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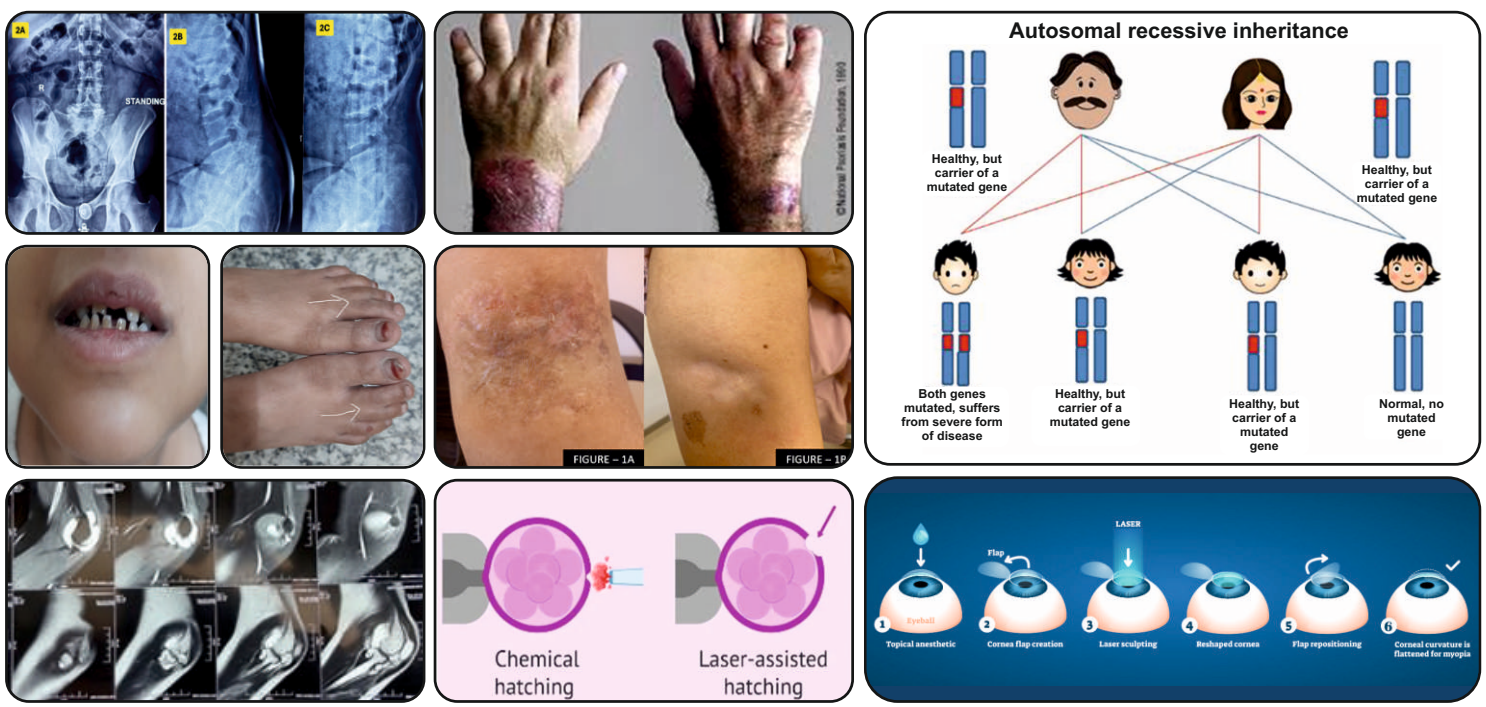
Largest Circulated Medical Journal in Gujarat

# I.M.A.G.S.B. News Bulletin

## GUJARAT MEDICAL JOURNAL

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# All India Medical Conference
# IMA NATCON 2025

27th & 28th December 2025

**HOSTED BY**

**IMA Gujarat State Branch & Ahmedabad Medical Association**

Dear Doctor,

Seasons Greetings!

The Indian Medical Association Gujarat State Branch & Ahmedabad Medical Association invite you to be a part of **IMA NATCON 2025**, a benchmark annual conference shaping the future of healthcare. This event, scheduled on December 27th and 28th, will feature impactful lectures and skill-enhancing workshops by leading national faculties.

Your participation as a vital member of the healthcare community will undoubtedly enrich the discussions and contribute to the collective knowledge shared. We look forward to welcome you for this prestigious event.

Together, let's make **IMA NATCON 2025** a landmark conference.

With anticipation,
**The Organizing Team**
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# I.M.A.G.S.B. NEWS BULLETIN

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## GUJARAT MEDICAL JOURNAL

INDIAN MEDICAL ASSOCIATION, GUJARAT STATE BRANCH



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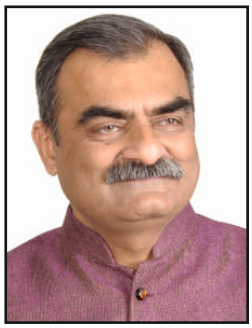

# STATE PRESIDENT AND HON. STATE SECRETARY'S MESSAGE

Every year on 5th September, we pause to celebrate "Teacher's Day". For us, it is not just about honoring the teachers of our classrooms but also about acknowledging the mentors we meet throughout our medical journey — professors, seniors, colleagues, and even patients who silently shape our understanding. Medicine is a profession where every day is a lesson, and every person we meet has something to teach us. This spirit of learning and gratitude forms the foundation of our work at IMA.

Carrying this motive forward, we now stand at a historic moment — the 100th NATCON. The preparations are in full momentum, and the excitement is visible across Gujarat. Like students preparing for their final examinations, our teams are working day and night to ensure that this centenary celebration shines as one of the finest in IMA's history. But just like in the classroom, success here too depends on teamwork. Every local branch has an important role, and we urge each one to come forward and make their contribution. When every branch adds its energy, the entire state will shine.

We have already plan the first steps on this journey. The pre-conference seminar, to be held with the collaboration of prestigious institutes, an excellent preview of what lies ahead. The discussions will be rich, the ideas innovative, and the participation heartening.

Looking ahead, we want NATCON 2025 to be more than just a conference. Our scientific sessions will highlight the latest advances in medicine, but they will also create space for medical students and young doctors. Their curiosity, their questions, and their fresh perspectives are what will carry our profession into the future. By giving them a platform, we are not only sharing knowledge but also nurturing tomorrow's leaders.

it is a journey of discovery and inspiration. Imagine halls abuzz with the wisdom of eminent national and international faculties, where doctors will explore everything from hospital start-ups and advanced management, strategies to business-to-business collaborations and international medical tourism. It will be a place where knowledge meets innovation, experience meets curiosity, and ideas transform into action. Every session, every discussion, and every interaction will echo the spirit of progress, leaving participants energized to take their practices, institutions, and patient care to new heights. NATCON 100 is not just an event; it is a living story of medicine, vision, and the relentless pursuit of excellence — a legacy we are proud to continue and share with the world.

So as we write to you this month, We see NATCON 2025 as a story we are all co-authoring. A story where gratitude for our teachers leads us to prepare diligently, where unity across branches ensures success, where students and young doctors find their voice.

Together, let us not just organize a conference but create a legacy that future generations will look back upon with pride.

**DR. MEHUL J. SHAH**
President, G.S.B.,I.M.A.

**DR. GARGI M. PATEL**
(Hon. State Secy., G.S.B.,I.M.A.)

# FROM THE DESK OF EDITORS

Dear Doctor Colleagues,

Greetings!

At the outset we would like to apologise for the delay in releasing the Gujarat State Medical Journal. It is heart warming to know that many of you are reading this Journal and updating your knowledge. It is very rightly said –"THE PERSON WHO STOPS LEARNING OR THINKS HE/SHE KNOWS EVERYTHING -IS THE MOST UNEDUCATED AND FOOLISH IN THIS WORLD"

Our journal is not an INDEX JOURNAL but that should not make a difference to most of us as those who are in private practice or want to share something new which has happened in their subject- this Journal is the best medium to reach out to 35000/- Doctors of Gujarat.

We are sure many of us might be coming across some very unique cases- out of the box cases- in your day to day practice- We want you to come forward and share such cases with us- i.e. send us and we will print- so that other doctors would learn and know about your excellence in your field. If there is some new innovation in your field you can also send us such articles and we would be pleased to print it. Basically Journals are not a ladder to your Success- but it is a medium through which you learn about the latest inventions and innovations in different fields.

So for next issue we expect lot of new case studies from your practice plus interesting articles. This Journal is also excellent medium to advertise about your institute or set up, as 35000 doctors are going to read and see it. So more advertisement is expected from all of you.

" A SINGLE PERSON CANNOT PLAY A SYMPHONY HE NEEDS A WHOLE ORCHESTRA"

Similarly we need all your support to improve the quality of our Journal

Hoping for a positive and energetic response from you all

With Regards,

**DR. MONA P. DESAI**
Editor-IMA-GSB-GMJ

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# I.M.A. G.S.B. NEWS BULLETIN (Gujarat Medical Journal)

**Vol. : 19** **AUGUST-2025** **Issue : 08**


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**ORIGINAL ARTICLE**

# Fetomaternal outcome in patients with Gestational Diabetes Mellitus

**Dr. Tushar M. Shah*, Dr. Pratiksha S. Bangde****

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**KEY WORDS :** Gestational Diabetes Mellitus, DIPSI, Maternal / Neonatal Complications, Dietary Control

## INTRODUCTION

Gestational Diabetes Mellitus is defined as "carbohydrate intolerance of variable severity with first onset or recognition during the present pregnancy" irrespective of treatment with diet or insulin. It excludes patients with previously diagnosed Diabetes.

The incidence of DM in pregnancy is expected to increase to 20%. Approximately 8% of all pregnancies are complicated by GDM. The prevalence may range from 1 to 14% of all pregnancies depending on the population studied and the diagnostic tests used.

Pregnancy is a diabetogenic state charecterised by hyperinsulinemia and decreased sensitivity to insulin at cellular levels. Glucose intolerance and gestational diabetes mellitus result when pancreatic beta-cell function cannot adequately compensate for the degree of insulin resistance in pregnancy.

### New 2015 NICE guidelines suggestions: Revised in 2020

Women who have had gestational diabetes in a previous pregnancy should be advised for early self-monitoring of blood glucose or a 75 g 2-hour OGTT as soon as possible after booking (whether in the first or second trimester), and further 75 g 2-hour OGTT should be repeated at 24- 28 weeks if the results of the first OGTT are normal, Offer women with any of the other risk factors for gestational diabetes a 75 g 2-hour OGTT at 24-28 weeks.

## AIMS & OBJECTIVES

Aim and objectives of study is to study the prevalence of gestational diabetes mellitus, demographic characteristics like age, parity, Past history in relation to GDM .

To evaluate the maternal outcome in terms of mode of delivery, intrapartum and post-partum complications.

To study the fetal outcome in terms of birth weight, maturity, presence of congenital anomalies, and neonatal complications in cases of gestational diabetes.

## MATERIALS & METHODS

Current study design is prospective observational study in which total 136 patients were studied during the period of January 2024 to June 2024 in department of obstetrics and gynaecology B. J. medical college and civil hospital Ahmedabad.

Their detailed data was obtained and their age, obstetric history, gestational age, past history of DM, mode of delivery, outcome of pregnancy, fetal weight, maternal and fetal complications and requirement of oral hypoglycaemic agents or Insulin were taken into consideration for the study.

An analysis is made based on these parameters.

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## INCLUSION CRITERIA

Pregnant women > 24 weeks of gestation with blood sugar levels 140-200mg/dl after 2 hours of 75 g oral glucose (DIPSI). Patients who willing to participate and could be followed up, investigated and those in which fetal outcome could be recorded are only included.

## EXCLUSION CRITERIA

Patients with pregestational or overt diabetes were excluded from the study.

## RESULTS & DISCUSSION

Out of total 3911 deliveries during January 2024 to June 2024 total 136 patients of gestational diabetes were studied. The prevalence of gestational diabetes mellitus in the present study was found to be 3.477 %. IADPSG(5) has reported prevalence of pregnancy with diabetes as 27%. Proportion of patients with diabetic pregnancy in present study was less, probably because universal screening was not feasible due to certain constraints, or probably because of improvement in medical and obstetric facility at secondary level, a smaller number of patients were referred to tertiary care hospital.

In the present study, 5.15% patients of gestational diabetes belonged to age less than 20 year, 33.82% patients belonged to age group 20-29 years. The maximum number of patients, that is, 54.41% were in the age group of 30-39 years. And 6.61% were in the age group of more than 40 years.

| Age (yrs) | N (136) | % |
|---|---|---|
| <20 | 7 | 5.15 |
| 20-29 | 46 | 33.82 |
| 30-39 | 74 | 54.41 |
| >40 | 9 | 6.61 |

In the present study, 11.76% patients were primigravida, 17.65% patients were second gravida, 26.47% patients were third gravida, 44.12% patients were above forth gravida. Study was compared to study of Binny Thomas et al and it was observed that pregnancy with diabetes was more in multigravida patients. Prevalence of pregnancy with diabetes was 47.7% in primi patients and 50.3% in multipara in a study by Binny Thomas et al. The association between parity and diabetes is strongly linked due to obesity & age.

| GRAVIDA | N (136) | % |
|---|---|---|
| Primi | 16 | 11.76 |
| Second | 24 | 17.65 |
| Third | 36 | 26.47 |
| Four and Above | 60 | 44.12 |

Out of all 120 multigravida patients 39.81% patients had past history of gestational diabetes and 60.19% patients were not having past history of diabetes. Thus, past history of GDM is a risk factor for recurrence of gestational diabetes mellitus in next pregnancy.

In the present study out of 136 patients of the gestational diabetes mellitus 26 patients were managed alone with the dietary control with life style modification and exercise. All registered patients had followed up diet and exercise as advised. 110 patients of gestational diabetes mellitus were not maintaining euglycemia with life style modification and exercise with dietary control, they were managed by insulin or oral hypoglycemic agents.

In the present study it was observed that, 28.68% patients were delivered normally, 8.82% patients required instrumental vaginal delivery. 47.06% patients delivered by emergency caesarean section; 15.44 %patients underwent elective caesarean section.

**Mode of management of the patients with the gestational diabetes**

| Management | N (136) | % |
|---|---|---|
| Life style modification and exercise with dietary control | 26 | 19.152 |
| Oral antidiabetic agents + lifestyle modification + dietary control | 20 | 14.70 |
| Insulin | 90 | 66.17 |

**Mode of delivery in cases of gestational diabetes**

| Mode of Delivery | | N (136) | % |
|---|---|---|---|
| Vaginal Delivery | Normal | 39 | 28.68 |
| | Instrumental | 12 | 8.82 |
| Cesarean Section | Emergency | 64 | 47.06 |
| | Elective | 21 | 15.44 |

In the present study it was observed that most common indications for caesarean section inpatients with gestational diabetes mellitus were previous 1 or more CS which includes 33 cases, 14 cases were having cephalopelvic disproportion, 13 cases were having induction failure and 13 cases were of fetal distress, 12 cases had uncontrolled GDM.

In the present study, most common association of gestational diabetes mellitus in pregnancy is PIH that is 23 cases developed preeclampsia and 11 cases had uteroplacental insufficiency, and 46 cases had abnormality in amniotic fluid (polyhydramnios), 4 patients developed septicaemia, 4 patients had wound gap after caesarean section, 7 patients had intra uterine fetal death, 2 patient developed diabetic ketoacidosis, 13 cases underwent pre term labor, 6 of them developed hypoglycaemia. Comparison of the present study with studies of Binny Thomas et al and Dahiya et al. It is evident from table that in patients with diabetic pregnancy, incidence of preeclampsia was 16.91% , polyhydramnios was present in 33.82 % of the patients, preterm labor was present in 9.56% patients, these results were comparable with the studies by Binny Thomas et al and Dahiya et al.

