

Supplemental Table 1. Scoping review summary of the landscape of molecular diagnostic and bioinformatics for pathology residents.

Citation, Setting	Literature Summary	Areas of Molecular Training	Curriculum Description or Study Results	Method of Training and Duration	Level of Resident Exposure and/or Involvement in Molecular Diagnostics and/or Bioinformatics Training	Barriers/ Gaps Identified	Comments/ Opinions
Moore, D.A., <i>et al.</i> ; 2018; UK 19	Provides 3 alternative models for pathology training to address challenges of genomics training	Genomic pathology	<ul style="list-style-type: none"> Model 1 (Belfast Model ²⁵) – delivers morphological-molecular training within current 5 year time frame of training Model 2 –Molecular pathology training with optional extension to allow a Masters or Fellowship Model 3 – Train morphological-molecular pathology in RCPATH-accredited centers that have capacity to train molecular pathologists 	<ul style="list-style-type: none"> Model 1 – molecular pathology lectures in the early stages of training, followed by required 2-3 month molecular component + optional final year of training in molecular diagnostics Model 2 – MSc or Fellowship Model 3 – modular training program that follows RCPATH training template 	<ul style="list-style-type: none"> Residents should have knowledge and skills in the following 5 key areas: understanding of complex data (e.g.: bioinformatics), molecular technologies its test results, molecular testing variables, clinical relevance of molecular test results Model 1 – provides basic level of molecular pathology training to all pathology residents (most programs use this model) Model 2 – provides research opportunities in molecular pathology Model 3 – in depth molecular training 	<ul style="list-style-type: none"> Little margin to change RCPATH's curriculum at this time Pathology training does not deliver the following 5 key skills and knowledge 	<ul style="list-style-type: none"> A combination of models 1 and 3 may work in today's landscape of molecular pathology training by providing in depth morphological-molecular diagnostic training over the 5 years of residency
Clay, M.R., <i>et al.</i> ; 2017; USA 4	Current scope, future direction, required skillset for bioinformatics training in AP and/or CP residency education	Bio-informatics, molecular pathology	-	<p>In a 4 year ACGME-accredited AP/CP residency program:</p> <ul style="list-style-type: none"> molecular pathology training - 1 month in year-3 of CP rotation bioinformatics training - at most 10 days during 4 year AP/CP residency training program (where bioinformatics comprise of 25% of clinical informatics, and clinical informatics comprise of only 1 of 27 ACGME pathology-specific milestones) 	<p>Pathology residents should be understand or be trained in the following regarding bioinformatics:</p> <ul style="list-style-type: none"> how data are generated, processed and stored complexities, errors and limitations to molecular pathology testing cycle and analytic factors that affect TAT knowledge of data structure, file formats, bioinformatics pipelines for effective communication of information to other healthcare providers and patients understanding bioinformatics and NGS workflow in clinical molecular pathology labs 	<ul style="list-style-type: none"> Most residents receive limited exposure to bioinformatics during medical school Limited and non-standardized training during residency, if available Ill-defined educational milestones by ACGME leads to 	<ul style="list-style-type: none"> Bioinformatics training in pathology residency programs possible, as long as there is motivation, institutional support, educational resources, adequate faculty expertise (more than 1 or 2 to

				<ul style="list-style-type: none"> • Case-based learning materials currently widely used for training pathology residents in molecular genomics and molecular pathology 	<ul style="list-style-type: none"> • gain exposure to testing parameters (depth of coverage, strand bias, quality scores) • how data are filtered prior to receipt by end-user (e.g.: when classifying oncologic-genomic variants) • how queries to internal or public databases are performed (e.g.: COSMIC) • sequence/variant databases – how are they populated and curated, which ones exist/best to utilize • how bioinformatics assists with medical context (e.g.: patient history, medical condition, clinical indication) • all aspects of patient genetic data protection <p>Fellowship training should incorporate advanced principles unique to each subspecialty, such as molecular pathology</p>	<p>varied training for residents</p> <ul style="list-style-type: none"> • Inadequate faculty expertise • Lack of standardized case-based learning materials for training pathology residents in bioinformatics 	<p>train all residents)</p> <ul style="list-style-type: none"> • Until formal guidelines can be determined by ACGME, each program/institution must tailor bioinformatics learning objectives to institutional mission, needs or expertise of faculty and fellows
<p>Forcucci, J.A., <i>et al.</i>; 2017; USA 26</p>	<p>Describe experience with, and results of, RISE review series that evaluates AP and/or CP residents' progress and medical knowledge</p>	<p>Molecular pathology, cytogenetics</p>	<p>Results:</p> <ul style="list-style-type: none"> • Although an improvement in RISE performance was observed after the review series for most topics, molecular pathology and special topics had lower scores on average following the review series (not statistically significant) • Of note, molecular pathology was among the top 5 highest performing sections before the review series began, suggesting there is less room for improvement in molecular pathology • RISE review series enjoyed by residents 	<ul style="list-style-type: none"> • RISE review series for molecular pathology – 30 minutes • RISE review for cytogenetics – 30 minutes • Lecture based for molecular pathology RISE review • <i>Jeopardy</i> style practice questions for cytogenetics RISE review 	-	-	<p>Molecular pathology RISE review series may be more useful for:</p> <ul style="list-style-type: none"> • senior residents before certification/board exams • year1 and year 2 residents as a taste of what's to come in rotations later in the training program

