

# *Dental Follicle*

The E- Journal Of Dentistry

ISSN 2230-9489 (e) | Dr. Syed Nabeel

**Dentistry**  
**United.com**

Complete Dentistry Information

**Contents**

Abstract: ..... 151

Introduction – ..... 151

Discussion – ..... 158

Conclusion – ..... 159

References – ..... 160

Ellis Van Creveld Syndrome with an unusual cardiac manifestation; A rare case report ..... 161

Abstract: ..... 161

Introduction..... 161

Case report: ..... 161

Discussion: ..... 165

References: ..... 167

## Implant placement after graftless Indirect Sinus lift –a case report

Dr.Sanjay Jamdade | Associate Editor: Dental Follicle - The E Journal of Dentistry

|INDIA

---

### Abstract:

**Objective** – To check the short and long term outcome and sequelae of graftless maxillary sinus lift procedures in a patient desiring a prosthetic restoration of a missing maxillary first molar whose maxilla has undergone pneumatization drastically reducing available vertical height of residual bone.

One interesting case is presented where a direct open sinus lift was performed, leading to sinus lining tear. The tear was repaired with sutures and Collatape. No graft was placed. Five months later an implant placement by indirect sinus lift was performed where the sinus was raised unusually high to six to eight millimeters instead of the traditional two millimeters and the Implant was used for tenting. Further four months later the Implant was restored traditionally.

**Materials and methods** - Collatape collagen membrane, Dentsply Xive Titanium implant, Vicryl sutures, direct Sinus lift instruments, indirect sinus lift osteotomes, and general implant surgery related equipments, instruments and materials

**Results** - Follow up after 4 years showed trabecular pattern around implant suggesting bone formation around the implant threads.

**Conclusion** - This confirms findings of other various other researchers elsewhere that even large graftless sinus lifts do lead to alveolar bone formation around the implant provided space is maintained between the sinus floor and sinus lining.

**Discussion** - literature on this topic with references is discussed as well as the finer points about the case are discussed.

**MeSH Key Words:** Maxillary Sinus, Schneiderian membrane, Maxillary Sinus Augmentation, Sinus lift complication, Dental implantation, open sinus lift, closed sinus lift, Graftless sinus lift, open maxillary sinus lift, maxillary Sinus lining tear, Tenting of sinus lining, Collatape, Xive dental implant, Pneumatization of Maxillary sinus, acid etched sand blasted dental implant, Risk of Maxillary Sinus surgery, Sinus lining suture, Sinus lining repair, Professor Lundgren, Professor Sohn, Dr Hanan Shokier, Dr Michael Pikos

---

### Introduction –

Graftless maxillary sinus lifts have been in practice and series of successful cases have been presented by researchers like Professor Dr Sohn of South Korea 1,2,3,4. In accordance with their findings they have published papers demonstrating their claim to be a viable option. Few others have also

presented similarly successful graftless maxillary sinus lift cases like Hanan m Shokier and Nagla 7. Professor Lundgren has also presented similar cases in his research paper 7. A case is being presented here which initially shows that sinus lining could be sutured as well as Collatape could

successful help repair it, then it also tests the hypothesis that graft less maxillary implant surgery is possible and osseoremodelling of new bone on the implant surface does occur in the absence of bone grafting material provided tenting is done.

**MATERIALS AND METHODS** – Collatape collagen membrane, Dentsply Xive Titanium implant, Vicryl sutures, direct Sinus lift, indirect both done.

A patient with partial edentulism presented with the desire to replace a missing upper left first molar tooth. (16 Dec 2008) The edentulous ridge span was unusually wide considering that the extraction was done few decades ago, as well as the neighboring premolars and molars had not tipped into the edentulous space very much which was unusual considering that the area had remained unrestored for many decades. All his teeth were attrited with flattened occluding surfaces. Restorative options were explored and the patient showed some inclination towards receiving an implant restoration. Accordingly an Intra Oral Periapical X-ray picture was exposed which showed that the maxillary sinus had descended coronally (pneumatization) and the available bone height was too less to support a regular threaded implant. Figure 1

The need of a maxillary sinus lift was explained to the patient to which he immediately agreed. So to be able to explore the maxillary sinus a CT scan was ordered.

The CT Scan (not available) showed that the Maxillary sinus lining at the floor was thick and the sinus had a small polyp like growth at the superior extent. These polyps did not trouble the patient in anyway. An ENT

opinion was sought and a clearance was received for a maxillary sinus lift procedure. A complete blood study was done and no negative findings were noted. The patient was put on Amoxicillin + Clavulanic Acid anti biotic combination as a preventive cover, along with Decadron. And Otrivin nasal drops and steam inhalation pre surgically. A completely disinfected air rotor air/waterline was kept ready along with a properly disinfected water bottle. The water bottle was then filled with normal saline from a fresh new bottle. A sterile autoclaved air rotor hand piece was first run to help remove any excess oil in cartridge to come out right at the onset.

On the day of the surgery (16/12/2008) the site of surgery was infiltrated with Lignocaine as a Local anaesthetic. Externally the entire face below the eye up to the upper neck was scrubbed with Povidone Iodine, Savlon and spirit. Intra-orally the part to be operated was scrubbed with Povidone Iodine and Chlorhexidene 0.02%. The scrubbed area was draped with surgical towels.