**Comparison of this study with other study with regard to maternal-complications.**

| Material Complication | Present Study | Binny thomas et al (2012) | K Dahiya et al (2014) |
|---|---|---|---|
| Preeclampsia | 16.91% | 14.40% | 14.30% |
| Polyhydramnios | 33.82% | 2.70% | 17.10% |
| Preterm | 9.56% | 4.80% | 8.60% |

In our study perinatal mortality was observed in 13 cases of gestational diabetes mellitus. Respiratory distress was present in 14 babies who required supplemental oxygen or positive pressure ventilation, 30 neonates were required NICU management and 2 babies had congenital malformation.

| Neonatal Complication | No. of Babies | % |
|---|---|---|
| Perinatal mortality | 13 | 9.56% |
| Respiratory distress | 14 | 10.29% |
| Hypoglycemia | 19 | 13.97% |
| Hyperbilirubinemia | 14 | 10.29% |
| NICU care | 30 | 22.06% |
| Congenital Malformation | 2 | 1.47% |
| IUD | 1 | 0.73% |

In the present study, 18.38% babies had birth weight <2.5kg, 64.7% babies had weight between 2.5-3.5kg, 16.91 % babies had weight between 3.6->4.0kg.

| Birth Weight | N | % |
|---|---|---|
| <2.5kg | 25 | 18.38 |
| 2.5-3.5kg | 88 | 64.70 |
| 3.6-4.0 kg | 23 | 16.91 |

## CONCLUSION

Gestational diabetes mellitus is one of the common medical disorders encountered in pregnancy. Clinical recognition of gestational diabetes mellitus is important because timely intervention by dietary measures and/or insulin can reduce the well-known maternal and fetal complications associated with it.

The findings of the present study confirmed that patients having pregnancy with GDM are liable to have adverse pregnancy outcomes. The study concluded that increased maternal age, high body mass index, multi gravidity, past history of diabetes and also family history were identified to be major risk factors. Patients with such factors should be

identified as early as possible and classified as a high-risk group and called for frequent antenatal check- up as required.

Neonates born to gestational diabetes mellitus mothers had increased rate of macrosomia and metabolic complications which can lead to increase in perinatal morbidity and mortality rates. But immediate intensive care after birth lead to reduced neonatal mortality.

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**ORIGINAL ARTICLE**

# 24-Hour Urine Metabolic Profile for Kidney Stone Disease: A Critical Tool for Prevention

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**KEY WORDS :** Urine Metabolic Profile for Kidney Stone Disease

## INTRODUCTION

Kidney stone disease (nephrolithiasis) is a **common and recurrent condition**, affecting approximately **8- 12 % of the global population**. Without preventive measures, **50% of stone formers** will experience a recurrence within **5–10 years**. One of the most effective ways to prevent kidney stone recurrence is through **24-hour urine metabolic analysis**, which helps identify metabolic abnormalities contributing to stone formation.

An essential component of this test is the **urinary supersaturation index**, which quantifies the risk of crystal formation and stone growth. Understanding urine composition and **supersaturation levels** allows for personalized treatment strategies, significantly reducing recurrence rates.

This article explores the **24-hour urine metabolic profile, the supersaturation index, and their role in kidney stone prevention**, incorporating the latest research and guidelines.

**What is a 24-Hour Urine Metabolic Profile?**

***24 hrs Urine Metabolic profile (Litho-risk) is only and most important test to know cause of stone and useful guide for focal, prophylactic treatment.***

A **24-hour urine test** measures the excretion of key substances that influence kidney stone formation. The patient collects all urine produced over a 24-hour period, which is analysed to detect metabolic abnormalities.

**Key Components Measured and Their Significance**

| Parameter | Role in Stone Formation | Abnormal Findings & Risk |
|---|---|---|
| **Urine Volume** | Dilutes stone-forming substances | Low volume (<2L/day) increases risk |
| **Calcium** | High levels promote calcium-based stones (e.g., calcium oxalate, calcium phosphate) | Hypercalciuria (>200-250 mg/day) |
| **Oxalate** | Binds with calcium to form calcium oxalate stones | Hyperoxaluria (>40 mg/day) |
| **Citrate** | Natural inhibitor of stone formation | Hypocitraturia (<320 mg/day) |
| **Uric Acid** | High levels cause uric acid stones | Hyperuricosuria (>750 mg/day in women, >800 mg/day in men) |
| **Sodium** | Increases calcium excretion, promoting stones | High sodium (>100 mEq/day) |
| **pH** | Influences stone type (acidic = uric acid stones, alkaline = calcium phosphate stones) | pH < 5.5 (uric acid stones), pH > 6.8 (calcium phosphate stones) |
| **Magnesium** | Inhibits crystal formation | Low magnesium (<50 mg/day) |
| **Creatinine** | Used for assessing collection adequacy | Deviations indicate errors in collection |

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## Urinary Supersaturation Index: A Predictor of Stone Risk

### What is the Supersaturation Index?

The **urinary supersaturation index (SI)** is a measure of how likely a person's urine is to form kidney stones. It represents the **degree to which stone-forming substances exceed their solubility threshold** in urine, leading to crystal precipitation.

The **SI is calculated** for different stone types:

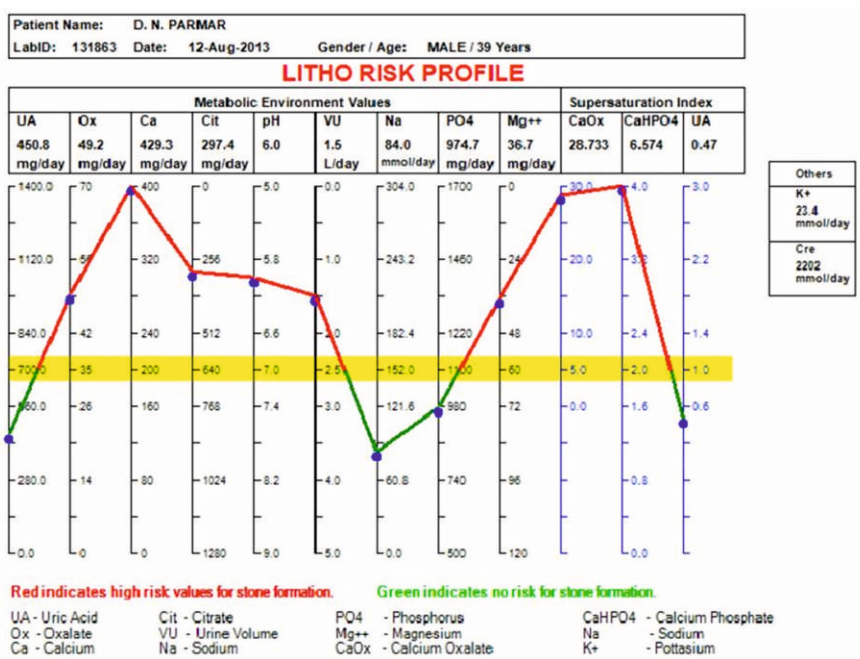
- **Calcium Oxalate Supersaturation (SSCaOx)** – Risk for calcium oxalate stones.
- **Calcium Phosphate Supersaturation (SSCaP)** – Risk for calcium phosphate stones.
- **Uric Acid Supersaturation (SSUA)** – Risk for uric acid stones.

**Report as unique graph: Results of a**ll urinary metabolic profile components can be reportedas unique graph. This is a unique and very useful way to explain metabolic status. Abnormal, high risk values which causes stone formation are shows above cut off values as red, while thosie

### Why is Supersaturation Important?

1. **More Accurate Risk Prediction**
   - The **higher the supersaturation level**, the **greater the risk of stone formation**.
   - It correlates better with stone formation than individual urine components alone.
   - High risk components in graph suggest they can promote stone formation and needs to be controlled.
2. **Guides Personalized Treatment**
   - If **SSCaOx is high**, interventions focus on **reducing calcium and oxalate levels, increasing citrate, and boosting fluid intake**.
   - If **SSUA is elevated**, treatment includes **urinary alkalization with potassium citrate and reducing purine intake**.
3. **Evaluates Treatment Success**
   - A follow-up 24-hour urine test helps determine if dietary or pharmacologic interventions have **lowered supersaturation levels**, reducing stone risk.

**Urine report presentation:**



**Clinical Evidence Supporting Supersaturation Index**

- **Moe et al. (2023)** found that supersaturation levels were **more predictive of recurrent stone formation** than individual urine parameters alone (Moe et al., 2023).
- **A 2022 Mayo Clinic study** demonstrated that patients who successfully reduced supersaturation levels had a **50–70% lower recurrence rate** (Curhan et al., 2022).

**How Does the 24-Hour Urine Test Help in Kidney Stone Prevention?**

1. **Identifies Metabolic Abnormalities**
   - Detects conditions such as **hypercalciuria, hypocitraturia, hyperoxaluria, and low urine volume**.
   - Distinguishes between **dietary and genetic** causes of kidney stones.
2. **Guides Personalized Dietary and Lifestyle Changes**

Based on urine findings, patients receive **tailored dietary advice**, such as:

- **Increase fluid intake** (>2.5L/day) to lower supersaturation.
- **Reduce dietary oxalate** (e.g., spinach, nuts, chocolate) in hyperoxaluria.
- **Lower sodium intake** to reduce urinary calcium excretion.
- **Increase citrate intake** (e.g., lemon juice) to prevent calcium stone formation.

3. **Optimizes Pharmacologic Therapy**
   - **Thiazide diuretics** (e.g., hydrochlorothiazide) for hypercalciuria.
   - **Potassium citrate** for hypocitraturia.
   - **Allopurinol** for hyperuricosuria in uric acid stone formers.
4. **Monitors Effectiveness of Treatment**
   - A **repeat 24-hour urine test** is often performed **6–12 weeks after intervention** to assess whether changes have **lowered supersaturation levels and reduced stone risk**.

**Latest Research and Guidelines Supporting 24-Hour Urine Testing**

1. **American Urological Association (AUA) 2023 Guidelines**
   - **Strongly recommends** 24-hour urine testing in patients with **recurrent stones or high risk factors** (e.g., family history, early-onset stones).
   - Reports that **tailored interventions based on urine analysis reduce recurrence by up to 50%** (AUA, 2023).
2. **European Association of Urology (EAU) 2024 Guidelines**
   - **Urinary supersaturation measurement** is recommended to **identify stone risk and monitor treatment response** (EAU, 2024).
3. **Harvard Kidney Stone Prevention Program (2023 Review)**
   - Found that **patients who reduced their urinary supersaturation index had significantly lower stone recurrence rates**, reinforcing its role in long-term prevention (Taylor et al., 2023).

## CONCLUSION

The **24-hour urine metabolic profile**, including **urinary supersaturation analysis**, is a critical tool for **preventing kidney stone recurrence**. It provides a **detailed assessment of metabolic abnormalities and stone risk**, allowing for **individualized treatment plans** that significantly lower recurrence rates.

Patients with a history of kidney stones should discuss **24-hour urine testing with their healthcare provider** to implement an effective, long-term prevention strategy.

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**CASE REPORT**

# Bioptics in Extreme Pathological Myopia: A Case Report of Vision Restoration Beyond Conventional Limits

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**KEY WORDS :** High Myopia, Bioptics, Phakic Intraocular Lens (ICL), LASIK

## ABSTRACT

**Purpose:** To report an exceptional case of extreme high myopia managed using the bioptics approach—a combination of phakic intraocular lens (ICL) implantation followed by LASIK— for achieving spectacle independence and visual rehabilitation.

**Methods:** A 32-year-old male with pathological myopia exceeding -29.00 D and associated astigmatism underwent detailed ophthalmic evaluation. Given the limitations of LASIK or ICL alone, a staged bioptics treatment plan was implemented. Retinal prophylaxis was followed by toric EVO ICL implantation in both eyes, and one month later, topography-guided femtosecond LASIK was performed to correct residual refractive error.

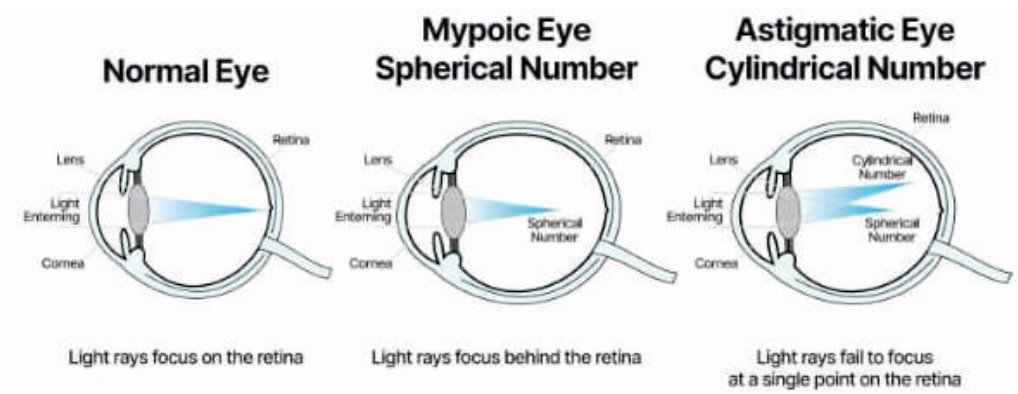
**Results:** Post-procedure, the patient achieved unaided visual acuity of 6/9P in both eyes with significant improvement in functional vision and quality of life.

**Conclusion:** The bioptics approach enabled safe and effective correction of extreme pathological myopia. This case underscores the potential of combining ICL and LASIK to extend refractive surgery boundaries and improve outcomes in highly myopic patients.

**Keywords:** Bioptics, pathological myopia, phakic IOL, LASIK, EVO Toric ICL, refractive surgery, high myopia correction.

## INTRODUCTION

Refractive errors are common vision abnormalities that occur when the eye fails to focus light accurately on the retina, leading to blurred vision. These include myopia (near-sightedness), hyperopia (farsightedness), astigmatism, and presbyopia. High myopia—especially in its pathological form—is a severe and progressive condition characterized

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by excessive elongation of the eyeball, often associated with degenerative changes in the retina and sclera[1].

LASIK (Laser-Assisted In Situ Keratomileusis) reshapes the cornea using an excimer laser and

is typically effective for myopia up to -10.00 D, limited by corneal thickness and safety margins[2]. Phakic intraocular lens (IOL) implantation, by contrast, involves inserting a synthetic lens into the posterior chamber without removing the natural lens and can safely correct myopia up to -20.00 D or more[3]. Bioptics—a sequential combination of phakic IOL implantation followed by LASIK—has emerged as a powerful strategy for extreme refractive errors that exceed the range of either procedure alone[4] [5]. In this article, we present an exceptional case of extreme high myopia where a bioptics approach was employed with remarkable, visionrestoring results.

## CASE REPORT

A 32-year-old male presented with complaints of poor vision with spectacles, difficulty in performing daily tasks, and a desire for spectacle independence. No history of contact lens use or systemic illness was reported.

On examination:

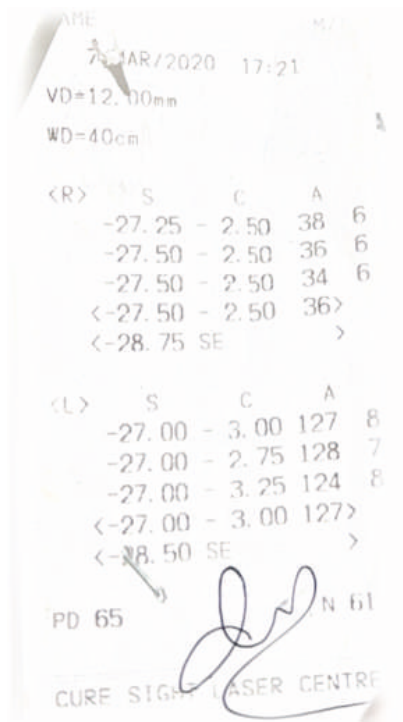
- Spectacle-corrected visual acuity was 6/60 in both eyes.
- Manifest refraction:
  - Right Eye (OD): -28.00/-3.00 × 30, BCVA 6/18P
  - Left Eye (OS): -28.00/-3.00 × 130, BCVA 6/18

Slit-lamp examination revealed normal anterior segments. Fundus examination revealed myopic discs, dull fovea, and multiple peripheral lattice degenerations.