<p>Aisner, D.L., <i>et al.</i>; 2016; USA 36</p>	<p>An updated and detailed suggested AP or CP curriculum on molecular pathology</p>	<p>Molecular diagnostic, molecular genetics</p>	<p>Molecular diagnostic and genetics training in the following 10 major subjects: 1. Basic molecular pathology and laboratory management 2. Basic concept in molecular biology and genetics 3. Technology for molecular pathology 4. Inherited disorders 5. Oncology 6. Infectious diseases 7. Pharmacogenetics 8. Histocompatibility and identify 9. Genomics 10. Information Management</p>		<ul style="list-style-type: none"> • Basic knowledge and concepts of each of the 10 subject areas and its specific tests, assays and software programs (see Tables for detailed list) • Understand the limitation of assays and diagnostic tests • Interpret molecular test results • Communicate molecular diagnostics to clinicians effectively • Residents should understand the potential biases introduced by bioinformatics pipelines 	<p>Many institutions do not have access to laboratories with extensive genomic testing to train residents.</p>	<p>Sharing of training materials should be encouraged</p>
<p>Black-Schaffer, W.S., <i>et al.</i>; 2016; USA 3</p>	<p>Propose a re-design of pathology residency training program</p>	<p>Molecular diagnostics, bio-informatics</p>	<p>The new model would:</p> <ul style="list-style-type: none"> • Provide a clear definition and acquisition of core knowledge and practice skills that would span year 1 and year 2 of AP and CP residency program • Assess competency based metrics with emphasis on critical thinking and skills • Intensive individual modular training during final years of training 	<ul style="list-style-type: none"> • Proposed molecular diagnostic and bioinformatics training in the advanced training section that follows core training, and/or as an elective for those interested in pursuing this career path in the new model of training • Propose an improved method of teaching and education, with a more flexible and balanced overall curriculum, elective tracks for each resident's career goals (e.g.: molecular pathology/diagnostics) 	<p>As part of the Pathology Milestone Project, residents should understand the following regarding molecular diagnostic tests:</p> <ul style="list-style-type: none"> • these tests may be applied to patient care (for genetic, neoplastic, infectious disorders) and population health • the roles of advanced diagnostic assays and how results affect patient diagnosis and prognosis • pre-analytic issues and QC • provide consultation as needed to clinicians about utilization and interpretation of molecular diagnostic testing • proficient in consultation regarding molecular test utilization and treatment decisions 	<p>-</p>	<p>Hematopathology training is a great example of how to integrate molecular pathology training into AP and/or CP</p>
<p>Haspel, R.L., <i>et al.</i>; 2016 USA but involves Canadian residents</p>	<p>Evaluated a one-day team-based learning (TBL) genomic pathology workshop</p>	<p>Genomic pathology for residents who have had completed a molecular</p>	<p>Results:</p> <ul style="list-style-type: none"> • 100% of residents would recommend this workshop to others • 94% of pathology residents felt this workshop would help them as practicing pathologists • Residents from the workshop conducted at the first national meeting 	<ul style="list-style-type: none"> • Workshop training totaled 8 hours, with each module being 90 minutes • Pre-readings and multiple choice prep questions – emailed to residents prior to workshop 	<p>Workshop modules used different genomic testing modalities: single gene testing, prognostic gene panel testing, cancer gene panels, and whole-exome sequencing</p>	<p>-</p>	<p>-</p>

