Basilar impression and osteogenesis imperfecta: a 21-year retrospective review of outcomes in 20 patients


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Object. Basilar impression (BI) secondary to osteogenesis imperfecta (OI) is a rare but debilitating condition that is often progressive unless it is halted. More recently, ventral decompression surgery has been advocated for this condition. This study is a retrospective review of the 21-year experience of ventral decompression surgery and dorsal occipitocervical fixation in patients with BI secondary to OI and is the largest patient series reported to date.

Methods. Twenty patients treated between 1982 and 2003 by the senior author at the authors’ institution were included in this study. All patients underwent ventral decompression surgery followed by dorsal craniocervical stabilization. Patients were followed up for a median of 10 years.

Results. There were no intraoperative or perioperative deaths. Postoperatively, 16 of 20 (80%) patients showed objective improvement or maintained their good preoperative level of function. After surgery, of the 15 patients admitted with Karnofsky Performance Scale (KPS) scores of 70% or less, 11 improved, two remained unchanged, one patient’s condition deteriorated, and one patient died of an unrelated cause. Of five patients admitted with a KPS score of 80% or greater, no patient’s condition deteriorated in the short- and midterm period, but one patient had recurrence 15 years after surgery. At the end of follow-up, 25% of the patients had recurrence of brainstem compression symptoms or had died, and 15% showed no improvement after surgery. All of the remaining patients (60%) had sustained a long-term benefit from surgery.

Conclusions. Aggressive ventral decompression surgery and dorsal stabilization for patients with BI secondary to OI can not only halt disease progression but can also produce a good and sustainable long-term functional outcome, even in those patients who present as severely symptomatic. Patients who presented early with minor symptoms had good long-term outcomes. (DOI: 10.3171/SPI-07/12/594)

KEY WORDS • basilar impression • basilar invagination • craniocervical surgery • occipitocervical fixation • osteogenesis imperfecta

Basilar impression is a rare and striking mechanical deformity of the CVJ that is characterized by migration of the upper cervical spine and the odontoid peg into the base of the skull. Patients with BI can present with an impressive array of features that arise as a consequence of brain stem and cerebellum compression, disturbance of cerebrospinal fluid circulation, and/or mechanical stretching of cranial nerves. These features are often progressive, and unless BI is halted in some way, can lead to rapid neurological deterioration, respiratory arrest, or even sudden death.

Basilar impression may arise as a congenital deformation but is more commonly an acquired disorder of bone-softening conditions such as OI, rickets, hyperparathyroidism, and Paget disease. Osteogenesis imperfecta is a rare inherited bone disorder that results in fractures caused by trivial bony injuries, and was first reported to be linked to BI in the 1940s. Curiously, only 8 to 25% of patients with OI eventually develop BI, and not all patients who develop BI become clinically symptomatic.

Basilar impression has been described in the literature for centuries and was first reported by Ackermann in 1790. The term “basilar impression” was first coined in 1855 by the anatomists Berg and Retzius. Basilar impression is sometimes referred to as basilar invagination, and these terms are used interchangeably; however, the term “platybasia,” a comparative anatomy term that describes the angle between the clivus and the neural axis, is no longer used to describe the condition.

In 1863 the discovery of BI in human skulls from ancient
European tombs confirmed that BI was not a modern disease as was initially believed. However, the diagnosis of BI remained a postmortem finding. Virchow produced a detailed description of BI in 1876. The first clinical diagnosis of BI based on clinical features in a living patient was made at the beginning of the 20th century by the neurologist Homen, and the first radiological image of BI was produced in 1911 by Schuller, who used the newly discovered x-ray technology. Thirty years later, Chamberlain, in a seminal publication, reported radiographic craniometric measurements as diagnostic criteria for BI, which led to increased attention on the condition. Despite detailed knowledge of BI existing since the 19th century, little was offered in the way of treatment, and the diagnosis of BI was considered fatal. Surgical decompression was first considered as a treatment option for BI in the 1930s in the form of dorsal decompression, but with mixed results.

Investigators from the authors’ institution noted the inadequacy of dorsal decompression for BI and stressed that, because ventral structures (such as the clivus, odontoid peg, and upper ventral cervical spine) in BI were primarily responsible for the most significant compression of the neural axis, ventral decompression (through extended or open-door maxillotomy) would be a more appropriate approach. This recommendation was consistent with the understanding that structures causing compression ventrally require decompression through a ventral route. Our group9 and others14 have reported good early postoperative results of ventral surgery for BI. However, there remains a paucity of long-term outcome reports in the literature of patients with BI treated surgically. In this paper, we report on a retrospective review of patients who underwent ventral decompression surgery of BI secondary to OI in a 21-year period for the purpose of sharing our relatively long experience in managing this condition.

