

# STATEMENTS OF COMPETENCE FOR CYTOGENETIC TECHNOLOGISTS

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A competent cytogenetic technologist working independently without constant supervision can interpret and implement established procedures to prepare appropriate specimens for cytogenetic analysis, perform that analysis, and prepare and describe the karyotypes.

Competence includes, but is not limited to, skill and knowledge in the following specific areas:

## **1. Collection, handling, preparation, and processing of various specimens**

- 1.1 Identify appropriate specimens for study, and methods of collection, preservation, and transport.
  - 1.1.1 Select appropriate containers, anticoagulants, collection media, antibiotics, and preservatives.
  - 1.1.2 Identify factors important for the transport of specimens, such as overnight delivery, transport media and containers, recommended temperatures.
- 1.2 Assess acceptability of specimen for study.
  - 1.2.1 Evaluate suitability of specimen for requested study, both for type and amount obtained.
  - 1.2.2 Judge quality of specimen noting presence of blood in amniotic fluid, blood clots or hemolysis in blood samples, syringe number and presence of spicules in bone marrow, etc.
  - 1.2.3 Describe methods for possible recovery of poor samples.
  - 1.2.4 Notify appropriate individuals of any unsatisfactory samples and document such notification.
- 1.3 Enter details of specimen into appropriate logbooks and computer systems.
  - 1.3.1 Record patient's name and all required and pertinent information including identification number, date of birth, sex, clinical history, indication for study, referring physician
  - 1.3.2 Record accurate and complete information concerning specimen including type of tissue, amount, appearance, collection date and time, anticoagulant etc.
  - 1.3.3 Record priority status of specimen, and identify as appropriate.
  - 1.3.4 Record notes of any special test requests, particularly those requiring transport of samples to other laboratories.
  - 1.3.5 Record chromosomal region of interest in high-resolution studies.
- 1.4 Follow protocols to ensure proper identification of patient materials through the complete process, from accession to final report.
- 1.5 Assist in maintaining necessary records and laboratory database, in logbooks or computers, as appropriate.

## **2. Appropriate culture techniques for submitted specimens**

- 2.1 Explain and use aseptic techniques.
  - 2.1.1 Use measures (such as Universal Precautions) that protect employees from real or potential exposure to infectious agents (e.g., protective clothing, gloves and masks, containers for sample delivery and waste disposal, biological safety cabinets).
  - 2.1.2 Use and document methods to detect, identify, control, and eliminate microbial or chemical contamination.
  - 2.1.3 Practice measures that prevent cross-contamination between samples.
- 2.2 Prepare appropriate media for specimens, giving consideration to the clinical indication for the study.
  - 2.2.1 Choose appropriate medium additives such as sera, antibiotics, buffers, mitogens, and growth factors.
  - 2.2.2 Select appropriate methods of preparation and storage of media to maintain pH, sterility, and ability to support growth
- 2.3 Employ appropriate culture techniques for specimen.
  - 2.3.1 Select culture equipment and vessels for closed or open culture system.
  - 2.3.2 Select culture technique for specimen taking into account type of tissue, methods of initiation, type of culture, and purpose for study.
  - 2.3.3 Adjust culture methods to minimize adverse effects of unwanted constituents in the culture (e.g., blood in amniotic fluid).
  - 2.3.4 Appraise the effect of cell density on rate of growth and adjust appropriately (e.g., cell count of leukemic specimens).
  - 2.3.5 Employ techniques for cryopreservation and recovery of cultures, and maintain

- documentation of all samples preserved.
- 2.3.6 Monitor and document the effectiveness of all solutions used in the procedures prior to use on diagnostic material.
  - 2.3.7 Record complete information for culture of specimen, including identification of technologist, lot numbers of media, sera, and other reagents, incubator used, and mitogen, if used.
- 2.4 Monitor cell growth and control variables.
- 2.4.1 Employ measures that will maintain optimum cell growth (e.g., feeding and spinning of cultures to correct for depleted medium).
  - 2.4.2 Evaluate status of cultures using assessment of growth and mitotic activity, pH of medium, and turbidity.
  - 2.4.3 Identify and document probable causes of poor growth and culture failure, such as inadequate specimens, or equipment failure, and corrective actions taken.
  - 2.4.4 Report to appropriate authority findings of culture failure or growth inadequate for analysis, and request new sample if appropriate.