41	that was refined from a curriculum for pathology residents in 3 national meetings	pathology rotation	<p>gave feedback on wanting more NGS related instruction</p> <ul style="list-style-type: none"> In the 2nd and 3rd workshop, the instructors expanded the introduction to NGS and included genomic alignment data analysis activity 	<ul style="list-style-type: none"> Workshop included a 15-30 minute presentation by the instructor to review answers to prep questions, an hour activity where teams of 3-6 residents (with a balance of junior and senior residents) answer a series of questions that requires the use of online genomic tools, and a 30 minute presentation by the instructor to discuss answers to questions. Lecture based - workshop ended with a 30 minute lecture to summarize major teaching points 			
<p>Murphy, M.J., <i>et al.</i>; 2016;</p> <p>USA</p> <p>33</p>	Curriculum development between the departments of pathology and dermatology for their residents	Molecular diagnostics in dermatopathology	<p>6 steps used by the two departments when collaborating on the development and implementation of educational objectives and teaching modalities for multidisciplinary curriculum:</p> <ol style="list-style-type: none"> 1. Problem identification 2. Needs assessment 3. Goals and objectives (see Table 1) 4. Educational strategies 5. Implementation 6. Evaluation and feedback 	<ul style="list-style-type: none"> Readings and review of teaching sets on clinical dermatology cases that required molecular diagnostics Two 1 hour lecture on molecular pathology and cytogenetic topics in dermatology and dermatopathology 1 week rotation in clinical molecular pathology and cytogenetics lab RISE Assessment and feedback Trainee presentations 	-	<p>Dermatology residents felt that they would like to understand more about the molecular diagnostic tests to better serve their patients</p>	-
<p>Naritoku, W.Y., <i>et al.</i>; 2016;</p> <p>USA</p> <p>16</p>	Surveyed US pathology residency program directors in 2013 to	Molecular pathology, genomics, cytogenetics	<p>Surveyed 143 pathology residency training program directors with 56% response rate:</p> <ul style="list-style-type: none"> 95% (72) had molecular pathology and genomics rotations in AP/CP 	<ul style="list-style-type: none"> Molecular pathology rotation duration increased from mean 5.2 weeks in 2005 to 5.8 weeks in 2013 Cytogenetics rotation decreased in rotation duration 	-	<ul style="list-style-type: none"> Limited pathology residency time Lack of detailed guidance on training by ACGME 	-

	identify trends in residency curriculum and how things have changed since 2005 survey		<ul style="list-style-type: none"> 75% (57) had cytogenetics rotation Large variation and standardization of training program across 143 ACGME accredited program due to variation in size, trained staff, patient populations, facilities and resources 	<p>from mean 3.9 weeks in 2005 to 3.1 weeks in 2013</p> <ul style="list-style-type: none"> Weak trend toward more weeks of molecular pathology training in larger programs (defined as > 16 residents) but there was a wide range in training duration across all program sizes 			
Laudadio, J., <i>et al.</i> ; 2015; USA 32	Developed a list of genomics competencies that a practicing pathologist needs to be successful, regardless of previous training and practice setting	Genomics	List includes 32 highest priority competency statements, organized by 6 competency areas: <ol style="list-style-type: none"> *Basic genetics and genomics principles Ethical, legal and social issues *Sample acquisition QA and regulatory *Testing and interpretation *Patient management and reporting *Highest competency priorities with subtopics and tasks for genomics education to further refine pathology curriculum 	Suggested that an ideal delivery of curriculum (to practicing pathologists) is performance-based and task-oriented education that is interactive and includes simulated cases	-	Weakness - curriculum does not include bioinformatics competencies	-
Domen, R.E., <i>et al.</i> ; 2014; USA 39	Propose re-design AP or CP pathology residency training program	Molecular pathology	Propose to shorten core AP or CP training program to 3 years and extend fellowship program from 1.5 – 2 years to 2.5 years for further subspecialization training in emerging topics such as molecular pathology	N/A, but proposed to incorporate a fast-track option in the residency program in molecular pathology as it is an emerging area of need	-	-	Private practice groups may need to hire AP-only graduates who have completed subspecialty training in molecular pathology fellowship in order to adjust to this proposed model of training pathology residents in only AP or CP to be