Clinical Material and Methods

Patient Data Collection

We retrospectively reviewed clinical data of all patients with a diagnosis of OI and BI who were treated surgically between 1982 and 2003. We obtained approval for the study from the ethics committee of the National Hospital for Neurology and Neurosurgery and Great Ormond Street Hospitals. Complete case notes and imaging records of 20 patients were evaluated for clinical presentation, radiological features, operative findings, response to treatment, and surgical complications. The diagnoses of OI and BI were established in accordance with guidelines provided by Sillence and based on patient clinical history and examination, as well as radiological findings (using MR images, plain radiographs, and/or computed tomography scans, or in earlier cases, using computed tomography myelograms). Patients were followed up to evaluate their postoperative course and level of daily functioning (which was converted to a KPS score). The median duration of follow-up was 10 years (mean 8.9 years). Whenever possible, follow-up was conducted at outpatient visits at our institution, but for patients unable to attend (often overseas residents), follow-up assessment was performed by the referring physician.

Clinical Status at Presentation

The majority of patients presented with severe physical disability and advanced symptomatic BI. Eight patients (40%) had sleep apnea, and one of these patients was already receiving oxygen at home. Twelve patients (60%) were regular wheelchair users, and 16 (80%) had muscular weakness with pyramidal signs. Nine patients (45%) had dysphagia and four (20%) had a diminished gag reflex. Five patients (25%) were found to also suffer from vertigo in accordance with data from a recent publication. Two patients presented with sudden quadriaparesis: one patient after a traffic accident with a flexion/extension neck injury and the other after waking up with paresis after using an unfamiliar hard pillow while on vacation. Mild hydrocephalus and asymptomatic syringomyelia were present in two patients. Fourteen patients (70%) had multiple previous skeletal fractures, nine (40%) had severe and progressive scoliosis, and seven (35%) had dentinogenesis imperfecta. Details of all patients’ presenting features observed in our study, and for comparison, those in the study of Sawin and Menezes, are summarized in Table 1. The three most frequent clinical features of these patients were nystagmus, headache, and ataxia. At presentation, 15 of the 20 patients scored 70% or lower on the KPS, whereas the remaining five patients scored 80% or higher (Tables 2 and 3).

Sensory Deficit and Pain

We found facial numbness to be a prominent feature in our patient cohort. During direct questioning, eight patients (40%) noted facial numbness; six had abnormal light touch and pinprick sensation in the maxillary division of the trigeminal nerve, and two (10%) had impaired sensation in the mandibular division of the trigeminal nerve. No patient had a sensory deficit in the ophthalmic division of this nerve. Three of the eight patients with facial numbness subsequently developed severe TN pain in the previously numb facial areas.

Clinical Features

<table>
<thead>
<tr>
<th>No. of Patients (%)</th>
<th>Current Study</th>
<th>Savin &amp; Menezes, 1997</th>
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<tr>
<td>nystagmus</td>
<td>11 (55)</td>
<td>7 (28)</td>
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<tr>
<td>ataxia</td>
<td>10 (50)</td>
<td>8 (32)</td>
</tr>
<tr>
<td>headache</td>
<td>10 (50)</td>
<td>19 (76)</td>
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<tr>
<td>dysphagia</td>
<td>9 (45)</td>
<td>15 (60)</td>
</tr>
<tr>
<td>scoliosis</td>
<td>9 (45)</td>
<td>5 (20)</td>
</tr>
<tr>
<td>facial numbness</td>
<td>8 (40)</td>
<td>—</td>
</tr>
<tr>
<td>dentino genesis</td>
<td>7 (35)</td>
<td>—</td>
</tr>
<tr>
<td>vertigo</td>
<td>5 (25)</td>
<td>—</td>
</tr>
<tr>
<td>hydrocephalus</td>
<td>4 (20)</td>
<td>—</td>
</tr>
<tr>
<td>hearing loss</td>
<td>4 (20)</td>
<td>4 (16)</td>
</tr>
<tr>
<td>facial pain (TN)</td>
<td>3 (15)</td>
<td>—</td>
</tr>
<tr>
<td>quadriaparesis</td>
<td>2 (10)</td>
<td>12 (48)</td>
</tr>
<tr>
<td>syrinx</td>
<td>2 (10)</td>
<td>—</td>
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* — = unknown.
Severe kyphoscoliosis was present in almost one half of our patients. Unlike the classical type of left-to-right or right-to-left kyphoscoliosis, in patients with BI, the scoliosis appears to curve the spine inward, perhaps best described as “internal scoliosis” (Fig. 1).