An incision was taken along the sulcus from the canine to the second Molar region. A small releasing incision above the canine and second molars was taken to make flap reflection easier. The flap was reflected with a periosteal elevator. Once the flap was reflected the height of the residual vertical height calculated from the Intraoral X ray was calculated and a mark was placed on the bone with a bur. The air rotor bur was used to mark a circular ring starting from the lowest point above the marking for the residual height. A round diamond bur was used to cut the window. As soon as the sinus lining was reached a couple of small punctures in the lining were noticed.

When the bone was cut through on all sides and the sinus lining was reached the window was gently tap with the back of a bone condenser. As the bony window flap moved inside the sinus a tear in the sinus lining was noticed. A sinus lining elevator was placed antero inferiorly moving along the sinus wall with the convex side towards the lining. The entire lining on the anterior and middle compartment of the maxillary sinus was elevated. However the sinus lining tear also propagated in an antero-posterior direction especially along the bony flap.

Considering that the direction was antero-posterior and the length of the tear was around 1.5 cm or so it was decided to suture the lining with 5.0 vicryl sutures. So with a very small ophthalmic Castroviejo needle holders and loupes 3 sutures in the lining were placed with great difficulty. Yet there were areas where the lining tear was gaping. So a Collatape\* collagen membrane was used to cover up the tear. The sinus lining was adequately covered with Collatape. It was decided to abandon the graft placement and close the wound up as risk of graft infection increases with large tears. 5 So before suturing the unused

Collatape was placed in double/ triple layers over the previous layer and excess completely tucked inside the Maxillary sinus chamber.<sup>2</sup> The Mucoperiosteal flap was then sutured back in place with 4.0 Vicryl sutures. Immediate post operative radiograph is shown in figure 2.

The patient was asked to continue with Otrivin nasal drops, antibiotics, anti inflammatory medicines, Decadron for the week. He was also advised against any deep breathing exercises, jogging, inverted positions in yoga. He reported after a week with no complaints what so ever. He had just one drop of blood come out of the left nostril and some drops of salty water on day one of surgery. There was no bleeding, no pain or any complaints of any origin. The intra Oral wound had healed very well without any issues what so ever. Sutures were removed on the 7th post operative day.

After 2 months the case was again followed up to check for healing. Radiograph as shown in figure 3 and 4 were taken. Note the bony window flap that was elevated in to the sinus has remained elevated after 2 months.

