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Editorial board

From the Editors' Desk

Dear Reader,

It gives us immense joy to place the first issue of the newyearin your hands!

The roughest of storms are followed by a placid calm. We wish that the COVID pandemic which taught the healthcare fraternity tough lessons make way to better weather & greener pastures.We also take this opportunity to thank all our patrons for the immense support we have garnered over the years.

In this first edition of this year we revisit tuberculosis an archaic nemesis which presents in uncommon forms. We have introduced 'master thought' with key physicians in the Apollo ecosystem, who share their vast experience from avantage point of view in their respective fields. We have included 2 articles which delve into the finer aspects healthcare management. The first article sheds light on lean management in healthcare & the other probes into 'error disclosure' in healthcare which is very much voiced in hushed tones in our country. We expect our literary nuances to grow as the year passes & gravitate towards biochemistry ballads, serology sonnets & musings from microbiology in the forthcoming issues.

Finally we have covered FAQ's on genetic counselingwhich has become an integral part of laboratory medicine as well in this issue. We thank the contributors for taking time to put pen to paper & covering a gamut of topics with aplomb. AD express has gained considerable impetus by the way of your contributions & we welcome more from you all.

We humbly request you to share your feedback on 'AD express' & we assure you that feedback from you will make each next issue ever more interesting !

Wish you all a fabulous new year ahead!

Best regards,

Dr. Marquess Raj M.D. Dip. N.B (Path), MBA (HM)
ZTC – TN & Pondicherry
marquess.raj@apollodiagnostics.in

Dr. Shalini Singh M.D (Path), IFCAP, DMSC (Endocrine)
Lab Director, Global reference lab, Hyderabad
shalini.singh@apolloclinic.com

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Contents

1. Pancytopenia in military tuberculosis

Dr.V.Kalyan Chakravarthy
ZTC – Andhra Pradesh & Lab head RRL,Vijayawada

An 87 year old woman presented with a complaint of weight loss since 2 months. Blood work up was requested -

History of Present Illness:

- H/O loss of appetite since 3 months
- H/O onset of swelling on right side of neck 2 months back which is associated with discharge of pus and followed by fever for 2 days .
- Fever is intermittent, at night and associated with chills and rigors.
- c/o cough with expectoration +

History of Past Illness:

- Mantoux: positive, patient was started on ATT and stopped after 10 days.
- No H/O DM/ HTN

Obstetric History:

- $G_3L_3A_0$
- Attained menopause 40 years back. No H/O AUB.
- **VITALS:** Normal
- **CVS**- S1+ S2+
- **RS**- B/L AE+, NVBS
- **CNS** - NAD

Radiological investigations

- **USG Neck:**
- Mildly prominent lymph node in level IV supraclavicular region measuring 11 x 5.8cm noted on right side.
- **USG ABDOMEN & PELVIS:**
- Grade-I fatty liver.

Hematological investigations :

Hb- 7.5 gm%, Total WBC count - 2,850/QL, N- 50%, L- 42%, E- 4%, M-4%, ESR- 44 mm/hr, PCV- 22.7 %

Platelet- 70,000/QL, TRBC- 2.13 million/cu mm,

MCV - 106.8 fL, MCH - 35.4 pg, MCHC - 33.1 gm/dl, RDW-CV – 18%

Peripheral smear examination:

RBC morphology: - Normocytic Normochromic (few microcytes, macrocytes & ovalocytes are seen)

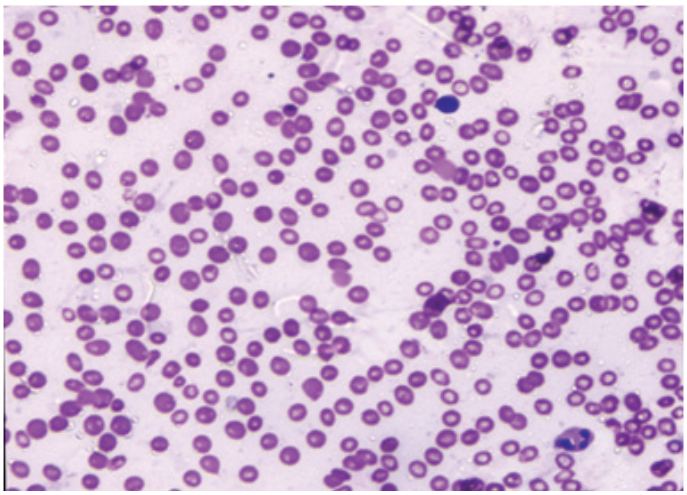
WBC morphology: - Decreased in number

Platelets: - Decreased in number, Hemoparasites - Absent

Impression: – Pancytopenia for evaluation

Reticulocyte count: – 4.5 %

Photomicrograph of peripheral smear is given below: -



LFT	RFT	CUE
• Total Bilirubin – 0.5 mg% • Direct Bilirubin- 0.2 mg% • Indirect Bilirubin – 0.3 mg% • ALP – 89 IU/L • SGOT – 46 IU/L • SGPT- 30 IU/L • Total protein- 7.3 gm% • Albumin – 3.8 gm% • Globulin – 3.5 gm% • A/G ratio – 1.0 : 1	• S.Creatinine – 0.8 mg/dl • S.Urea – 31 mg/dl • S. Ferritin – 123 ng/ml • Vit B12- 532.9pg/ml • **Thyroid function tests** • TSH – 4.51 QIU/ml • Free T3 – 2.32 pg/ml • Free T4 – 0.86 ng/dl	• pH - 1.020 • Specific gravity - 6.0 • RBCs – ++ • WBCs – 20/HPF • Squamous epithelial cells - 4 / HPF • Bacteria - ++/ HPF • Blood - +++ • Protein - ++ • Leukocytes - ++ • Stool occult blood - negative

