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AD Express

DECEMBER 2022

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From the Editors' Desk

It gives us immense joy to place the December issue of AD Express in your hands! We thank you profusely for the continuous support & the feedback from all of you.

While we gingerly await an immanent declaration by the WHO on the COVID pandemic, we thank the clinical as well as the laboratory fraternity for keeping the patient care ship afloat.

In this edition of AD express, we have taken care to cover the commonplace as well as the 'cutting edge' aspects of laboratory medicine. We thank the contributors for taking time to put pen to paper & covering a gamut of topics with aplomb. In this issue we 'revisit diabetes' from a practicing biochemist's point of view. We have an article shedding light on the importance of clinicians divulging relevant clinical history so that it contributes to positive patient outcomes.

This will be the first edition in which an interview between a lab medicine practitioner & a physician titled 'interview with AFP (Apollo family physician) is being introduced. Our AFP's view disease prevalence in the community from a vantage point & it is important that their views are circulated.

We are ever committed to our contribution to advanced diagnostics & an article on novel PCR techniques has been covered. In addition to a common benign tumour occurring in an uncommon location being showcased as a case report, the importance & significance of reporting 'giant platelet predominance' also features in our fifth issue.

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

Wish you all a Merry Christmas & a great year ahead!

Best regards,

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Contents

1. Peculiar location of leiomyoma – a case study of 2 cases

Dr. Abhik Banerjee, Dr. Debajyoti Singha Roy, Dr. Zishan Akhtar

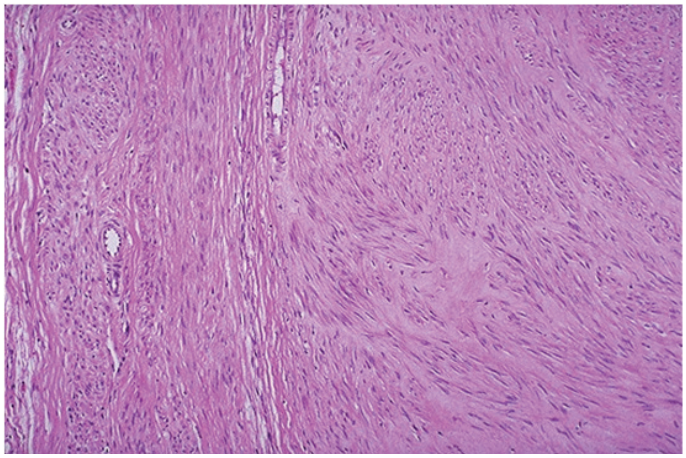
Introduction

A **leiomyoma**, also known as a **fibroid**, is a benign smooth muscle tumour that very rarely becomes cancer. They can occur in any organ, but the most common forms occur in the uterus and small bowel. It literally means smooth muscle tumour. Here we present 2 case reports of Leiomyoma presenting in unusual locations.

Case – 1

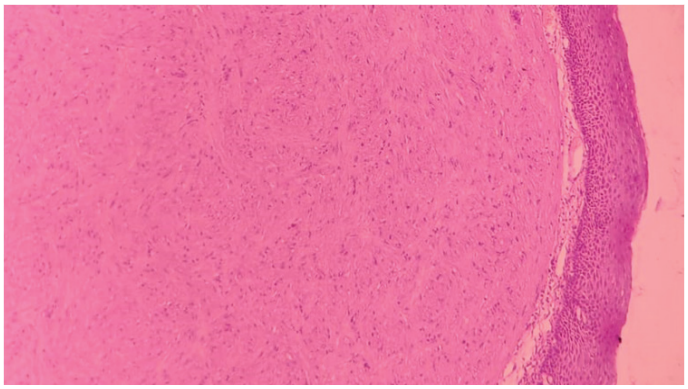
A 35 year old male presented with complaints of testicular mass, upon examination the mass was found to be separate from the testes and subsequently the mass was excised and sent for Histopathologic examination. On microscopy, the mass consisted of intersecting fascicles of monotonous spindle cells with moderate eosinophilic cytoplasm and thick walled blood vessels.

The morphology was typical for a Leiomyoma, but considering the rarity of such a tumour in this location, a diagnosis of Benign spindle cell lesion – favouring Leiomyoma was made and IHC was advised.



Case – 2

A 42 year old female patient presented with complaints of gradual increase in difficulty in swelling, followed by relevant radiologic examinations and excision of the mass, the tissue was sent for histopathologic examination. On microscopy, the mass was lined by stratified squamous epithelium and underneath, a well-encapsulated mass consisting of intersecting fascicles of monotonous spindle cells with moderate eosinophilic cytoplasm and thick walled blood vessels. Thus, a diagnosis of Leiomyoma – Esophagus was made.