**Findings**

A comprehensive preoperative refractive workup showed:

- **IOP (NCT):** 11 mmHg OU
- **Pachymetry:** OD 521 µm, OS 527 µm
- **Steep K:** OD 45.59 @ 105°, OS 45.48 @ 76°
- **ACD:** OD 3.17 mm, OS 3.14 mm

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- **Axial Length:** OD 29.08 mm, OS 29.12 mm
- **White-to-white:** 11.30 mm OU

Slit-lamp examina on showed clear cornea, quiet anterior chamber, reac ve pupils, and clear lenses. Posterior segment evalua on revealed myopic discs and dull foveal reflexes, with mul ple la ce degenera ons in the peripheral re na. The findings confirmed high pathological myopia with as gma sm, and a bioptics approach using Toric EVO ICL (STAAR Surgical) implantation followed by LASIK was advised.

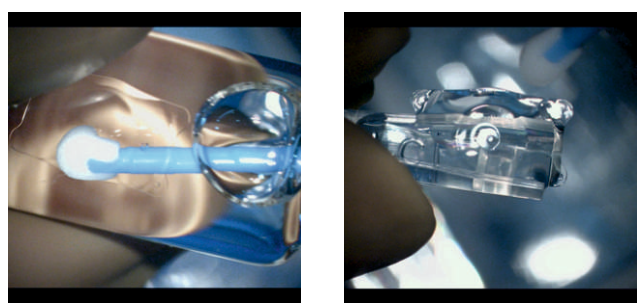
## METHOD

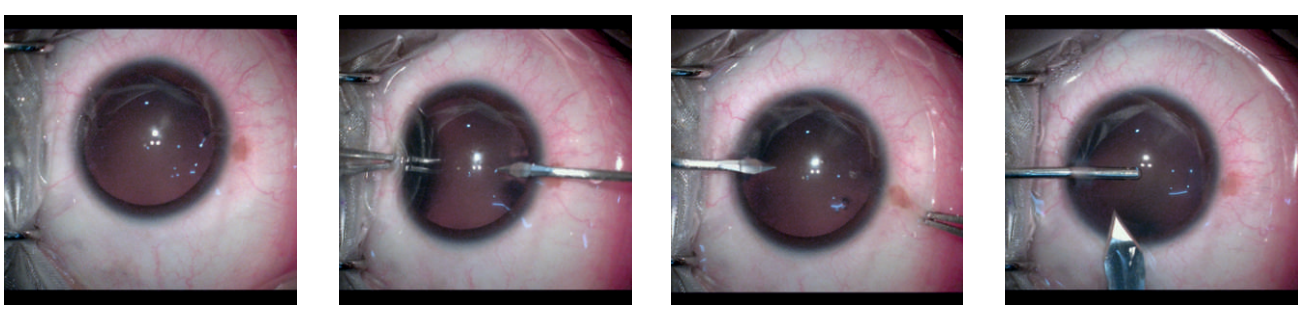
Given the presence of peripheral lattice degeneration, the patient was referred to a retina specialist and underwent prophylactic laser barrage in both eyes. Following retinal stabilization, EVO Toric ICL lenses were ordered based on biometric parameters. After informed consent, ICL implantation was carried out under topical anesthesia. Using the VERION digital marking system, toric ICLs were implanted through a 2.8 mm clear corneal incision, ensuring precise alignment with the astigmatic axis. All four haptics were carefully positioned in the posterior chamber. Anterior chamber washout and wound hydration completed the procedure.

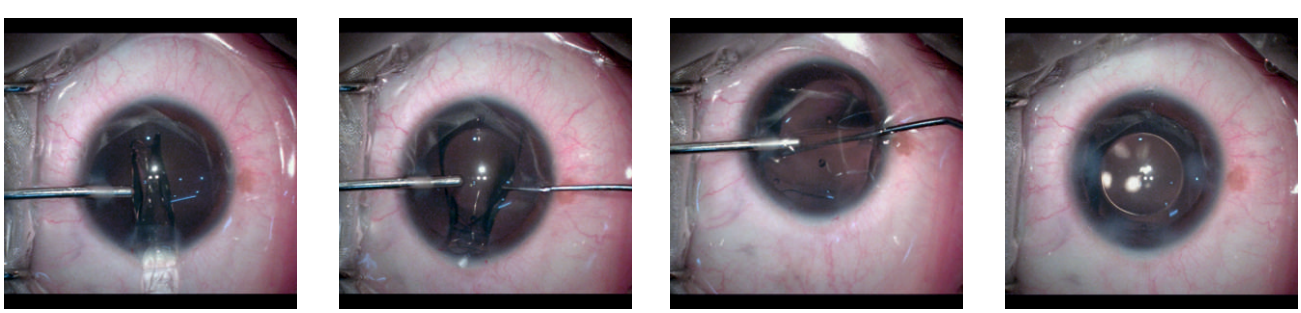
The same procedure was performed for the fellow eye the following day. One week post-operatively, residual refractive errors remained: -8.00 DS (OD) and -9.50 DS (OS), with BCVA of 6/12P in both eyes. One month later, femtosecond-assisted LASIK (FS200) with topography-guided Contoura Vision correction (EX500, Alcon) was performed to address the residual refractive error.

**THE ICL PROCEDURE**

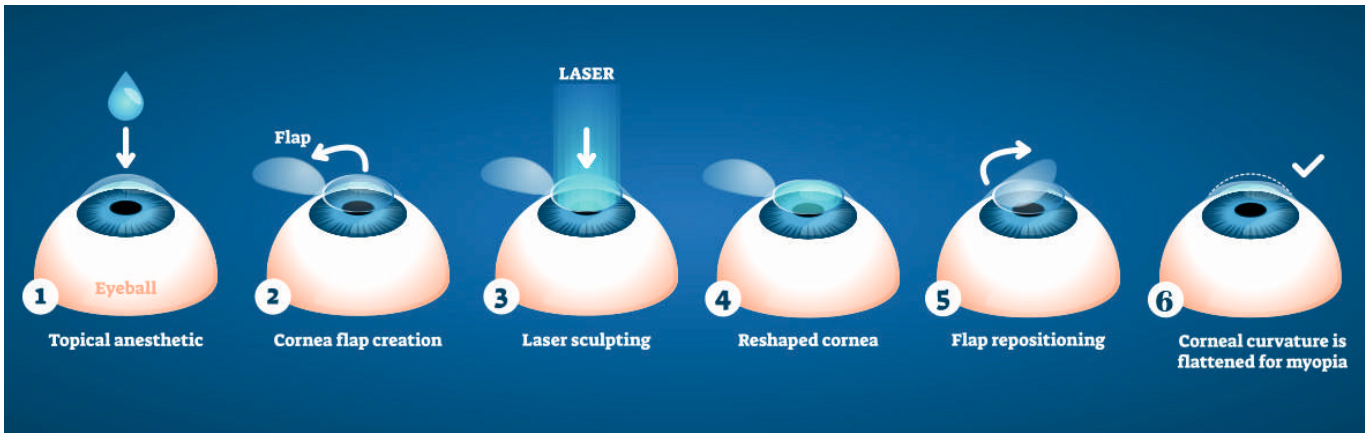
1. Extract the lens
2. Place it in the injector
3. Dilated eye
4. Create first side port
5. Create second side port
6. Create main port
7. Inject lens
8. Adjust lens
9. Tuck in the lens
10. Eye with ICL lens







**Steps for the LASIK procedure**




As a result of this layered procedure, one day post-LASIK, the patient achieved unaided visual acuity of 6/9P bilaterally. There were no postoperative complications. The patient reported significant improvement in visual clarity and confidence in daily life activities. This case illustrates how bioptics enables personalized and comprehensive refractive correction, even in extreme scenarios once considered untreatable.

## ANALYSIS AND DISCUSSION

This case illustrates the expanding horizons of refractive surgery in the context of extreme pathological myopia. The patient's refractive error exceeded -29.00 diopters, well beyond the correction limits of either LASIK or phakic intraocular lenses (IOLs) when used independently. LASIK is typically contraindicated in such high myopia due to insufficient residual stromal bed and elevated risk of postoperative ectasia, especially in the presence of thin corneas and steep keratometry. Similarly, while the EVO Visian ICL has a broader correction range (up to approximately -20.00 D in standard practice), it is often unable to neutralize extreme refractive errors completely when approaching the upper physiological limits of the eye's axial length and anterior chamber depth.

Bioptics—a strategy first introduced by Zaldivar et al.—was developed precisely to address this gap, enabling staged correction of extreme myopia through a combination of phakic IOL implantation and subsequent LASIK refinement[4]. In this case, the Toric EVO ICL was used to debulk the majority of the myopia and astigmatism, preserving the cornea's structural integrity and allowing for precise, lower-powered LASIK correction later. This approach leverages the benefits of each modality: the safety and reversibility of phakic IOLs and the fine-tuning capabilities of topography-guided LASIK.

A significant clinical consideration was the presence of lattice degeneration in the peripheral retina, a known risk factor for retinal detachment in high myopia. This was appropriately addressed with preoperative retinal laser barrage, reducing the risk of intraoperative or postoperative retinal complications. The delay of 15 days post-laser treatment allowed for retinal stabilization before proceeding with anterior segment surgery.

The use of advanced technologies such as the VERION digital axis marking system played a critical role in ensuring optimal Toric ICL alignment, which is paramount for effective astigmatic correction. Misalignment by even 10 degrees can reduce astigmatic correction efficacy by up to 33%[6]. Additionally, the decision to use Contoura Vision with the EX500 excimer laser allowed for

customized topography-guided treatment, improving higher-order aberrations and optimizing postoperative visual quality[7].

Another key aspect of this case was patient counselling and staged consent. In patients with extreme myopia, expectations must be managed realistically. The patient was made aware that spectacle independence would likely require a two-step approach and that postoperative residual refractive error was to be expected after ICL implantation alone. This collaborative approach between surgeon and patient is essential for success in such complex cases.

Overall, this case highlights the clinical utility and safety of the bioptics approach in patients who would traditionally have been considered unsuitable for refractive surgery. It showcases the strength of a modular, individualized treatment paradigm that combines structural preservation with high-precision refractive correction. It also reflects the growing consensus in ophthalmology that treating extreme refractive errors requires not just surgical expertise but also the integration of high-resolution diagnostics, advanced surgical planning tools, and careful postoperative management.

## CONCLUSION

Bioptics offers a versatile and powerful approach for the correction of high pathological myopia. Combining the strengths of phakic IOLs (high correction range, reversibility, preservation of accommodation) with LASIK (refinement and precision) allows for enhanced patient outcomes. As diagnostic and laser technologies evolve, the scope of bioptics will continue to expand, offering hope and spectacle independence to patients with extreme refractive errors. This case serves as a compelling example of the transformative potential of bioptics in modern refractive surgery.

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CASE REPORT

# Ectopic Tooth in Ethmoid Sinus-A Rare Case Report.

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**KEY WORDS :** Ectopic Tooth in Ethmoid Sinus

**ABSTRACT**

Ectopic eruption is a disturbance in which the tooth does not follow its usual course. Ectopic eruption of tooth occurs in a variety of locations. Commonly seen in palate and maxillary sinus, they have also been reported in nasal cavity, para-nasal sinus, orbit, mandibular condoyle, coronoid process, and facial skin. Eruption in sino-nasal cavity is a rare clinical entity occurring in only 0.1-1% of the general population. (7) We report a rare case of right maxillary swelling and sinusitis caused by ectopic tooth in right maxillary and ethmoid sinus and successful removal. The symptoms of the patient disappeared after surgery. Here we present a manuscript is to provide a brief review regarding the incidence, symptoms and management technique use for correcting ectopically erupted tooth.

## INTRODUCTION

The development of deciduous tooth starts in the sixth week of intra-uterine life with the development of dental lamina. Then the ectodermal layer proliferation begins to form the permanent dentition between 5$^{th}$ and 10$^{th}$ post natal months. It is a multistep event in which interaction between the oral epithelium and the underlying mesenchymal tissue plays a vital role. Abnormal interaction at any step may result in ectopic eruption of permanent tooth development.[3] Ectopic teeth are those located in the jaw bones at regions other than the alveolar arch ectopic placement of tooth bud occurs due to genetic and environmental relationship factors which cause a budding tooth to congenitally migrate in the initial stages of embryogenesis, or is the result of a change in the displacement of teeth owing to local factors. Although etiology of the eruption of a tooth is unclear, developmental disturbances such as cleft palate, displacement of teeth by trauma or cyst, infection, genetic factors, crowding and dense bone are the suspected clinical conditions.

## CASE REPORT

A male 13 yr old patient presented to us with a complain of swelling over right side cheek region and nose block for 2 years. The swelling was small in size which gradually increased with right side nose block.

Patient had no any other complain. Clinically swelling was 3×3cm$^2$ size firm, fixed non tender over

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right maxillary region at canine fossa. Patient had high arched palate and protrusion of upper teeth.

CT-PNS confirmed radio-opaque erupted 2nd molar in anterior wall of maxillary sinus and another tooth in posterior ethmoid with odontogenic sinusitis changes.

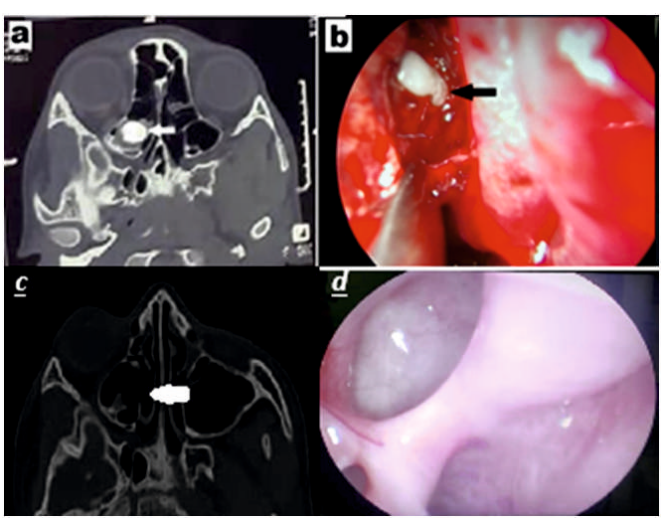
Oro-maxillofacial surgeons opinion was taken and combined approach surgery was planned.

The patient underwent surgical excision under GA. A tooth in posterior ethmoid was excised by doing F.E.S.S along with the cyst layer and soft small tissue covering it. The 2nd molar in anterior wall of maxillary sinus was excised by Caldwell Luc approach. Specimen was verified as tooth with cystic wall. Patient recovered well and had no recurrence.