Surgical Procedure

Both an elective tracheostomy and ventral decompression surgery were performed in all cases; the decompression was performed through an open-door or extended maxillotomy approach, removing the “knuckle” of the clivus, anterior rim of foramen magnum, odontoid, and basal axis as described previously. An electrophysiologist exclusively monitored intraoperative somatosensory and (more recently) motor-evoked potentials to detect any inadvertent distress caused to the neural axis. Patients were kept under sedation for 24 to 72 hours after ventral decompression and were fed only through the PEG tube for as long as 1 week.

The second surgical stage involving dorsal occipitocervical fixation, from the occiput down to the C-7 or T1–2 level, was undertaken to prevent further progression of BI. Posterior cervical fixation was made difficult by the short neck and the severely distorted CVJ anatomy of the patients. Various fixation instruments have been used through the years, including a Ransford loop and the more recently devised occipitocervical fixation instruments. The head position for fixation was neutral or at a slight extension. Various types of bone grafts were used, such as bone harvested from the iliac crest, ribs, and outer table of the occipital skull. Although it was possible to perform both ventral decompression and dorsal occipitocervical fixation in exceptionally fit patients, a delay of a few days or as long as 1 to 2 weeks without cervical skeletal traction was the usual practice. Before decompressive surgery, patients under-
went lung function tests and a sleep test to document the level of respiratory dysfunction. A PEG tube was also inserted in patients electively in preparation for feeding in the immediate postoperative period.

**Results**

*Demographic Data*

There were 11 men and nine women in the study cohort, with a mean age of 27 years (range 12–52 years; only one patient was younger than 18 years of age). Ten patients presented with Sillence Type III OI, five with OI Type IV, and another five with OI Type I. There were no patients with Sillence Type II OI, which is the fatal form of the disorder. Most patients were from the United Kingdom, but two patients were from Australia, and one patient each from Italy, New Zealand, Norway, and the US. Details of the patients’ radiological features at presentation and the correlation of clinical signs with radiological findings is the subject of another paper (unpublished data).

Patients who were offered surgery were categorized into two groups: those who presented with rapid clinical deterioration of symptoms but were relatively well and those who presented with established severe disabling features of brainstem compression. Fifteen patients were admitted with a KPS score from 50 to 70%, and five were admitted with a KPS score of 80% or greater.

*Short-Term and Long-Term Functional Outcome*

Full ventral decompression was achieved in all patients. There were no intraoperative or perioperative deaths (within 30 days of surgery). However, one patient died 2 months after undergoing surgery for uterine sepsis because of retained gestational products from a previously terminated pregnancy. At discharge, 16 (80%) of the 20 patients showed an objective improvement of one or more KPS score levels or maintained a good level of preoperative function (KPS score ≥ 80%). Of the 15 patients admitted with a preoperative KPS score in the range of 50 to 70%, 11 improved, two remained unchanged, one patient’s condition deteriorated, and one patient died of causes unrelated to BI and OI. All five patients admitted with a KPS score of at least 80% maintained or improved their preoperative clinical status. Patients with preoperative TN continued to have severe pain after ventral decompression surgery. Five of nine patients showed improvement in their preexisting dysphagia after surgery.

At the 5-year follow-up, most patients remained at their postoperative status noted previously, but there was a significant change in status in two patients. One of these two patients developed a rapid generalized deterioration of symptoms of BI and subsequently died, whereas the other patient developed a recurrence of signs and symptoms of brainstem compression. All five patients who presented with a preoperative KPS score of 80% or more maintained this level of functioning at 5 years.

At the end of the follow-up period, nine patients scored 80 to 90% on the KPS, three scored 70%, and five patients scored 60% or lower. A total of three patients (15%) had developed a recurrence of clinical BI features at 2, 10, and 15 years after surgery, respectively. Those patients who developed recurrence at 2 and 15 years subsequently died, bringing the total number of deaths in this cohort to three (15%) at the end of follow-up. Of the five patients with a preoperative KPS score of 80% or more, one patient’s condition deteriorated 15 years after undergoing surgery. At the end of the study, 15% of the patient cohort did not show any improvement after surgery, and 25% had either recurrence of symptoms or had died after rapid or long-term deterioration.