### **3. Principles and techniques for harvesting specimens or cell cultures**

- 3.1 Determine optimal time sequence and method for harvest (manual or robotic).
  - 3.1.1 Apply knowledge of cell cycle for various cell types and culture conditions (e.g., PHA stimulated lymphocytes, unstimulated leukemic cells, synchronized cultures, chorionic villus or amniotic fluid cells, fibroblast or solid tumor cultures) to determine time for harvest
- 3.2 Use appropriate harvest procedures for specimen or culture.
  - 3.2.1 Use synchronizing or intercalating agents, such as amethopterin, fluorodeoxyuridine, bromine deoxyuridine, ethidium bromide, or actinomycin D, at appropriate concentration, temperature, and duration.
  - 3.2.2 Use spindle fiber inhibitor (e.g., Colcemid, Velban) at correct concentration, temperature, and duration.
  - 3.2.3 Use recommended procedure for removing cells from culture vessels.
  - 3.2.4 Use appropriate hypotonic solution (KCl or sodium citrate), at correct concentration, temperature, and duration.
  - 3.2.5 Use cell fixative (acetic acid/alcohol) at correct concentration, temperature, and duration.
  - 3.2.6 Control mechanical damage to chromosomes by proper mixing, shaking, pipetting, centrifuging, or other handling of the cells.
  - 3.2.7 Record complete information for harvest of specimen including date, spindle fiber inhibitor, intercalating or synchronizing agents, conditions used for harvest, and identification of technologist.
- 3.3 Prepare slides with analyzable metaphases.
  - 3.3.1 Select method of slide preparation that will produce high quality metaphases with optimum spreading (e.g., control variables such as wet or dry slides, air flow, humidity level and temperature during air drying to regulate slide drying rate.)
  - 3.3.2 Employ techniques that control concentration and distribution of cellular and other debris on slides.
  - 3.3.3 Evaluate quality of slides with phase contrast microscope and adjust variables as necessary.
  - 3.3.4 Employ techniques that control the aging of slides to produce optimal banding conditions (e.g., storing at various temperatures, such as 37°C, 60°C, 90°C, for various times, such as 20 min to 2 hours, or overnight; UV exposure or microwave.)
  - 3.3.5 Use slide storage methods that best maintain chromosome quality for banding and staining procedures, with protection from humidity, light, chemicals, or mechanical damage.

### **4. Principles and techniques of chromosome banding and staining**

- 4.1 Select banding methods that permit identification of each chromosome pair, at an appropriate band level; describe the advantages and disadvantages of each method.
  - 4.1.1 Use G-banding pretreatment and staining methods, utilizing trypsin, 2XSSC, urea, or other chemicals.
  - 4.1.2 Use fluorescent banding methods with stains such as quinacrine mustard or Hoechst stain.
  - 4.1.3 Use R-banding methods, with heat treatment of slides, and staining with acridine orange, chromomycin A3, or other dyes.
- 4.2 Use and interpret additional staining procedures, such as C-banding and NOR staining that yield nonbanded chromosomes or enhance specific chromosome regions, if indicated to identify specific abnormalities.
- 4.3 Perform and interpret results for other specialized staining procedures when needed (e.g.,

- DAPI/Distamycin A, sister chromatid exchange, etc.).
- 4.4 Select mounting materials, hydration/dehydration methods, and destaining techniques when necessary for multiple staining procedures on the same slide.
    - 4.4.1 Select the appropriate type of specimen and type of probe for both interphase or metaphase FISH analysis.
    - 4.4.2 Perform appropriate slide pretreatment, denaturation, dehydration, hybridization and detection for both direct and indirect labeled probes for both interphase and metaphase FISH analysis.
    - 4.4.3 Understand the use and interpretation of controls for FISH analysis.
  - 4.5 Select slide cleaning and storage methods that maintain quality of chromosome preparations for period of time required by regulatory agencies.
  - 4.6 Troubleshoot unacceptable or unanalyzable results for all banding/ staining procedures.