## DISCUSSION

Intranasal ectopic teeth are rare. Yeung and Lee reviewed the literature and found a total of 41 well-documented cases.[1][2] The age of the patients ranged from 3 to 62 years with mild male predominance. In regard to location, there was no side predilection in the literature. Eruption in Sinonasal cavity is rare clinical entity occurring in only 0.1-1% of the general population.[7]

**Image I**



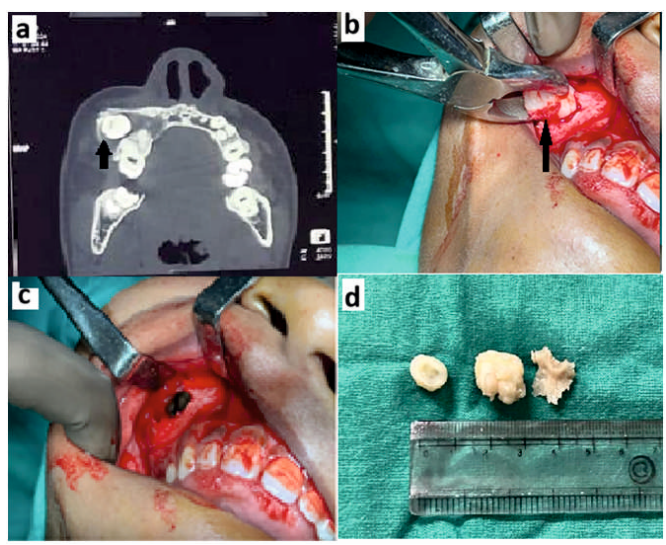

*(a) Axial CT showed ectopic tooth erupted inside right posterior ethmoid sinus surrounded by inflammatory tissue.*

*(b) Intra operative nasal endoscopic view with 0° rigid scope shows ectopic tooth in right posterior ethmoid sinus.*

*(c) Post-operative axial CT showed empty space in right posterior ethmoid sinus. [as compared to image 1(a)]*

*(d) Post-operative nasal endoscopic view with 0° rigid scope showed well epithelised right posterior ethmoid sinus.*

**Image II**




*(a) Axial CT showed ectopic (!) erupted tooth in right anterior wall of maxillary sinus.*

*(b) Intra-operative(!) picture of erupted tooth on right maxilla-anterior wall via Caldwell Luc approach.*

*(c) Post removal picture of anterior maxillary wall.*

*(d) Extracted teeth from posterior ethmoid sinus and anterior maxillary sinus with cyst layer.*

The teeth in question were said to have arisen mostly from the permanent dentition or to have been supernumerary. Ectopic eruption of teeth occurs in a variety of locations including the nasal cavity, para-

nasal sinus, coronoid process, mandibular condyle, facial skin, and orbit. Although the cause of ectopic teeth remains unclear, some theories have been proposed.

Very rarely the tooth may be present in the maxillary sinus and ethmoid sinus with or without symptoms. The displacement of the tooth may be due to the pressure caused by the cystic enlargement.

Other etiology may include developmental disorders such as cleft palate, trauma causing displacement of the teeth, maxillary infection, crowding, genetic factors and high bone density. Symptoms may include recurrent or chronic sinusitis or purulent discharge from the nose and sometimes elevation of the floor of the orbit.

Many surgical approaches have been reported in the literature including the endoscopic removal of such ectopic teeth and associated lesions. Di Pasquale P and Shermetaro C[4] used a nasal endoscope to create a middle meatal antrostomy and deliver the ectopic tooth and its cystic contents.

The endoscopic techniques are being used for removal of an intranasal ectopic tooth obstructing osteomeatal complex. Ectopic maxillary tooth was removed by Caldwell Luc approach. Postoperative follow-up with radiographic examination at regular intervals is mandatory.

Although traditional CaldwellLuc procedure provides a direct view into the maxillary sinus, it is associated with more morbidity than transnasal endoscopy. Transnasal removal of the tooth may be attempted if the tooth is small and sited near the maxillary ostium.

## CONCLUSION

In summery, ectopic tooth is an uncommon cause of sinusitis. Some differential diagnose of a radiopaque mass in sinuses are dentigenious cyst ,osteoma, foreign body, and rhinolith.

However otolaryngologists should be aware of this disease entity when encountering patient presenting with maxillary swelling and recurrent sinusitis unresponsive to medical treatment.

In our case, there was cystic change within the sinus and the opacification in the anterior wall of right maxilla. A FESS and conventional CaldwellLuc operation was performed. The tooths crown and root were removed separately.

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**CASE REPORT**

# Role of Preimplantation Genetic Testing (PGT) in Prevention of Hemoglobinopathy before Birth

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**KEY WORDS :** Hemoglobinopathy, Preimplantation Genetic Testing (PGT)

**ABSTRACT**

Hemoglobinopathies are among the most common monogenic disorders worldwide, affecting millions annually. Preventive strategies are crucial, especially in high-risk populations. This article explores the use of Preimplantation Genetic Testing (PGT) as a proactive reproductive option for carrier couples, preventing the transmission of hemoglobinopathies such as sickle cell disease and thalassemia. We present a case report illustrating the clinical application of PGT-M (for monogenic diseases) and PGT-A (for aneuploidies) in achieving a successful, disease-free pregnancy

A young, nulligravida couple presented in 2023 with the aim of having a genetically healthy child.

- **Genetic Screening:**
  - o **Female:** Sickle cell trait
  - o **Male:** β-thalassemia trait

**IVF and PGT Procedure:**

- **Oocyte Retrieval:** 9 metaphase II oocytes
- **Embryo Culture:** 5 blastocysts developed
- **Biopsy:** All 5 embryos biopsied for PGT-A and PGT-M

**Genetic Results:**

- **PGT-A:** 2 embryos normal, 3 abnormal
- **PGT-M:**
  - o 1 embryo: mutation-free
  - o 1 embryo: β-thalassemia carrier

The mutation-free embryo (PGT-A + PGT-M normal) was transferred in a frozen embryo transfer (FET) cycle. The patient achieved a positive β-hCG, followed by an uneventful pregnancy and the delivery of a healthy baby.

**Results of PGT-A and PGT-M**

| Embryo No. | PGT-A Result | PGT-M Result | Outcome |
|---|---|---|---|
| 1-3 | Abnormal | Not Tested | Discarded |
| 4 | Normal | β-thalassemia carrier | Not transferred |
| 5 | Normal | Mutation-free | Transferred – Healthy baby delivered |

## CONCLUSION

- PGT-M is a proven, reliable, and safe method for the prevention of hemoglobinopathies in carrier couples.
- Technological advances, including whole-genome approaches, have significantly improved the scope and accuracy of PGT-M.
- Clinical implementation of novel techniques requires rigorous validation aligned with international best practices

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## DISCUSSION

Hemoglobinopathies are among the most common monogenic disorders worldwide, affecting millions annually. Preventive strategies are crucial, especially in high-risk populations. This article explores the use of Preimplantation Genetic Testing (PGT) as a proactive reproductive option for carrier couples, preventing the transmission of hemoglobinopathies such as sickle cell disease and thalassemia. We present a case report illustrating the clinical application of PGT-M (for monogenic diseases) and PGT-A (for aneuploidies) in achieving a successful, disease-free pregnancy.

Hemoglobinopathies, the most prevalent class of inherited monogenic disorders globally, affect approximately 300,000 to 400,000 newborns annually. It is estimated that 7% of the global population are carriers of pathogenic variants associated with these conditions[1].

These disorders include structurally abnormal hemoglobins (e.g., HbS, HbD, HbE) and thalassemias, typically inherited in an autosomal recessive manner. The conditions manifest due to either structural hemoglobin variants—such as sickle cell disease (SCD)—or due to reduced synthesis of hemoglobin chains, as seen in thalassemia syndromes.

Clinical severity varies widely, with the most severe forms including:

- Thalassemia major (TM)
- Sickle cell syndromes
- Hemoglobin E-thalassemia
- Hb Bart's hydrops fetalis syndrome[2]

### Sickle Cell Disease (SCD)

SCD is a hemoglobin disorder that demands lifelong management and significantly increases childhood morbidity and mortality. It arises from inheritance of:

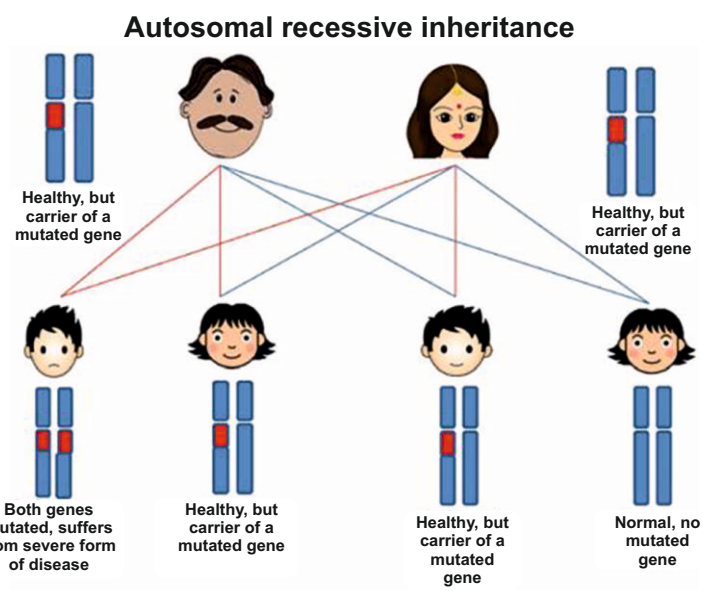
- Two HbS alleles (HbSS)
- HbS + HbC (HbSC)
- HbS + β0 thalassemia
- HbS + β+ thalassemia

### Thalassemias

Thalassemias are classified as:

- Thalassemia Major (TM)
- Thalassemia Intermedia (TI)
- Thalassemia Minor (Trait)

**Figure 1: Understanding the Inheritance of Autosomal Recessive Disorder (citation: Prevention and Control of hemoglobinopathies in India; National Health Mission; 2016)**



While TM and TI require lifelong transfusions and iron chelation, Thalassemia Minor (β-Thalassemia Trait) is asymptomatic. The disease results from inheriting abnormal β-thalassemia alleles from both parents or one β-thalassemia allele and one structural hemoglobin variant (e.g., HbE or HbS).

**Preventive Reproductive Options**

For carrier couples, reproductive options include:

1. **Prenatal Diagnosis (PND):**
   - **Chorionic Villus Sampling (CVS):** ~12 weeks gestation
   - **Amniocentesis:** ~16 weeks gestation
2. **Preimplantation Genetic Testing (PGT):**
   - Conducted on embryos produced via in vitro fertilization (IVF).
   - Embryos free of disease-causing mutations are selected for uterine transfer, minimizing the risk of an affected pregnancy and the ethical dilemmas associated with late-term termination[3].

*Figure 1: Understanding the Inheritance of Autosomal Recessive Disorders (Source: NHM, India, 2016)*

**Preimplantation Genetic Testing (PGT)**

**Types of PGT:**

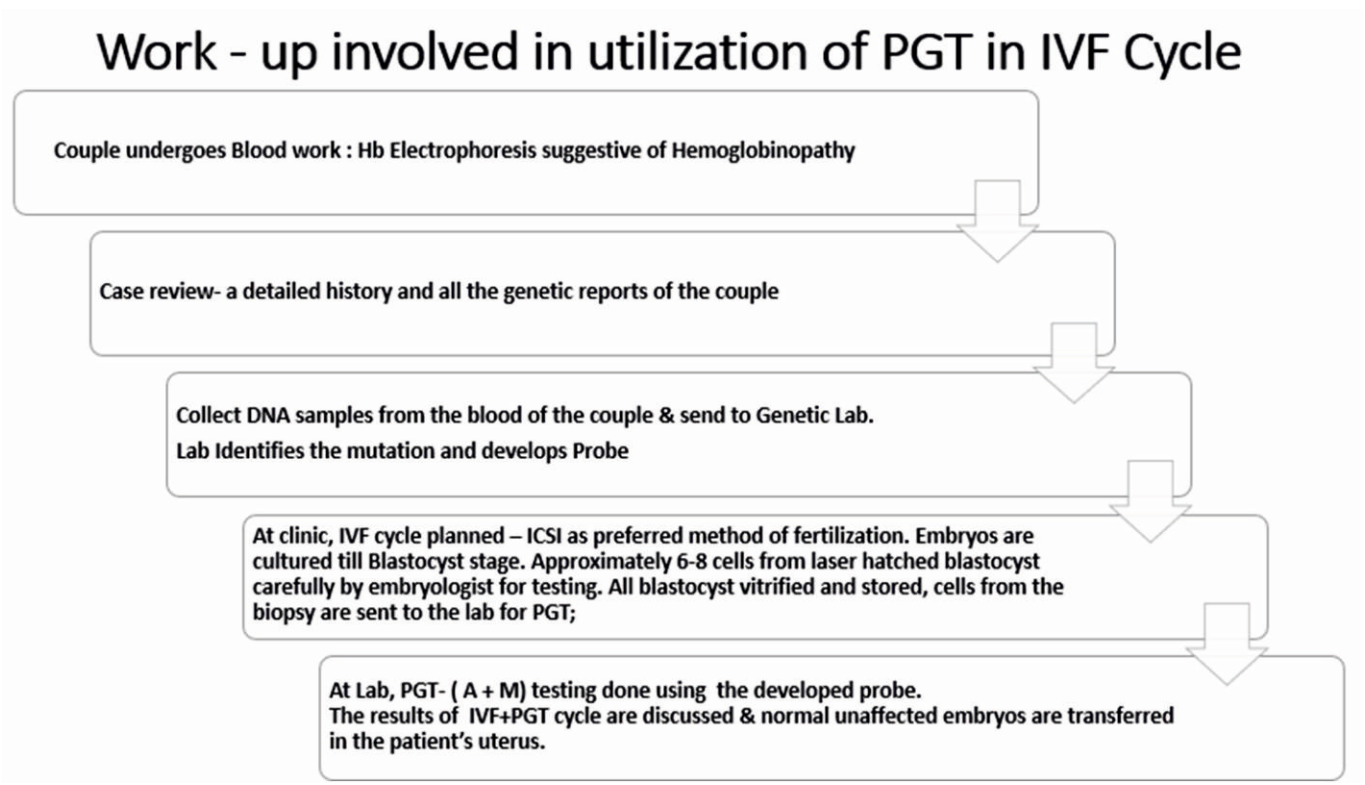
- **PGT-A** (for aneuploidies)
- **PGT-SR** (for structural rearrangements)
- **PGT-M** (for monogenic disorders)

The **ESHRE PGT Consortium** provides global best practice guidelines on the organization and execution of PGT.

**PGT-M for Hemoglobinopathies and HLA Typing**

Hematopoietic stem cell transplantation (HSCT) offers a potential cure for hemoglobinopathies when a matched donor is available. In families with an affected child, **PGT-M with HLA typing** enables the

**Figure 2: Work-up involved in utilization of PGT in IVF Cycle**



selection of embryos that are both disease-free and HLA-matched to the affected sibling.

- The chance of finding an embryo that is both unaffected and HLA-matched is approximately **18.8%** for autosomal recessive conditions[7].
- PGT-M with haplotyping is commonly applied for **β-thalassemia**.

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CASE REPORT

# Minimally invasive spine surgery in high grade listhesis : A case report

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**KEY WORDS :** Minimally invasive spine surgery, High-grade listhesis

## ABSTRACT

**Introduction:** High-grade listhesis is challenging to treat due to anatomical deformities and instability. While open surgery is standard, minimally invasive spine surgery (MISS) offers reduced morbidity but is limited by visualization constraints.

**Case Presentation:** A 35-year-old male presented with progressive low back pain and intermittent left leg radiculopathy. Imaging showed L5–S1 Grade IV listhesis with left foraminal stenosis. Conservative management for six weeks failed.

**Surgical Technique:** We operated the case using MISS technique to reduce and

**Outcome:** The patient was mobilized on postoperative day two. Recovery was uneventful, and imaging showed satisfactory reduction and alignment.

**Conclusion:** MISS with navigation is a feasible and effective technique in selected cases of high-grade listhesis, combining reduced surgical morbidity with adequate reduction and instrumentation accuracy.