Bone marrow aspiration

Bone marrow aspiration was advised to rule out primary hematological malignancy in view of the patient's history.

- **Site:** Posterior superior iliac spine
- **Cellularity:** Hypercellular
- **Adequacy:** Adequate
- **Marrow to fat ratio:** 60:40

Photomicrograph of bone marrow aspirate is given below: -

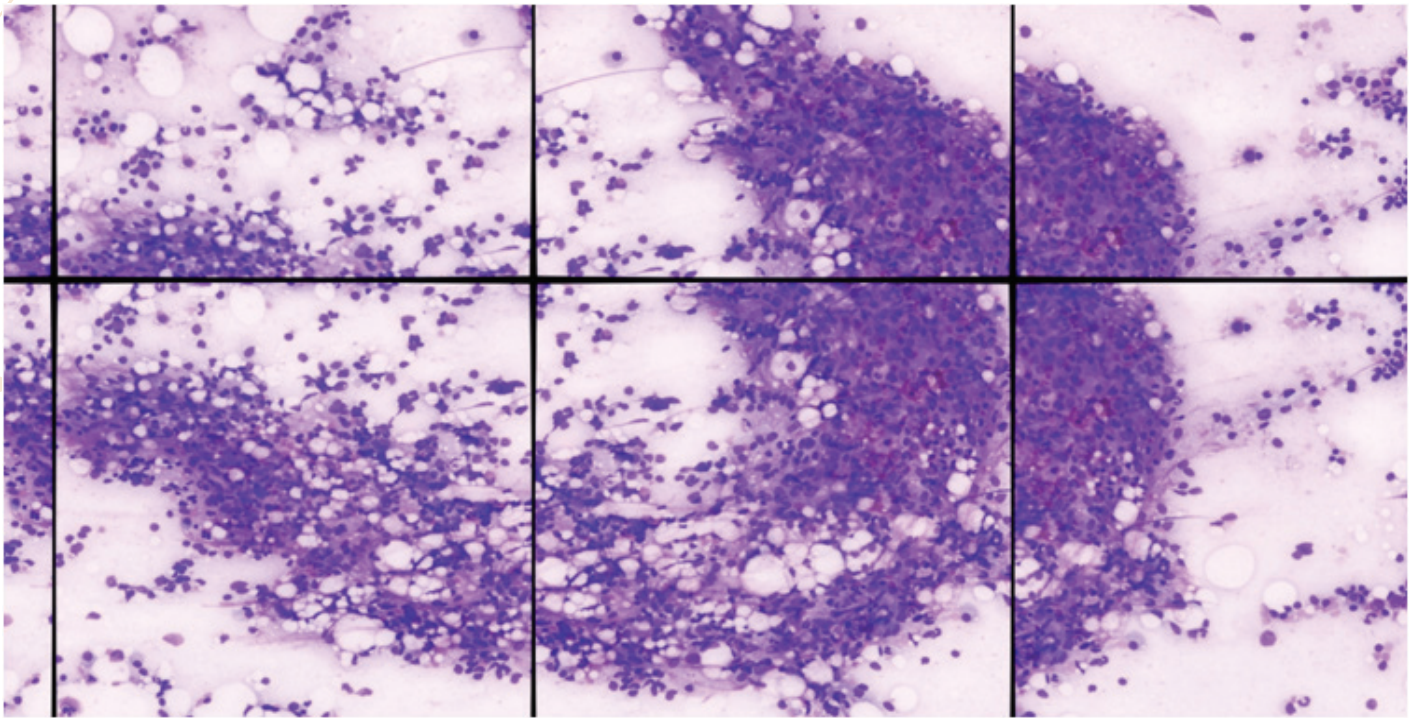


Fig. A. Leishman stained bone marrow aspiration smear under low power magnification