Discussion

Although Leiomyoma is a fairly common lesion encountered in specimens of female genital tract, Paratesticular leiomyomas are extremely uncommon tumours affecting the male genitalia. Most of such tumours run a benign course and are indolent showing a slow growth and are non-invasive. Timely diagnosis and appropriate treatment would largely preclude the need for orchidectomy in most of the patients.

Although leiomyoma is the most common benign esophageal tumour, malignant carcinoma is still 50 times more likely. Thus a timely diagnosis is essential to avoid unnecessary medical interventions that would be required in case of carcinomas.

Note – Both these cases were received in Apollo Diagnostics, as cases needing a second opinion (owing to the peculiarity of the locations).

2. Interview with Apollo family physician (AFP)

Dr. Yashod Reddy M

(MBBS, Dip diabetology,Fellowship in clinical cardiology) - AFP Chennai- Apollo Family physician, Chennai.

Interviewed by Dr. Marquess Raj, Co- editor of AD Express.

Apart from being an abbreviation for a certain tumour marker, AFP is well-known amongst the Apollo fraternity as Apollo family physician. The Apollo hospitals group was the first healthcare institution to introduce the 'family physician' concept in India. On an average every AFP is consulted by 15 to 10 patients per day & hence the AFP gets not a snapshot but an in depth understanding of communicable as well as non-communicable disease (NCD) prevalence on a community level.

It was a pleasure meeting Dr.Yashod Reddy M, who practices in Chennai & learn from his perspective. Excerpts from the interview are as follows -

Q: Why do you think there is an increase in the brunt of diabetes & hypertension in the urban population?

Although stress, sedentary lifestyle & unhealthy eating habits are common factors that lead to both diabetes & hypertension, the importance of quality sleep is often not taken into account. Adults require at least 8 hours of uninterrupted sleep, while children, infants &newborn require an even longer duration.

Another overlooked fact is that genetic mechanisms are often present in patients who present at a younger age.

Q: Can you explain this better?

It is important to elicit family history if any of the close relatives had a heart attack, stroke or cancer at less than 40 years of age. In such patients it is important to screen them earlier for the non-communicable diseases (NCD) such as heart attack, stroke & cancer.

In case a heart attack or stroke has occurred in the family of the patient in individuals less than 40 years of age, tests for lipoprotein –A, homocysteine, HsCRP & lipid profile have to be taken apart from the ECG, ECHO & the treadmill test.

Q: It is well known among the urban population that a high blood pressure can cause heart attack & stroke both. But are all hypertensives symptomatic?

No, not all patients with hypertension are symptomatic. In fact 90 % of patients are asymptomatic. It is also prudent to say that we should not conclude that a patient is hypertensive based on a single reading.

Symptoms such as giddiness, blurring of vision, early morning nausea & headache are commonly encountered.

Q: Then how do we confirm that an individual is hypertensive?

Ideally the blood pressure should be monitored for 24 hours with demonstration of normal 'nocturnal dipping.'

Q: What is nocturnal dipping?

The physiological decrease in nocturnal BP relative to daytime BP is referred to as 'nocturnal BP dipping'. A decrease of 10–20% in nocturnal BP relative to daytime BP is considered normal.

This is not the case in individuals who have hypertension. As discussed earlier inadequate sleep can have a detrimental effect as well.

Q: What is the recommended line of testing once a patient is diagnosed with hypertension?

It is important to diagnose whether the patient has primary or secondary hypertension. In patients less than 40 years if the systolic blood pressure is over 180 & the diastolic BP is over 100 in 3 successive readings then secondary causes such as endocrine causes, renal artery stenosis, coarctation of the aorta & adrenal pathology have to be ruled out.

Baseline investigations ECG, ECHO, thyroid function tests (TFT), routine urine examination, serum urea, creatinine & electrolytes are recommended. As hypertensive retinopathy can damage the eyes a fundus examination by an ophthalmologist is also suggested.

Q: How do you recommend that diabetic patients monitor their sugar levels?

It is important that patients monitor their blood glucose at least 2 to 3 times a week either using a capillary blood glucose monitoring device or in venous blood. Whereas the HbA1c can be monitored once in 3 months. The target should be to maintain a random blood sugar between 80 to 170 mg/dl & a Hba1C between 7 & 7.5%. Urine spot microalbumin or even a 24-hour urine test is a useful tool to assess if the kidneys are functioning properly in diabetics.

