Craniofacial Microsomia: Goals of Treatment, Staged Reconstruction and Long-Term Outcome

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Background: Craniofacial microsomia (CFM) is a complex congenital condition and includes a spectrum of malformations primarily involving structures derived from the first and second branchial arches.

Objective: to present a review of the clinical presentations, assessment, treatment and outcome of the patients with CFM who were treated by the authors in Srinagarind hospital, Khon Kaen University.

Material and Method: Medical records were reviewed of patients with CFM, seen and managed by the authors at Srinagarind Hospital between 1993 and 2011; for an analysis of the clinical and radiologic assessments, treatments and outcomes.

Results: There were 23 patients (14 males and 9 females), 4 of whom had bilateral deformities, 19 unilateral deformities, and 1 syndromic CFM. The clinical presentation ranged from microtia to more severe deformities involving the mandible, zygoma and maxilla. Surgical treatments included: correction of macrostomia, distraction osteogenesis, staged ear reconstruction, orbit-maxillary-mandibular surgery, soft tissue reconstruction with dermis fat graft, vascularized free tissue transfer, cleft lip-cleft lip nose repair and palatoplasty. Most of the patients were satisfied with the medical treatment and many patients were still in the growing skeletal age and had staged reconstruction planned.

Conclusion: The goals of treatment for CFM are to (a) manage respiratory insufficiency and feeding problems (b) maximize hearing and communication (c) improve facial symmetry and proper facial growth and (d) optimize dental occlusion. These patients require staged reconstruction and coordinated care by an experienced multidisciplinary craniofacial team in order to ensure a thorough, comprehensive assessment of the nature and extent of the problems and to provide better treatment planning and long-term outcome adapted to the respective anatomic and functional deformities of each patient, and the needs of their family and other involved stakeholders.

Keywords: Craniofacial microsomia, Goals of treatment, Staged reconstruction, Long-term outcome

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Craniofacial microsomia (CFM) is a complex congenital condition and includes a spectrum of malformations primarily involving structures derived from the first and second branchial arches. The phenotype of CFM is highly variable in severity, ranging from almost unnoticeable to severely disfiguring. Bilateral involvement, usually asymmetrical, may be found in 15-30% of patients. Associated craniofacial anomalies may include macrostomia, micrognathia, cleft lip with or without cleft palate, pre-auricular skin tags or pits, ear deformities (varying from microtia, anotia to aural atresia), hearing loss and mandibular deformity. Non-craniofacial anomalies may include the cardiac system, the vertebral or central nervous system and/or the limbs(1,2).

A diagnosis of CFM is based on clinical findings and the proposed minimal diagnostic criteria is asymmetric hypoplasia of the facial structures with pre-auricular skin tags or pits, ear deformities (varying from microtia, anotia to aural atresia), hearing loss and mandibular deformity. Non-craniofacial anomalies may include the cardiac system, the vertebral or central nervous system and/or the limbs(1,2).

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Coursley(5) discussed research findings which affect all aspects of CFM, with an emphasis on facial anomalies which are important for diagnosis, classification and treatment. Heike and Hing(2) used the term CFM to include hemifacial microsomia, first and second branchial arch syndrome, otomandibular dysostosis, oculo-auriculo-vertebral
spectrum, facio-auriculo-vertebral syndrome, Goldenhar syndrome and lateral facial dysplasia.

The incidence of CFM is not known but it represents the second most common group of birth defects after cleft lip and palate\(^,\)\(^6,\)\(^7\). Kay et al advocated that microtia should be considered a microform of CFM\(^7\). About 45% of patients have affected relatives, and 5-10% have affected siblings\(^8,\)\(^9\).

The objectives of the study are to review the clinical presentations, assessment, treatment and outcome of the patients with CFM who were treated by the authors in Srinagarind Hospital, Khon Kaen University between 1993-2011. The results of the analysis may be useful for setting appropriate guidelines for diagnosis and treatment of these patients, goals of treatment and long-term outcome.

**Patients and Method**

**Setting**

Srinagarind Hospital is the only university hospital and the main tertiary referral center for northeastern Thailand. The geopolitical region has a population of about 22 million and is the poorest region and one which has numerous endemic and genetic diseases.