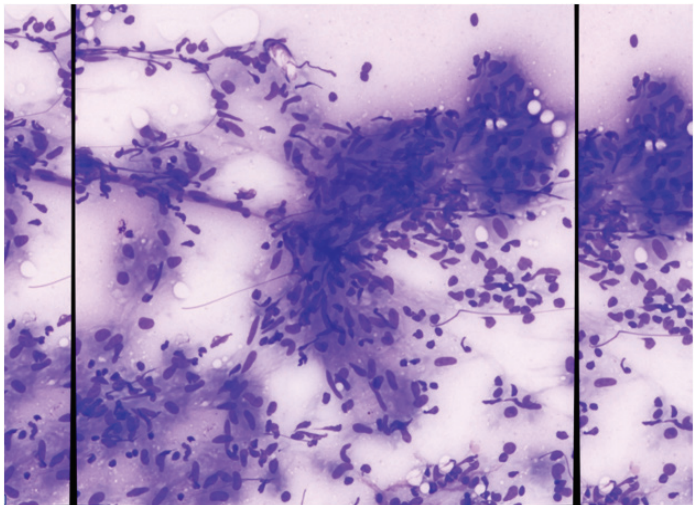


Fig. B. Leishman-stained bone marrow aspiration smear under high power magnification showing epithelioid cell granulomas

Impression

- Features are suggestive of Hypercellular marrow with Erythroid hyperplasia with megaloblastic maturation and Epithelioid Granulomas.

Note: In correlation with previous history Tuberculous etiology is to be considered

Case discussion

- Overwhelming infection can produce pancytopenia, and therefore is not immediately distinguishable on clinical grounds from aplastic anaemia in which sepsis has developed as a consequence of inadequate neutrophil production.
- Bone marrow examination is capable of differentiating between these two conditions, as the cellularity is greater in the former, even though the more mature cells of the neutrophil series tend to be depleted.

Conclusion

- In miliary tuberculosis, leucocytosis and leucopenia both can occur. There may be either thrombocytosis or thrombocytopenia. Pancytopenia can occur but is uncommon.
- In mycobacterial infections, bone marrow is usually hypercellular but a markedly hypocellular bone marrow can also occurs
- There may be dyserythropoiesis
- In miliary tuberculosis, majority have granulomas and half of them have caseation
- Disseminated tuberculosis is a less fulminant cause of pancytopenia, and is overlooked unless bone marrow is subjected to ZN staining and culture for mycobacteria.

2. NIPT: The Boon to Low Risk Pregnancies Shielding from Infection

Eswari Dodagatta-Marri
Molecular biologist, GRL, Hyderabad

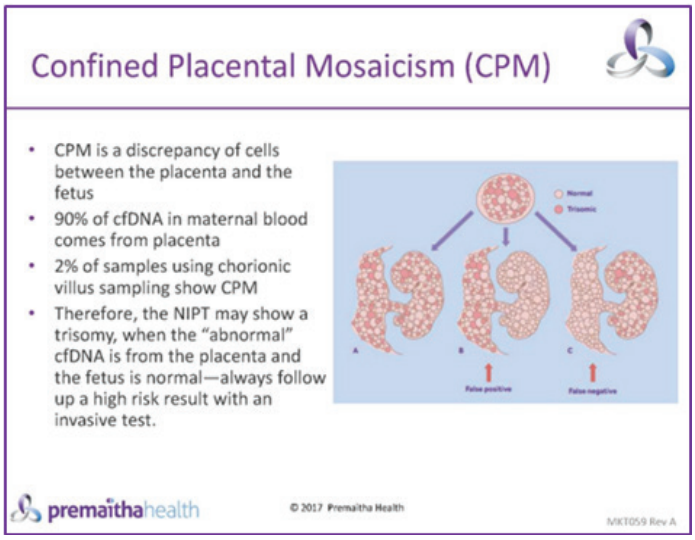
Cell-free DNA (cfDNA)-based, Non-invasive Prenatal Testing (NIPT) also known as non-invasive prenatal screening (NIPS), utilizing next generation sequencing (NGS) is a highly sensitive and specific approach designed to screen fetal aneuploidy. This screening test requires less invasive procedures on the pregnant woman as it is based on cell-free fetal DNA (cfDNA) in the maternal blood and has no associated risk for miscarriage. Small cfDNA provides screening for trisomy 21 (Down's syndrome), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome) and sex chromosome aneuploidies (45, X-Turner syndrome, 47, XXX-triple X, 47, XXY-Klinefelter Syndrome) [1] and Jacob's syndrome (XYY). It can also be used to screen for additional chromosomal disorders of fetal microdeletions, duplications and single-gene diseases such as DiGeorge syndrome (22q11 deletion) [2], Duchenne muscular dystrophy [3] and autosomal recessive nonsyndromic hearing loss [4].

About one of every 150 live births has chromosomal abnormalities, the most common being Down syndrome and the incidents increase by maternal age. The first trimester screening (FTS) between 11+0 weeks and also 13+6 weeks of gestation has become the basis for decision-making about further diagnostic and therapeutic concepts in early pregnancy [5]. Combined FTS is based on maternal age, fetal nuchal translucency (NT) and two maternal serum parameters; free β-hCG (human chorionic gonadotrophin) and PAPPA-A (pregnancy-associated plasma protein A) leading to false positive rate of 5%. Clinical implementation of NIPT is a remarkable advanced screening for fetal aneuploidies with a false positive rate of 0.35% and is a widely accepted routine. NIPT with its significantly improved screening performance for all types of trisomy while combined with FTS, consisting of a detailed fetal ultrasound examination together with maternal serum biochemistry could be considered as gold standard. NIPT being a screening method and not a confirmatory test, it is advisable to correlate the data and when positive results are obtained, always recommended for further investigations like ChorionicVillus sampling, Amniocentesis.