## CASE REPORT

A 35-year-old man presented with complaints of back pain associated with a feeling of instability and intermittent left leg pain. He had been working as a bank employee for the past 10 years. He started noticing back pain when sitting for prolonged durations and needed to frequently change positions while at work for the past 6 months. The back pain increased in intensity over the last 3 months, such that the patient was unable to attend work and could not ride a two-wheeler due to the pain. He also developed radiating pain extending to the left lower limb up to the knee. This radiating pain was intermittent and occurred while sitting cross-legged.

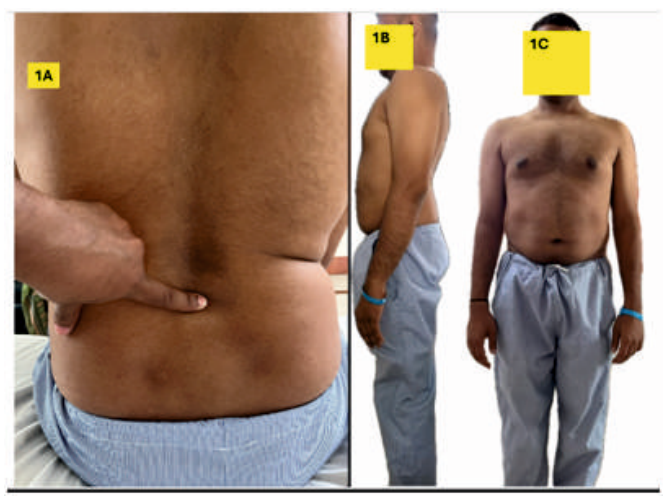


**Figure 1: 1A: Clinical image showing a palpable step deformity in the lower lumbar spine.**

**1B, 1C: Standing clinical images demonstrating good sagittal and coronal alignment.**

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On examination, there was a palpable step at the lower lumbar spine. The range of motion (ROM) of the lumbar spine was restricted. Straight leg raise test (SLRT) was positive on the left side; however, neurological examination was intact. From the anterior aspect, the head is centered over the pelvis, and the lateral view shows the external auditory meatus (EAM), shoulder, hip, and malleoli aligned in a straight line, indicating appropriate coronal and sagittal alignment.

## IMAGING

Imaging showed L5–S1 high-grade listhesis (Grade IV) with foraminal stenosis, more on the left than the right, and an78 degree lumbosacral slip angle. A 6-week trial of conservative management was done, following which a decision to proceed with surgery was made.

## REVIEW OF LITERATURE

Epidemiological studies show the rates of isthmic spondylolisthesis to be 7–8% of the total population.[1] The principles of surgical management in these cases are decompression, reduction, and interbody fusion. However, high-grade dysplastic listhesis presents many challenges, including the dome-shaped sacrum and dysplastic pedicles in L5, which cause difficulty in instrumentation of pedicle

**Figure 2: 2A: Anteroposterior (AP) radiograph of the lumbosacral spine.**

**2B, 2C: Lateral radiographs in flexion and extension, respectively.**

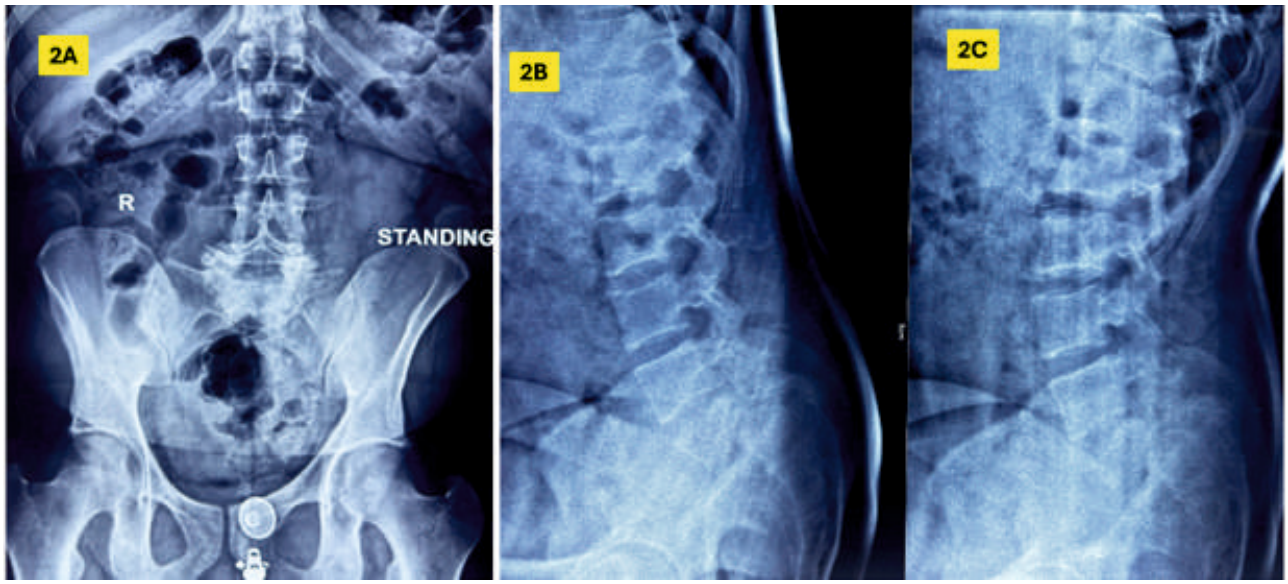



**Figure 3: 3A, 3B, 3C: MRI images showing right parasagittal, mid-sagittal, and left parasagittal views, respectively, along with axial cuts at the L5–S1 level.**

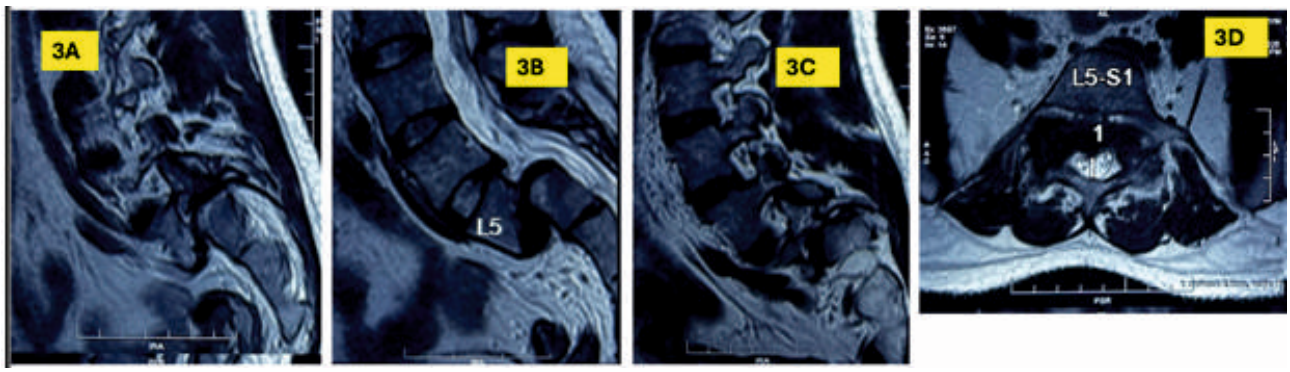
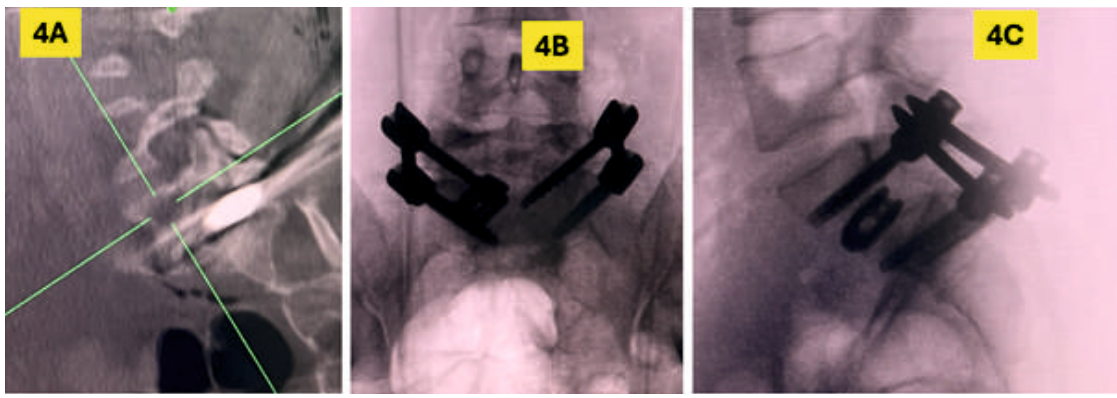


**Figure 4: 4A: Intraoperative CT image showing dysplastic pedicles of L5.**

**4B, 4C: Postoperative AP and lateral radiographs showing partial reduction achieved following MISS and instrumentation.**




screws.[2–4] In such cases of high-grade listhesis, the decision on whether to reduce or not depends on the sagittal alignment. In a well-aligned spine, in-situ fusion can be attempted. However, in an imbalanced spine, reduction and fusion is preferable.[5] In cases where reduction of listhesis is attempted, it is imperative to have perfectly placed intra-pedicular screws, overcoming the dysplastic anatomy. With the advent of 3D navigation, this difficult task is made simpler. The exact trajectory can be identified and placed accurately and with ease under direct visualization.[6]

Studies comparing the outcomes of minimally invasive versus open surgery in low-grade listhesis show that minimally invasive spine surgery (MISS) results in reduced blood loss and shorter hospital stay, with similar functional and pain outcomes.[7] However, the use of MISS in high-grade listhesis carries a high risk of implant-related complications and inadequate decompression due to limited visualization compared to open procedures. When comparing reduction achieved in open versus MISS techniques, the aggressive disarticulation possible during open surgery contributes to better correction and sagittal alignment. Even though open surgery has advantages in high-grade listhesis, MISS significantly reduces the morbidity associated with spinal procedures. Other advantages include reduced intraoperative blood loss, decreased postoperative pain, shorter recovery time, and a reduced duration of bed rest.[8]

In order to retain these advantages, multiple minimally invasive techniques for the reduction of high-grade listhesis have been described.[9–11] The objective of this case report is to describe the authors' method of reduction of high-grade listhesis and fusion using a navigated MISS technique.

## TECHNIQUE

The patient is placed in the prone position on a radiolucent table. A tubular retractor is docked over the ipsilateral facet, and facetectomy is performed. Using an osteotome, entry into the disc space is achieved. Serial reamers are used to dilate the disc space. Leaving the reamer in situ in a vertical position to open up the disc space, docking is then performed on the contralateral facet. Following contralateral facetectomy, a comparatively easier entry into the disc space is achieved, allowing for disc space preparation. After cage insertion, pedicle screws are placed using navigation assistance. The use of navigation greatly improves the accuracy and ease of placing screws in the frequently encountered dysplastic pedicles. Bilateral rods are connected, and fixation is completed.

## OUTCOME

Postoperative recovery was unremarkable. The patient was mobilized on the second postoperative day. Postoperative X-ray showed satisfactory reduction.

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CASE REPORT

# Ectodermal Dysplasia Case Series with One Rare Variant of Ectodermal Dysplasia with cutaneous Toe only Syndactyly (EDSS1 A -Novel NECTIN 4 Variant)

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**KEY WORDS :** Ectodermal Dysplasia, Syndactyly, Hypo Hidrotic, Anhidrotic, Pili Torti, Palmer Keratosis Nectin 4

**ABSTRACT**

Background- Ectodermal Dysplasia are group of rare inherited genetic disorders with structural and functional defects of more than one ectodermal appendage.

## CASE CHARACTERISTICS

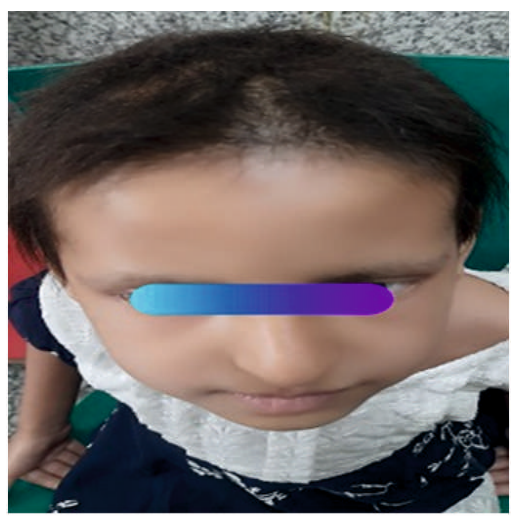
Presenting 4 patients of Ectodermal Dysplasia of which one female patient is with rare EDSS1 Novel NECTIN 4 Mutation with cutaneous toe only syndactyly probably 13th reported case till now and second in India. One male patient with EDA mutation whose mother also has mild features of Ectodermal Dysplasia and two male patients who belong to same family and are siblings,

## CASE REPORT

### 1. Ectodermal Dysplasia with cutaneous toe only syndactyly with Novel EDSS1 (NECTIN 4) Mutation

A Six years old female child, second in order of 2, born out of non-consanguineous marriage, visited OPD for complaint of fever. On examination, she had dry skin especially on palms and soles, brittle, light colored, scanty, frizzy hair, scanty eyelashes and eyebrows, conical, peg shaped teeth, dystrophic nails, patient also had gross caries, gingivitis, pulpitis on dental examination. On questioning, parents gave history of absent sweating and intolerance to heat. On careful examination, she also had bilateral cutaneous syndactyly between 2nd and 3rd toes. Patient's NGS panel was sent for gene mutation studies. It confirmed EDSS1/ NECTIN 4 gene mutation with AR inheritance. (Table 1) Parents and patient did not consent for skin biopsy and hair examination. Elder sibling was not brought for examination. (Image No 1.1, 1.2, 1.3, 1.4)

**Case 1.1 Ectodermal dysplasia with sparse, brittle, frizzy. Light hair on scalp and eyelashes, Eyebrows**



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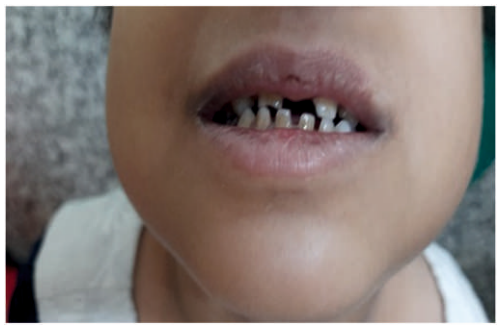
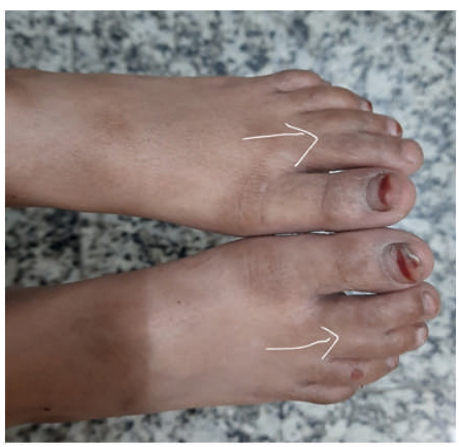
Emai : drchitram@yahoo.co.in

**Case 1.2 Ectodermal Dysplasia with Hypodontia, peg shaped teeth with wide gaps with caries**



**Case 1.3 Ectodermal Dysplasia with Bilateral cutaneous Toe only syndactyly and nail dystrophy.**



**Case1.4 Ectodermal Dysplasia panel- gene mutation study.**

harbour mutation in the NECTIN4 gene and has been evaluated for pathogenic variations in the genes listed in appendix 1.