							ready for private practice
<p>Haspel, R.L., <i>et al.</i>; 2014; USA</p> <p>20</p>	<p>A needs assessment of the current landscape of molecular diagnostic and genomic training for AP/CP pathology residents</p>	<p>Molecular diagnostics, genomics</p>	<p>Needs assessment:</p> <ul style="list-style-type: none"> • There is a need to incorporate genomic topics during pathology training • Pathology residents need to understand fundamental NGS approaches used in clinical genomics • Trainees will need to understand core principles as practicing pathologists will need to design, develop and validate most clinical genomic tests and assays • Trainees will need some understanding of bioinformatics pipelines in order to ensure optimal clinical performance • Trainees must be able to use various online sequence variance catalogues, learn efficient search strategies and understand how to integrate sequence results with other clinical and laboratory findings in order to report to other healthcare providers 	-	<ul style="list-style-type: none"> • Trainees will need some level of understanding of different bioinformatics pipelines for detection of single nucleotide variants, small insertions/deletions, copy number variants and structural variants 	<ul style="list-style-type: none"> • There are only a few program that perform genomic testing routinely • Many programs lack faculty expertise to train pathology resident to prepare them for practice • Time constraints of pathology residency training limited to 4 years, but advancement in technologies and information are growing rapidly • Further educational tools and support in genomic medicine are needed 	-
<p>Haspel, R.L., <i>et al.</i>; 2014; USA, but involves international residents</p> <p>21</p>	<p>Evaluation current state of AP/CP residency training using RISE</p>	<p>Molecular pathology, genomic pathology</p>	<ul style="list-style-type: none"> • RISE has 3 sections: AP, CP and Special topics common to AP and CP • Special topics section contained 19 (5%) of molecular pathology questions (e.g.: single genes), 5 genomic pathology questions (e.g.: whole genome NGS analysis, GWAS, terminology, test interpretation, ethical issues) <p>RISE results:</p>	<p>RISE results regarding training duration:</p> <ul style="list-style-type: none"> • Molecular pathology training: <ul style="list-style-type: none"> • Mean time: 2-4 weeks • 68% = 2-4 weeks • 19% = 2-6 months • 7% = zero training • 1% had > 6 months training 	<p>RISE results regarding level of exposure during training for PGY4 pathology residents:</p> <ul style="list-style-type: none"> • Use of molecular pathology during training: Often = 39% (Seldom = 28%, Sometimes = 26%, Never = 6%, Always = 2%) • Use of genomic pathology during training: Sometimes = 32% (Seldom = 29%, Never = 24%, Often = 14%, Always = 1%) 	-	<p>RISE results show that high quality, not just any level, of molecular pathology and genomic pathology training is essential for residents</p>

			<ul style="list-style-type: none"> • In 2013, 2506 AP/CP residents from 143 programs in USA, and 22 programs in other international countries took the pathology RISE • By PGY4, 90% of residents taking the RISE had received some molecular pathology training, 58% had some genomic pathology training • RISE scores in molecular pathology knowledge questions by PGY, showing that AP/CP residents did better year after year: <ul style="list-style-type: none"> • PGY1-4: 470, 504, 530, 554 • RISE scores in genomic pathology knowledge questions by PGY, showing that AP/CP residents' knowledge leveled off at PGY3: <ul style="list-style-type: none"> • PGY1-4: 488, 502, 503, 506 • PGY4 pathology residents' predicted use of molecular pathology as practicing pathologists RISE results: <ul style="list-style-type: none"> • 47% often (21% sometimes, 18% seldom, 9% never, 6% always) • PGY4 pathology residents' predicted use of genomic pathology as practicing pathologists RISE results <ul style="list-style-type: none"> • 29% sometimes (28% often, 25% seldom, 14% never, 4% always) 	<ul style="list-style-type: none"> • Genomic pathology training duration RISE results <ul style="list-style-type: none"> • Mean time: < 1 week • 42% had zero training • 35% had 2-4 weeks • 20% had < 1 week • 3% had 2-6 months • 0% had > 6 months 	<ul style="list-style-type: none"> • Ability to discuss molecular pathology results with clinicians/other providers: 36% good (32% fair, 15% poor, 13% very good, 5% excellent) • Ability to discuss genomic pathology results with clinicians/other providers: 35% good (24% poor, 29% good, 9% very good, 4% excellent) <ul style="list-style-type: none"> • Note, of the 42% of residents who had no genomic pathology training, 25% still reported their ability as being good to excellent when discussing results with clinicians • Note, of the 58% of residents who had some genomic pathology training, 53% of them felt good to excellent when discussing results with clinicians • Rated self as being able to interpret molecular pathology or genomic pathology data in the very good/excellent range: <ul style="list-style-type: none"> • 25% PCR data (molecular pathology) • 18% DNA sequencing data (molecular pathology) • 11% SNPs (combined topics) • 11% microarray data (combined topics) • 9% NGS data • 6% GWAS 		
Mandelker, D., <i>et al.</i> ; 2014; USA 24	Describes pathology informatics fellowship curriculum with a focus on molecular pathology	Molecular pathology, genomics and bioinformatics	<ul style="list-style-type: none"> • Structured curriculum developed for pathology informatics fellows in: • Molecular pathology – customizable component of curriculum • Bioinformatics – required component of curriculum 	2/3 of fellow's time (totaling 1-2 years) for specialization in molecular pathology consists of: <ul style="list-style-type: none"> • Didactic activities • Rotations • Research projects • Mentorships • Clinical activities 	Subtopics of molecular pathology during didactic curriculum of pathology informatics: <ul style="list-style-type: none"> • information fundamentals (e.g.: molecular and genomic information principles/ bioinformatics, info architecture, data manipulation and analysis) • information systems (e.g.: specimen identification systems, EMR, results reporting principles, billing and coding) 	-	<ul style="list-style-type: none"> • Recommend joint fellows in pathology informatics and molecular pathology to collaborate with an institution's bio-informatician to