Q: When will you suspect a genetic link in a patient with diabetes?

A genetic basis such as MODY (Maturity onset diabetes of the young) can be suspected if the patient has uncontrolled hypoglycemia. The glucose tolerance test can help as an initial line of investigation. Fasting insulin & fasting c-peptide also help in ascertaining the cause.

Many a time presence of autoantibodies against GAD (Glutamic acid decarboxylase) help rule out Type 2 diabetes. There is a stronger genetic link present in individuals with type 1 diabetes.

Q: What is insulin resistance? In which type of diabetes is insulin resistance common.Can you elaborate on symptoms encountered apart from the usual 3 P's – polyuria, polydipsia &polyphagia?

Either hyperglycemia or hypoglycemia can happen when there is a mismatch between the endogenous insulin circulation& the blood glucose.

90 % of individuals with type 2 diabetes develop insulin resistance. Patients notice darkening of the skin in certain parts of the body or spots over the shins. One of these lesions is termed acanthosis nigricans. Many patients with type 2 diabetes also have centripetal obesity & grade-1 fatty liver.

Q: Do we see an increase in the number of patients presenting with fever in the monsoon season?

Urban malaria is well documented in medical literature. Dengue is also quite common & patients can present with bleeding sometimes. Scrub typhus is actually more common than we think.

Q: What are the first line investigations in a patient with fever?

Baseline investigations include CBC, CRP, Urine routine & blood culture, which will bring to light if the etiology is infectious. Specific antibodies or PCR can be suggested to rule out various pathogens depending on the symptoms. For example, a PCR test (H1N1 or COVID) is often the first line of investigation in a flu not responding to conventional treatment.

Q: Is there any general advice you give to all your patients?

Eat healthy, on time. Exerciseevery day. Enjoy responsibly are the 3 E's that I tell all my patients

3. Is human touch losing the battle when it comes to eliciting clinical history?

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

Foreword:

The digital era has ushered in technology which is considered to be more precise and trustworthy. The COVID pandemic has brought about a palpable change in patient experience as online consultations & tools to record clinical history through algorithm's that operate on AI were put to use on a never before scale. While it will be difficult for hospitals & health care providers to 'wean off' the patients from AI based technology, we as healthcare professionals definitely need to ponder if we are losing the human touch in key areas of healthcare delivery.

The importance of clinical history: -

A common assumption prevails among clinicians who believe that the laboratory medicine professional when given only a tiny fragment of a patient's tissue or a few millilitres of blood, have the entire foresight necessary to produce a statement of absolute truth at the end of his or her report. Sometimes, it is also presumed that any additional clinical history might prejudice the laboratory into producing a biased report. However, incomplete communication between the clinician and the pathologist may make diagnosis difficult or impossible.

When hospitals & health care professionals work on the lines of feeding history to a computer which uses artificial intelligence to give a consistent output, it is possible that we could lose vital information that could many a time come to light only by personal connect. Are we as health care professionals losing the -

Art of extracting history and its application?

Art to listen, win trust & communicate effectively between teams?

The health care system which comprises of many vital cogs might not be able to serve its purpose of working to the patient's benefit when it relies too much on processes that employ artificial intelligence. Hence interaction between the various personnel with a 'human touch' setting aside differences is vital for good patient outcomes.

The laboratory point of view:

A. Reducing process waste: The numbers of personnel & processes involved in the pre-analytical have grown by leaps and bounds. The number persons handling the sample have become more and more important than the patient to whom the sample belongs.

   Samples entering the laboratory without test request forms (TRF's) or with incomplete TRF's invite second time scrutiny. The process of identifying & rectifying errorsby connecting with the clinical team to gather vital information such as clinical history takes plenty of time & effort. Much of this 'process waste' can be avoided if relevant history is furnished for each & every test ordered.

   For example :When a patient or a clinician does not record reason for repeating a test such as platelet count, how does it matter if an instrument has given a report or if it has been given after interpretation of a pathologist.

   The role of the lab medicine expert is to put bits and pieces of clinical history together with the results & to arrive at a logical and scientifically acceptable report. Inaccurate or misunderstood clinical history can be equally problematic as having no clinical history at all. Following are the potential consequences of inadequate clinical information and correlation:

1. Inappropriate treatment or management
2. Inadequately specific diagnosis
3. Inappropriate specimen triage for ancillary studies
4. Delayed TAT
5. Delayed notification of significant or unexpected results to treating personnel
6. Diagnostic over-commitment based on sampling error
7. Misdiagnosis
8. Medico legal consequences
9. Misdiagnosis
10. Contradictory diagnoses in patient record.