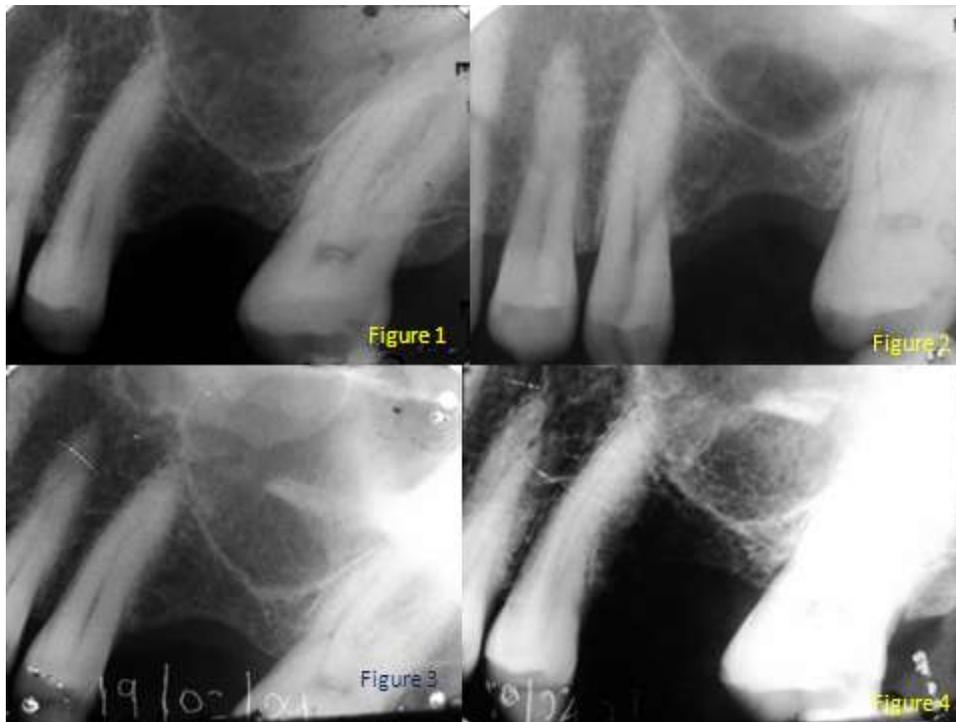


Figure 1 Preoperative intraoral periapical X ray picture

Figure 2 Immediate post operative after first surgery

Figure 3 Two month post operative

Figure 4 Two month post operative

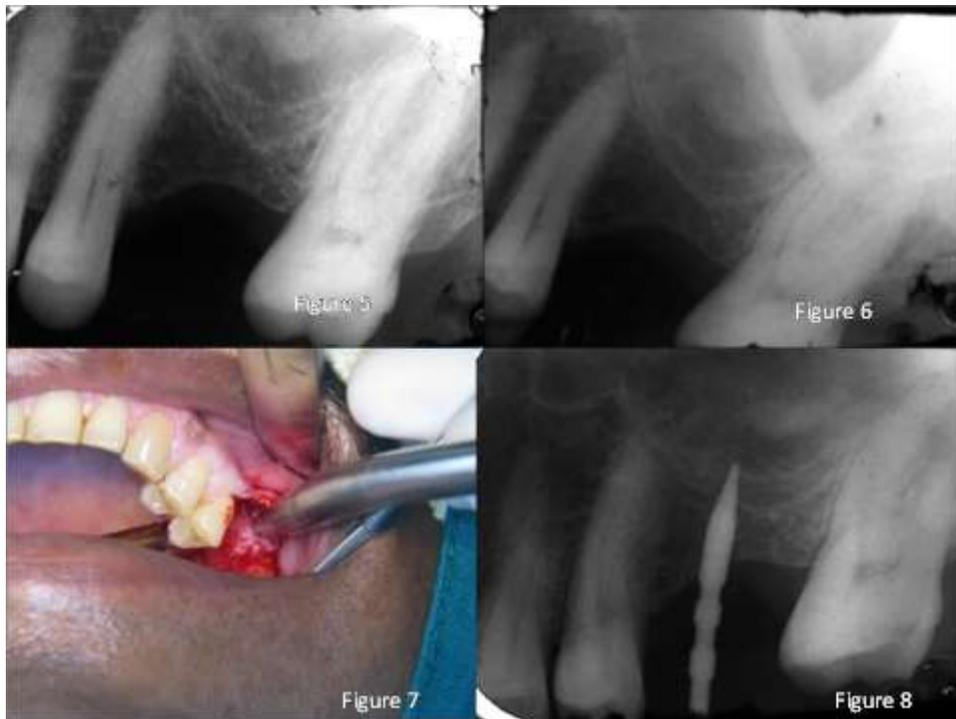


Figure 5 and Figure 6 – 4 to5 month healing showing the bony window hasn't completely descended

Figure 7 - Mid crestal incision and buccal and palatal flaps elevated to show the width of the ridge

Figure 8 – Paralleling pin in the osteotomy

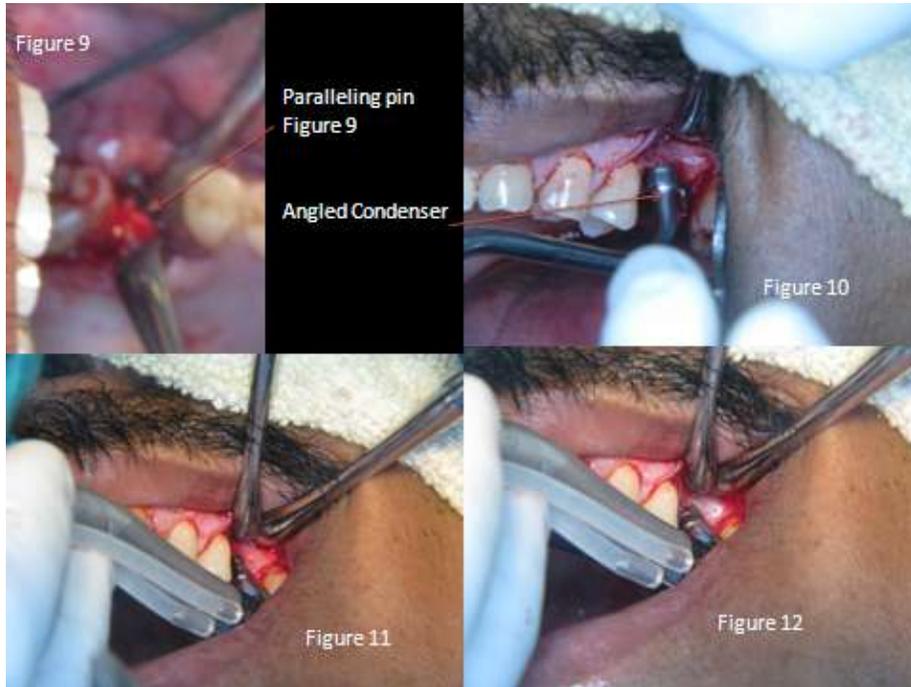


Figure 9 Picture of a paralleling pin inside the pilot osteotomy Figure10. The round condensing angled osteotome of 4.2mm diameter completely driven inside the Maxillary sinus up to 13 mm. Figure 11 and Figure 12 showing twist drill and crestal drill being used respectively



Figure 13 Osteotomy ready for implant placement Figure 14 Dentsply Xive Implant 4.5/11 mm Figure 15 Dentsply Xive Implant being ratcheted in Figure 16 Ridge after Implant Placement

Another round of follow up was done in 5 months time when 2 fresh IOPA

radiographs were taken. As shown in figures 5 & 6. The bony window has reduced in

opacity as compared to the previous x rays. However it is still up in the sinus suggesting that it hasn't come down even in 5 months. At 5 months it was decided to redo the maxillary sinus lift. However due to the unexpected delay in the treatment execution and completion, it was contemplated that if direct sinus lift along with tenting of sinus lining was done simultaneously some treatment time would be saved. It was guessed that since the bony window flap was still raised the bone bony window was either open partially, with some soft tissue interspersed or the new cortical plate formed was soft or thin, even some soft bone may have formed which was not noticeable on the X ray. Accordingly in the second surgery a crestal incision was taken with releasing sulcular incision in the crevices of neighboring teeth was taken. The buccal and palatal flap was raised. The Buccal incision would be

extended mesially and distally after checking the status of healing of the buccal bony window. The Buccal flap was raised and the bony window was found completely formed and hardened cortical bone albeit a little pinkish looking with mild porous surface texture unlike the untouched neighboring cortical bone which had a smooth surface.

