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Case Report

A Case of Crouzon's Syndrome with a True Human Tail

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Abstract
A human tail complicated by Crouzon syndrome is extremely rare, with only eight reported cases of human tails associated with Crouzon syndrome. A human tail is defined as a true human tail or a pseudo-human tail according to the presence or absence of the bone tissue. A 4-year-old boy had a true caudal sacral protrusion from birth and was also diagnosed with Crouzon syndrome. There were no neurological symptoms or vesico-rectal disturbances. The patient underwent a human tail resection at 4 years and 7 months. The postoperative course was uneventful, and there was no recurrence at 6 months postoperatively.
Some reports suggested that Crouzon syndrome and the human tail are associated with FGFR2 gene mutation, but the details are currently unknown. Further genetic searches for cases of the combined human tail and Crouzon syndrome in the future are likely to reveal further associations.

Keywords
human tail, Crouzon syndrome, FGFR2

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Introduction

A human tail complicated with Crouzon syndrome is extremely rare. A human tail is a protrusion located at the midline of the sacral coccyx from birth. It could be either a true human tail with tailbones or a pseudo-human tail lacking them. In this study, we report a case of Crouzon syndrome complicated by a true human tail with a literature review.

Case

A 4-year-old boy was referred to our hospital with a chief complaint of a caudal sacral mass.

Patient's history

At birth, he had a bilateral ocular protrusion, hypoplasia of the maxilla, and a skin mass in the caudal sacral region. He was diagnosed with Crouzon syndrome and a human tail. He underwent occipital cranial enlargement surgery at 1 year and 11 months old and monoblock type craniofacial bone lengthening at 3 years and 2 months old. The mass in the caudal sacral region showed an increase with growth. The family requested surgery because of mild tenderness.

Family history

No family history of the human tail, Crouzon syndrome, or other congenital diseases in parents or siblings.

Preoperative findings

There was a 3 × 4 cm dome-shaped skin mass with a central depression in the caudal sacral region (**Figure 1**). A bone-like mass was palpated subcutaneously. There was no adhesion between bone and skin, no neurological symptoms of the lower extremities, or vesico-rectal disturbances.

Laboratory findings

Blood and biochemical tests were normal.

Imaging

Preoperative plain X-ray and computed tomography (CT) revealed the presence of two fused tailbones. There was no

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indication of either spina bifida or spinal nerve extension to the tailbone (**Figure 2a, b**). The tailbone was curved in a dorsal concave shape, which was different from the normal tailbone, which is curved in a dorsal convex shape.

Diagnosis and treatment

We diagnosed a true human tail based on the physical findings and the imagings. We performed an excision of the human tail when the patient was 4 years and 7 months.

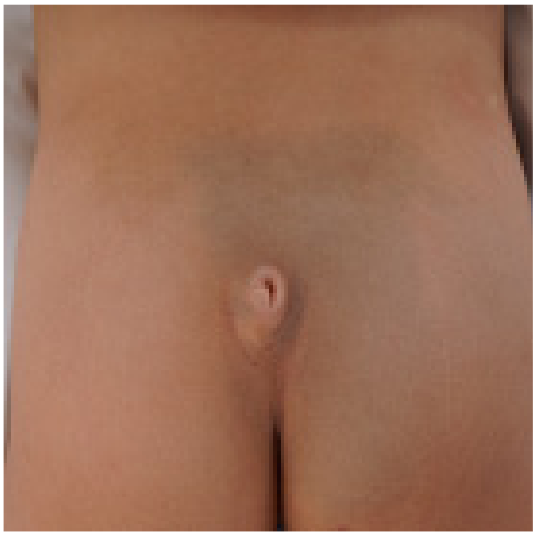

Figure 1. A photograph at the initial examination.
A mass with a central cutaneous depression is seen in the sacral area.

Surgical findings

The patient received general anesthesia in the supine position. After a subcutaneous injection of 0.5% lidocaine solution containing 200,000x epinephrine, an incision was made over the spindle-shaped design. The subcutaneous fatty tissue was dissected, and the protruding tailbone was exposed. The protruding tailbones and overlying excessed skin were resected. After washing with saline solution, the periosteum was sutured to cover the exposed bone, and the surgery was completed with skin sutures (**Figure 3a, b and c**).

Pathology

There was collagen fiber hyperplasia under a normal epidermis with adipose tissue interposition (**Figure 4a**). There was normal bone tissue with bone marrow surrounded by cartilage without atypia (**Figure 4b**). The findings were consistent with a true human tail.