B. Exchange between the lab & the clinical team:

We can definitely not belittle that fact that both the clinical team & the lab medicine expert gain significantly by exchanging ideas & information. People like me get professional satisfaction when we share our knowledge while interacting with our clinical peers. Also when a doctor approaches us for clarification in a report we refer to hallowed literature & finally when we give the doctor feedback, a sense of professional satisfaction prevails. In addition camaraderie is established between both the teams, much to the benefit of the patient.

Final thoughts:

"Those Who Do Not Know History are Destined to Repeat It" - Edmund Burke

One would expect a clinician's narrative case report to contain complete record of past events and circumstances that are or may have led to a patient's current state of health. A lab medicine expert's narrative report should have scientific evidence and explain events logically rather as a programmed monotone. Tools such as AI can be a welcome addition to process improvement but they have to be reviewed with rigor. The 'human touch' that involves interaction between the patient & the health care provider & interaction between health care personnel can never be supplanted.

4. Prediabetes revised - It's never too late to start

Dr. E. Maruthi Prasad PhD
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Introduction:

Diabetes is a serious health concern with an increasing prevalence. It can be a serious condition if the patient does not receive a timely diagnosis, and also increases the risk of developing type 2 diabetes, and heart disease/ stroke. Several research studies revealed that it can be reversed by modifying simple lifestyle changes. The present article discusses diabetes risk factors, diabetes diagnosis (HBA1c, fasting glucose, glucose tolerance test (GTT)), urinemicro albumin and simple preventive measures.

Background:

As per the American diabetes guidelines, a higher-than-normal range of blood glucose levels is prediabetes (100-125 mg/dL) (1). Having prediabetes means having blood sugar levels that are higher than normal, typically, this means that blood sugar is higher enough to progress to a type 2 diagnosis. The symptoms of prediabetes aren't always obvious, so it's important to have your blood sugar levels tested if you're at high risk for it. Diabetes and prediabetes complications can only be prevented by maintaining a normal glucose level (1). A person with prediabetes has a subclinical impairment in fasting plasma glucose levels or impaired glucose tolerance (2). Understanding the pathophysiology of diabetes may contribute to appropriate prevention for patients with prediabetes (2).

Risk factors:

Obese individuals can progress prediabetes to type 2 diabetes. Prediabetes checks are essential as prediabetes often has no symptoms, and one can have it for years and not even know it. Age (20 or older), high blood pressure or cholesterol, parental or hereditary with Type 2 diabetes, ethnicity, previous gestational diabetes, polycystic ovary syndrome, certain medications (steroids, antipsychotics, HIV medications), hormonal conditions (Cushing's syndrome and acromegaly), and sleep apnea are also considered as risks for diabetes. It is imperative to get your blood sugar& A1C checked regularly if you have risk factors for prediabetes.

Diagnosis:

The fasting blood glucose test (FBS) is used to measure blood sugar after not having eaten for at least 8 hours. This test is preliminary used to detect diabetes or prediabetes. Second, the glucose tolerance test (GTT) measures blood sugar after going at least eight hours without eating and two hours after drinking a glucose-containing drink. To determine how quickly glucose is cleared from the blood, a GTT is taken. This test detects diabetes, insulin resistance, impaired beta-cell function, reactive hypoglycemia, and acromegaly, among other disorders (4). Third, the random plasma glucose test is used to investigate blood sugar without regard to fasting/postprandial. The postprandial blood sugar test (PPBS) measures your blood sugar level precisely two hours after you start eating. It is challenging to have adequate diabetes control unless and until the PPBS, in addition to the measurement of hemoglobin A1c (HBA1c), is monitored and is within the recommended range. The HBA1c test, along with an assessment of symptoms, is used to diagnose diabetes. Furthermore, the HBA1c test does not require any fasting, and it can give an over the past eight to twelve weeks average diagnosis or endorse either diabetes or prediabetes. Up to 5.7 is normal, 5.7-6.4 is prediabetes, and greater than 6.5 are diabetic (Table 1)

Table 1: Blood Sugar Chart for healthy, prediabetes, and diabetes (12).			
FBS	**PPBS**	**HBA1c**	**Result**
<100 mg/dL	<140 mg/dL	<5.7%	Healthy
101-125 mg/dL	141-199 mg/dL	5.7-6.4%	Prediabetes
>126 mg/dL	>200 mg/dL	>6.5%	Diabetes