It was decided to do a crestal osteotomy first and try and check the internal of the sinus to see if any bone had formed. As shown in figure 7. After the penetration of the pilot to the depth of 3-4 mm a paralleling pin was placed. The pin went up to a depth of about 5 to 7mm, a rush film was exposed to see where it went up to as shown in Figure 8

The X ray revealed that the sinus floor had already been breached either by the pilot drill, the paralleling pin or both. Figure 8.



Figure 17 Radiograph immediately after Implant placement Figure 18 Radiograph at four month healing Figure 19 Implant restored with a Porcelain-fused to non precious metal crown. 5 months later Figure 20 Buccal view of restored Implant.

Any puncture in the sinus lining was checked by asking the patient to blow the nose with mouth closed, (the Valsava technique). No air bubbling was seen. It was decided that the widening of the osteotomy should be carried out with round condensing osteotomies rather than drills. After the working part of first osteotome went completely inside very easily the lining integrity was again checked. All the osteotomies up to the 4.2mm osteotomies were used till the second last marking (13 mm). At no step was any evidence of a lining tear experienced. Shown in Figure 10. A 4.5 mm twist drill was used followed by a crestal widening drill was used as recommended in the Xive implant placement protocol. Figure 11, Figure 12.

Osteotomy free of any perforation of sinus lining was thus attained. Figure 13.

A Dentsply Xive 4.5 diameter and 11mm length implant was selected for implantation at the site. Figure 14. The Implant was ratcheted in and at placement there seemed to be adequate resistance to placement (insertion torque). Figure 15. The implant was placed flush with the alveolar crest. This placement was done slowly to avoid any sinus lining tear at a late stage during implant placement. Figure 16. A

radiograph was taken to confirm angle of implant placement. Figure 17.

After a period of 4 months the implant was checked for healing status by taking a radiograph. The radio graph showed neat remodeling of the bone at the crestal extent. Figure 18. Some very faint trabecular remodeling of bone was noticed inside the sinus around the implant.

The Implant was then exposed, a gingival former placed and restored prosthetically according to recommended protocol. A porcelain-fused to metal crown was delivered. Figure 19 and figure 20.

At the end of the treatment a radio graph was taken to confirm the fit of the prosthesis as well as the condition of the sinus was reconfirmed. The bony window that had been originally seen as elevated was present at the apical extent of the implant.

Three years later a follow up radiograph was taken as well as a follow up radiograph was taken. The 4 th year it was decided to do full mouth rehabilitation for the case as there was generalized attrition. So while checking the fit of the abutment a radiograph was taken.

Follow up X rays showed new bone formation with the old bone floor level and as well as the new floor level and the newly formed bone.

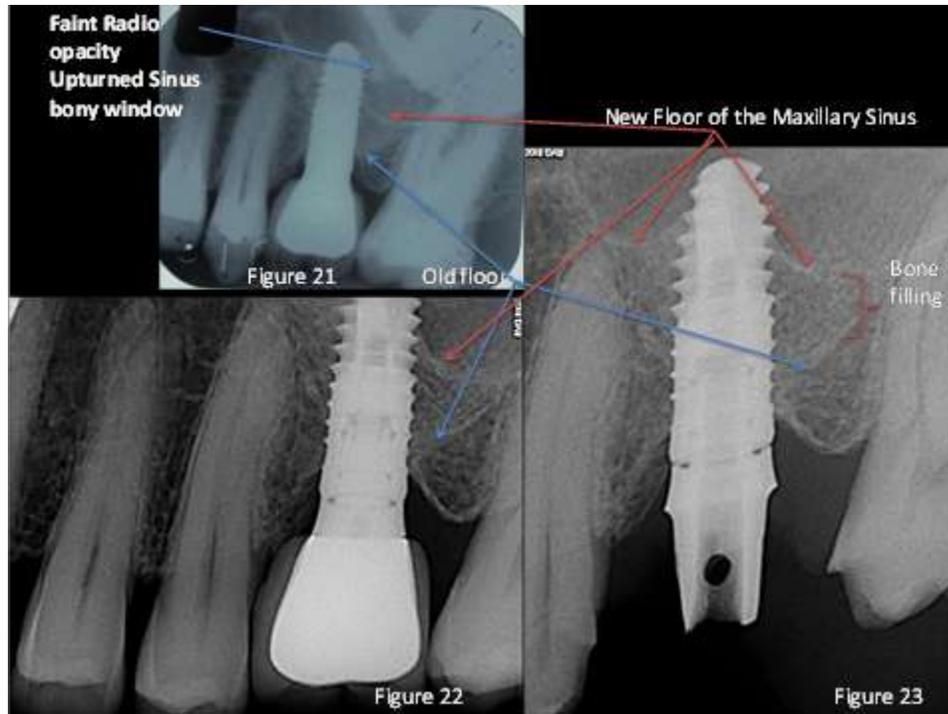


Figure 21 immediate post-op after crown delivery Figure 22 a 3 year follow up Figure 23 a 4 year follow up showing bone filling and crestal bone remodeling, old sinus floor and new sinus floor and trabeculae in the new bone fill. Old crown was being changed to a Zirconia crown, radiograph taken to check the fit of the abutment.