Why NIPT is a Screening Test?

The test analyses the small fragments of cffDNA that are circulating in a pregnant woman's blood, which is used to determine the risk of certain genetic abnormalities. These fragments are free floating unlike most DNA which are inside a cell's nucleus, hence called cell-free DNA. These small fragments (~200 base pairs) are formed during the cell lysis and released into the bloodstream along with other contents. Mother's bloodstream consists of placental along with hers as the placental tissue in the uterus links the fetus and the mother's blood supply which is shed in the bloodstream (mix of cfDNA). The test is screening test as it can only give the estimation of the risk of having certain genetic condition (has decreased or increased) but not a definitive answer. Discordant NIPT results could be because of the following reasons;

1. Blood transfusion and organ transplant
2. Low fetal fraction
3. DNA sample contamination
4. Vanished twin
6. Mosaicism (See Figure below)

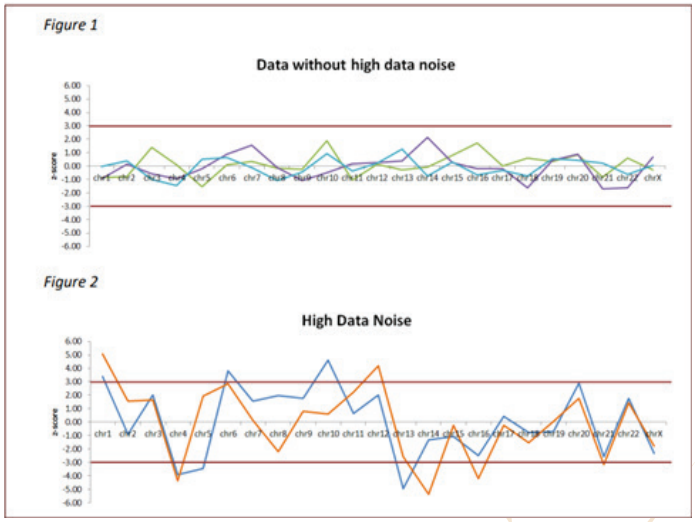


7. Other reasons: Maternal Copy Number Variation (CNV), Supernumerary ring, Single gene disorder, Maternal malignancy, inflammation may cause false positive results.

cffDNA in the mother's bloodstream known as fetal fraction should be above 4% (sufficient enough) to be able to identify fetal abnormalities, which is generally obtained around the tenth week of pregnancy. Unaffected fetus indicating abnormality is false positive report and vice versa is a false negative report because of the involvement of the mother's (cfDNA) genetic conditions. Low fetal fractions can lead to inability to perform the test or concluded as false negative result. Both fetal and maternal cfDNA fragments count gives the percentage of cfDNA fragments from each chromosome. If the percentage of cfDNA fragments from a particular chromosome is more than expected, then the fetus has an increased likelihood having a trisomy condition. A positive screening result indicates further investigations.

What is Z-Score and how does it work?

Z-score indicates the difference between the given values or the standard deviations (of that particular chromosome) to that of mean measured (control diploid samples) by performing the Gaussian distribution. The results of NIPT are based on the Z-score values which are calculated based on the comparison of the control group (diploids) samples. Individual Pregnant mother's sample is compared to a relative surplus or shortfall for that particular chromosome compared to normal diploid samples. Z-score between -3 and +3 are expected with the statistical assumptions from 99.7% plasma samples from pregnant women in the second trimester (10+ Weeks). The more the deviation from the zero for the Z-score, the more the more the sample deviates from the control sample indicating aneuploidies and thus the reliability of NIPT. (Figure1 and Figure2)



The accuracy of the results depends on various factors like sample preparation, fetal fraction, reference samples chosen and sequencing method.

NIPT for aneuploidies is not a diagnostic test at present and so positive NIPT test still requires invasive testing to confirm. Because of high accuracy of NIPT, fewer women will be able to go through the process of confirmatory invasive tests once there is any high risk for genetic variation

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3. Master thoughts (much better than after thought) - Diabetes

Dr. Harpal Singh DGM & Head
Medical Services, Apollo Sugar Clinics

All those who are overweight/having high BMI should be tested for Diabetes. Also those, who are having family history of diabetes. Apart from above two important reasons for getting tested for Diabetes, all having a stressful occupation, physically less active (less than 3 times a week), having Non-Alcoholic Liver Disease should be tested for Diabetes. Females having hormonal disorder like PCOS & those with history of Gestational Diabetes, should be regularly tested for Diabetes. Since we Indians are prone to Diabetes, all Corporate Employees above the of 30yrs, especially those with desktop jobs and long sitting hours, having sedentary lifestyle should be tested for Diabetes. All hypertensives & suffering from dyslipidaemias should be regularly tested for Diabetes.

1. What are my goals regarding blood Glucose levels?

Fasting Blood Sugar levels should be less than 100mg/dl. Post prandial Blood Sugar levels should be less than 140mg/dl and HbA1c should be less than 5.7%.