**Material and Method**

**Study design**

The medical records of the patients with CFM seen and managed by the authors in Srinagarind Hospital between 1993 and 2011 were reviewed. The diagnosis of CFM was accomplished using clinical assessments and radiology reports. The details of these, the treatment and outcomes were reviewed.

The protocol of this study was reviewed and approved by the Ethics Committee of Khon Kaen University, using the standards set out in the Declaration of Helsinki. Additionally, written informed consent was obtained from each patient or from the parents (in which case assent was sought from the pediatric patients).

**Results**

Table 1 shows detail of the 23 patients, which included 14 males (M) and 9 females (F). Four patients had bilateral deformities and 19 unilateral deformities. The clinical presentation ranged from microtia to more severe deformities involving the mandible, zygoma and maxilla. Syndromic CFM presented in only one patient. Fig. 1 shows the geographic distribution of the 23 patients (using Geographic Information System-GIS).

Khon Kaen, Maha Sarakham and Sakon Nakhon were the provinces in which the highest number of patients was found.

The surgical treatments included (a) correction of macrostomia in 3 patients (b) distraction osteogenesis in 1 (c) staged ear reconstruction in 10 (d) orbit-maxillary-mandibular surgery in 6 (e) soft tissue reconstruction with dermis fat graft in 1 (f) vascularized free tissue transfer in 2 (g) cleft lip-cleft lip nose repair in 1 and (h) palatoplasty in 1. Almost all of the patients were satisfied with treatment and many of them were still in the growing skeletal age (at the time of this writing) and had plan for further staged reconstruction.

**Patient reports**

**Patient No. 6**

A female patient, born in 1973 in Sakon Nakhon province, presented with hypoplasia of the maxilla and mandible with left microtia. The surgical reconstruction included LeFort I osteotomy with bilateral sagittal split, sliding genioplasty and soft tissue reconstruction with a parascapular free flap. At 27 years of age, she was satisfied with her facial appearance and no longer came in for follow-up.