Results: Bone formation around the implant below the sinus lining was noted in the post operative period and afterwards up to a follow up period of four years

## Discussion –

Maxillary sinus lining is one cell thick. It can tear very easily 6, 8, 10, 11 at times, in spite of great operator care. Most common cause is the in fracturing of the bony window, it has been reported that it can happen when the membrane is being elevated off the antero inferior bony wall of the sinus. This could be due to bony spicules on the floor of the sinus.

But it can be repaired too. Some authors have recommended suturing of lining<sup>7</sup>, some have recommended a collagen membrane like Collatape,<sup>6,7</sup> whereas some have recommended fibrin Glue.<sup>8</sup> Sinus lining repair with Collatape has been

researched by Dr Michael Pikos<sup>7</sup> in which the Pikos pouch has been discussed as suitable for cases where there is no lining left or a complete tear occurs.

After having repaired the tear some do not recommend grafting at the same sitting as the success rate drops drastically<sup>5</sup>. The expected problems with grafting a large tear are

- 1) Greater risk of post operative infection
- 2) Loss of graft through the superior side of the lining and escaping through the nose
- 3) Greater risk of failure

Sinus lining tear management with suture has been mentioned by that perforation

and repair of sinus lining may compromise new bone formation.<sup>5</sup> Hence the author felt justified in repairing and abandoning the procedure as it has been cited in the article that perforated and repaired sinus tear sites had a lower potential to convert graft to bone and a higher risks of infections.

However Dr Michael Pikos in his article has shown with the help of many cases that in fact even sinuses with partially absent lining could be recreated with a collagen membrane (Pikos Pouch) and he has shown success with his technique too.

In the presented case bone formed was retained through-out the period of follow up providing adequate anchorage to a molar implant and withstanding all chewing loads. As an incidental finding no crestal bone loss was noted in the 4 years of follow up.

The four year follow up on the graftless indirect sinus lift shows that alveolar bone can form below the maxillary sinus lining as

long as the lining is tented and the lining is intact.

Professor Sohn of Seoul University has established that graft less maxillary sinus lifts could lead to alveolar bone formation around implants provide as long as tenting is achieved and provided sinus lining is maintained intact.<sup>1,2,3,4</sup> This has been done by Stefan Lundgren et al <sup>7</sup> as well as Hanan M.R Shokier and Naglaa Shawky. <sup>9</sup> Animal studies also confirm graftless sinus lifts also can form bone below the lining<sup>3</sup>.

Lundgren also showed that though tenting with machined implants may encourage bone formation in the sinus, an implant with specially treated/ created implant surface attracted more bone and encouraged bone apposition all around it <sup>7</sup>. Prof. Lundgren over and above attributed the success of bone formation to what he calls “replaceable bony window” in which he replaced the bone window flap into the place from where he had removed it. Though this was not done in the case presented above bone formation did take place quite well.

## Conclusion –

- Sinus lining tear is repairable and healing does occur with resorbable sutures and collagen membrane, individually and collectively both
- Repaired Sinus lining can withstand pressures of an indirect sinus lift after complete healing
- Repaired sinus lining will not interfere in alveolar bone formation
- The maxillary sinus cavity has enough osteogenic and osseoinductive potential which can lay new alveolar bone simply after creation of a space between the lining and the floor and walls of the Maxillary Sinus
- Since the implant is functioning uneventfully for the last 4 years it can be assumed that the bone formed inside the Sinus has enough density and can be hypothesized to be good quantity clinically useful quality dense bone
- Tenting is very important when it comes to graftless sinus lifts as it will help maintain the distance between the lining and maxillary sinus walls and floor where a space is created for bone cells to deposit new bone.

- Professor Lundgren, Professor Sohn and Dr Hanan and Nagla and other researchers findings on graftless sinus lifts was duplicated. Especially Dr Lundgren's work perfectly matched the author's observations with the exception that a replaceable bone window was not done.