Discussion:

Your pancreas produces the hormone insulin, which functions as a key to allow blood sugar to enter cells for use as fuel. When you have prediabetes, your body's cells typically don't react to insulin. To try and stimulate a response from cells, your pancreas produces more insulin (5). Your blood sugar levels eventually rise as a result of your pancreas' inability to keep up, which can lead to prediabetes and type 2 diabetes in the future. It is advised that FBS and PPBS be interpreted independently of one another and in relation to their respective biological reference ranges (4). Reactive hypoglycemia, dietary meal composition, timing or duration of sampling after food digestion and absorption, medications like insulin preparations, sulfonylureas, and amylin analogues, or conditions like overproduction of insulin can all cause PPBS levels to be lower than FBS levels. Patients who are meeting treatment objectives (and who have stable glycemic control) should have an A1C test done at least twice a year. It has been demonstrated that lowering A1C to under or near 7% can lessen the microvascular and neuropathic consequences of type 1 and type 2 diabetes (5). Patients with clinical disorders that reduce mean erythrocyte age or limit erythrocyte life span may experience falsely low HbA1c levels (below 4%) (5). When there are clinical factors that impact erythrocyte survival, HbA1c may not adequately reflect glycemic management. As an alternative to glucose, fructosamine can be used to measure glycemic control. Another useful tool that clinicians use is urine micro albumin, which can help judge renal involvement in diabetes.

Preventive measures: Some risk factors for prediabetes are modifiable, for instance, extra weight, less exercise, controlling blood pressure, and avoiding nicotine products/ smoking. Increasing a more fiber-rich diet helps or avoiding more sugary foods helps you to prevent prediabetes (5). A method that is frequently employed to encourage better blood sugar control is the glycemic index. A food's glycemic index depends on a number of things, such as the nutrients it contains, how it was prepared when it was picked, and how much processing it had. The glycemic index can aid in weight loss, lower blood sugar levels, and lower cholesterol in addition to raising your awareness of what you're placing on your plate.

Higher body mass index (BMI) and waist circumferences increase the prevalence of diabetes. Weight maintenance according to height is essential for leading a normal healthy life and controlling BMI can prevent diabetes from progressing from prediabetes. Both men and women have inverse associations between prediabetic prevalence and educational levels. The use of pharmacotherapy should follow a case-by-case approach. Biguanides, Thiazolidinediones, C-Glucosidase Inhibitors, GLP-1 analogs, and non-antidiabetic drugs and therapies including anti-obesity drugs and bariatric surgery have been studied in the context of prediabetes (4). During prediabetic pharmacotherapy, the physician should define the treatment plan's goals and endpoints in advance. In the case of children and adolescents, pharmacotherapy should be utilized with caution.

Concluding remarks:

Diabetic prevention strategies that detect and treat prediabetes can delay the onset of diabetes. People with prediabetes shouldn't panic but should start building changes in their diet and lifestyle to prevent their blood sugar from increasing and turning into type 2 diabetes. Prediabetes can be reversed and Type 2 diabetes prevented by losing weight, exercising regularly, and eating healthy.

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5. The paradox of a giant platelet predominance

Dr.Marquess Raj
ZTC TN & Pondicherry

Foreword:

Platelets are anucleate cells which are essentially pinched off fragments of megakaryocyte cytoplasm. In a way platelets are akin to space orbs, 2 to 4cm in size that detach themselves from the giant megakaryocytespaceship that is 50 to 100cm in size.Yet for a small anucleate cell, platelets possess a complex canalicular system & plenty of organelles to help play their part in haemostasis. In Romanowsky stained smears platelets appear as pale blue structures with purple granules.

In normal individuals there should be one platelet for every 10 to 25 RBC's. A significant fall in platelet count affects the haemostatic process & can predispose to bleeding. Thrombocytopenia refers to decrease in the number of platelets in peripheral blood below the lower range of platelets (1.5 lakhs/cu.mm). The most common causes of thrombocytopenia are tabulated below -

Abnormal platelet morphology: Many a time platelets are viewed only from a count perspective & subtle morphological changes that might be vital clues to underlying disease are under-reported.

Examples:

- Hypogranular platelets or agranular platelets can be seen in myleodysplasticsyndrome (MDS).
- Giant platelets along with neutrophilic inclusions are described in May Hegglin anomaly.
- Giant platelets can also be artefactual, when smears are made from EDTA samples which have crossed 24 hours.
- In several pathological states ranging from DIC to rare genetic mutations such as GATA – 1 related disease, giant platelets are noted.