## **5. Maintenance and use of microscopes and computer-generated imaging techniques and equipment**

- 5.1 Operate a standard compound microscope, inverted microscope, stereo microscope, and computerized karyotype equipment.
  - 5.1.1 Clean, adjust, focus, and use appropriate illumination systems, eyepieces, objectives, condenser systems and filters, for bright field, fluorescent, and phase contrast microscopes.
  - 5.1.2 Operate microscopes and computerized image capture system equipment for optimal resolution of specimen. Demonstrate appropriate use of cover slips and immersion oil.
  - 5.1.3 Maintain computer image analysis equipment in optimal working order.
- 5.2 Select methods that produce optimal chromosome images.
  - 5.2.1 Produce electronic images with appropriate clarity.

## **6. Chromosome analysis**

- 6.1 Select suitable metaphases/interphase cells for analysis.
  - 6.1.1 Select metaphases according to morphology, spreading, length, and banding detail.
  - 6.1.2 Assess difficulties in microscopic analysis and computer imaging posed by overlapping chromosomes, debris, poor stain, etc.
- 6.2 Perform accurate microscopic counts and analyses of banded and nonbanded chromosomes or FISH signals
  - 6.2.1 Determine modal number.
  - 6.2.2 Analyze chromosomes at the microscope, and identify normal/abnormal karyotype.
  - 6.2.3 Document the analysis of separate colonies on amniotic *in situ* cultures.
  - 6.2.4 Document the detection of signal numbers for specimens and controls by cell percentages.
- 6.3 Record microscope Identification, stage coordinates, and cell analysis data on all cells selected.
  - 6.3.1 Document analysis in an organized manner (e.g., patient information, modal number, sex chromosome constitution, aberrant chromosomes, slide identification and verniers, identification of technologist, and date of work).
  - 6.3.2 Use a method that allows rapid retrieval of any cell analyzed, on the same or another microscope, (e.g., use of a calibrated microscope stage, microlocator slide, conversion chart).
- 6.4 Prepare accurate karyotypes from computer images.
  - 6.4.1. Organize chromosomes according to a systematic and approved format (e.g., ISCN).
- 6.5 Identify numerical and structural chromosome abnormalities, and relate their implications (e.g., phenotype and relationship to disease).
  - 6.5.1 Determine numerical abnormalities of the autosomes and sex chromosomes.
  - 6.5.2 Differentiate between the presence of multiple cell lines, and random gain or loss of chromosomes in slide preparation of specimens and /or controls.
  - 6.5.3 Identify constitutional structural abnormalities such as translocations, deletions, inversions, ring chromosomes, isochromosomes, and fragile sites.
  - 6.5.4 Identify sporadic structural abnormalities such as chromatic breaks, chromatic exchanges, fragments, and endoreduplication.
  - 6.5.5 Recognize and identify heteromorphic chromosomes with different variable regions, by band number, code letters, size, and banding intensity.
  - 6.5.6 Assess band level for high resolution studies.
- 6.6 Summarize the results and report to appropriate authority.
  - 6.6.1 Recognize and avoid hazards implied in oral reporting of results.
  - 6.6.2 Draft a neat, accurate report using standard nomenclature established by the current ISCN, summarizing the findings in understandable text and incorporating the patient identification, and all relevant clinical and laboratory data; forward to the appropriate individual for review and signature.

- 6.6.3 Document oral and preliminary reports on final written report
- 6.7 Report to the appropriate authority the need for additional studies to complete the diagnosis (e.g., repeat the culture, perform additional staining techniques, analyze other tissues, request family studies, molecular or biochemical studies).