RESULTS

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

| Gene[#] (Transcript) | Location | Variant | Zygosity | Disease (OMIM) | Inheritance | Classification |
|---|---|---|---|---|---|---|
| NECTIN4 (-) (ENST00000368012.4) | Exon 3 | c.499del (p.Leu167TrpfsTer15) | Homozygous | Ectodermal dysplasia-syndactyly syndrome-1 | Autosomal recessive | Pathogenic |

[#]Genetic test results are reported based on the recommendations of American College of Medical Genetics [1].

ADDITIONAL FINDINGS: NO VARIANT(S) OF UNCERTAIN SIGNIFICANCE (VUS) DETECTED

No other variant(s) that warrants to be reported was detected. For any further technical queries please contact invitromdx@gmail.com

The coverage of ectodermal dysplasia panel genes is given in appendix 1.

VARIANT INTERPRETATION AND CLINICAL CORRELATION

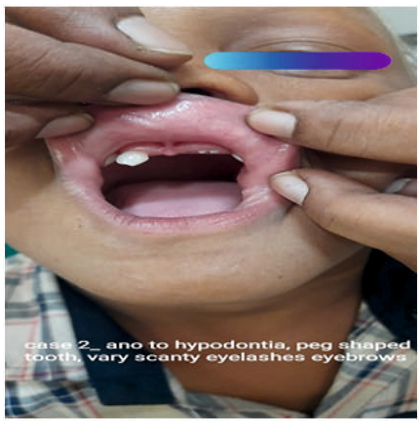
**Variant description:** A homozygous single base pair deletion in exon 3 of the NECTIN4 gene (chr1:g.161077684del; Depth: 163x) that results in a frameshift and premature truncation of the protein 15 amino acids downstream to codon 167 (p.Leu167TrpfsTer15; ENST00000368012.4) was detected (Table). This variant has not been reported in the 1000 genomes, gnomAD and our internal databases. The in silico prediction# of the variant is damaging by MutationTaster2. The reference region is conserved across species.

**2. Ectodermal dysplasia with EDA gene mutation with Mother having mild features of ED.**

A 7 years old male patient visited Paediatric OPD with complaint of very high-grade fever and absence of sweating. This child was 3rd in order of 3 born out of non-consanguineous marriage. The child had dry sweat free skin, dystrophic nails, scanty to absent eyelashes and eyebrows, very scarce light colored golden frizzy, brittle hair, delayed dentition with only few peg shaped teeth. Again, patient and parents did not consent for skin biopsy and hair examination. Patient's NGS panel was sent for gene mutation studies. It confirmed EDA gene mutation with XR inheritance. It was also observed that patient's mother has scanty thin hair, scanty eyelashes and eyebrows, widely spaced conical teeth and history of delayed dentition and less sweating. Elder 2 sisters were not brought for examination. (Image No. 2.1, 2.2, 2.3, 2.4)

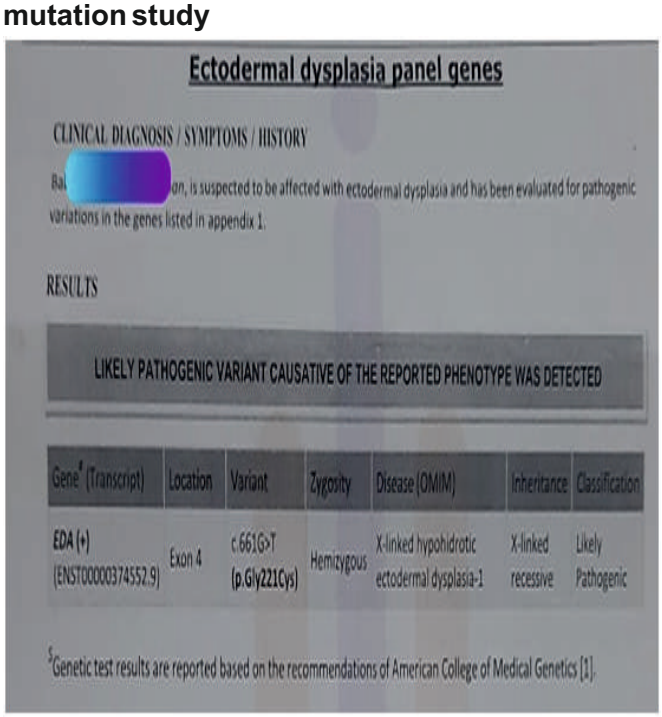
**Case 2.1 Ectodermal Dysplasia with Hypotrichosis, sparse, brittle, light hair on scalp, eyelashes and eyebrows**

**Case 2.2 Ectodermal Dysplasia with hypodontia, peg shaped teeth, scanty eyelashes and eyebrows**




**Case 2.3 Mother of case 2 with hypotrichosis on scalp, eyelashes eyebrows, wide spaced teeth.**

**Case 2.4 Ectodermal Dysplasia Panel -gene mutation study**



Ectodermal dysplasia panel genes

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

[redacted] on, is suspected to be affected with ectodermal dysplasia and has been evaluated for pathogenic variations in the genes listed in appendix 1.

RESULTS

LIKELY PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

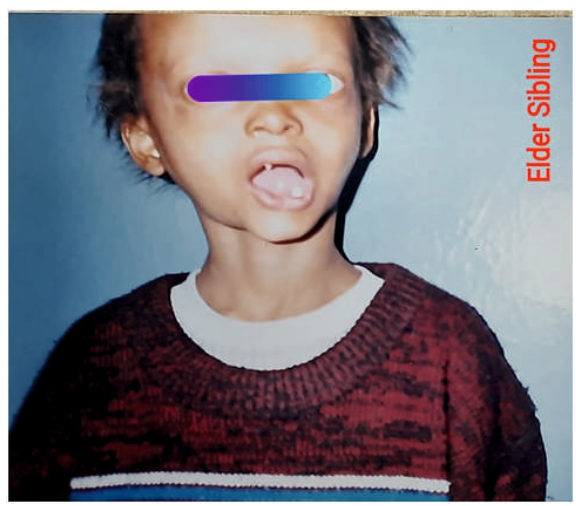
| Gene* (Transcript) | Location | Variant | Zygosity | Disease (OMIM) | Inheritance | Classification |
|---|---|---|---|---|---|---|
| EDA (+) (ENST00000374552.9) | Exon 4 | c.661G>T (p.Gly221Cys) | Hemizygous | X-linked hypohidrotic ectodermal dysplasia-1 | X-linked recessive | Likely Pathogenic |

*Genetic test results are reported based on the recommendations of American College of Medical Genetics [1].

**3. Ectodermal Dysplasia in 2 siblings.**

Two siblings, 9 and 7 years old, visited Paediatric OPD for high grade fever and intolerance to heat and absent sweating. Both brothers had severe features of Ectodermal Dysplasia in form of thin, sparse to absent golden brown hair, palmer keratoses, absent to minimal dentition, peg shaped teeth, absent eyelashes, sparse eyebrows, intolerance to heat and absent sweating. Both patients did not follow up for any investigations so further work up could not be done. (Image No. 3.1, 3.2)

**Case 3.1 Elder sibling with Ectodermal Dysplasia with hypotrichosis, hypodontia, absent eyelashes and eye brows, peg shaped teeth.**




**Case 3.2 Younger sibling with Ectodermal Dysplasia with hypotrichosis, anodontia, absent eyelashes and eyebrows.**

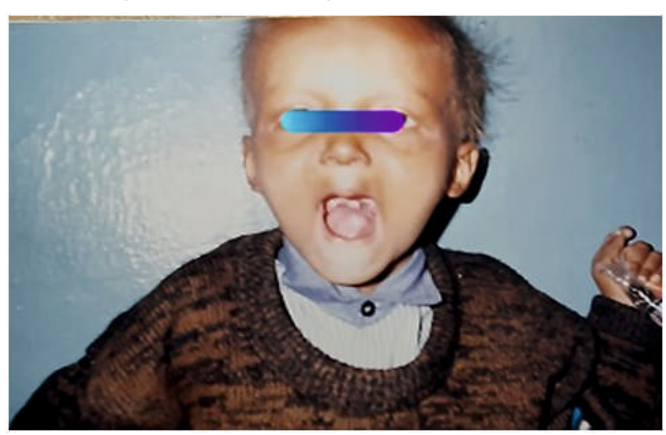


**Table- 1 NECTIN 4 Variants and countries of origin**

| S.N. | Country of Origin | NECTIN 4 Variant | References |
|---|---|---|---|
| 1 | Algeria | c.851G>A, p. Arge284Gln | 1 |
| 2 | Italy | c.554C>T, p. Thr185Met; c.906delT, p. Pro304HisfsTer2 | |
| 3 | Pakistan | c.635C>G, p. Pro212Arg | |
| 4 | Afghanistan | c.724G>A, p. Val242Met | |
| 5 | Denmark | Exon 2 in- frame deletion | |
| 6 | Jammu and Kashmir | c.181C>T, p. AspGln61Ter | |
| 7 | Turkey | c.247C>T, p. His83Tyr | |
| 8 | Pakistan | c.242T>C, p. Leu81Pro | |
| 9 | Turkey | c.229C>T, p. Gln77Ter | |
| 10 | Italy | c.1150delC, p. Gln384ArgisTer7 | |
| 11 | Pakistan | C.163C>T; p. Arg55* | 4 |
| 12 | Japan | c.400C>T; p. Arg134* | 5 |
| 13*Our Case | India | c.499 del, p. Leu167TrpfsTer15 | Our case |

## DISCUSSION

Ectodermal Dysplasia are rare inherited genetic disorders. They are characterized by various defects in one or more ectodermal appendages like hair, skin, teeth, nails etc. The incidence of Ectodermal Dysplasia (EDS) is 7:10,000.[1]

One of case reported here Ectodermal Dysplasia with cutaneous Toe only syndactyly due to Novel NECTIN 4 gene mutation (AR) is still more rare with prevalence of 1 < 1000000. This is probably 2nd reported case in India as per review of literature.

Ectodermal Dysplasia are classified into more than 10 subgroups depending on involvement of ectodermal appendages. Anhidrotic or hypo hidrotic variant is most common. Common variant is of XR inheritance2 Patients with Ectodermal Dysplasia will have various degrees of hypotrichosis with thin, sparse, light colored, brittle hair on scalp, eye brows, eyelashes. Pili Torti (twisted hair) is common feature, they may have hypodontia or Anodontia, delayed dentition, peg or conical shaped teeth, widely spaced teeth, dry skin especially of palms and soles with absent sweating, dystrophic nails. Because of decreased or absent sweat glands, these patients have intolerance to heat and may develop malignant hyperpyrexia on exposure to heat especially in summer. Few patients may have ocular manifestations like hypermetropia or crowding of lens, few may have high arched palate and few with cleft lip and cleft palate [3,5]

Ectodermal Dysplasia with cutaneous toe only syndactyly (EDSS1) is very rare. It has Autosomal Recessive inheritance and there is mention of reported very less cases in literature. Literature review identified 3 sporadic EDSS1 cases and 6 families with more than 2 affected members. The cases are reported from countries like Turkey, Denmark, Algeria, Italy, Pakistan, Afghanistan and Jammu Kashmir India (Table1) Most of the cases reported have bilateral cutaneous syndactyly of both fingers and toes. There is one girl reported to have toe only cutaneous syndactyly and second one with barely visible 2nd 3rd toe cutaneous syndactyly.[1]

Ours is probably 3rd reported case with bilateral cutaneous toe only syndactyly and 2nd reported case in India.

For our patients, Hypotrichosis, light colored brittle hair was present in all cases, anodontia, hypodontia, delayed dentition and peg shaped teeth present in varying degrees, Palmer Keratoses was present in all patients, all patients had absent or decreased sweating and heat intolerance.

## CONCLUSION

Our cases are submitted here to understand that Ectodermal Dysplasia is mostly a clinical diagnosis supported by Gene Mutation studies. EDSS1 (NECTIN 4 mutation) is presented for its rarity and to best of our knowledge it is a Novel mutation, so ours is probably second reported case in India. Also, it is observed that patients with EDA mutation have more severe features of disease than patient with EDSS1 mutation.


## ACKNOWLEDGEMENT

The authors thank the patients and their parents for giving consent to publish their case details and photos.


## DECLARATION OF CONFLICTING INTERESTS

The authors have no conflicts of interests.

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## CASE REPORT

# Localized Lupus Profundus

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**KEY WORDS :** Lupus profundus, Lupus panniculitis, Autoimmune diseases

**ABSTRACT**

Lupus profundus is the inflammation of dermis and subcutaneous fat. It is characterized by tender subcutaneous nodules and plaques mainly involving face, upper arm, hips and trunk. This patient is a 34 year old female presented to OPD with a two erythematous to skin coloured atrophic plaques over lateral aspect of both arms for last six months. Patient had no other oral or systemic involvement. Blood investigations including ANA profile were negative. Dermoscopy and skin biopsy from lesion was suggestive of lupus profundus. Patient was treated with oral and topical , intralesional corticosteroid and hydroxychloroquine 400 mg daily. Patient had improvement in lesion after 1 month. Lupus profundus which is an inflammatory disease which is very rarely presented variant of lupus erythematosus. It can present with other autoimmune diseases so regular follow up is required to check for development of systemic manifestations.

## INTRODUCTION

Lupus profundus is also known as lupus panniculitis. Lupus profundus is the inflammation of dermis and subcutaneous fat. It can present as a separate entity or can be present with lupus erythematosus. Clinical characteristics of lupus profundus were first described by Kaposi in 1869.[1] In 1956, It was described as lupus panniculitis by Arnold.[2]Lupus profundus is characterised by tender subcutaneous nodules and plaques mainly involvingface, upper arm, hips and trunk.

## CASE REPORT

A 34 year old female patient came to OPD with a two erythematous to skincoloured atrophic plaques over lateral aspect of both armsfor last six months. The plaque over left forearm was approximately 7*5 cm2 with tender hidebound skin with erythema, scaling and telangiectasia, while the plaque over right forearm was approximately 4*5 cm2 with hard and atrophic skin. Patient had no history of photosensitivity, oral ulceration, arthralgia, neuropsychiatric manifestations or trauma. Family history was negative. Dermoscopy of lesion showed scaling, dilated linear blood vessels and red and white structureless areas.

Blood investigations including complete blood cell count, differential cell count, ESR, Liver function tests, Renal function tests, serum electrolytes were normal. X-ray chest was normal. ANA Profile was negative. Skin biopsy showed lobular panniculitis with intense lymphocytic infiltration with perivascular infiltration, vacuolar degeneration ofbasement membrane. Patient was diagnosed as having localized lupus profundus.

Patient was started on oral corticosteroids and tab hydroxychloroquine 400 mg daily with intralesional

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triamcinolone acetateand topical potent corticosteroid and tacrolimus 0.1% cream. Patient had significant improvement in symptoms after 1 month of treatment. After treatment telangiectasia was decreased, skin became softer and patient had relief in pain.