**Patient No. 10**

A male patient, born in 1997 in Khon Kaen province, presented with macrostomia, hypoplasia of the right maxilla and eight mandibles, right microtia and hand anomalies. Aural atresia with right conductive hearing loss was noted. Correction of macrostomia, hand deformities, and staged ear reconstruction were performed. At the time of the last follow-up at the age of 14, his parents and he were satisfied with his appearance. Additional maxillary and mandibular surgeries and soft tissue reconstruction were, However, planned once he reached the age of facial skeletal
<table>
<thead>
<tr>
<th>Patient No.</th>
<th>Sex</th>
<th>Birth year</th>
<th>Province</th>
<th>Clinical finding(s)</th>
<th>Treatment(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>M</td>
<td>1987</td>
<td>Maha Sarakham</td>
<td>Right microtia</td>
<td>-Staged ear reconstruction</td>
</tr>
<tr>
<td>2</td>
<td>F</td>
<td>1989</td>
<td>Khon Kaen</td>
<td>Left lateral facial cleft</td>
<td>-Correction of macrostomia</td>
</tr>
<tr>
<td>3</td>
<td>M</td>
<td>1981</td>
<td>Khon Kaen</td>
<td>Bilateral CFM</td>
<td>-Left ear reconstruction</td>
</tr>
<tr>
<td>4</td>
<td>M</td>
<td>1978</td>
<td>Khon Kaen</td>
<td>Left microtia</td>
<td>-Left ear reconstruction, prefabricated technique</td>
</tr>
<tr>
<td>5</td>
<td>M</td>
<td>1985</td>
<td>Si Sa Ket</td>
<td>Bilateral CFM, asymmetry of mandible</td>
<td>-Mandibular onlay bone graft</td>
</tr>
<tr>
<td>6</td>
<td>F</td>
<td>1973</td>
<td>Sakon Nakhon</td>
<td>Right CFM</td>
<td>-LeFort I osteotomy + sagittal split-parascapular free flap -dermis fat graft-sliding genioplasty</td>
</tr>
<tr>
<td>7</td>
<td>M</td>
<td>1985</td>
<td>Kalasin</td>
<td>Right microtia</td>
<td>-Staged ear reconstruction</td>
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<td>8</td>
<td>F</td>
<td>1973</td>
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<td>Right CFM</td>
<td>-LeFort I osteotomy + sagittal split</td>
</tr>
<tr>
<td>9</td>
<td>F</td>
<td>1995</td>
<td>Chaiyaphum</td>
<td>Right CFM</td>
<td>-Correction of macrostomia</td>
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<tr>
<td>10</td>
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<td>1997</td>
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<td>Right CFM, asymmetry of mandible</td>
<td>-Staged ear reconstruction</td>
</tr>
<tr>
<td>11</td>
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<td>1978</td>
<td>Nong Bua Lam Phu</td>
<td>Right microtia</td>
<td>-Staged ear reconstruction</td>
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<tr>
<td>12</td>
<td>M</td>
<td>1994</td>
<td>Maha Sarakham</td>
<td>Right microtia</td>
<td>-Staged ear reconstruction</td>
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<td>13</td>
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<td>-Distraction osteogenesis</td>
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<tr>
<td>14</td>
<td>M</td>
<td>1983</td>
<td>Maha Sarakham</td>
<td>Left CFM and Left unilateral cleft lip</td>
<td>Cleft lip-nose revision + alveolar bone graft + calvarial bone graft + genioplasty</td>
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<tr>
<td>15</td>
<td>M</td>
<td>1981</td>
<td>Khon Kaen</td>
<td>Bilateral CFM</td>
<td>-dermis fat graft to upper lip LeFort I osteotomy + bilateral sagittal split -Parascapular flap free flap for soft tissue augmentation</td>
</tr>
<tr>
<td>16</td>
<td>F</td>
<td>1989</td>
<td>Khon Kaen</td>
<td>Left microtia</td>
<td>-Staged ear reconstruction</td>
</tr>
<tr>
<td>17</td>
<td>M</td>
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<td>Roi Et</td>
<td>Right microtia</td>
<td>-Staged ear reconstruction</td>
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<td>18</td>
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<td>1985</td>
<td>Khon Kaen</td>
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<td>-</td>
</tr>
<tr>
<td>19</td>
<td>M</td>
<td>2005</td>
<td>Kalasin</td>
<td>Bilateral CFM</td>
<td>-</td>
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<tr>
<td>20</td>
<td>F</td>
<td>2000</td>
<td>Sakon Nakhon</td>
<td>Left CFM</td>
<td>-Correction of macrostomia</td>
</tr>
<tr>
<td>21</td>
<td>F</td>
<td>2004</td>
<td>Nong Khai</td>
<td>Bilateral microtia, bilateral hearing loss and cleft palate</td>
<td>-Palatoplasty</td>
</tr>
<tr>
<td>22</td>
<td>F</td>
<td>1996</td>
<td>Khon Kaen</td>
<td>Syndromic right CFM with torticollis, scoliosis and right upper extremity anomalies.</td>
<td>-Orbital, zygomatic, and temporomandibular joint reconstruction</td>
</tr>
<tr>
<td>23</td>
<td>F</td>
<td>1991</td>
<td>Nakhon Ratchasima</td>
<td>Left microtia</td>
<td>-Staged ear reconstruction</td>
</tr>
</tbody>
</table>
Patient No. 12
A male patient, born in Maha Sarakham province in 1994, presented with right microtia. Staged ear reconstruction was performed with a costochondral cartilage graft (at the age of 6). He was lost to follow-up but revisited the hospital in 2011, at the age of 17. His parents and he were satisfied with the results, however, further revision was planned.

Patient No. 13
A male patient born in Khon Kaen in 1998,
presented with hypoplasia of the left zygoma, orbit and mandible. Distraction osteogenesis of the left mandible was performed when the patient was 2 years old. At the time of last follow-up in 2011 (when the patient was 13), he was satisfied with his facial appearance.

**Patient No. 20**
A female patient, born in 2000 in Sakon Nakhon province, presented with left macrostomia, hypoplasia of the maxilla and mandible and left microtia. A CT scan was performed which revealed the hypoplasia. Surgical correction of the macrostomia was performed when the child was 6 years of age (Fig. 6). She was lost to follow-up after correction of the macrostomia.