## **7. General laboratory skills, quality control, and quality assurance**

- 7.1 Prepare reagents at the proper concentration and pH, with proper labeling and dating, using required grades of water and chemicals.
- 7.2 Select, operate, clean, and maintain all laboratory equipment and instruments, as appropriate.
  - 7.2.1 Monitor the need for service or repair on any equipment and report this to appropriate authority.
  - 7.2.2 Document usage of gas tanks, and replace as necessary.
  - 7.2.3 Record equipment temperatures with reference thermometers, and adjust controls if necessary.
  - 7.2.4 Monitor centrifuge speed, using a tachometer and adjust if necessary.
- 7.3 Demonstrate principles of sterilization and decontamination procedures (e.g., use of disinfectants, steam, dry heat, gas, U.V. irradiation, and membrane filtration).
- 7.4 Maintain adequate stocks of laboratory supplies and chemicals.
  - 7.4.1 Employ limits on stock usage imposed by shelf life and expiration dates.
- 7.5 Employ appropriate cleaning procedures for laboratory glassware and Instruments.
- 7.6 Practice established procedures for general laboratory safety.
  - 7.6.1 Use Universal Precautions as established by Centers for Disease Control (CDC) and individual state or local governments.
  - 7.6.2 Use appropriate procedures for laboratory emergencies (e.g., fire, accidental injury, natural disaster, chemical spill, or power failure).
  - 7.6.3 Use correct procedures for storage, handling, and disposal of different types of materials and waste: biological and chemical, volatile or stable; radioactive; sharps and glass.
- 7.7 Maintain a system to ensure laboratory quality control in all areas, to comply with all regulatory requirements.
  - 7.7.1 Maintain a system to ensure accuracy of chromosomal results, including appropriate documentation, throughout all steps of laboratory procedures.
  - 7.7.2 Maintain a system to ensure confidentiality and security of patient records.
  - 7.7.3 Maintain system to appropriately label, store, and monitor shelf life, sterility, and quality of all media, sera, reagents and chemicals.
  - 7.7.4 Maintain an easily accessible collection of current Material Safety Data Sheets (MSDS) for all chemicals used in the laboratory procedures.
  - 7.7.5 Maintain a system of records for equipment and instruments (serial numbers, date of purchase, maintenance checks, gauge readings, dates and type of service or repair).
  - 7.7.6 Practice the techniques, procedures and policies used in the laboratory, as documented in the laboratory manual.
  - 7.7.7 Assist in reviews and revising the laboratory manual.
  - 7.7.8 Participate in laboratory proficiency testing, as appropriate.

## **8. General principles of biology and genetics**

- 8.1 State the principles of general biology and genetics that relate to cytogenetics.
  - 8.1.1 Describe cell structure and their function.
  - 8.1.2 Summarize the stages of the cell cycle, and of mitosis and meiosis (both spermatogenesis and oogenesis).
  - 8.1.3 Describe DNA structure (base sequence, complementarity, etc.), and function (genetic code, replication, transcription and translation, and mutations) chromosome ultrastructure: telomeres, centromeres, nucleosomes, histones, loop domains, scaffolding, DNA packing, etc.
  - 8.1.4 Review basic embryology and the origin of various tissues: blood, skin, CVS, and amniotic fluid.
  - 8.1.5 Describe basic principles of inheritance (dominant or recessive, autosomal or sex linked, multifactorial, polygenic, Lyon hypothesis, etc.).
  - 8.1.6 Describe mutagenicity and principles of genetic toxicology.
- 8.2 State the principles of clinical cytogenetics.
  - 8.2.1 Describe etiology of chromosomal abnormalities such as anaphase lag, non-disjunction, dispermy, breakage and repair, uniparental disomy, and the influence of these processes of maternal age effect, clastogens, inherited breakage syndromes and imprinting.
  - 8.2.2 Discuss basic principles of genetic counseling including pedigree analysis and risk calculations for inherited conditions.
  - 8.2.3 Discuss basic principles of cancer cytogenetics including hematopoiesis, clonal evolution,

and findings during remission and relapse.

8.2.4 Correlate molecular genetic results with cytogenetics for prenatal diagnose, family studies, and cancer cytogenetics.