				<p>Bioinformatics education in core curriculum:</p> <ul style="list-style-type: none"> • 1 week didactics 	<ul style="list-style-type: none"> • workflow process (e.g.: molecular and genomics workflow, software, data storage principles) • governance and management (e.g.: leadership, management, regulations relating to molecular pathology) 		gain valuable practice experience
<p>Flynn, C., <i>et al.</i>; 2014;</p> <p>Ireland</p> <p>22</p>	<p>Describes molecular diagnostic training currently offered to their histopathology trainees in the fellowship program</p>	<p>Molecular pathology/ diagnostics, bio-informatics</p>	<ul style="list-style-type: none"> • Fellowship program incorporates training of histological tissue hybridization-based tests and molecular tests in a lab structure that is well equipped, and has a critical mass of training staff (pathologists, clinician scientists, biomedical scientists, technicians, and bioinformaticians) • 5 year fellowship program where the last year is an optional, further subspecialization year 	<p>Molecular diagnostic topics:</p> <ul style="list-style-type: none"> • Year 1: Introductory lectures (weekly) • Years 2 – 3.5: A mandatory 2-3 month attachment in molecular diagnostics (interactive sessions, practical experience of core competencies, hands-on experience, discussions, trainee performance evaluations) • Year 5: Option 1 – 1 year full time tissue molecular diagnostics; Option 2 – 1 year subspecialty attachment with part time practice and part time reporting molecular tests; Option 3 – mix of molecular diagnostics and research <p>Bioinformatics topics:</p> <ul style="list-style-type: none"> • Year 2 – 3: A 2-3 month attachment in bioinformatics training 	<p>Topics covered during the 2-3 month attachment:</p> <ul style="list-style-type: none"> • Basic principles and knowledge of molecular pathology/biology/diagnostics • Molecular testing – PCR, NGS, DNA/RNA microarrays, whole genome sequencing • Molecular oncology • QA, QC • Regulation • Core skills in DNA extraction <p>Topics covered in 1-year optional fellowship option:</p> <ul style="list-style-type: none"> • Knowledge and skills in core molecular technologies and techniques • Expertise in molecular pathology in various cancers • Sign-out all molecular diagnostic tests requests from the lab • Research, development and innovation in molecular pathology • Laboratory and management of molecular diagnostic laboratory • Training and education 	<p>Concerns remain on how to teach high quality molecular pathology to pathology residents within the 5 year time constraints</p>	-
<p>Schrijver, I., <i>et al.</i>; 2013;</p> <p>USA</p> <p>35</p>	<p>Created the Stanford Open Curriculum in Genomic Medicine for AP/CP residents</p>	<p>Genomic pathology, bio-informatics</p>	<p>Developed the Stanford genomic medicine core curriculum and elective course in advanced genomic medicine developed according to ACGME core competencies</p>	<ul style="list-style-type: none"> • Didactic series of lectures (core) • Interactive learning in small groups (elective) 	<p>Didactic series of 10 lectures in year 1 and 2:</p> <ul style="list-style-type: none"> • Overview of fundamental principles of molecular biology (e.g.: DNA, RNA), clinical genomics (e.g.: microarrays, hybridization analysis, HLA genetics, oncology, pharmacogenetics), commercial personalized medicine, research and clinical applications, bioinformatics 	<p>Most pathology trainees and faculty do not have strong computational programming background/skills required for bioinformatics</p>	<p>Development of subspecialty fellowship training in bioinformatics may become necessary in the</p>