**Figure: 1A and 1B – Photographs of left and right arm before treatment**

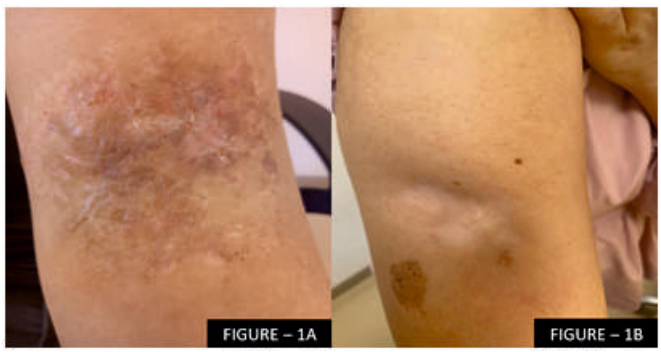
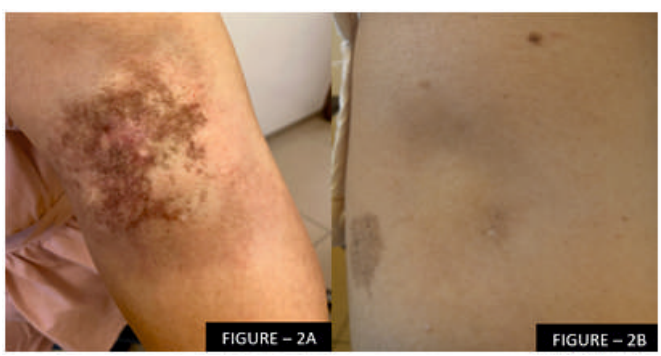



**Figure: 2A and 2B – Photographs of left and right arm after treatment**




## DISCUSSION

Lupus profundus is an inflammatory disease involving dermis and subcutaneous fat, usually seen in patients of cutaneous lupus erythematosus. It usually occurs in adult female patients with median age of 30 to 40 years.[3] It can also present in children with neonatal lupus. The pathogenesis includes interplay between ultraviolet irradiation, autoantibody generation, and dysregulation of T cells, dendritic cells, and other immune cells. Deficiency of complements caused dysregulation of immune complexes and causing disease.[4] Lupus profundus is characterised by deep, tender, subcutaneous nodules and plaques. Overlying skin may show changes like scaling, follicular plugging, dyspigmentation, telangiectasias, atrophy, scarring and tethered appearance. It can occur prior to other manifestations of LE and in the absence of other autoimmune connective tissue diseases. Histopathology shows lobular lymphocytic panniculitis with an infiltration of the fat lobules with lymphocytes, histiocytes, plasma cellsand hyaline fat necrosis. It also shows thickening of the basement membrane, mucin deposition, calcification, and vascular changes such as lymphocytic vasculitis, fibrin thrombosis, and perivascular fibrosis.[5] Laboratory analysis may show presence of anti-nuclear antibodies, lymphopenia, anemia, decreased C4 levels, and positive rheumatoid factor. The differential diagnosis includes dermatomyositis, scleroderma, lipodystrophies, inflammatory diseases of subcutaneous fat such as erythema nodosum, erythema induratum of Bazin, subcutaneous panniculitis-like T-cell lymphoma and traumatic fat necrosis, sarcoidosis.[6]

Treatment includes antimalarials like hydroxychloroquine, systemic steroids in initial treatment, topical potent corticosteroid, intralesional steroids. Other systemic therapies include dapsone, mycophenolate mofetil, cyclophosphamide, thalidomide, and IVIg, biologicals and autologous fat transfer or dermal filler for atrophy.[7]

## CONCLUSION

Lupus profundus is a very rare variant of lupus erythematosus. This case is being reported in view of its localized involvement in the absence of any systemic manifestations and absence of any other autoimmune disease. Regular follow up is required to observe the development of any other skin and systemic manifestations.

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**CASE REPORT**

# Case report of Endometrial Rejuvenation in a nulligravida woman with Thin Endometrium

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**KEY WORDS :** Endometrial Rejuvenation, Nulligravida Woman, Thin Endometrium

## INTRODUCTION

Endometrial receptivity is critical for implantation success in IVF. In women with a history of uterine surgeries or intrauterine adhesions, endometrial development can be compromised. Asherman's Syndrome, characterized by partial or complete obliteration of the uterine cavity due to adhesions, is a common cause of poor endometrial response and implantation failure. Traditional estrogen therapy may not be sufficient. Platelet-rich plasma (PRP), rich in cytokines and growth factors, has emerged as a regenerative treatment promoting endometrial repair and angiogenesis.

**Patient Profile**

- **Female Age:** 33 years
- **Male Partner Age:** 38 years
- **Type of Infertility:** Primary
- **Marital Life Duration:** 13 years
- **Menstrual History:** Scanty flow
- **Obstetric History:** Nulligravida
- **Surgical History:**
  - o Diagnostic Laparoscopy (2019): Revealed **bilateral tubal block**

The patient wascounselled at multiple centers for surrogacy due to **poor endometrial lining and intrauterine adhesions**. She presented to our clinic seeking one final attempt at achieving biological motherhood. After counselling, the couple consented for IVF post regenerative endometrial therapy.

- o Diagnostic Hysteroscopy at our centre: Confirmed **Asherman's Syndrome. Endometrium Pale. Adhesiolysis done. SubendometrialPRP instillation done in anterior wall, posterior wall & both lateral walls with hysteroscopic needle.**
- o **Procedure:**
  - **-** Subcutaneous G – CSF administration was done2 days before hysteroscopy
  - **-** 60 ml blood aspiration from peripheral vein on the day of hysteroscopy
  - **-** Five doses of Sub - Endometrial administration of PRP prepared
  - **-** First 3 Dosages: Same day Sub–endometrial administration of PRP was done on all four walls of the uterine cavity under hysteroscopic guidance
  - **-** Rest 2 doses were frozen for further during endometrial preparation for embryo transfer.

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**IVF and Laboratory Outcome**

Ovarian stimulation was done using Antagonist protocol that resulted in:

- **10 oocytes retrieved**
- **07 oocytes** at metaphase II
- **3 embryos** developed to **blastocyst stage and were frozen**:
  - o 2 blastocysts **(2*4AA)** & 1 blastocyst **(2AA)**

Endometrial Preparation for Embryo Transfer:

- Hormone Replacement Therapy with Estrogen support was started from 2nd Day of menstrual cycle.
- Fourth Dosage of Frozen PRP was thawed and instilled in to uterine cavity using IUI catheter on day 9th of menstrual cycle.
- Endometrial thickness was 5 mm on 14th day of menstrual cycle.
- Progesterone support added according to blastocyst transfer.
- Fifth Dosage of frozen PRP was thawed and instilled into the uterine cavity using IUI catheter2 days before ET.

**Embryo Transfer and Outcome**

- Embryo transfer was performed.
- Two blastocysts (2 x 4AA) were transferred.
- **β-hCG was positive** 14 days post-transfer (3626 mIU/L).
- Ultrasound confirmed a **singleton intrauterine pregnancy and Cardiac activity was seen in subsequent follow – up.**

## DISCUSSION

Asherman's Syndrome, can drastically reduce fertility potential by impeding endometrial regeneration and receptivity. Platelets and multiple growth factors (e.g., PDGF, TGF-β, VEGF), plays a pivotal role in tissue healing and neovascularization. In our patient, where conventional treatments failed, Endometrial Rejuvenation therapy enabled optimal endometrial growth and subsequent successful implantation.

This case also emphasizes the importance of not prematurely ruling out biological pregnancy in patients with complex uterine histories. With a combination of advanced IVF laboratory support and regenerative medicine, we were able to facilitate a positive outcome.

## CONCLUSION

Endometrial rejuvenation therapy is a promising adjunct in patients with Asherman's Syndrome and poor endometrial development.Even in cases of severe endometrial compromise like Asherman's Syndrome, **therapy can restore receptivity** and enable self-pregnancy. **Surrogacy is not the only option—regeneration is possible.**

CASE REPORT

# Synovial Chondromatosis – A Rare Cause of Elbow Pain and Stiffness

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**KEY WORDS :** Synovial Chondromatosis, Elbow Arthroscopy, Elbow Stiffness, Soft Tissue Tumour.

**ABSTRACT**

The elbow is prone to stiffness due to its unique anatomy and capsular reaction to inflammation. The resulting movement impairment can significantly interfere with a patient's activities of daily living. Usual causes of stiffness include Trauma (including surgery for trauma), posttraumatic arthritis, and heterotopic ossification (HO). Synovial chondromatosis is an extremely rare cause of elbow stiffness and there are only case reports in literature. Early diagnosis is critical in preventing long term complications. Open and arthroscopic arthrolysis are the primary surgical options. This paper reports a rare case, and early outcome following elbow arthroscopy.

## INTRODUCTION

Elbow stiffness can be a debilitating condition that significantly impacts a patient's ability to perform activities of daily living. The elbow joint is particularly susceptible to stiffness due to its highly congruent bony anatomy, relatively confined joint space, tightly stabilizing collateral ligament complex, and the close relationship of the surrounding muscles acting as secondary stabilizers[1]. Morrey *et al.* reported that most of our daily activities could be accomplished within a range of 30°–100° of elbow flexion and 50% of pronation or supination[2]. While trauma, surgery, systemic or posttraumatic arthritis, and heterotopic ossification (HO) are among the most common causes of stiffness, Other rarer causes like infection and tumours must be considered. We report an extremely rare cause of elbow pain and stiffness due to synovial chondromatosis.

## CASE REPORT

A 19-year-old boy presented with complaint of progressive pain and stiffness in his elbow in the last 18 months. He denied any history of trauma or constitutional symptoms like weight loss, fatigue or fever. Multiple Orthopaedic Surgeons had seen the patient and treated in the form of physiotherapy, painkillers and antibiotics. Clinical examination showed restricted movements with 50-80 degrees flexion-extension with inability to reach mouth due to limited flexion. Pronation was full and supination was restricted to 50 degrees. Overlying skin was normal and no neurovascular deficit was seen. X-rays were normal.

Patient was advised for an MRI scan that showed increased signals in the joint fluid suggestive of synovitis of possibly infective aetiology (Fig 1, 2). However, all his blood reports were normal and there was no history of fever. Extended arthritis profile was also normal. Hence, elbow arthroscopic biopsy and synovectomy was planned.

## SURGICAL TECHNIQUE

As described by Brach et al[3,4], patient was put under GA. Tourniquet was applied and he was positioned

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in lateral decubitus position with the affected extremity up. Medial and lateral epicondyle were marked out and position of the ulnar nerve was marked[5] (Fig 3). Anteromedial portal was established as a viewing portal and anterolateral portal as a working portal. Surprisingly there was no excess fluid in the joint but a soft and friable shiny layer was seen lining the whole joint with occasional loose piece (Fig 4). A 4mm shaver was introduced and carefully some of the lining was shaved out. The particles were examined in vivo and noted to have a waxy consistency. Patiently, the whole anterior compartment synovium was shaved until the joint surfaces were clearly visible (Fig 5). Anterior capsule was released from the humerus. Posterolateral and direct posterior portals were then established to debride the posterior compartment. Now the waxy pieces were noted to be loosening up and were carefully removed in blocks of 1-2 cm size. After debridement was completed all the deposits were put together and estimated to be around 8 sq cm in size (Fig 6). At the end, full range of movements was achieved on table. All the portals were closed using 3-0 ethilon. The tissue was sent for histology and genexpert testing to rule out TB.

**Fig 1**

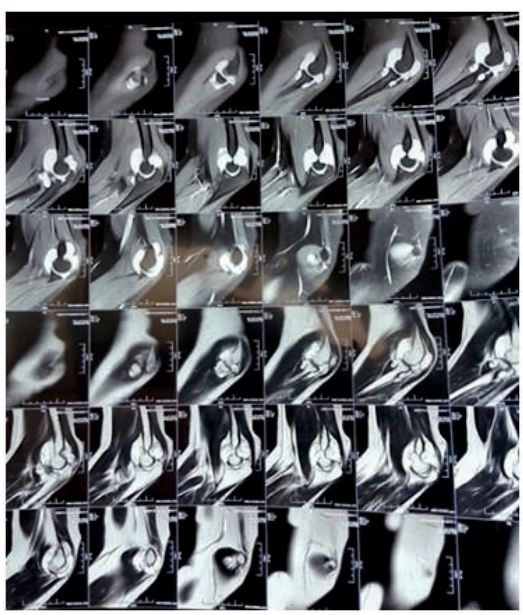


**Fig 2**

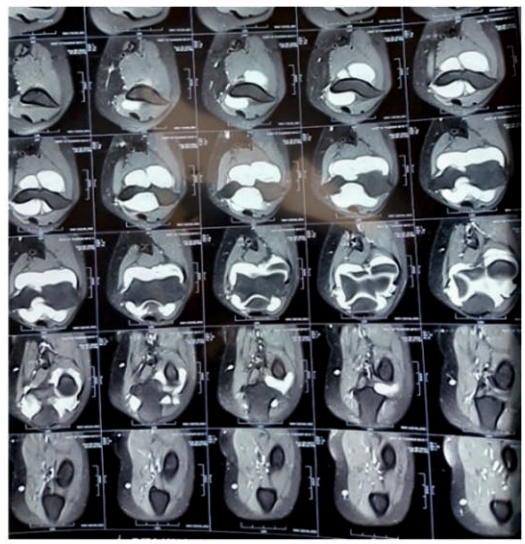


**Fig 3**

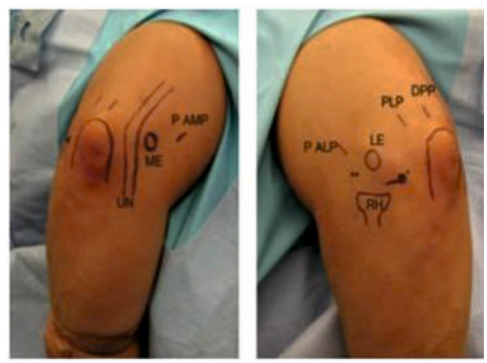



**Fig 4**

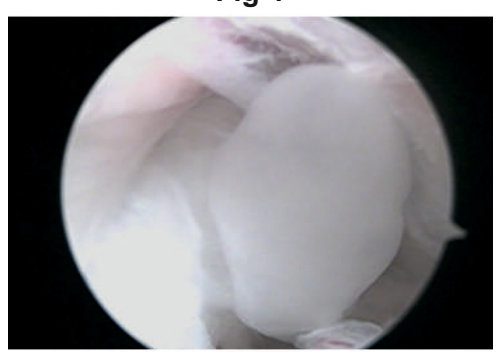


**Fig 5**

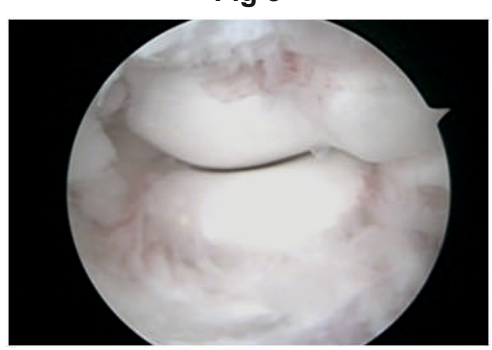
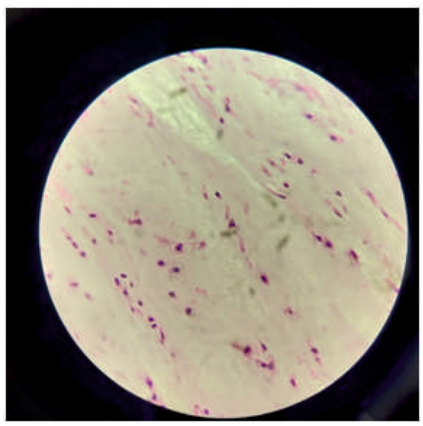


**Fig 6**

**Fig 7**



## DISCUSSION

Synovial chondromatosis is an extremely rare soft tissue tumour. It has a prevalence of less than 1:100000 and knee is the commonest joint to be affected[6]. The aetiology remains uncertain but FGF-2 and TGF-β3 are being investigated for its role[6]. Generally, it presents as multiple loose bodies in the joint but in this particular case it was a uniform sheet of hyaline cartilage type tissue as seen during arthroscopy. If not treated early, it can cause irreversible damage to cartilage and joints, and can even be transformed into synovial chondrosarcoma.