Postoperative course

The postoperative course was uneventful, with no recurrence at 6 months postoperatively (**Figure 5**).

Discussion

The human tail was first described by Miller in 1881 when he reported a cutaneous protrusion of the sacrococcygeal region as tailed humanity[1]. According to subsequent reports by other authors, the human tail is a protrusion found on the midline of the sacrococcygeal region from birth, with no apparent gender or racial differences[2]. The human tail is

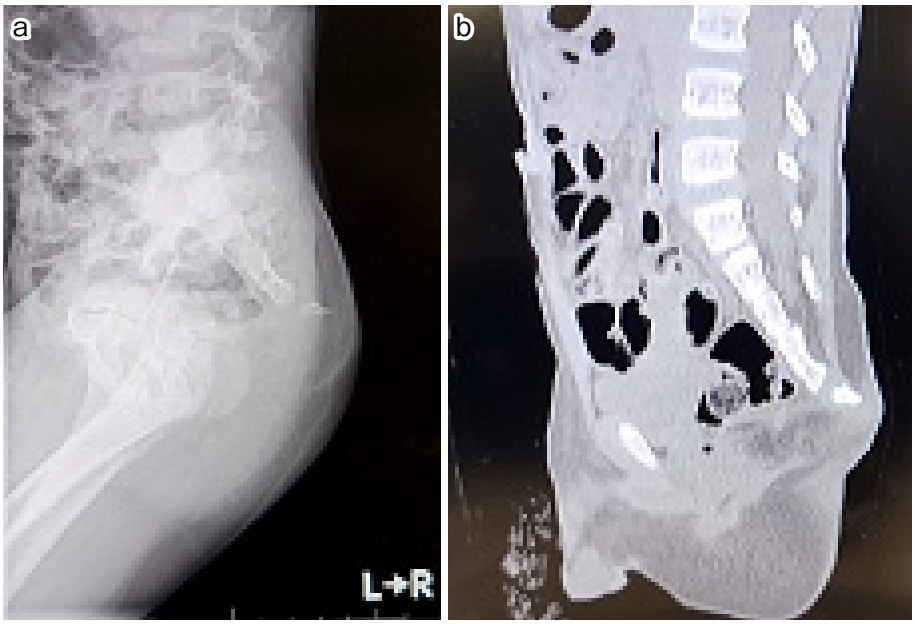

Figure 2. Preoperative imagings.
a Plain X-ray of the sacral region (lateral view), b Simple CT of the sacral region (sagittal section).
The coccyx is separated from the sacrum and curved into a dorsal concave shape.
b The soft tissue at the tip of the tailbone has a high CT value and is thought to be fibrous tissue.

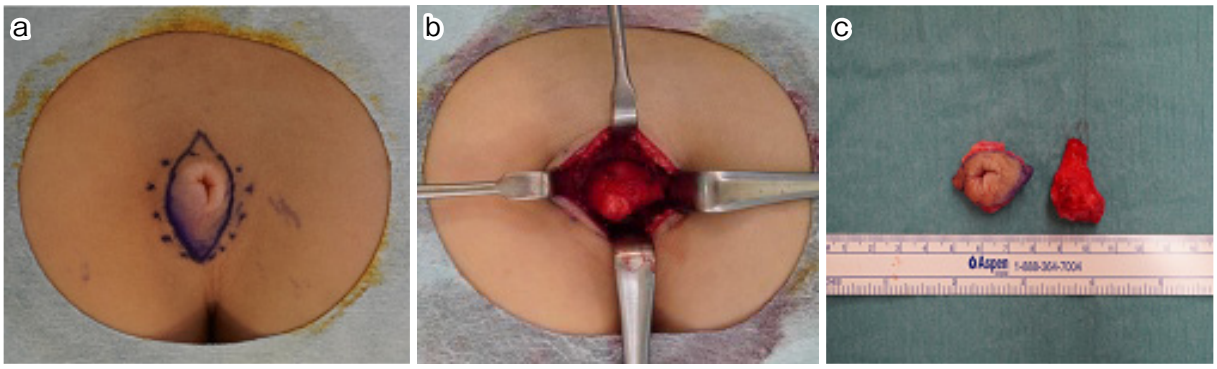

Figure 3. Surgical photograph.
a Resection design for the mass.
b Protruding tailbone.
c Resected excess skin and tailbone.