2. What tests would you recommend for diagnosing Diabetes?

OGTT is gold standard for Pre-Diabetes. And HbA1c is Gold Standard test for Diabetes.

But we can rely on Fasting Blood Sugar, Post Prandial Blood Sugar and HbA1c for diagnosing Diabetes. Even Random Blood Sugar levels clubbed with HbA1c – can assist/support in diagnosing Diabetes.

3. What test results tell me if I have diabetes or Prediabetes?

If your HbA1c values are in range of 5.7% - 6.4%, you are Prediabetic. And if your HbA1c is more than 6.5% or higher in two separate incidents, you are Diabetic.

Similarly, if your fasting blood sugar is in between -100 mg/dl to 125 mg/dl, you are pre-diabetic. Similarly, if your fasting blood sugar is more than 125 mg/dl in two occasions, you are Diabetic. OGTT values in range of 140 -199mg/dl are considered Prediabetes & OGTT values of more than 200mg/dl are signifying Diabetes.

4. What is the gold standard for diabetes diagnosis?

Majority of bodies consider HbA1c as Gold Standard for Diabetes diagnosis. Many bodies also consider OGTT (Oral Glucose Tolerance Test) as Gold Standard especially in Pre-diabetes & Gestational Diabetes.

5. What are diagnostic criteria?

If your values are more than – normal range, in two separate tests/ incidents e.g., more than 6.5% of HbA1c and more than 125mg/dl of fasting blood sugar values and more than 140mg/dl of post prandial values in two separate tests/incidents or OGTT values of more than 200mg/dl in two separate tests/ incidents – its considered Diabetes

6. Which tests help to know what kind of diabetes I Have?

For further differentiating it into Type I Diabetes of MODY, we need to C -Peptide & GAD.

Please do consult your Endocrinologist/ Diabetologist for same.

7. How quickly can HbA1c change?

It all depends on diet, exercise & medication. 1% HbA1c change is noticed in 2weeks in various studies. But if HbA1c values are very high, it takes up to 12wks to bring it to baseline with help of diet, exercise and medications.

8. How do other factors such as high cholesterol and high blood pressure affect me if I have diabetes?

Of course, they do. Dyslipidaemia or Hypertension along with Diabetes increase your chances of Cardio Vascular Diseases, Stroke, Nephropathy etc. multiple times. So, its advisable to get your BP, Lipid profile and Renal Function Tests done periodically and keep all

parameters under control by diet, exercise, weight control & medication, if required.

9. Should I stop eating sugar altogether?

If possible, yes. You can switch over natural source of sweetness – like fruits.

Guava, papaya, apple are very healthy choices being a Diabetic.

Apart from Sugar, its more about Carbohydrate intake in general. One needs to reduce Carbohydrates, if Diabetic. Stop drinking aerated sugary drinks & sugary juices. Avoid trans-fat & junk foods.

10. Do I need to take Insulin?

It depends on control & blood sugar values. Taking Insulin should not be considered a taboo.

Adding Insulin to your medication/management regime, can help you getting the blood sugar levels managed & we can avoid many complications. But all medications & Insulin shall be taken on advice/consultation of an Endocrinologist, a Diabetologist or a Physician.

11. Can I reverse Diabetes?

This is main topic discussed on social media. Many new start-ups have mushroomed & taking advantage of this. As per medical fraternity & various research studies – remission of Diabetes is definitely possible. This has been seen practically by us as well. Reversal is not possible in majority of cases. So, one has to be very careful. While in pre-diabetes, if one works on his or her diet, weight control and exercise. The progression to full blown Diabetes can be delayed or prevented.

12. How can I lower my blood glucose?

Blood Sugar can be lowered by diet, exercise & medication. A disciplined life, with home cooked balanced diet at regular intervals definitely helps. Portion control is definitely helpful. Regular consultation with Physician & Dietician is definitely helpful. Also regular check-ups are advisable.

13. What lifestyle modifications do I need to make?

Diet is first and foremost lifestyle modification, one need to focus on. Exercise & regular walks (30mins/day) is very helpful. One should adopt at least one sport and involve in some hobbies like gardening, music etc. Sleeping at a particular time & a sound sleep of 6hr-8hr is very important. Yoga & meditation is also very important lifestyle modification, which can make a huge difference.

14. What long term complications can diabetes cause?

Uncontrolled Diabetes can lead to Cardio-Vascular Diseases, Neuropathy, Nephropathy, Retinopathy, Peripheral Vascular Diseases, Podiatric problems, Gum & Teeth problems, Skin problems, Mental health issues and even amputation of feet, blindness & stroke etc. Considering complications of Diabetes, we shall manage Diabetes with all possible means including Diet & Exercise (lifestyle modification), managing Stress & Medication (including OHA & even Insulin) etc.

