrognathia, poorly developed and depressed philtrum, apparently widened nasal bridge, mandibular joint sagon both sides resulting in mandibular retraction, tightly pursed lips with apparent swollen skin ranging from nasal root to superior margin of lips, a broad square nose accompanied by anteverted nostrils, and soft tissue layers of facial skin revealing diffuse edema. Besides, low-set ears, and remarkably short neck were also noticed.

Conclusion

Smad Nuclear Interacting Protein1 mutation, maybe cause craniofacial dysmorphism and the bone morphogenetic proteins originate abnormality, including wide mouth, broad jaw, a bulbous nose, short hands, and tapered fingers, together with broad thumbs.