**Patient No. 22**
A female patient, born in 1996 in Khon Kaen province, presented with syndromic CFM with clinical features of hypoplasia of the orbit, maxilla and mandible, lagophthalmos, right microtia, bilateral hearing loss, torticollis, scoliosis, and right upper extremity anomalies. A CT scan revealed hypoplasia of the zygoma, orbit, maxilla and mandible. Diagnosis of syndromic CFM was made. When the patient was 11 years of age, orbital reconstruction was performed with a calvarial bone graft and TMJ reconstruction with a costochondral graft. At the time of follow-up in 2011 at the age 15, the patient was feeling more satisfied with her facial appearance. Facial analysis and planning for further bony surgery have to wait until after the age of facial skeletal maturity (Fig. 7-9).

**Discussion**
Craniofacial microsomia (CFM) is characterized by facial asymmetry resulting from a unilateral or bilateral congenital deficiency of the affected skeletal and soft tissue structures, derived from the first and second branchial arches. The etiology can be divided into environmental, heritable, multifactorial and unknown. It is considered the second most common craniofacial anomaly after cleft lip and palate and has an estimated prevalence between 1:5,600 and 1:26,550 live births. The male to female ratio is 3:24.

The diagnosis of CFM can be based on clinical examination and the common findings are: macrostomia, cleft lip and/or palate, ankylosis, asymmetric mandible, mid-face hypoplasia, malocclusion, epibulbar dermoid, asymmetric shortening of the palpebral fissure, microphthalmia or anophthalmia, coloboma of the upper eye lid, vertical displacement of the orbit and vertebral anomalies. The clinical findings in individuals with craniofacial microsomia can be associated with: syndromes, developmental anomalies,
and/or sequences such as VATER, CHARGE, MUECS, and OEIS; which led to the hypothesis that these conditions may represent developmental abnormalities resulting in anomalies that may be a part of a broad spectrum\(^{(11)}\).

Classification of patients with CFM is difficult and the most effective and universally-accepted and used in treatment plans are (a) the earlier classification by Pruzanski\(^{(12)}\) and (b) Kaban’s modification\(^{(13)}\). Later, two more comprehensive and modified classifications were introduced, including (c) the Orbital Mandible Ear Nerve Soft tissue (OMENS) classification\(^{(14)}\), and (d) the OMENS-Plus which included associated extracranial manifestations\(^{(15)}\). Birgfeld et al developed (e) a modified pictorial OMENS-Plus and (f) a standardized photographic protocol for a more comprehensive and structured phenotypic assessment tool for CFM\(^{(16)}\).

A multidisciplinary craniofacial team comprising a medical geneticist or physician who specializes in craniofacial disorders, a plastic and craniofacial surgeon, a pediatric dentist, an orthodontist, an audiologist/otolaryngologist, a speech pathologist, a social worker and a nurse coordinator is needed to ensure a more thorough comprehensive assessment of the problems and provide a better treatment planning\(^{(17)}\). The analysis and record of a patient’s anatomic and functional deficiencies should include facial asymmetry, severity of soft tissue and skeletal deformities, ear findings and hearing evaluation, ophthalmologic examination, as well as the functional requirement, and the subsequent growth and development.
development\(^{(18)}\). Additionally, other non-craniofacial associated anomalies such as the neck, heart, spine, limbs, and kidneys should be evaluated.

Though radiologic imaging is not necessary for making a diagnosis of the patient with CFM, a 3-D CT scan is helpful for refining the anatomy and morphology for pre- and post-surgical treatment planning of both bony and soft tissue deficiency and outcome evaluation of these patients. Radiographic evaluation of patients with CFM may reveal asymmetric hypoplasia of the zygoma, maxilla and mandible. Additionally, other imaging studies may be indicated, including x-rays of the cervical spine, echocardiogram, renal ultrasound examination, and CT scan of the temporal bone in patients with significant hearing impairment.

Airway and feeding are among the first priorities during early management of these patients and perinatal endotracheal intubation, tracheostomy, mandibular distraction, and gastrostomy may be indicated. The options for surgical techniques for the correction of the skeletal and soft tissue deformities of the patient with CFM include (a) correction of macrostomia (patient No. 2, 9 and 20) (b) bone reconstructions (patient No. 5, 6, 8, 14, 15 and 22) (c) distraction osteogenesis (patient No. 13) (d) staged ear reconstruction (patient No. 1, 3, 4, 7, 10, 11, 12, 16, 17 and 23) (e) dermis-fat grafts (patient No. 14) and (f) microvascular free tissue transfers (patient No. 6, and 15).