Surgery can be done both open or arthroscopically. However, due to proliferation of abnormal tissue seen all around the elbow joint and uncertainty of diagnosis, it was decided to approach arthroscopically as otherwise the joint exposure would need extensive dissection both anterior and posteriorly, and subsequent risk of postop stiffness and Heterotopic ossification. Elbow arthroscopy is challenging compared to other jointsas the joint space is small, and there is a risk of damage to ulnar or median nerves if care is not taken.It also has a higher learning curve compared to other joints. This patient was started on physiotherapy immediately after surgery and night splinting was done to maintain the extension. At 3 month follow up, his swelling and pain had completely settled, and range improved to 20-100 degree flexion/extension and full pronation – supination. He is able to all activities of daily living. The patient needs to be constantly motivated to continue stretching exercises and be counselled about risk of persisting movement deficit despite surgery.

The histology in this case showed features typical of synovial chondromatosis (Fig 7) and genexpert was negative for TB. At 6 months there is no recurrence but there is a reported recurrence rate of upto 25% after synovial chondromatosis[5] and so regular follow up is essential in the long term along with yearly MRI scans.

## CONCLUSION

Elbow swelling and stiffness is quite common in orthopaedic practice. Clinician must be aware to the possibility of rarer causes like synovial chondromatosis after common causes have been ruled out. The longer an elbow remains stiff, the more difficult it is to retain movements even after surgical release. Patient should be explained about the possible risk of recurrence as well as some degree of persisting stiffness in such cases.

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**ARTICLE**

# New Innovations in IVF:

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## 1. Electronic Witnessing System : Safer and Smarter Fertility Treatments

IVF is a delicate and deeply personal journey—and when it comes to handling eggs, sperm, and embryos, accuracy and safety are absolutely essential. To make this process safer, faster, and more reliable, one of the latest innovations being adopted in modern fertility centers is the Electronic Witnessing System.

**What exactly is a witnessing system and why is it a game changer in fertility treatments?**

This system is a smart technology that uses barcodes, tablets, and scanners to make sure every step in the IVF lab is done correctly—without any mix-ups. It replaces the old method of having a second person manually double-check details and brings in a more advanced, automated, and error-proof system.

In a fertility center, there are many critical steps where eggs, sperm, and embryos are handled. Even a small error—like mislabelling—can have serious consequences. This is where Witnessing system steps in. It ensures that every item, from the start of treatment to embryo transfer, is correctly identified and tracked using barcodes.

It even alerts embryologists before a mistake can happen, such as during freezing, thawing, or before even starting ovum pick up if the patient's name doesn't match. That's a big leap forward in safety.

**How does it work?**

Each patient's samples (like eggs or embryos) are given special cards & barcode labels.

Embryologists use handheld devices or tablets to scan and confirm the correct items before every step.

It even keeps a record of who did what, when, and where, giving a complete history of the treatment.

There's no need for a second staff member to manually check, saving time and reducing workload.

**How is it beneficial for patients?**

**No more manual errors**: The system prevents mistakes by alerting embryologists instantly.

**Saves time**: Embryologists can focus more on patient care instead of paperwork.

**Improved data and insights**: The system gives real-time updates and useful reports to improve clinic performance.

**Regulatory ready**: Automatically meets many quality and legal standards.

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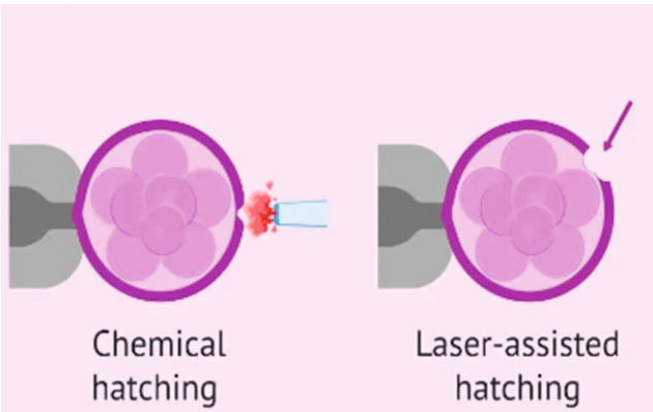
## IVF SAFETY, ONE STEP AT A TIME

At a time when patients are trusting us with their most precious hopes, Witnessing systems are helping raise the bar in care and accountability. With this innovation, patients can feel reassured knowing that technology is working silently in the background—keeping everything safe, secure and on track.

### 2. Laser Assisted Hatching – Boost to Embryo Implantation




The process of Assisted Hatching (AH) has evolved remarkably over the past two decades, emerging as a vital tool in improving implantation outcomes, particularly in select subgroups of patients undergoing IVF.

In natural conception, the embryo hatches from its surrounding protein coat—the zona pellucida (ZP)—before it can implant into the endometrial lining. However, in some IVF cycles, especially in patients with repeated implantation failures, advanced maternal age, or when embryos exhibit a thickened or hardened zona, this natural hatching may not occur effectively, reducing the likelihood of implantation.

Traditionally, assisted hatching was performed manually using mechanical or chemical techniques. These methods, though pioneering, were technically demanding and carried an inherent risk of trauma to the embryo due to variability in operator skill and lack of precision.

Laser-assisted hatching (LAH) uses a precisely focused infrared laser beam to create a small opening in the zona pellucida. This procedure is fast, consistent, and significantly reduces the risk of mechanical or chemical injury to the embryo.

At our center, the implementation of LAH has enabled embryologists to execute the procedure with controlled depth and timing, thereby improving safety and reproducibility. The laser system is non-contact and integrated into the micromanipulation workstation, ensuring embryo handling is minimized.

Clinical Indications for Laser-Assisted Hatching

- While not routinely applied to all embryos, LAH is particularly beneficial in certain groups, including:
- Women above 35 years of age, where zona hardening is more common
- Patients with repeated IVF failures, where implantation has previously not occurred despite good-quality embryos
- Frozen-thawed embryos, which may have a tougher zona due to the cryopreservation process
- Embryos with a visibly thickened zona pellucida
- Use in PGT (Preimplantation Genetic Testing), where biopsy access is improved by prior zona opening

## IMPACT ON IVF OUTCOMES

While studies continue to evaluate the broader applicability of LAH, several trials and meta-analyses have shown improved clinical pregnancy rates, especially in the above-mentioned patient populations. Importantly, LAH has not been associated with an increased risk of congenital anomalies, underscoring its safety when performed in experienced hands.

In practice, we ensure that LAH is performed only on embryos that meet the clinical criteria and on Day 3 or Day 5 embryos, depending on the developmental stage and treatment plan.

Patient counseling is essential, as is proper documentation and quality control during the procedure.

**ARTICLE**

# Treat to Target – A new paradigm shift in the treatment of Rheumatological Diseases

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**KEY WORDS :** Rheumatological Diseases

The management of rheumatological diseases has undergone a revolutionary transformation inrecent years. One of the most significant advancements is the adoption of the "Treat to Target". (T2T) strategy — a structured, goal-oriented approach that emphasises on regular monitoring andadjustment of treatment based on predefined clinical targets. This paradigm shift marks a departurefrom traditional symptom-based management to a more proactive, personalised, and outcome-driven methodology.

## What is "Treat to Target"?

"Treat to Target" is a clinical strategy in which therapy is adjusted at regular intervals to achieve andmaintain a clearly defined treatment goal — typically **remission** or **low disease activity**. Originating in diabetes and cardiovascular care, this approach has now been widely adopted in**rheumatology**, particularly for diseases like **rheumatoid arthritis (RA), psoriatic arthritis (PsA),axial spondyloarthritis (axSpA)**, and **systemic lupus erythematosus (SLE)**. The success of this model is intertwined with the development of **novel targeted therapies** andbiologics that allow precise modulation of disease pathways.

## Key Principles of the T2T Approach

1. **Define the Target**: The ultimate goal is either **remission** or **low disease activity**, depending on the disease andpatient-specific factors.
2. **Regular Monitoring**: Disease activity is assessed at frequent, pre-specified intervals using validated tools (e.g., DAS28, CDAI, BASDAI).
3. **Timely Adjustment of Therapy**: If the target is not achieved, treatment is escalated or modified promptly.
4. **Shared Decision-Making**: Patients are actively involved in setting treatment goals, enhancing adherence andsatisfaction.
5. **Individualized Care**: The strategy is adapted based on co morbidities, age, patient preference, and risk factors.

## Why Now? The Role of Newer Drugs in Enabling T2T

Traditional **conventional synthetic DMARDs (csDMARDs)** like methotrexate, sulfasalazine, andleflunomide laid the foundation, but the true leap came with **biologicb DMARDs)** and **targetedsynthetic DMARDs (tsDMARDs)** that precisely block disease pathways.

Below is a disease-wise overview of the **newer drugs** that support the T2T strategy.

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### 1. Rheumatoid Arthritis (RA)

RA is the prototype disease where T2T has shown profound benefits. Studies like the **TICORA** and **COMET** trials demonstrated that a tight control and goal-oriented approach leads to:

- Higher rates of remission
- Improved functional outcomes
- Reduced radiographic progression
- Better quality of life

**Newer Therapies Enabling T2T in RA:**

| Drug Class | Examples | Target |
|---|---|---|
| **TNF Inhibitors** | Adalimumab, Etanercept, Golimumab,Certolizumab | TNF-α cytokine |
| **IL-6 Inhibitors** | **Tocilizumab**, **Sarilumab** | IL-6 receptor |
| **Costimulation Blockers** | **Abatacept** | CD80/86–CD28interaction |
| **B Cell Depleters** | **Rituximab** | CD20+ B cells |
| **JAK Inhibitors (OraltsDMARDs)** | **Tofacitinib, Baricitinib, Upadacitinib,Filgotinib** | JAK-STAT pathway |

These agents allow precise tuning of immune responses, often leading to remission within 3–6months when monitored and adjusted as per T2T protocols.

### 2. Psoriatic Arthritis (PsA)

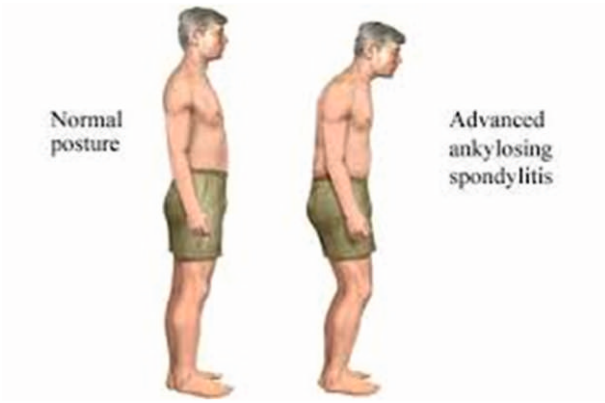
Figure 1. Dactylitis of the digits is highly predictive of psoriatic arthritis.

PsA is heterogeneous, affecting joints, skin, entheses, and axial skeleton. T2T here requires multi-domain control. The **TICOPA trial** established that early, aggressive treatment using a T2T strategysignificantly improves joint and skin outcomes, although at the cost of more frequent adverse eventsdue to intensified treatment.

**Newer Therapies Enabling T2T in PsA:**

| Drug Class | Examples | Target |
|---|---|---|
| **TNF Inhibitors** | Adalimumab, Infliximab | TNF-α |
| **IL-17 Inhibitors** | **Secukinumab, Ixekizumab** | IL-17A |
| **IL-12/23 & IL-23Inhibitors** | **Ustekinumab, Guselkumab** | IL-12/23 or IL-23 |
| **PDE4 Inhibitors** | **Apremilast** | PDE4 enzyme |
| **JAK Inhibitors** | **Tofacitinib, Deucravacitinib** | JAK/STAT or TYK2 pathways |

### 3. Axial Spondyloarthritis (axSpA)

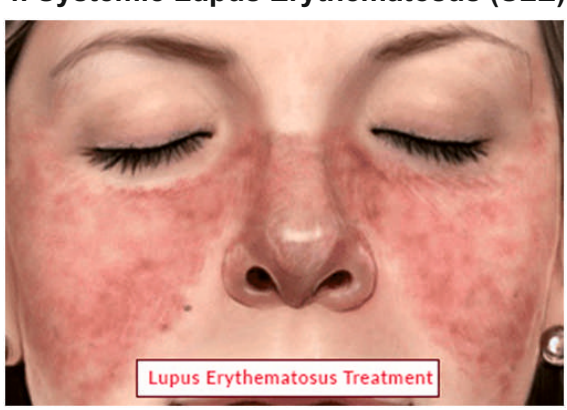



AxSpA, including ankylosing spondylitis, benefits from early T2T-based intervention to preventstructural damage. While defining targets like remission is more complex in axSpA, indices such as **ASDAS** help guide therapy. Early T2T adoption is associated with better symptom control and reduced structural damage.

**Newer Therapies:**

| Drug Class | Examples | Target |
|---|---|---|
| **TNF Inhibitors** | Etanercept, Infliximab | TNF-α |
| **IL-17Inhibitors** | **Secukinumab, Ixekizumab** | IL-17A |
| **JAK Inhibitors** | **Upadacitinib** | JAK-1 |

### 4. Systemic Lupus Erythematosus (SLE)




Although complex, the T2T approach in SLE is evolving, with emerging consensus aroundachieving **low disease activity (LLDAS)** or **remission**, thereby reducing flares, damage accrual,and minimizing steroid use.

**Newer Drugs Supporting T2T in SLE:**

| Drug Class | Examples | Target |
|---|---|---|
| **BLyS Inhibitors** | **Belimumab** | B-lymphocyte stimulator |
| **Type I IFN Inhibitors** | **Anifrolumab** | Type 1 interferon receptor |
| **CalcineurinInhibitors** | **Voclosporin** | T-cell activation |

**Advantages of the T2T Strategy**

- **Improved Clinical Outcomes**: Higher remission and response rates.
- **Reduced Long-term Complications**: Less joint damage and disability.
- **Patient Empowerment**: Increased participation and satisfaction.
- **Efficient Resource Utilization**: Early control may reduce hospitalizations and surgeries.

**Advantages of Newer Drugs in T2T:**

- **Faster onset of action** (JAK inhibitors, IL-17 blockers)
- **Oral options** available (e.g., Tofacitinib, Apremilast)
- **Targeted action** ! fewer systemic side effects
- **Better domain control** (e.g., skin + joint in PsA)

**Challenges and Limitations**

- **Resource Intensive**: Requires regular visits, labs, and monitoring.
- **Variable Access to Biologics**: Economic and systemic barriers may impede aggressive therapy.
- **Heterogeneity of Diseases**: Not all rheumatologic conditions have clearly defined targets.
- **Patient Diversity**: Comorbidities, pregnancy, or age may necessitate flexible targets.

**Challenges in Implementing T2T with Newer Drugs**

- **Cost and Access**: Many advanced therapies are expensive and not widely accessible in low-resource settings.
- **Monitoring Requirements**: Regular assessments using validated scores can be time-intensive.
- **Safety Concerns**: Long-term immunosuppression risks (e.g., infections, malignancies).
- **Patient Hesitancy**: Fear of escalation or injection-based therapies.

**The Future of T2T in Rheumatology**

With the advent of **biologics**, **targeted synthetic DMARDs**, and **precision medicine**, T2T is poisedto become even more personalised. Future directions include:

• Incorporating **biomarkers and imaging** for target assessment.

• Use of **digital health tools** for remote monitoring.

• Enhanced **multidisciplinary care models** to support T2T execution.

## CONCLUSION

The "Treat to Target" strategy has fundamentally changed the landscape of rheumatology. Coupledwith **next-generation biologics and small molecules**, it allows rheumatologists to aim for **deepremission**, **sustained quality of life**, and **halting disease progression**. While challenges in cost andaccess remain, the future promises **personalized, goal-directed care** for every rheumatologypatient.

**KEY MESSAGE** *"Treat to Target, empowered by cutting-edge therapies, is not just a strategy — it is the newstandard of excellence in rheumatologic diseases.*

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