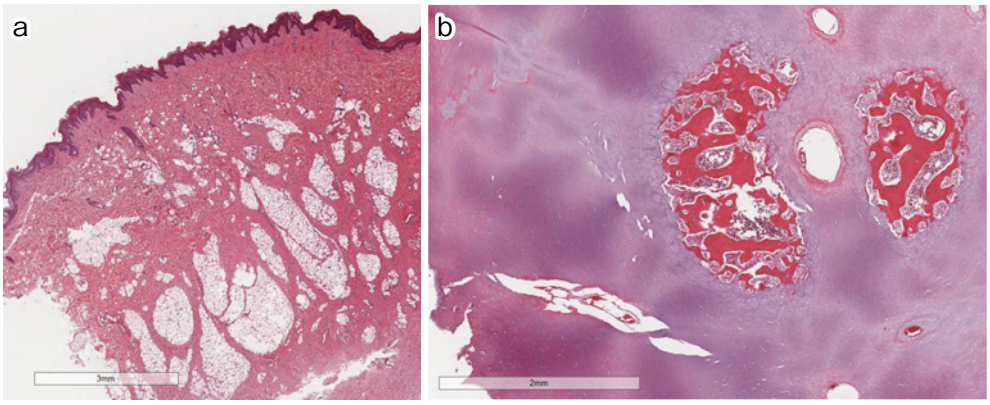

Figure 4. Pathological photographs.
a Hematoxylin and eosin stain, weak magnification.
Adipose tissue interposition under the squamous epithelium without atypia. Collagen fiber hyperplasia is observed.
b Hematoxylin and eosin stain, strong magnification.
Normal bone tissue, bone marrow, and cartilage are seen.

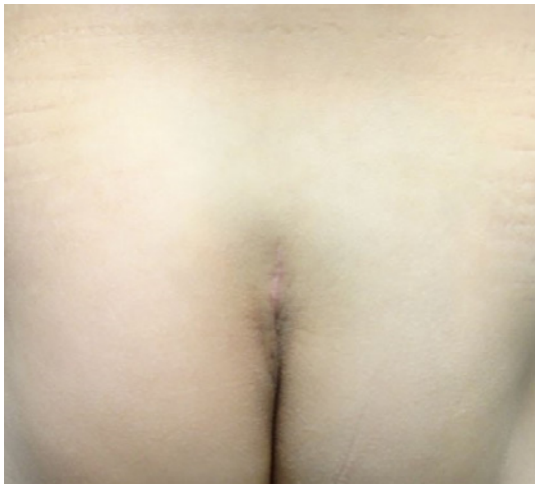

Figure 5. No recurrence was observed at 6 months postoperatively.

reportedly a remnant of the fetal tail present during the embryonic period. The embryonic tail is most developed around the 6th week of gestation, then gradually degenerates and is lost by the 9th week. Harrison et al. reported that the most developed fetal tail is divided into a vertebral portion (including the tailbone) and a caudal filament (not including the tailbone). These degenerations are somehow disrupted, resulting in the human tail. Tojima et al., however, denied the existence of caudal filament in the embryonic tale, as somites, which contain embryonic tailbone anlage, are present throughout the entire length of the fetal tail. According to their report, the number of embryonic tailbone anlages reaches a maximum of about 15 at around 6 weeks of gestation and then rapidly declines. The human tail as a congenital anomaly arises when the degeneration is impaired for some reason[3,4].

In the overlying skin of the human tail of our case, there was a depressed area in the center, but as far as we could find in the literature, there were no reports noting a concavity in the skin of the human tail. In this case, we speculated that the central depression was caused by the skin adhering to the terminal of the embryonic tailbone while decreasing

Table 1. Cases of Crouzon Syndrome with Human Tails.

Reporting person	Reporting year	Gender	Age	Complicated congenital abnormality	Tail nature	Central nervous system complications	Treatment	Recurrence	Follow-up period
Tsutsumibashi et al.	1990	Female	8 months	corneal ulcer, upper airway stricture	unknown	none	resected	none	died during treatment
Tsuji et al.	2001	Female	2 years old	arteriovenous duct patency	true	spina bifida	resected	none	unknown
Lapunzina et al.	2005	Unknown	unknown	tracheomalacia, facial asymmetry, Umbilical hernia	true	paresthesia of toes	unknown	unknown	12 months
Akai et al.	2006	Male	unknown	contractures of both elbow joints, Respiratory failure	pseudo	hydrocephalus	unknown	unknown	died during treatment
Endo et al.	2007	Male	4 week	contractures of both elbow joints, hydrocephalus, Single umbilical cord Artery	true	none	resected	unknown	unknown
Shanske et al.	2008	Male	1 year and 9 months	tow-headed, mental retardation ventricular and atrial septal defect	pseudo	none	resected	none	unknown
Sureka et al.	2010	Unknown	unknown	unknown	true	none	unknown	unknown	unknown
Self-examined case	2022	Male	4 years	none	true	none	resected	none	9 months

the number of tailbone primordia.