Those patients who did not improve immediately or shortly after surgical decompression did not show significant improvement in neurological function in the long term. Of those patients who had good immediate neurological improvement, only one patient’s condition deteriorated 15 years after initially undergoing surgery.

*Morbidity and Special Problems*

Posterior wound infection occurred in two patients who had very short necks, which posed particular difficulties when obtaining access for instrumentation. Significant upper airway problems such as dysphagia, aspirations, nasal air escaping, and others occurred in six patients (30%). Almost one half of the patients developed chest infections, two had posterior surgical wound infections, and one developed meningitis that fully responded to systemic antibiotic treatment. The only patient whose condition deteriorated did so after a complication of loosening of dorsal metal work while undergoing otolaryngology surgery. Two patients required prolonged tracheostomy support (for 1 month and 1 year, respectively). A summary of postoperative complications is detailed in Table 4.

Three patients who had severe upper airway problems with nasal speech, recurrent aspirations, and progressive dysphagia were treated in three different ways. One of these patients was successfully treated with a reduction glossotomy for a subjectively perceived, progressively enlarging tongue, caused by a reduction in oral volume as a result of the progressive invagination; another patient was treated with soft palate lengthening. The last patient, who had severe recurrent aspirations and who had required prolonged tracheostomy with PEG feeding support, finally underwent closure of the larynx to prevent the overspill into the trachea. After this treatment, the patient was able to

<table>
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<th>Complication</th>
<th>No. of Patients (%)</th>
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<tr>
<td>postop chest infection</td>
<td>9 (45)</td>
</tr>
<tr>
<td>dysphagia (new onset)</td>
<td>4 (20)</td>
</tr>
<tr>
<td>recurrence of brainstem compression</td>
<td>3 (15)</td>
</tr>
<tr>
<td>surgical wound infection</td>
<td>2 (10)</td>
</tr>
<tr>
<td>nasal air escaping, nasal speech</td>
<td>2 (10)</td>
</tr>
<tr>
<td>prolonged tracheostomy</td>
<td>2 (10)</td>
</tr>
<tr>
<td>loosening of metal work</td>
<td>1 (5)</td>
</tr>
<tr>
<td>neck kyphosis</td>
<td>1 (5)</td>
</tr>
<tr>
<td>meningitis</td>
<td>1 (5)</td>
</tr>
<tr>
<td>depression</td>
<td>2 (10)</td>
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swallow and was weaned off PEG tube feeding. The three patients with TN continued to suffer from severe pain, and unfortunately, successful treatment of these patients has eluded us thus far.

Discussion

Symptomatic BI is a relentlessly progressive condition that can lead to catastrophic complications and even death, if left untreated. Affected patients are often young and require definitive long-term treatment. To date, this is the longest follow-up study of outcome of aggressive surgical management of BI secondary to OI. We have shown that early clinical improvement after ventral decompression surgery in this group of patients is achievable, including in those who present with rapidly deteriorating signs or severe disability, and that the clinical improvement is sustainable over the long term. No patient died intra- or perioperatively, and one patient died 2 months after surgery because of causes unrelated to BI. In particular, patients who had presented with early disease status benefited from surgery both in the short and long term.

In the immediate postoperative period, the vast majority of our patients improved, and in the long term, 15% developed symptomatic recurrence. Thus, in patients with severe BI, adequate ventral decompression together with a secure, primary posterior, instrumented craniocervical fixation using a bone graft can markedly improve symptoms of BI and reduce long-term recurrence. It is difficult to comment on the formation on pseudoarthrosis in the patients who have a severely distorted CVJ anatomy in the short term. Although we do not have complete radiological long-term follow-up data for all of our patients, Sawin and Menezes have reported a radiological recurrence of disease in 80% of their patients after ventral decompression and posterior fixation, with 30% of these patients developing clinical symptoms. One of the reasons Sawin and Menezes have observed a higher symptomatic recurrence rate is because they essentially described the progressive BI changes associated more with pediatric patients; the mean age of the patients in their study was 11.9 years, whereas we reviewed older patients with a mean age of 28.6 years.