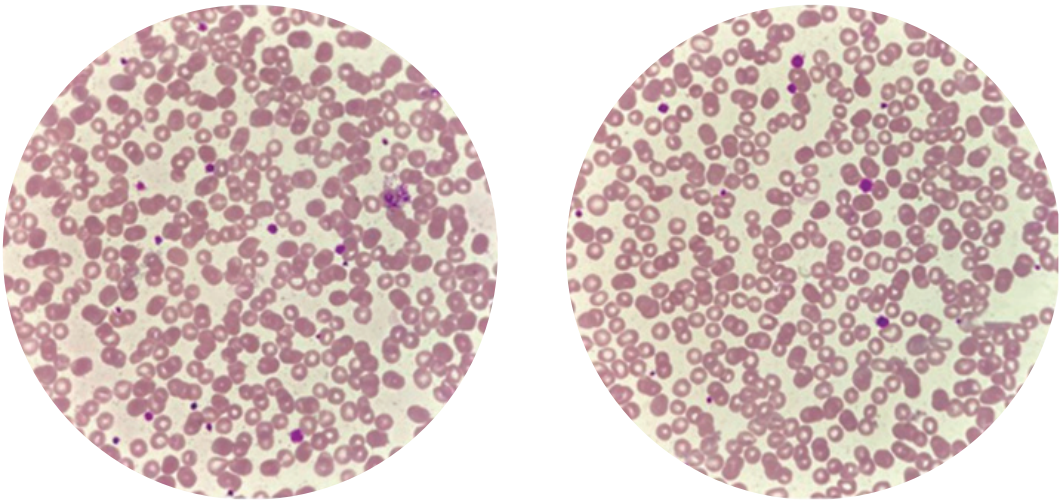
Causes of thrombocytopenia

Increased destruction of platelets	Decreased production of platelets	Dilutional thrombocytopenia
A.Immune	A.Hereditary	Massive blood transfusion
ITP	Fanconis anemia	
SLE	Wiskott- Aldrich syndrome	
B.Drugs - Penicillin, Gold salts		Increased sequestration
C.Infectious causes - Malaria, Dengue	B.Acquired	Hypersplenism
D.Non Immune	Aplastic anemia	Giant hemangioma
DIC	Megaloblastic anemia	
HUS	Marrow infiltration by tumour	

Body of the matter:

Increased destruction of platelets is one of the mechanisms of thrombocytopenia in autoimmune disease as well as thrombocytopenia due to infectious agents such as malaria & dengue. Serial monitoring of platelets is often required in latter & interventions such as platelet transfusion may be based on the platelet counts.

Thrombocytopenia is often marked in thrombocytopenia due to infectious agents such as malaria & dengue. However in immune thrombocytopenia such as ITP the thrombocytopenia is often moderate (around the lower limit of the normal reference range) & not very severe. Another important morphological feature that has to be reported in peripheral smears is the presence or predominance of giant platelets.



Images: Showing predominance of giant platelets (Image courtesy: - RRL,Chennai)

The importance of reporting giant platelet predominance:

Giant platelets are larger in size than their more conventional counterparts. Giant platelets are larger than 7 microns & hence can be as large as normal RBC's or even larger. The presence of giant platelets in thrombocytopenia reflects that the marrow is trying to overcompensate for the increased destruction of platelets. The presence of giant platelets suggests a positive response from the marrow irrespective of aetiology.

Also, it should be borne in mind that though the peripheral blood might show thrombocytopenia significant number of platelets might be sequestered in the spleen. This is the reason why majority of the patients even with severe thrombocytopenia don't developing bleeding. In conditions such as ITP where in there is an autoimmune aetiology, the thrombocytopenia can be mild or in some instances the platelet counts can be near normal. However, the morphology of these platelets largely tends to take a 'giant' morphology. Hence it is important to report the subtler aspects of platelet morphology such as a preponderance of giant platelets in the peripheral smear rather than only mentioning their presence. It is also prudent to convey this significant finding to the clinical team & request them to probe further for autoimmune thrombocytopenia's.

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6. Something to catch on- DNA Sequencing technologies

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Decoding the genetic information using DNA sequencing was underway as 'Sanger Sequencing' for more than four decades. With the advent of massive parallel sequencing technology i.e., Next Generation Sequencing (NGS) it is feasible to sequence large amounts of DNA in lesser time, while reducing the cost. NGS has expanded access to the information for the entire genome, however the ongoing challenge will be able to expand the knowledge about utilization of information for our health and well-being.