4. Lean management in healthcare

Dr. Niranjan Naik
ZTC – Maharashtra

Foreword

If we perform an Internet search on lean healthcare, within 0.29 sec there are 9,403,000 hits.Healthcare organizations have seen their customers shop for services, with access to information at their fingertips. Customers today seek out treatment and provider options and soon will be able to seek out services that offer the most value. Information is also starting to be available and understandable. Today, we live in a competitive economy. The market drives the selling price as customers seek to find the best value at the lowest cost. The value of our healthcare in service, quality, and cost is being compared to healthcare around the world; therefore, the global market, in turn, will impact the customer's (patient's) value proposition. With healthcare reform and increasing healthcare costs always on the horizon, it appears that the only way to be effective in this competitive environment will be for organizations to embrace 'lean'in the near future.

So what is lean?

Lean is a term that originated in manufacturing to describe a way of managing enterprises in the most efficient way. Becoming Lean requires the continual pursuit to identify and eliminate waste and establish efficient work flow within the organization. The term "value stream" is used today to describe what is involved in producing a product or service from raw material, manufacturing, distributing, wholesaling, and retailing to recycling. Even though healthcare and service organizations may not begin with raw material or even manufacture a product, they still have value streams related to providing healthcare services to their customers.Lean concepts were originally developed around manufacturing, but lean principles have now been adapted to meet the needs of healthcare.

"Lean" is not just a scientifically tested process but a philosophy and way of thinking. Lean has its roots in the United States, but what we know today as Lean was taken to another level and given a new dimension by Toyota. Lean, if implemented properly, is an enterprise-wide initiative that requires a cultural change. It will not be successful if it is directed only at the frontline staff. This means if Lean or Lean/Sigma (the combination of Lean and Six Sigma) is to be truly successful it must include all functional areas (i.e., finance, marketing, information systems, etc.), all levels, from the board of directors to the patient, and all value streams within the enterprise. Lean principles are based on what is known today as the Toyota Production System.

Typical Lean results, when given sufficient time to work, can result in:

- 20–80% productivity improvement
- 50–90% reduction in inventory
- 50–99% throughput time reduction
- 30–50% reduction in space requirements
- 10–30% reduction in overheads

Lean and healthcare organisations

Healthcare organisations are not factories. Factories deal with products while healthcare organisations deal with people. While our products in factories cannot talk to us, patients do talk to us before, during, and after they go through our processes. Organizations and processes within the healthcare environment are not all that different from organizations within factories.

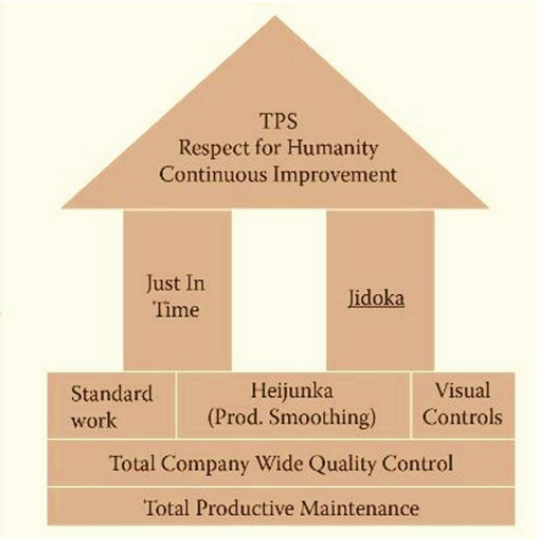
What is a lean business delivery system?

The goal of the lean Enterprise is to supply the best value to the customer, at the right time, with the highest quality at the lowest cost. This means creating a culture of continuous improvement and an environment where everyone in the organization participates in eliminating waste and streamlining processes in order to supply the best value to the customer. Best value means meeting or exceeding the customer's expectations for delivery and service for both

product quality and customer-desired quality before and after their encounter. A company's primary mission is not solely to satisfy its customer, but exceed customer's expectation and promote customer loyalty, while making a profit to stay in the reckoning.

Understanding the value of the lean business delivery system

The Lean Business Delivery model begins with understanding customer expectations. It is difficult, if not impossible, to achieve or exceed customer expectations if we don't have a good understanding of what the customer views as value-added. If there is waste in the process then there will always be opportunities for variability and inconsistency thus leading to opportunities for customer dissatisfaction. We find healthcare organizations don't routinely survey "customer value." Today's healthcare customer does not necessarily pay fully or directly for the majority of care received. At a minimum, the customer would expect a positive outcome (high quality, error or mistake-free), in a courteous (caring) environment, and in a timely manner. This is rapidly changing as the consumer is becoming more attuned to healthcare reform and may be directly paying for a higher percentage of his or her healthcare costs to one that is more value-based.



Toyota House

Just in time (JIT): The first pillar of the Toyota production system model

Companies need to be more responsive to customers, provide what the customer orders, in the quantity and quality the customer orders, at just the right time that the customer wants it. This is called Just in Time. JIT is one of the two pillars of the Toyota system. The goal behind JIT is to use the minimum amount of inventory, equipment, time, labour, and space necessary to deliver the product JUST IN TIME TO THE CUSTOMER!

In Healthcare it means "Giving the Right patient, the Right care, the Right way, while providing a Great Patient Experience." It includes providing the staff with the necessary and proper tools and supplies in the right locations, when they need them, and in the right amount with shortest travel distances within efficient layouts.