					<p>Elective courses in advanced genomics (not available online)</p> <ul style="list-style-type: none"> • Interactive learning in small groups on computational and statistical methods of DNA sequencing and genomics analysis (NGS, sequence alignment, scripting programming languages, genome assembly) • Targeted to residents who plan on practicing genomic pathology 		future of pathology training
<p>Wieringa, G., <i>et al.</i>; 2012;</p> <p>European Union (includes 27 EU member states)</p> <p>42</p>	<p>Described an updated common curriculum and competencies for clinical chemistry and laboratory medicine residency training across the 27 EU member states</p>	<p>Molecular diagnostics in clinical chemistry</p>	<p>EU curriculum regarding molecular diagnostic competencies for clinical chemists:</p> <ol style="list-style-type: none"> 1. Laboratory medicine – basic knowledge requirements in core knowledge, skills 2. Analytical principles and techniques –core knowledge, innovation, creativity in implementation, maintain up to date knowledge in new development areas in molecular diagnostic technologies and methodologies <p>Research & development, and audit – ability to perform research and development, follow regulations and governance</p>	-	<p>Core knowledge, skills and competencies in: qPCR, mutation detection, cytogenetic analysis, array technologies, DNA sequencing, FISH (see article for a complete and detailed list)</p>	-	-
<p>Haspel, R.L., <i>et al.</i>; 2012;</p> <p>USA</p> <p>17</p>	<p>A needs assessment conducted for a national pathology residency curriculum on molecular pathology</p>	<p>Molecular pathology and genomic pathology</p>	<p>Surveyed 185 pathology residency program directors in the US to conduct a needs assessment for genomic pathology training (23% response rate):</p> <ul style="list-style-type: none"> • 93% of pathology residency programs offered molecular pathology training • 31% of programs had developed curricula in genomic medicine 	<ul style="list-style-type: none"> • Current genomic pathology curriculum consists of 7-10 hours of lectures and exercises to be incorporated into molecular pathology programs • Lectures or journal clubs were dominating teaching methods for genomic medicine in pathology 	<p>Programs that provide genomics training cover the following topics:</p> <ul style="list-style-type: none"> • GWAS, DTC genotyping (e.g.: 23andMe), NGS/ whole-genome sequencing, ethics, genomic variation types, genotype-phenotype correlations, role of genomic medicine in pathology 	<p>Genomic medicine education barriers:</p> <ul style="list-style-type: none"> • Lack of time in current resident schedule (76% of pathology program directors) • Lack of faculty expertise (52% of respondents) 	<p>Pathologists play a key role in patient care and must be prepared to practice genomic medicine, which includes interpreting NGS results and GWAS</p>

	and genomic training		<ul style="list-style-type: none"> 91% of those who do not have genomic medicine training wanted to initiate training in their programs 	<ul style="list-style-type: none"> Pathology residency program directors rated online modules as the most helpful tool in implementing or improving a genomic medicine curriculum 			
McClintock, D.S., <i>et al.</i> ; 2012; USA 25	Describes core curriculum of pathology informatics, which includes a special topic on molecular/genomic pathology	Molecular pathology, genomics, bio-informatics	<ul style="list-style-type: none"> Core didactic course curriculum taught over 2 years during fellowship training, with one subtopic taught per week 	<ul style="list-style-type: none"> Core didactic course format includes primary readings (published in the last 5 years), subtopics for discussion Weekly discussion of readings ranged from 60 – 90 minutes Special pathology information and process domains - Molecular pathology/genomics principles and workflow = 2 weeks total Bioinformatics information process = 1 week 	-	-	-
Tonellato, P.J., <i>et al.</i> ; 2011; USA, but also targets Canada 40	Propose to define and implement 7 “Blue Dot” pilot projects in order to push the national agenda forward to ensure the future of pathology in genomic and personalized medicine	Molecular genomics in pathology	<p>The following are Blue Dot Project relates to molecular diagnostic training for pathology residents</p> <ol style="list-style-type: none"> To establish a nationwide pilot program to ensure that every ACGME approved pathology residency program will include a mandatory genomics and personalized medicine curriculum. 	-	-	-	All practicing pathologists should acquire and demonstrate expertise in molecular genomic medicine technologies, such as NGS and whole genome analysis

<p>Haspel, R.L., <i>et al.</i>; 2010; USA 29</p>	<p>Description of curriculum to train AP/CP pathology residents in NGS and other molecular diagnostic tests</p>	<p>Molecular diagnostics and genomics</p>	<p>Training program includes the following components:</p> <ul style="list-style-type: none"> • Series of didactic presentations focused on basic aspects of NGS and their diagnostic applications, current state of genomic analysis, how genetic counsellors use genetic information to counsel patients and physicians • Pathology residents have opportunity to voluntarily have their own genomes sequenced analyzed using NGS <p>For full details of curriculum, see online supplement ³⁰</p>	<ul style="list-style-type: none"> • Didactic presentations on NGS • Hands-on - practice on one-self by using NGS to voluntarily sequence their own (residents') genomes for potential disease associations • Ensure residents understand DNA and RNA sequencing data and analysis in the clinical laboratory • Research - Residents are encouraged to engage in translational research activities that apply NGS technologies in molecular diagnostics 	<p>-</p>	<p>Molecular genomics curriculum created for pathology residents to address the large gap in this training area in medical education</p>	<p>The ultimate goal is to help ensure by July 2012 that every pathology residency program in USA and Canada will implement a personal genomics curriculum so that pathology residents will develop core competencies in this field</p>
<p>Haspel, R.L., <i>et al.</i>; 2010; USA 30</p>	<p>Full description of NGS and molecular genomics curriculum for AP/CP pathology residency training programs</p>	<p>Molecular diagnostics and genomics</p>	<p>Curriculum objectives/ teaching modality:</p> <ul style="list-style-type: none"> • Lecture 1 – clinical and educational genomics (e.g.: DTC genotyping) • Lecture 2 – NGS • Lecture 3 – genetic counselling and personalized genomics • Offering personalized genotyping kits to residents for those who voluntarily want to participate (discussion of implications regardless) <p>Resident presentations on genome-wide association studies</p>	<ul style="list-style-type: none"> • Orientation session and lectures (3 hours total) occur in the 1st month • Residents given opportunity to perform personalized genotyping testing and present risk factors in months 2 – 3 • Residents prepare presentation with faculty mentor in the 4th month • Resident presentation and discussion of genomics and personalized medicine occur in the 5th month of the curriculum • Lecture based – residents attend three 1-hour lectures on personalized genomics, high-throughput sequencing, 	<p>-</p>	<p>-</p>	<p>-</p>