## References –

1. New Bone Formation in the Maxillary Sinus Using Only Absorbable Gelatin Sponge Dong-Seok Sohn, DDS, PhD, Jee-Won Moon, DDS†, Kyung-Nam Moon, DDS, MSD, Sang-Choon Cho, DDS, Pil-Seoung Kang, DDS J Oral Maxillofac Surg. 2010 Jun;68(6):1327-33.
2. A Method of Sealing Perforated Sinus Membrane and Histologic Finding of Bone Substitutes: A Case Report Shin, Hong-In DDS, PhD, Sohn, Dong-Seok DDS, PhD Implant Dent. 2005 Dec;14(4):328-33.
3. Comparative Histomorphometric Analysis of Maxillary Sinus Augmentation With and Without Bone Grafting in Rabbit Sohn, Dong-Seok DDS, PhD\*; Kim, Woo-Sung DDS‡; An, Kyung-Mi DDS, MSD‡; Song, Kyung-Jin DDS§; Lee, Jae-Mok DDS, PhD||; Mun, Yong-Suk PhD¶ Implant Dent. 2010 Jun;19(3):259-70.
4. New Bone Formation in the Maxillary Sinus Without Bone Grafts
5. Sohn, Dong-Seok DDS, PhD\*; Lee, Ji-soo DDS‡; Ahn, Mi-ra DDS‡; Shin, Hong-In PhD§ Implant Dent. 2008 Sep;17(3):321-31.
6. Repair of the perforated Sinus lining membrane with a resorbable collagen membrane Perkliffs Prosaeffs DDS MS, Jaime Lozada DDS, Jay Kim PhD, Michael D Rohrer DDS MS
7. Maxillary Sinus Membrane Repair: Update on Technique for Large and Complete Perforations Michael A. Pikos, DDS (Implant Dent 2008;17:24–31)
8. Sinus membrane elevation and simultaneous insertion of dental implants: a new surgical technique in maxillary sinus floor augmentation STEFAN LUNDGREN, GIOVANNI CRICCHIO, VINICIUS C. PALMA, LUIZ A. SALATA & LARS SENNERBY Periodontology 2000, Vol. 47, 2008, 193–205 Printed in Singapore.
9. The use of autologous fibrin glue for closing sinus membrane perforations during sinus lifts Byung-Ho Choi, DDS, PhD,a Shi-Jiang Zhu, MD,b Jae-Hyung Jung, DDS,c Seoung-Ho Lee, DDS, PhD,d and Jin-Young Huh, DDS,e Seoul, South Korea YONSEI UNIVERSITY, EWHA WOMANS UNIVERSITY, GANGNEUNG HOSPITAL (Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2006;101:150-4)
10. Long term follow up for Closed Technique of sinus lift with simultaneous implantation with and without grafting Using Direct Digital Radiography and CT scan Hanan M.R Shokier and Naglaa Shawky Cairo Dental Journal (25) Number (1), 43:52 January, 2009
11. Management of the Schneiderian membrane perforation during the maxillary sinus elevation procedure: a case report - Deborah Meleo, DDS, PhD,1 Francesca Mangione, DDS,1 Sergio Corbi, MD, DDS,2 and Luciano Pacifici, MD, DDS Copyright ©2012 CIC Edizioni Internazionali, Rome, Italy
12. Simultaneous Sinus-Lift and Implantation Using Microthreaded Implants and Leukocyte- and Platelet-Rich Fibrin as Sole Grafting Material: A Six-Year Experience - Simonpieri, Alain DDS\*; Choukroun, Joseph MD†; Corso, Marco Del DDS‡; Sammartino, Gilberto MD, PhD§; Ehrenfest, David M. Dohan DDS, MS, PhD Implant Dentistry: February 2011 - Volume 20 - Issue 1 - pp 2-12doi: 10.1097/ID.0b013e3181faa8af Clinical Science and Techniques

---

\*Collatape is the registered trademark of Zimmer dental implants

## **Ellis Van Creveld Syndrome with an unusual cardiac manifestation; A rare case report**

Dr. Sharath Chandra.B |MDS |Prof & Head |Dept of Oral Medicine & Radiology  
|Panineeya Mahavidyalaya Institute of Dental Sciences & Research Centre |Hyderabad

Dr. Abhinethra.M.S |MDS |Senior Lecturer |Dept of Oral Medicine & Radiology  
|Vokkaligara Sangha Dental College & Hospital |Bangalore |abhinetra@gmail.com

Dr. Arun KP |MDS |Senior Lecturer |Dept of Oral & Maxillofacial Surgery |Kannur Dental  
College |Anjarakandy |Kerala.

---

### **Abstract:**

Chondroectodermal Dysplasia also known as Ellis Van Creveld Syndrome is a rare autosomal disorder that was first described by Richard W B Ellis and Simon van Creveld in 1940. It is a rare mesenchymal – ectodermal dysplasia. It is also known as mesvectodermal dysplasia. The disease is characterized by disproportionate dwarfism, polydactyly, and congenital heart disease. Reported incidence is one in 1500000 live births. Incidence in India is very rare.

Here we report a case of a child aged 7 years who reported with all the features of the syndrome. but she presented with coarctation of aorta, as a cardiac manifestation which is not been reported in the literature so far.

**Key-words:** Ellis Van Creveld syndrome, polydactyly, coarctation of aorta, Chondroectodermal dysplasia.

---

### **Introduction**

Access cavity preparation is the most Chondroectodermal Dysplasia also known as Ellis Van Creveld Syndrome is a rare autosomal disorder that was first described by Richard W B Ellis and Simon Van Creveld in 1940. It is a rare mesenchymal – ectodermal dysplasia. It is also known as mesoectodermal dysplasia. The disease is characterized by disproportionate dwarfism, bilateral postaxial polydactyly, and congenital heart disease. Reported incidence is one in 1500000 live births. Incidence in India is very rare.

Here we report a case of a child aged 7 years who reported with all the features of

the syndrome. but she presented with coarctation of aorta, as a cardiac manifestation which is not been reported in the literature so far.

### **Case report:**

A female patient(fig 1) aged 7 years reported with a complaint of missing teeth. Her history was non contributory. Her general examination revealed the plump extremities with presence of polydactyly(fig 3) in both the upper extremities, and there was hypoplasia of the terminal phalanges, the nails of both the fingers and toes were hypoplastic, thin wrinkled and spoon

shaped. There was presence of genu valgum of the legs.



Fig 1: Short stature with plump extremities.



Fig 2: Oral findings showing missing teeth, conical teeth and macroglossia



Fig 3: Clinical & radiographic appearance of hands showing polydactyly & spoon shaped fragile nails.

The craniofacial measurements were within the normal limits and the intelligence was in normal range

Her upper limbs were warm and the lower limbs were cold and clammy. Her BP measured to be 144/86 mm of Hg.