The Sanger sequencing - 'first generation' DNA sequencing is a robust method targeting specific DNA regions including point mutation, small deletion/duplication, mutational spectrum of a tumour, constitutional variant and so on in diagnostic testing. Most importantly, it serves as an orthogonal method for confirming sequence variants identified by NGS. Sanger is "gold standard" in molecular diagnosis with >99% test accuracy as first line approach. Now-a days Sanger method is automated to make it faster but it would take years to sequence all of a person's DNA/genome. Next-generation sequencing has sped up the process taking only days to weeks to sequence a human genome as it can sequence parallel large number of fragments of DNA (Figure 1).

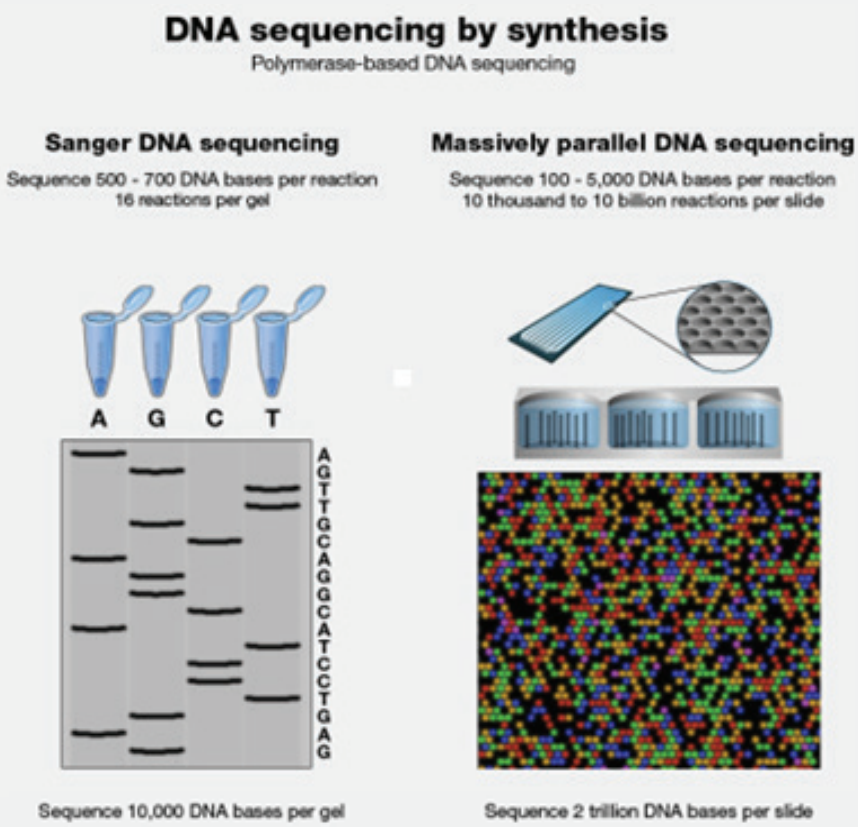

Figure 1: Sanger and NGS methods of DNA Sequencing.

Recently, two methods of NGS (Massive Parallel sequencing) i.e., whole exome sequencing and whole genome sequencing are used in healthcare, are catching up rapidly to identify genetic variations. In Prenatal screen testing market, Non- Invasive Prenatal Test (NIPT) by whole genome sequencing approach is in demand and is surging in India, thanks largely to various factors: a desire to delay pregnancy, more pregnancy-related complications in the second or third trimester, and an increasing number of doctors opting for advanced genetic testing for high-risk pregnancies. With increasing availability of the test as per Global Data's report, "Prenatal Screening Tests– Global Market Analysis and Forecast Model," reveals that NIPT tests market in India is expected to exceed $20 million in 2025.

Based on the Human genome data availability from Human Genome project and automated sequencers like NGS has allowed fast analysis of the whole genome and helped in various fields like oncology, namely development of liquid biopsies or companion diagnostics for precision therapies. Among other applications in the diagnosis of hereditary hearing and vision loss, genetic cardiomyopathies and autosomal dominant polycystic kidney disease. NGS also permits single cell sequencing, which allows the discovery of biomarkers at the level of an individual cell. NGS techniques are, at this point, well codified. Research studies have grown in multitudes and the horizons of genomics has expanded because of the invention, implementation and wide adoption of NGS technology and immensely upgraded the genetic testing tool. International Cancer Genome Consortium (ICGC)-icgc.org, The Cancer Atlas (TCGA)-cancergenome.nih.gov, University of Cambridge, Genomics England, and Illumina to sequence 10,000 whole genomes of children and adults with rare disease were all possible. The horizons of clinical diagnostics are developing immensely based on all the research carried out for decades and there is a clear expansion of clinical diagnostics over the years (as seen in the graphical representation in figure 2). Inherited testing in panels of genes, exomes, whole human genomes, diagnostics in solid tumour, infectious diseases, NIPT is all possible with accessible cost with general acceptance of genetic tests in the clinical sciences.