				<p>and genetic counselling (lectures available on www.genomicmedicineinitiative.org)</p> <ul style="list-style-type: none"> • Interpretation of NGS and GWAS data and results 			
<p>Talbert, M.L., <i>et al.</i>; 2009; USA 23</p>	<p>Detailed report of guidelines, goals and objectives for competency-based molecular genetic pathology fellowship training in US</p>	<p>Molecular genetic pathology</p>	<p>Targets ACGME requirements for competency and evaluation of molecular pathology fellows (but also works for pathology residents exposed to molecular pathology techniques) and includes:</p> <ul style="list-style-type: none"> • Description of core competency • Summary of goals • Tools to assess each competency (e.g.: testing, evaluation instrument, checklist evaluation, portfolio, critically appraised topics, presentation skills) • Sample evaluation questions for assessment 	-	<p>Patient care</p> <ul style="list-style-type: none"> • Through use of molecular genetic tools, diagnostics, patient information, and consultation <p>Medical knowledge</p> <ul style="list-style-type: none"> • Acquire and apply principles and practice of molecular genetic pathology (basic science, methodologies (PCR, NGS, FISH), lab management, clinical correlations) <p>Practice-based learning and improvement</p> <ul style="list-style-type: none"> • Ability to investigate and evaluate diagnostic and consultative practices, review scientific evidence, improve patient care practices <p>Interpersonal and communication skills</p> <ul style="list-style-type: none"> • Perform effective communication regarding molecular diagnostics with other healthcare providers, patients and their families, lab personnel <p>Professionalism</p> <p>Systems-based practice</p> <ul style="list-style-type: none"> • Provide guidance to clinicians and genetic counsellors to ensure molecular testing is used and integrated into patient care appropriately, and in a cost-effective manner 	-	<ul style="list-style-type: none"> • Design and structure of evaluation tools and programs will depend on the program itself, but institutions should consult with Pathology Residency Coordinators or Program Directors to obtain specific examples and formats
<p>Genzen, J.R. and Krasowski, M.D.; 2007; USA</p>	<p>Surveyed faculty practicing clinical chemistry, CP or AP regarding</p>	<p>Molecular pathology in clinical chemistry</p>	<p>In the US, clinical chemistry is trained one of two ways: as an MD (AP/CP can enter clinical chemistry after completion of residency), or PhD</p> <p>Survey results:</p> <ul style="list-style-type: none"> • 38.5% response rate 	-	<p>At some of the institutions surveyed, clinical chemistry residents review and sign-out molecular diagnostic tests</p>	-	<ul style="list-style-type: none"> • One respondent commented on that teaching pathology residents has become easier as molecular

27	future of pathology training in subspecialties of clinical chemistry		<ul style="list-style-type: none"> 35% of 52 respondents surveyed commented that expanding clinical chemistry training to include molecular techniques and genomic technologies will pose both challenges and opportunities for clinical chemistry residents 				<p>testing has been observed in many areas of pathology and medicine</p> <p>Another respondent does not believe there is a future for clinical chemistry training for pathology residents, but rather for science graduates with a molecular background</p>
Haller, B.L.; 2007; USA 28	Discussion of challenges, rewards and assessing methods of competence in clinical microbiology training in CP residency programs	Molecular diagnostics in infectious diseases	In the US, training and certification in clinical microbiology can follow the completion of MD (upon CP or AP + CP residency completion) or PhD, where the ABP offers subspecialty certification in medical microbiology	-	<ul style="list-style-type: none"> Instruction on new technologies, such as molecular testing, use of rapid tests in diagnosis of infectious diseases, point-of-care testing, and advantages/ limitations of each of these molecular diagnostic tests would contribute to resident learning Clinical microbiologists must be up to date on new molecular methodologies, including qPCR, microarrays, rapid assays for detection of antigens or antibodies 	-	-
Peerschke, E.I., <i>et al.</i> ; 2007; USA 34	Propose research training guidelines for AP and/or CP residents in laboratory medicine	Molecular biology, genomics, bioinformatics	Regarding subspecialty molecular training, propose didactic lecture series and practical laboratory exercises in addition of structured research experiences that CP residency programs do not normally offer in the areas of genomic pathology and bioinformatics	<ul style="list-style-type: none"> Didactic lectures Hands-on practical lab experiences Research 	<p>Didactic lecture series topics:</p> <ul style="list-style-type: none"> Genomics (microarray, gene expression profiling) Molecular biology Bioinformatics <p>Practical lab exercises/hands-on experience that apply to clinical practice and/or patient management:</p>	<ul style="list-style-type: none"> Requires experienced faculty A high faculty-trainee ratio Access to large and diverse patient population Requires advanced diagnostic facilities 	-