Oral examination revealed oligodontia of the upper and lower incisors (fig 2) with notching of the lower alveolar process giving a serrated appearance. The maxillary central incisors were conical. There was also the presence of macroglossia.

Complete skeletal survey was done.

OPG showed oligodontia in both the jaws, and altered morphology of the erupting maxillary canines.

PA of both hand and wrist (fig 3) showed bilateral post axial polydactyly and short and stout middle phalanges, cone shaped phalangeal epiphysis, short metacarpals, fusion of capitates and hamate seen bilaterally.



Fig 4: Valgus deformity of the knees.

Chest PA (fig 5) showed mild cardiomegaly. PA knee joint (fig 4) showed valgus deformity of the knees.

Ultra sound of abdomen and pelvis (fig 6 & fig 7) revealed the presence of monophasic flow pattern with low velocities suggestive of occlusion or narrowing in the thoracic

aorta. CT aortic angiography (fig 8) was

done which revealed coarctation of aorta.



Fig 5: Chest PA showing cardiomegaly.



Fig 6 & 7: Ultrasound of abdomen & pelvis showing monophasic flow pattern with low velocities. Hormonal assay was within normal limits. Chromosomal analysis (fig 9) revealed a normal female karyotype 46,XX. Echocardiography revealed a discrete coarctation of aorta.



Fig 8: CT Aortic angiography revealing coarctation of Aorta.

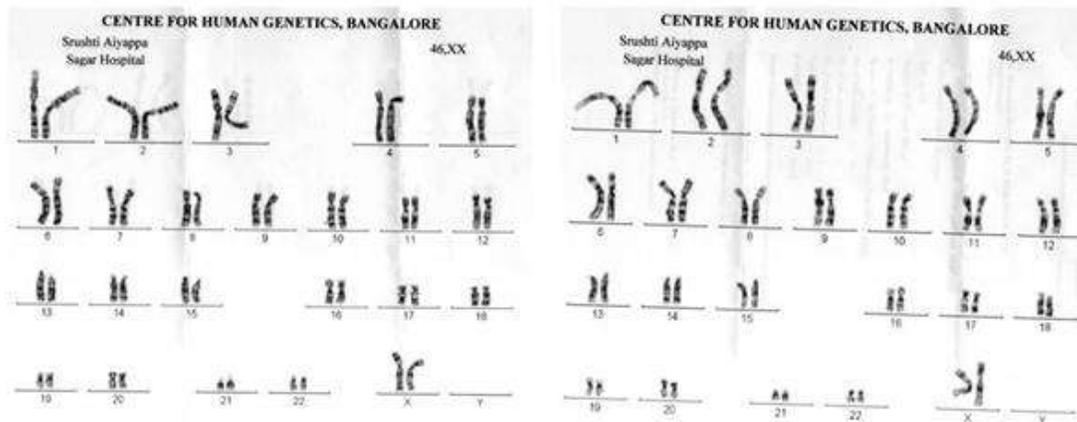


Fig 9: Chromosomal analysis showing a normal female karyotype.

Considering all the above factors a diagnosis of Ellis Van Creveld syndrome was considered.

## Discussion:

The first case of Ellis-van Creveld syndrome (chondroectodermal dysplasia) was reported by McIntosh<sup>1</sup> in 1933, but only in 1940 was it defined as a syndrome by Ellis and van Creveld<sup>1,2</sup>.

Combination of bony & cutaneous abnormalities is inherited as an autosomal recessive gene, which means that a child with Ellis-Van Creveld Syndrome (E.V.C.) is born to average sized parents who both carry the genes for E.V.C. The parents have 1 in 4 or 25% chance with each pregnancy

of having children with E.V.C. Association with parental consanguinity has been found in 30% of cases<sup>3,4</sup>. Reported incidence is one in 150000 live births. Incidence in India is very rare<sup>3</sup>.

Etiology: Recent studies have found 2 genes in head to head configuration along the chromosome. Mutations in EVC gene at 4p16{EVC1} is purported not to cause the syndrome while as the mutations in the other gene at 4p20{EVC2} are currently thought to be implicated. The cytogenetic

techniques have shown segmental uniparental disomy of chromosome 5.

Caffey described constant and inconstant features. The constant features include hypoplasia of teeth and nails; progressive shortening of the bones of the arms and legs ; bilateral polydactyly, polymetacarpalia and synmetacarpalia; hooklike bilateral fusion of the capitate and hamate bones; delayed maturation of the primary ossification centers of phalanges with accelerated maturation of the secondary epiphyseal cartilages ; and characteristic deformities of the proximal end of the tibiae<sup>6</sup>.

The inconstant features are alopecia; joining of the upper lip and gum ; congenital cardiac anomalies ; dislocation of the head of the radius ; delayed maturation of sesamoid bones in the hand ; and pedal polydactyly, synndactyly, and polymetatarsalism<sup>6</sup>.

The patient is achondroplastic dwarf with shortening of the limbs. The skull is normal with no depression of the base of nose. The teeth are dysplastic with marked alterations in their morphological and chronological development. The trunk is of normal size and shape for the patient's age, although some patients have been described with keel-shaped or barrel-shaped chests. The shortening of the limbs is acromelic in character, rather than rhizomelic, as in achondroplastic dwarfism<sup>15</sup>. There is a supernumerary cubital sixth finger, the nails are fragile and fluted and rarely extend out to the end of the digit. The malformation of the proximal epiphysis of the tibia causes genu valgum<sup>1,6</sup>.