Macrostomia and pre-auricular skin appendages are the most obvious deformities that may need to be corrected at an early age, especially since early repair of macrostomia can significantly improve feeding. The surgical technique should include (a) the correct position of the oral commissure for measuring the distance from the midline to the oral commissure on the non-cleft side (b) the repair of the underlying muscle and (c) the use of a Z-plasty to reduce scarring (patient No. 20, Fig. 7). For removal of a pre-auricular skin tag, it is important that the cartilage remnants be completely removed to avoid a depression deformity of the scar.

Assessment of hearing by an audiologist should be done as early as possible- that is, in the first two weeks of life- using a brain stem auditory response test. Parents of patients with CFM who have hearing impairment should receive guidance regarding recommendations for hearing aids, prevention of further hearing loss, and close monitoring for speech and language development.

Prior to planning surgical correction of microtia, it is recommended that a CT scan of the temporal bone be done between 5 and 6 years of age to assess the external ear canal, middle- and inner-ear structures in order to identify candidates for whom surgical correction would improve hearing.

For skeletal reconstruction, most patients with CFM with mandibular hypoplasia can be managed conservatively during the early years, except for the patients who are functionally affected by the mandibular hypoplasia asymmetry or absence of ramus, condyle and temporomandibular joint fossa\(^{(17)}\) who may require a bone graft and/or mandibular distraction osteogenesis to lengthen the mandible and/or create a functional TMJ. If indicated, the reconstruction of ramus can be performed by the use of a costochondral graft reconstruction between the ages of 3 to 4 years. Rib or iliac bone grafting has been used for mandibular reconstruction (Pruzansky type III-absence of the ramus, condyle and temporomandibular joint)\(^{(18)}\). The reconstructed ramus can later be distracted to provide more length of ramus. Correction of the hypoplastic orbit (or distropia) should be delayed and may be considered when the patient is between ages of 5 and 7 years.

A costochondral graft is used for ear cartilage framework reconstruction of microtia and may be performed around the age of 8 years or older which allows for better positioning, particularly in patients with a more severe deformity.

Mandibular distraction osteogenesis may be indicated in patients with grade I, IIA and IIB mandibular hypoplasia, and can be applied at any age from neonates to adults\(^{(19)}\). In the patient with unilateral craniofacial microsomia, the surgical goal is to increase the vertical or superoinferior dimension of the ramus and movement of the chin point to the midline\(^{(17)}\).

Any definitive skeletal reconstructive procedure should be delayed until growth of the facial skeleton is complete; that is, after the age of 18 years, depending on orthodontic and orthognathic assessment of the deformities.

Minimal or moderate soft tissue deficiencies may not require any treatment. Consideration of any soft tissue augmentation may be performed after the age of 18 years; that is, at the time of, or during, orthognathic surgery. Treatment by fat injections has produced significant post-treatment atrophy which may be from insufficient skin expansion\(^{(20)}\). Dermis fat graft from the lower abdomen or thighs may be used. However, in large soft tissue deficiencies, free vascularized tissue transfers are recommended for soft
tissue augmentation[21,22]. Objective assessment of the extent and magnitude of soft tissue and bony deficiencies is important for better surgical planning; such as soft tissue expansion and free flap transfer. The combination of distraction osteogenesis and microvascular free tissue transfer has been reported to provide more functional and esthetic outcomes[23].

Conclusion
The goals of treatment for CFM are to (a) manage respiratory insufficiency and feeding problems (b) maximize hearing and ability to communicate (c) improve facial symmetry and proper facial growth and (d) optimize dental occlusion. These patients require staged reconstruction and coordinated care by an experienced multidisciplinary craniofacial team for (a) comprehensive assessment (b) optimum treatment with planning, execution and long-term outcome adapted to the anatomical and functional deformities (c) and consideration of the needs of patients and their family and other involved stakeholders.

