Minutes of meeting SIG on Lab Techniques  
ISPD 17th International Conference, Lisbon, Portugal  
June 5, 2013

Chairs: Brigitte Faas / Vincenzo Cirigliano

The 2013 SIG Laboratory Techniques was scheduled on June 5, at 8.30, and was well attended. The list of attendees with their email addresses will be compiled and posted on the ISPD website.

Minutes 2012:  
From the minutes of the SIG meeting in Miami Beach, June 3 2012:  
“It was suggested by Joyce Harper and Lia Knegt to compose a “Data Collection Report”, similar to the PGD consortium. Joyce and Lia volunteered to initiate this.”

The chairs sent out a mail to both Joyce and Lia, but at the time of Lisbon meeting, they did not respond yet.

Activities 2012 / 2013:  
The SIG participated in several activities:  
In July 2012, Brigitte Faas participated as speaker in the XIV CORSO RESIDENZIALE DI GENETICA MEDICA: LE MALATTIE GENETICHE COME MALATTIE SOCIALI in Chieti (Italy). It was a very nice initiative to have ISPD involved in this course. The course is organized bi-annually, and other speakers on behalf of the ISPD were Peter Benn and Howard Cuckle.

In 2013, the ISPD Board approved a Position Statement developed by a Working Group, the Aneuploidy Screening Committee. Some members of the Laboratory Techniques SIG, actively involved in NIPT, were also part of this group. This updated policy document includes both conventional screening and non-invasive testing using cell-free fetal DNA in maternal plasma. The position statement is posted on the ISPD website.

Courses at the 17th ISPD conference in Lisbon:  
Two preconference course were organized this year:  
Course 1, “Array in prenatal diagnosis”, chaired by dr Brigitte Faas, took place on Sunday morning, June 2nd, and course 5, “Noninvasive prenatal testing”, chaired by prof Lyn Chitty, took place on the same day in the afternoon. Both were very well attended and so far, without having read the evaluation forms yet, reactions were very positive.

Points that were raised during the SIG meeting:  
In the minutes of the Miami Beach meeting it was raised that:  
“NIPT is coming up very quickly now. It was agreed that we, as SIG for Lab Techniques, could play a major role in setting up guidelines for this. But isn’t it too early?”

The chairs mentioned they discussed setting up laboratory guidelines for NIPT, but felt this should not be started up, as all laboratory handlings are carried out by companies now, that have their own protocols. ISPD cannot make lab protocols for companies and force them to follow this.

The option of putting together a database with results from individual caregivers offering NIPT was discussed. All results of sensitivities and specificities are now derived from data from the companies. There is no external, objective, non-commercial overview. Will it be possible to put together such a database? In that case we need to know who is providing NIPT in which country. This will probably be a huge task.

A discussion now started including several issues that go along with NIPT.

In several countries NIPT is offered via more than one caregiver. In Spain, Portugal and Italy all companies are present apart from Sequenom, also the U.K., Belgium and Germany are offering the test while more strict regulations on sample outsourcing are hampering its implementation in France.
Depending on the company providing the service and country, NIPT has different prices. If someone wants to offer NIPT, how can they actually choose? This depends on several issues:
- The reliability / accuracy of the test
- The price of the test
- The service of the company
  - The reliability / accuracy of the test is “more or less” comparable for all companies.
  - The prices differ between the companies, targeted tests are mostly somewhat cheaper.
    Matthias Ehrich was the only attendee from a company, in this case Sequenom. He explained that, at least in the US, prices are not fixed but negotiable and this holds for all companies. Prices can depend on expected numbers and what is included. E.g. if counseling is included, prices will be higher than without counseling.
    It was mentioned that especially, but not exclusively, for the patient it is very confusing that even within one country or region prices can differ, depending on the company and on how good one can negotiate.
  - Service level of the company. How much service does a company provide? Perhaps a company offers a reliable test and has attractive prices but what about the service? We can only find out by contacting others who offer the test and ask for their experiences. But how do we know who offers the test and to whom?
    It was agreed that it would be very helpful to have a database with at least contact details of care givers who offer NIPT and the name of the company via which they offer it, so these persons can be contacted and asked for their experiences. ➔ action point

Another issue that was raised was how to deal with the patents. If a lab is technically and logistically capable of performing NIPT, is it allowed to do so because of the patents? This question was also raised during the preconference courses and the patent issue is still pending. No one can comment on that now. Matthias Ehrich mentioned the possibility of companies offering help in implementing NIPT in a lab, providing standard operating procedures and training.

Another important issue that was raised was patient informed choice and counseling. How to pre- and posttest counsel the pregnant women? This topic was not included in the conference but there appears to be a tremendous need for NIPT counseling education. Companies are not reporting in the same way (risk or not), so how to counsel this?
Matthias Ehrich explained that at least Sequenom offers all-in packages, in which counseling is included. They have trained a number of counselors specifically for NIPT. But most care givers do not want the all-in offer, but only the test, and do the counseling themselves, mostly because they are from outside the US.
In those situations, SQNM cannot be responsible for correct counseling. They do not train or educate other counselors. They also do not audit their customers to make sure all aspects of the total NIPT service, including counseling, are sufficiently well covered. This is probably also the case for other companies, but there were no employers from other companies to confirm this.
It was suggested to organize a course under the umbrella of ISPD, dealing with more practical aspects, especially counseling, of NIPT. The upcoming conference, next year in Brisbane, was considered too late and too far away for quite a large group of highly interested attendees. Is it possible to organize a course in Europe on this, somewhere in 2013 or beginning 2014.
  ➔ Action point, to discuss with ISPD board.

All the above mentioned issues gave rise to a very lively informal discussion that was highly appreciated by the attendees.

Before closing the meeting at 9.30, BF shortly mentioned the absence of a external quality control scheme for NIPT. For all other tests, schemes like these are available. With the likely future expansion of testing laboratories, perhaps ISPD can play a role in this?

VC / BF, July 2013