Regarding the classification of the human tail, Bartels classified human tails into 5 types according to the shape and the presence or absence of the tailbone, Virchow classified them into 3 types according to the projection shape, and Harrison classified them into 2 types according to the presence or absence of the tailbone, Dao et al. classified them into 2 types according to the presence or absence of the tailbone and the other tissues, and Tojima et al. classified them into 4 types according to the position and the presence or absence of the bone, respectively[4,5]. In Japan, most reports use Harrison's classification, in which human tails are divided into true human tails, including the tailbone and pseudo-human tail containing no bone. The frequency of each is reported to be 1:9[6]. This classification is considered helpful because of its simplicity and direct link to surgical planning[6]. In our case, we classified it as a true human tail according to Harrison's classification, as the tailbone existed.

We found 33 Japanese cases of the human tail in 1984 or later on the literature review using Ichushi-Web. Among 34 cases, including ours, 16 were male, and 18 were female, with no clear gender difference. Among 34 cases, 9 were true tail, 16 were pseudo-tail, and 9 were unknown. This ratio of the true tail to the pseudo-tail is quite different from the ratio reported by Harrison of 1:9. We speculate that this may come from the overdiagnosis of normal os coccygis as an excess embryonic human tail bone.

There are a variety of concomitant congenital anomalies with a human tail. Searching for these anomalies in treating patients with this syndrome is essential. Common complicating congenital anomalies reported in Japan are those related to the central nervous system, including spina bifida (8/34, 24%), spinal lipoma (4/34, 12%), and spinal meningocele (2/34, 6%). Among these, the complication rate of spina bifida is the highest (24%), and great care must be taken to the adhesion of the spinal nerves to the human tail during removal. It is necessary to determine whether there are tailbones in the human tail and whether there is the adhesion of the cauda equina in the human tail by X-ray, CT, or magnetic resonance imaging before surgery[7]. If the penetration of the cauda equina into the human tail is observed, motor, sensory, and cysto-rectal disturbances of the lower extremities should be closely examined[7]. In our case, there was no evident penetration of the spinal nerve into the human tail in both preoperative examination and surgical findings.

The first line of treatment of the human tail is surgical resection. If there are no central nervous system complications, a simple resection, including the tailbone, is best. However, if a central nervous system complication is suspected in the preoperative examination, or if the imaging show the continuity between the cord of the human tail and the spinal canal, it is necessary to perform surgery in collaboration with neurosurgeons[5]. In our case, no central nervous system association with the tail was suspected in the preoperative examination. A simple tailbone resection was performed, resulting in an excellent postoperative course.

Table 1 Cases of Crouzon syndrome complicated by the human tail

Reports of human tails associated with Crouzon's syndrome are rare, and as far as we could find, there are only 8 cases in English and Japanese literature, including our case

(**Table 1**). In Crouzon syndrome, in addition to the cranial deformity associated with premature cranial suture fusion, various other complications, including patent ductus arteriosus, spina bifida, corneal ulcer, upper airway stenosis, and contractures of both elbow joints, have been observed in reports. In our case, there were no other complications other than craniofacial abnormalities.

The association between the human tail and Crouzon syndrome is unclear. However, there is a possible linkage by FGFR2 abnormality. FGFR2 is the causative gene of Crouzon's syndrome. Moreover, the FGFR2 gene abnormality may cause abnormal development of the human embryonic tail[8]. In Crouzon syndrome with a human tail, a missense mutation in the amino acid sequence of FGFR2 proteins(amino acid substitution of Cys342Arg) was reported, whereas this missense mutation was not found in Crouzon syndrome without the human tail. In our case, however, an FGFR2 gene examination was not performed. A further association may be revealed in the future by genetic study for cases with human tail and Crouzon's syndrome.

Conclusion

We experienced a rare case of the true human tail associated with Crouzon syndrome, and the tail was successfully treated by surgical resection. Though the association between the human tail and Crouzon syndrome is still unclear, a genetic study in the future may reveal the linkage between them.

Author Contributions: Nobuhiro Ando, Nobuyuki Mitsukawa designed the study; Minoru Hayashi, Koichi Higaki provided critical reagents; Yoshitaka Kubota supervised the experiments.

Conflicts of Interest: There are no conflicts of interest.

Consent to Participate: The patients provided written informed consent to participate in this study.

Consent for Publication: The patients provided written informed consent to participate in this study.

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