Acknowledgement
The present study was supported by the Tawanchai Foundation for Cleft Lip-Palate and Craniofacial Deformities and the Center of Cleft Lip-Cleft palate and Craniofacial Deformities, Khon Kaen University, in Association with the Tawanchai Project. The authors thank (a) all the patients with their families and the staff of the Foundation (b) the Cleft Center and the Audio-Visual Unit of Faculty of Medicine, Khon Kaen University, for their supportive participation and (c) Mr. Bryan Roderick Hamman and Mrs. Janice Loewen-Hamman for their assistance with the English-language presentation of the manuscript.

Potential conflicts of interest
None.

References

มิตร บวรศิลป์, ชวนชื่น พูนศักดิ์, ประชาธนา เชาวานิช, สงวนศักดิ์ ธนวัฒนาภูมิ

ภูมิหลัง: ครานิโอเฟเชียลไมโครโซเมียเป็นภาวะความพิการแต่กำเนิดที่มีความซับซ้อนและมีความเกี่ยวข้องปัญญามากมาย ทำให้การรักษาเป็นเรื่องที่มีความท้าทายและยากลำบากได้

วัตถุประสงค์: เพื่อสำรวจการรักษาและการประเมินผลที่ได้รับจากการรักษาโดยผู้นิพนธ์ในโรงพยาบาลศรีนครินทร์ มหาวิทยาลัยขอนแก่น

วัสดุและวิธีการ: การศึกษานี้เป็นการทบทวนทั้งการпущของครานิโอพฤศจิกและอาการจากการรักษาของผู้ป่วยที่ได้รับการรักษาโดยผู้นิพนธ์ในโรงพยาบาลศรีนครินทร์ มหาวิทยาลัยขอนแก่นในระหว่างปี พ.ศ. 2536-2554.

ผลการศึกษา: มีผู้ป่วยทั้งหมด 23 ราย เป็นเพศชาย 14 ราย และเพศหญิง 9 ราย 4 รายเป็นความพิการข้างเดียว 19 ราย เป็นความพิการข้างเดียว และ 1 ราย เป็นกลุ่มอาการ การตรวจพบทางคลินิกพบได้ตั้งแต่อาการกดทับหน้าท้องของอยู่ไปจนถึงความพิการรุนแรงของกระดูกขากรรไกรต่าง กระดูกโหนกแก้ม และกระดูกขากรรไกรบน การรักษาทางการศึกษาประกอบด้วย การแก้ไขภาวะการเจริญเติบโต การยืดถ่างที่ดี การเตรียมการรับผิดรับ การเสริมสร้างใบหูแบบเป็นระยะการผ่าตัดกระดูกขากรรไกรต่าง-กระดูกขากรรไกรบน-กระดูกขากรรไกรต่าง การเสริมสร้างใบหู โดยใช้การปลูกถ่ายและการทำหนังแท้ การผ่าตัดและผ่าท้องต่าง เกิดขึ้นในผู้ป่วยทั้งหมด 18 ราย ผู้ป่วยทั้งหมดได้รับการรักษาอย่างละเอียดอยู่ในระหว่างการผ่าตัดและรักษาขั้นตอนต่างๆ ผู้ป่วยส่วนใหญ่จะพอใจผลการรักษาและมีการกล่าวถึงความสามารถในการใช้ชีวิตที่ใกล้ชิดกับที่เดิมมากขึ้นในระยะยาว

สรุป: การประชุมของผู้ป่วยครานิโอเฟเชียลไมโครโซเมียเป็นความพิการที่ซับซ้อนและยากลำบากในการรักษาและการประเมินผล แต่ผลการผ่าตัดและการรักษาที่ได้รับการดำเนินการอย่างละเอียดและมีประสิทธิภาพสูงทำให้ผู้ป่วยสามารถใช้ชีวิตอยู่ได้ตามปกติในระยะยาวได้ การรักษาต้องมีการวางแผนที่ดีที่สุดในการรักษาและการผ่าตัด รวมถึงการระดมทีมที่มีประสบการณ์และทักษะในการรักษาขั้นตอนต่างๆ ที่ต้องการเพื่อให้ผู้ป่วยสามารถใช้ชีวิตอยู่ได้ตามปกติในระยะยาวได้