Case Report

Donor Site Calcification and Deformation Following Microtia Repair in a Pediatric Patient With Mosaic Trisomy 22

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Abstract
A microtia, or small or abnormally formed pinna, is an uncommon congenital abnormality of the external ear which can present as an isolated defect or as part of an underlying clinical syndrome. External ear reconstruction is a possibility, with either an autologous or non-autologous framework. The Nagata type of autologous reconstruction is a multistage process whereby costal cartilage, temporoparietal fascia, and a full thickness skin graft are used to form a new pinna. Here, we present the unique case of a young female born with mosaic trisomy 22, an extremely rare genetic condition, and a right-sided microtia. Between the first and second stages of her reconstruction, an anterior chest wall deformation was observed, coupled with unusual dystrophic calcifications over the cartilage near the ribs and sternum.

Keywords
congenital microtia, mosaicism, reconstructive surgical procedures, thoracic wall, trisomy

Introduction

Microtia
Congenital abnormalities of the ear stem from either the defects in embryogenesis or environmental factors which affect intraterine growth.1 Microtia and anotia occur only 1 to 3 times per 10 000 births.2 They may occur as isolated defects or be part of a genetic syndrome.3

Trisomy 22
Trisomy 22 is the second most common aneuploidy implicated in cases of spontaneous abortion;4 the associated organ deformation is so severe that affected fetuses rarely survive pregnancy. Only 15 to 20 cases of live-born children with trisomy 22 have been reported in the medical literature5 and almost all had a mosaic distribution.6 Known associated phenotypic features include delayed physical and intellectual growth and development, hemidystrophy, unilateral hearing loss, craniofacial malformations, as well as a number of findings classically associated with Turner syndrome.6

Purpose
We present a case of a young female with mosaic trisomy 22 and a right-sided microtia. Over the course of her recovery from autologous costal cartilage ear reconstruction, she developed an anterior chest wall deformity with dystrophic calcifications near the sternum and costal cartilage harvest site.

Case Report
The patient was first referred to our clinic when she was 21 months old, presenting with right-sided microtia, mild hemifacial microsoma, and a small ventricular septal defect (VSD). Although a previous diagnosis of Goldenhar syndrome was noted in her chart, a number of negative findings (absence of colobomata, epibulbar dermoids, cervical abnormalities, or macrostomia) and the presence of normal temporomandibular function provided uncertainty regarding the original diagnosis. The right ear was a classic lobular microtia, with an absent external auditory meatus. The left ear was normally shaped and

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mildly outstanding. At initial consultation, future staged reconstruction of the right external ear with the Nagata technique\textsuperscript{7-9} was planned for when her rib cartilage size would be appropriate, expected to be 10 to 12 years of age.

By 10 years of age, when the surgery was undertaken, it was noted that she had developmental delay, academic difficulties, and behaviour and learning patterns suggestive of fetal alcohol effects, though she displayed no other physical features. She was prepubescent. She had unilateral conductive hearing loss associated with her microtia, variable regions of cutaneous hypopigmentation, and extensive dental caries. The degree of hemifacial microsomia had remained consistent throughout growth. Her height and weight were in the 25th percentile.

During reconstruction, a cartilaginous framework was created from costal cartilage harvested from the patient’s fifth, sixth, and seventh ribs and placed into a Nagata skin envelope. A small segment of cartilage for use in the second stage was left in the subcutaneous tissue of the chest prior to skin closure. At the request of her geneticist, skin biopsies from areas of both normal and hypopigmented skin were taken, and this later confirmed mosaic trisomy 22, with the abnormal pigmentation being attributed as expected to hypomelanosis of Ito. No intra-operative or immediate post-operative complications arose. Two months later, she reported a prominence on her right chest: a firm palpable mass with an anterior hump over the right rib cage. A computed tomography scan revealed dystrophic calcification of the cartilage related to her right fourth, fifth, sixth, and seventh rib, and her sternum (Figures 1–3). She had no functional symptoms associated with this deformity.

She underwent the second stage of reconstruction about 1 year later, elevating the new ear with the stored cartilage and placing a small flap and full thickness graft. No significant complications occurred. A minor revision of her second stage repair was performed under general anesthesia 12 months later.

Follow-up with her pediatrician later that year revealed a 15° (mild) thoracolumbar scoliosis and a mild left-sided hydronephrosis. The VSD noted earlier had closed spontaneously and a magnetic resonance imaging of her brain was assessed to be normal. Despite significant obstacles including borderline cognitive ability, attention deficit, distractibility, unilateral hearing loss and speech issues, she had been making progress academically in an individualized program.

**Discussion**

Microtia has been reported in some live-born mosaic trisomy 22 patients.\textsuperscript{10} Though there are too few cases in the literature to demonstrate anything more than a potential association, trisomy 22 can present with craniofacial defects.\textsuperscript{10} To our knowledge, this is the first case report to outline the treatment of microtia in an individual with trisomy 22 and to report the specific complication that arose.

Hypomelanosis of Ito is a related pigmentary mosaicism which presents as streaks running along Blaschko lines;\textsuperscript{11,12} these may represent chromosomally aberrant melanocytes with a growth disadvantage.

Though chest wall deformity as a complication of microtia reconstruction is historically not uncommon,\textsuperscript{13} it is usually related to absence of the costal margin, and not dystrophic calcification. We have not found a report of this in the context of the Nagata ear reconstruction procedure or in the context of trisomy 22. Our consultants in medical genetics could not identify any genes on chromosome 22 related to bone healing, making it unclear if the complication can be specifically associated with the cytogenetic defect or was simply incidental. Mosaicism in general can lead to differential growth capacities between multiple populations of cells. Perhaps this predisposes...
a patient to create excessive calcification at sites of bone and cartilage healing. The patient’s mild scoliosis may have contributed to the deformity, but with the calcifications, it was thought unlikely to be the primary cause of rib asymmetry. It is unclear whether alcohol effects contributed to our patient’s condition; certainly, some of her facial features secondary to mosaicism overlap with the facial features typically seen in fetal alcohol syndrome. Because a history of maternal alcohol intake was not available, she was given a diagnosis of static encephalopathy with unknown alcohol exposure.

We were previously unaware of the possibility of bone and cartilage healing issues in this patient with a rare mosaic chromosomal aberrance. Because the clinical constellations of findings described in this report manifest from a young age, knowledge of further surgical risks may influence both decision-making and the informed consent process. Due to the uncommon nature of both trisomy 22 and microtia, we cannot conclude that there is a specific causal relationship demonstrated by this single case, but a report of our finding was felt to be important to add to the base of literature.

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Statement of Human Rights
All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised on 2008 (5).

Statement of Informed Consent
Informed consent was obtained from the study patient.

References