We observed a high rate of postoperative upper airway problems and aspiration-related chest infection. Adverse affects of ventral decompression surgery on swallowing and breathing is something that we have previously noted, particularly in a pediatric population. This group of patients already has a precarious swallowing function, and the surgical incision along with the subsequent healing process can further affect swallowing adversely. A number of other factors may also lead to upper airway dysfunction. First, the deformity of BI results in both compression of the central respiratory center and mechanical stretching of the lower cranial nerves, leading to a central cause of dysphagia. Second, the process of distortion that is evident in severe forms of BI also affects the maxilla, reducing the oral volume by displacing the tongue forward and giving a false sense of a progressively enlarging tongue. This distortion also impairs both the oral and laryngeal phases of swallowing, speech, and upper airway function. We did not observe a relationship between the type of posterior instrumentation and dysphagia in this cohort. In selected patients, we found reduction glossotomy, soft palate lengthening, and even closure of the larynx to be effective.

Basilar Impression and TN

Basilar impression is an unusual cause of TN. In our experience, successful surgical decompression of BI did not lead to resolution of TN symptoms. In addition, conventional TN treatment with radio frequency or injection techniques has been singularly unsuccessful in these patients. We postulate that the base of the skull deformity begins at the epicenter of the skull base and spreads outward (Fig. 2), mechanically stretching cranial nerves including the trigeminal nerve (Fig. 3). The maxillary division of the trigeminal nerve (which exits the skull through the more medial foramen rotundum) seems to be preferentially prone to these distorting forces, rather than the mandibular division (which exits the skull through the more lateral foramen ovale). The ophthalmic (first) division located furthest from the distorted base of the skull (which exits through the superior orbital fissure) does not seem to be affected, even in cases of severe BI.

Etiology of BI Secondary to OI

The etiology of BI in OI remain unclear. There is some evidence to support the hypothesis that BI in OI develops as a result of the failure of weight-bearing parts of the skull base exacerbated by early childhood erect posture. Inaba and Wakisaka, however, have recently reported BI, Chiari malformations, and syringomyelias in Weiser–Maples guinea pigs, which are quadrupedal animals. Variation in skull shape and repeated CVJ microfractures that heal in an abnormal position are among the other hypotheses that have been suggested. However, recent biomechanical studies have indicated that the lower C4–C6 segments bear the greatest cranial biomechanical stress load, not the CVJ, which is much more severely affected in BI than the...
lower cervical column segments that are spared by the disease.

The question that arises is not why BI develops in patients with OI, but why does it not develop in all of them? Our impression is that many patients with OI have a variable degree of BI, but only a few become symptomatic, which is consistent with observations by Sillence, who reported that of patients with OI Type III/IV who attended a random clinic, 30% had radiological evidence of BI, but only one third of the 30% were symptomatic.

In the last decade, we have seen a decline in the number of referred cases of BI to our unit. Around the same time, changes in the treatment of OI saw the introduction of bisphosphonates, which are pharmacological agents that have been shown to reduce bone resorption and increase bone mineral density, decreasing both pain and rates of fracture by altering osteoclast and osteoblast activities. Results of studies to determine whether these drugs have a role in delaying and modifying BI in patients with OI are yet to be published.

**Conclusions**

In our experience, aggressive ventral decompression surgery combined with dorsal occipitocervical fixation produces a good long-term symptomatic outcome in patients with BI secondary to OI. Patients who present early in the course of this disease, before prolonged establishment of compression of the neural axis, have recovered particularly well after surgery. Given the relatively high morbidity risks associated with surgical intervention, maximizing the time before undergoing surgery can also have merit. Patients who become symptomatic and begin to deteriorate rapidly, however, should be offered decompressive surgery without delay. The benefits of surgery remain unclear for patients with radiologically confirmed BI but who have minor or stable symptoms and signs. There are patients in our unit with radiologically confirmed BI who we follow up with on a regular basis clinically and, if symptoms arise, radiologically.

There is a significant short- and midterm surgical morbidity associated with treatment that has a protracted recovery period, and both patients and caregivers should be made aware of this potential morbidity. It also should be remembered that BI is not the only complaint of these patients with OI. Patients with OI have a systemic bone disorder and may be using a wheelchair and be severely disabled because of limb problems associated with OI alone.

Management of BI is best undertaken by a specialist multidisciplinary team, which will also need the ability to deal with the complications of breathing, swallowing, speech, and aspiration that may arise with this disease. Treating these patients is not inexpensive for the treating center, where patients spend up to 6 weeks in the hospital, 3 weeks of which is spent in an intensive care unit setting, and a further 1 to 2 months are spent in rehabilitation wards.

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Accepted August 27, 2007.

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