JIDOKA—THE SECOND PILLAR OF THE TOYOTA PRODUCTION SYSTEM

The closest Engish translation of Jidokais 'autonomation' or automating with a human touch. Creating "smart machines" or machines that can stop themselves if they make a mistake is a central principle to the Lean system. The goal is to prevent the error so that a defect does not result. With Jidoka, machines are designed to detect a defect prior to passing it on to the next process. Machines should check items before they work on them and after they work on them.

Another idea associated with Jidokais mistake proofing or "poka yoke," meaning never pass on a bad part. In order to mistake proof or foolproof processes, we need a way to take the humans out of the equation. Mistakes and errors not only cause human tragedy, but also add expense to an already overburdened healthcare system.

LEAN IS A JOURNEY

You will know you are further down the Lean culture path when building a culture of continuous improvement every day outweighs the insistence on implementing only those perceived large ROI projects first. Thousands of small improvements turn into large overall returns. Sometimes, a project with a lower ROI can lead to a project with a higher ROI. Many times, jumping to the highest ROI project is not the best strategy.

In summary, although there may be some differences between manufacturing and providing patient care, the Lean Business Delivery system has been shown to have application within healthcare organizations, will continue to grow as more organizations realize the benefits of practicing Lean.

5. Error disclosure in healthcare

Dr. Marquess Raj
ZTC Tamil Nadu & Pondicherry & Dr. Abhik Banerjee ZTC West Bengal & North East

Prologue:

The essential nature of science is to quest to find the truth. Successive additions to a stock of tested truths are where scientific progress lies. The practice of medicine is essentially conformation to medical science. But is there really a way to put this to test in every given situation be it a lab test or a patient given situation.Or Is there a mechanism to confirm that the best is being done in every clinical situation given the best of resources. Albeit as giant strides are being made in medicine & with the application of QMS in every crevice in health care,does medicine stand on the high pulpit of infallibility?

Error disclosure in healthcare.

The truth has been misunderstood, difficult to deal with & is elusive at times. In the process of seeking the truth science has perpetually run into several stumbling blocks ranging from cassock clad clergymen to conceited persona from the scientific community itself. For instance, the reticence Virchow to accept the germ theory of disease & fervent rejection of the laws of the universe by the catholic church bear testimony to the, 'I/We can't be wrong' attitude.These are instances where in credibility is based on face value & assumption but not inductive justification.

To subscribe to a practice which might seem perfectly reasonable now, or to conform to what is the norm now need not be essentially correct. Many a time a valid change in medicine comes after being faced with stiff opposition. Ignac Semmelweis being labeled a maniac when he proposed that hand washing could save women from puerperal sepsis is but one example of empirical evidence throttling scientific reason.

In an era where we have unlimited access to data & with our computational capabilities ever stronger than in any time in human history, the tendency to rationalize everything thing exists. The very nature to delve into logical explanations for everything ranges from probing the reason for a lab result becoming suddenly abnormal or a patient suddenly lapsing to coma is itself a fallacy. In many ways loosely accessible information about healthcare has given birth to the 'google' patient & has but complicated the way patients are being handled & responded to. Omnipresent data forces upon health care professionals a tendency to avoid trouble & be defensive before treating or reporting. This also applies to personnel who don the 'technical' or 'quality' robes. With an ever increasing need to do everything right the first time around & burgeoning expectations from the patient,practitioners of medicine are forced to view every patient as a potential pitfall.

We also should bear in mind that we are perennially offered with the opportunity of being healthy & have never had such limitless access to healthcare in medical history. However attaining the goal of hale & hearty cornucopia should but fill us with doubt rather than mirth. We constantly are faced with the tumult & uncertainty pertaining to what we know & what we don't know. The following situations can be agonizingly delicate especially when they are related to expectations of healthcare & truth disclosure.

Given these four situations in the patient – practitioner (lab medicine included) relationship, the first scenario is ideal with due consideration to patient rights. But it is worth noting that the possibility of the other 3 situations existing cannot be dismissed from the outcome in a clinical narrative. Whilst we practice evidence based medicine how can we be sure of the diagnosis/es,or the truth behind the values we get in the best of laboratories after applying the best of QC practices.

Moreover the truth holds true only for 'what' questions & not for 'why' questions.

The chance of ensuring infallibility relies on processes. However it is known that the best of processes can fail (like all security checks don't evade bomb explosions, plane crashes,etc). The same applies to healthcare as well. Clinical excellence does not translate to healthy bliss. This does not undermine the importance of keeping a robust process in place.Focus on that tiny fraction of error that we could have sneaked past person or process could also be avoided. Like an inadequately mixing a sample or not cleaning a puddle leading to a fall.Hence the onus is on preventive action that could have checked the error from happening in the first place.

Holier than thou!

Lab medicine has come a far way from being a branch which housed live rabbits a century ago. Current laboratory practice in the organized sector is incomplete without the application of QMS,QA processes & stringent accreditation guidelines. Hospital accreditation guidelines when compared with lab (ISO 15189) guidelines are but copiously watered versions. For instance the yardstick applied for the comparison of the quality of radiology reporting when compared with the evidence required for documenting histopathology reporting quality would put a wry smile on the face of most lab practitioners.

