



News Cerba

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Analysis info... Analysis info... Myelograms

Cytological analysis of blood smears is becoming an essential test in combination with a myelogram in the diagnosis of myelodysplastic syndromes (WHO 2008).

Myelodysplastic syndromes constitute a varied group of clonal disease of the haematopoietic stem cell that generally affect elderly subjects and are characterised by the following:

- **cytopenia** either **alone** (anaemia, thrombocytopenia, neutropenia) or **in various associations** (bicytopenia or pancytopenia) in the haemogram;
- **normal or enriched bone marrow**;
- morphological abnormalities affecting one or more of the three myeloid lines characteristic of **dysmyelopoiesis**;
- occasionally, **excess medullary blast cells** but in any case < 20% (a value of ≥ 20% indicates acute leukaemia);
- progression towards marrow insufficiency and in some cases (less than one-third), acute myeloid leukaemia.

The diagnosis and classification of myelodysplastic syndromes is based on cytological data (blood and bone marrow) and cytogenetic data (medullary karyotype). Since the publication in late 2008 of the new WHO classification, blood smears have become a new and essential element in the classification of different categories of myelodysplastic syndromes. For example, diagnosis of refractory anaemia with excess of type 1 blasts (AREB-1), previously defined in terms of the percentage number of medullary blast cells, can now be made in certain cases simply on the basis of the percentage number of blood blast cells.

Thus, cytological analysis of the myelogram, required for the diagnosis and classification of myelodysplastic syndromes, must now be combined with cytological analysis of blood smears.

Béatrice Caron Servan - bservan@lab-cerba.com

- IN PRACTICE -

- Sample: 3 unfixed unstained **bone marrow smears** + 3 unfixed unstained **blood smears** using venous blood taken on EDTA the same day.
- Run frequency: 5 d/week
- Turnaround time: 1 day
- Include haemogram data and "haematology" clinical data sheet.



Note...

New!!!

Ordering your supplies by internet

To simplify the ordering of supplies, we have created a special internet order form. Simply insert your laboratory address and the quantities of the products you wish to order. Your order will be forwarded by e-mail to the appropriate department, who will take care of your request.

<http://www.lab-cerba.com>

(section: analysis information
subsection: sampling).

Calendar July/August 2009

PUBLIC HOLIDAYS

On Tuesday **14 July** 2009, the **customer hotline** will be **closed** and there will be **no collections** or **transportation**.

On Monday **13 July**, please **do not take any samples** for the following analyses: HLA Class I, lymphocyte sub-populations, cellular allergy tests, CMV blood antigens and lysosomal enzymes.

SUMMER OFFICE HOURS

From Monday **3 August** to Friday **28 August**, the **customer hotline** will be open between **7.30 am and 6.30 pm** and on **Saturdays** between **8 am and 4.30 pm**.

New development...New development...

SHOX gene mutations and short stature.....

Short stature is a condition affecting between 2 and 3% of children and it is an important cause for concern. One of the genes involved in short stature is the SHOX gene.

The SHOX gene codes for a transcription factor expressed during development of skeletal tissue. It is located on the short arms of chromosomes X and Y, in the pseudoautosomal region (a region shared by chromosomes X and Y, thus being found in two copies in males and females alike), and the mode of transmission is comparable with dominant autosomal transmission. It comprises 6 exons (only exons 2 to 6 code), measures 35 kb, and produces two transcription factors by alternative splicing, SHOXa and SHOXb.

SHOX gene abnormalities can cause several types of short stature. Homozygous abnormalities (affecting both alleles) are responsible for Langer dwarfism: an extreme form of dwarfism involving severe deformity of the limbs and severe mesomelia (shortening of the intermediate limb segments).

Heterozygous SHOX gene abnormalities (affecting only one allele) are seen in 50 to 80% of cases of Leri-Weill syndrome, i.e. mesomelic short stature and Madelung deformity of the wrist, which in some cases is visible only on wrist x-ray. The short stature seen in Turner syndrome (monosomy X) is due to the absence of one of the alleles of the SHOX gene. Haploinsufficiency of the SHOX gene is also seen in 2 to 3% of cases of idiopathic short stature.

The chief indication for SHOX gene analysis is thus short stature, particularly where there is a familial history of short stature and/or bone signs affecting the intermediate segments of the limb or involving Madelung deformity.

Abnormalities in the SHOX gene, other than changes in the X chromosome seen in a standard karyotype, consist of microdeletions in 70 to 80% of cases and of mutations in 20 to 30% of cases. The strategy adopted by the laboratory consists of systematic testing by MLPA for microdeletions and by sequencing for mutations.

Short stature associated with a SHOX gene abnormality responds to growth hormones, allowing delayed growth to be reduced significantly if treatment is initiated before puberty; diagnosis of SHOX gene abnormality can therefore be useful in guiding suitable and effective treatment.

Dr Pascale Kleinfinger – pkleinfinger@lab-cerba.com

- IN PRACTICE -

- Sample: 5 ml of whole blood taken on EDTA at ambient temperature.
- Run frequency: 2/wk for MLPA, 1/wk for sequencing.
- Turnaround time: two weeks

Reminder ... Reminder ... Reminder ...

Urine sample bottles.....

We can now provide 30ml bottles (code: U14) for the collection of 24-hour urine samples.

When using these bottles, please transfer part of the HOMOGENISED 24-hour urine sample indicating diuresis. In order to ensure a tight seal, the bottles should be closed by screwing the top tightly shut without clipping.

You can obtain these bottles:

- by internet: <http://www.lab-cerba.com> (section: Analysis information; subsection: sampling device)
- by post: simply send us a **duly completed materials request form**
- by telephone: 01.34.40.20.20

We thank you in advance for using only these bottles.