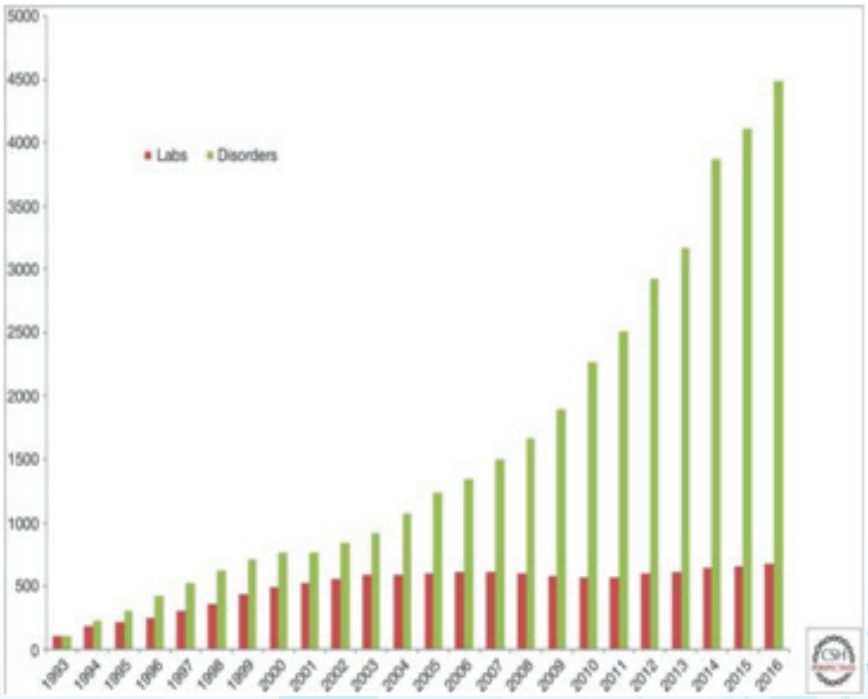

Figure 2: Number of Clinical diagnostics labs as per year.

1. Metastasis of which of the following benign tumors is described?

A. Lipoma
B. Leiomyoma
C. Osteoma
D. Fibroma

2. Which of the following degenerations is described in leiomyoma?

A. Calcareous
B. Hyaline
C. Red
D. All of the above

3. Which of the following diseases can run in families?

A. Breast cancer
B. Thyroid cancer
C. Colon cancer
D. All of the above

4. Which of the 2 types of diabetes can have a autoimmune etiology?

A. Type 1
B. Type 2
C. Both A & B
D. None of the above

5. Which of the following tests help physicians monitor renal impairment in diabetes?

A. Urine micro albumin
B. HbA1C
C. Blood sugar
D. All of the above

6. Which of the following organs is affected in diabetes?

A. Eye
B. Blood vessels
C. Nerves
D. All of the above

7. Hypertension is found in approximately in___% individuals with diabetes?

A. 10 %
B. 25 %
C. 75 %
D. 100 %

8. Which of the cells does not possess a nucleus?

A. RBC
B. Platelet
C. Both A & B
D. None of the above

9. Which of the following precursor cells produces platelets?

A. Hepatocyte
B. Neurons
C. Megakaryocyte
D. Endothelium

10. Which of the following is not a part of DNA?

A. Thymine
B. Uracil
C. Adenine
D. Cytosine

11. In which of the following diseases is increased turnover of purines noted?

A. Goiter
B. Gout
C. Hypertension
D. Osteoarthritis

12. Which scientist is known as 'the dark lady of DNA'?

A. Marie Curie
B. Rosalind Franklin
C. Virginia Apgar
D. Henrietta Lacks

November 22 issue quiz – Answers

1. B. Chromoblastomycosis is a fungus.
2. D. Folic acid deficiency causes macrocytic anemia.
3. C. Rickets is caused by deficiency of vitamin D
4. D. All of the above. Correct disinfection & volume are of utmost importance while collecting blood culture samples.
5. D. All of the above. Droplet PCR has multiple applications in disease diagnosis.

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