Even more silhouetted are the mechanisms of gauging competency in the closeted air conditioned recesses of healthcare. Evidence for the same in places such as operating theatre,delivery rooms,ICUsetc. are not in par with the lab medicine. When confronted by consent forms in dull print & litigable language in times of vicissitude, patients can only but hope on the scrub clad saviors. The high reputation & the many accolades that a clinician might carry along with the documented evidence need not bear much resemblance to what transpires as clinical outcome.

Epilogue:

All perceptions are a mix of the accurate & inaccurate. Though we practice science what we are faced with in practice at first is perception of a situation. The same could be a bleeding artery or a hemolyzed blood sample or a value reported by an analyzer.There is a tiny fraction of error that creeps even when the best of processes are in place. The choice of error disclosure & expert concealment lie in our very hands. Moreover the subject of error disclosure is faintly touched upon as it has a bearing on individual,as well as organizational reputation. Practitioners of healthcare walk a thin line & are often faced by the classic Hamlet situation.

6. FAQs regarding Genetic counselling

Dr. Vasavi Narayanan
Consultant Geneticist, Department of Cytogenetics, AHLL

1. What is the importance of a genetic counsellor in a clinical set –up?

With largely evolving genomic science and genetic technologies, a genetic counsellor is the much sought-after professional in various clinical specialities, The counsellor helps communicate effectively with the patient after identifying their relevant needs, helps them with Information pertaining to empathic understanding of their condition, testing requirements, coping strategies and reaching an informed decision with respect to the health of the person/s involved.

2. What areas of medical practice can take help from the presence of a genetic counsellor?

Most areas of medical science can benefit from the services of a GC. To mention a few, couples seeking help in case of infertility or repeated abortions can understand the appropriate reproductive options after relevant tests are performed to have a successful pregnancy. Those with affected children from previous pregnancies can be made to understand their risks for future pregnancies and strategies to avoid such anomalies. Unmarried couples with familial syndromes and other diseases can take the help of a genetic counsellor not only to identity potential risk of disease genes in their offspring but also communicate the same effectively with the family members involved.

3. What is the role of counselling in cancer syndromes?

Pre- and post-test genetic counselling will be useful especially in hereditary cancer syndromes to evaluate required tests for assessing risk and have due follow-up for the affected member as well as others in the family. The counsellor does this in sync with the oncologists and the surgeons, to also help in making important surgical and treatment decisions leading to a desirable outcome.

4. Much of the prenatal screening is done by obstetricians, would genetic counselling help?

In a lot of prenatal set-ups, plan for screening is done by the obstetricians with little time to explain potential outcome of the screening (NIPS) process to the patients. In these cases, it would help to have trained geneticist and counsellor render their expertise to pre-test counselling followed by post-test counselling where required, more so in cases with abnormal findings to help families make informed decisions on the further testing and pregnancy options.

5. By delivering genetic counselling, is there an increase in the uptake of genetic testing services?

There is no doubt that genetic counselling has been linked with genetic testing services for patient care. Now, there is a greater increase in the technologies for testing available and patients can avail of testing facilities via direct to consumer testing. However, it must be understood that testing may not always be the solution to a patient's query – it is the counsellor's role to help the patient understand whether a genetic test will help their scenario, and what is the likelihood of getting to a solution with respect to their health queries.

6. What kind of support tools would enhance working of a genetic counsellor?

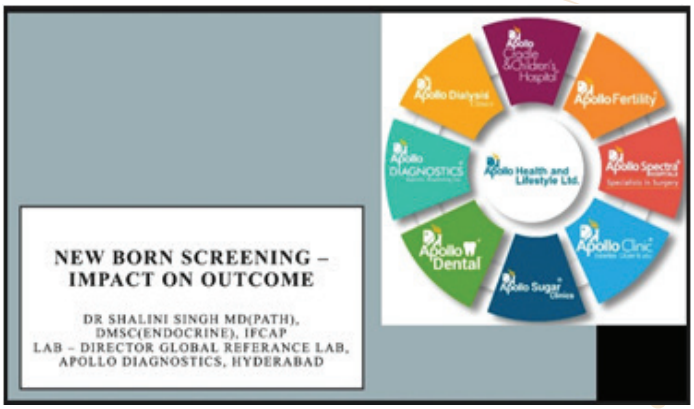
Equipping the GC with good educational tools to communicate the condition of a patient with them and their families in addition to maintaining good records of the cases and counselling reports for future follow-up, will help build a trusting relationship with patients and delivering satisfactorily to patient care.

Recent Events:

Webinar: Utility of the RCP Guidelines for reporting Breast FNACs.

As per the Apollo Diagnostics policy of Continuing Medical Education, two webinars were arranged in December and attended by many Doctors

Webinar: The guidelines issues by RCP 2016 for Breast Cytopathology: Dr. Marquess Raj



Webinar: New Born Screening- Impact of Outcome: Dr. Shalini Singh

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