The reported congenital cardiac anomalies include auricular and ventricular septal defects and hypoplasia of aorta<sup>5</sup>. Mental

retardation is not characteristic of this syndrome<sup>6</sup>.

Others include<sup>5</sup> :

CNS: Dandi Walker malformation

Thoracic: Narrow chest, pectus carinatum, short and poorly developed ribs

Renal: Glomerulopathy with nephritic syndrome

Genital : Epispadias, hypospadias, cryptorchidism

The constant oral findings are hypoplasia of deciduous and permanent teeth, with defective excessively friable enamel. The other oral manifestations include submucous clefts or notching of the maxillary and mandibular alveolar processes, continuous or broad labial frenula, dystrophic philtrum, hypodontia, teeth of abnormal form and malocclusion. Presence of natal teeth have been reported in 25 – 30 % of cases<sup>11</sup>.

The laboratory data in the cases to date are of little value in diagnosis<sup>1,6</sup>, but prenatal diagnosis in regard to growth retardation, skeletal malformations and cardiac defects can be depicted on ultrasound images. Diagnosis is also positive using chorionic villi or amniotic fluid using linked – microsatellite markers if a previously affected sibling has been identified<sup>2</sup>. The primary pathological lesion appears to reside exclusively in the epiphyseal cartilage<sup>6,9</sup>.

Infant mortality rate is higher mainly due to cardiorespiratory failure, if they survive morbidity is significant<sup>3</sup>.

Our case presented with a unique feature of coarctation of aorta, which explains the reason for the cold clammy lower limbs and increased blood pressure<sup>7</sup>.

EVC requires a multidisciplinary therapeutic approach. The dentist plays a fundamental

role in the control and treatment of oral and dental manifestations. A combination of cardiac surgeon, dentists and orthopedists are essential in the complete management of the patient.

Dental treatment should be made under low antibiotic prophylaxis due to the high incidence of heart pathology<sup>10</sup>.

The differential diagnosis includes Orofaciodigital syndromes

## References:

1. DA Silva EO, Janovitz D and Albuquerque SCD; Ellis-van Creveld syndrome: report of 15 cases in an inbred kindred. *Journal of Medical Genetics*, 1980, 17, 349-356
2. K Kurian, Shanmugam S, Harsh Vardah T, Guptha S; Chondroectodermal dysplasia (Ellis Van Creveld Syndrome): report of three cases with review of literature; *Indian J Dent Res*;18(1);2007.
3. Sharma OP, Saraf R, Gupta B; Ellis-Van Creveld's Syndrome (a Case Report) *Ind J Radiol Imag* 2006 16:3:325-327.
4. Vinay C, Reddy S; Uloopi KS, Chandrashekhar R; Clinical manifestations of Ellis Van Creveld syndrome; *J Indian Soc Pedod Prevent Dent*; Oct-Dec 2009;27(4).
5. Javaid C, Bhat A, Bhat T, Bhat T, BHat R, Feroze A, Jan S; Ellis Van Crevald Syndrome; *JK-Practitioner* 2007;14(2): 100-101
6. Narciso A. Ferrero, Osvaldo O. Pozo and Emilio S. Morresi; Chondro-Ectodermal Dysplasia (Ellis-van Creveld Syndrome): Report of a case and review of literature; *J Bone Joint Surg Am* 1961;43;1230-1236.
7. Smitha Ramegowda, Nallur B Ramachandra; An understanding of genetic basis of congenital heart disease; *Indian Journal of Human Genetics*; January – April 2005;11(1).
8. B. J. Cremin, and P. Beighton; Dwarfism in the newborn: the nomenclature, radiological features and genetic significance; 1974, *British Journal of Radiology*, 47, 77-93
9. Cesur Y, Yuca SA, Üner A, Yuca K, Arslan D; Ellis-Van Creveld Syndrome; *Eur J Gen Med* 2008; 5(3):187-190.
10. Pereira DA, Aytés LB, Escoda CG; Ellis-Van Creveld Syndrome. Case report and literature review; *Med Oral Patol Oral Cir Bucal*. 2009 Jul 1; 14 (7):E340-3.
11. Shah B, Ashok L, Sujatha GP; Ellis Creveld syndrome: A case report; *J Indian Soc Pedod Prevent Dent*; supplement 2008.
12. Polymeropoulos MH, Ide SE, Wright M, Goodship J, Weissenbach J, Pyeritz RE, Da Silva EO, Ortiz De Luna RI, Francomano CA; The gene for the Ellis-van Creveld syndrome is located on chromosome 4p16. *Genomics*. 1996 Jul 1; 35(1):1-5.
13. Ruiz-Perez VL, Ide SE, Strom TM, Lorenz B, Wilson D, Woods K, King L, Francomano C, Freisinger P, Spranger S, Marino B, Dallapiccola B, Wright M, Meitinger T, Polymeropoulos MH, Goodship J. Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrofacial dysostosis. *Nat Genet*. 2000 Mar;24(3):283-6.
14. Dhanrajani, PJ; Sami AA; Management of Severe Hypodontia; *Implant Dentistry*: December 2002 - Volume 11 - Issue 4 - pp 338-342
15. Syndromes of head & neck; Gortner RJ, Cohen MM Jr, Hennekam RCM. Oxford University Press USA 2010, 4th edition, 239-242.