					<ul style="list-style-type: none"> • Common molecular techniques (PCR, microarray) • Data analysis • Evaluation or development of new methodologies <p>Research – focus on practical clinical-based research, 4-6 month block in 4th year</p>		
Smith, B.R., <i>et al.</i> ; 2006; USA 38	Propose CP curriculum that defines goals and objectives for training, as they align with ACGME guidelines	Molecular pathology, cytogenetics	Propose the last 6 months of training in 24 months of level I and II CP on molecular pathology/diagnostics (which includes cytogenetics) that aligns with ACGME guidelines that states CP training must include molecular biological techniques, cytogenetics and other advanced techniques as they become available	1-2 months of molecular diagnostics (includes cytogenetics) rotation during the last 6 months of elective time in CP	<p>Molecular pathology</p> <ul style="list-style-type: none"> • knowledge of specific molecular pathology tests (skill level I and II) • analytical and technical training (skill level I and II) • consultation and presentation of cases (skill level II) • ACGME core competencies <p>Cytogenetics</p> <ul style="list-style-type: none"> • knowledge of specific cytogenetic tests (skill levels I and II) • analytical and technical training (skill level I) • consultation and presentation of cases (skill level II) 	-	Individual pathology programs may have different approaches as to how to educate these areas, depending on their structure
Association of Directors of Anatomical and Surgical Pathology; 2003; USA 37	A suggested curriculum and competency proposal for AP residency	Molecular diagnostics and cytogenetics	<p>The proposed curriculum and guidelines to assess competence in the form of a template for others to use and includes the following areas:</p> <ul style="list-style-type: none"> • Basic learning objectives • Basic programmatic expectations of resident • Specific skills that apply to all areas of AP (including molecular pathology and cytogenetics) • Basic schedule of rotation in AP • Competencies in pathology <p>Documentation in resident performance</p>	Molecular diagnostics and cytogenetic training begins after residents enter into AP track, and are incorporated into other rotations (e.g.: during advanced cytopathology)	<p>Molecular diagnostics and cytogenetics training in skill level I:</p> <ul style="list-style-type: none"> • Fundamental principles of molecular biology and cytogenetics in a variety of subjects (e.g.: infectious disease, oncology, hematology) • Describe various disorders commonly studied by molecular biology and cytogenetic techniques • Knowledge of routine molecular pathology and cytogenetic methods • Understand QC, quality improvement, lab management relating to molecular pathology and cytogenetics • Perform scientific literature search and critically assess new molecular tests 	Lack of published guidelines on how to monitor resident performance each year	Anticipate improvement and refinement of formal assessments in the future, similar to how other institutions have already developed comprehensive evaluation forms that incorporates competencies listed (completed by faculty at the

					<ul style="list-style-type: none"> • Observe and perform molecular biology tests (PCR, FISH, karyotyping) • Communicate with and consult requesting physicians on utilization of molecular pathology tests • Review and interpret molecular pathology and cytogenetic test results, prepare reports 		end of each rotation)
Katzman, P.J., <i>et al.</i> ; 2003; USA 31	Description of pediatric pathology residency rotations, including molecular diagnostic education, that involves innovative multidisciplinary approaches to education	Molecular diagnostics in pediatric pathology	Since the fall of 1999, every 6 months, 1-2 pathology residents are scheduled for 2 month antenatal rotation, where molecular diagnostic training is included	Lab bench exercises, didactics and clinical experience	<p>Lab bench exercises:</p> <ul style="list-style-type: none"> • PCR to identify cytomegalovirus in paraffin-embedded tissue samples • Conventional karyotyping • Evaluation of FISH in cytogenetics lab • Molecular testing for genetic diseases <p>Didactics:</p> <ul style="list-style-type: none"> • Molecular testing for cystic fibrosis <p>Clinical experiences:</p> <ul style="list-style-type: none"> • Cytogenetics lab to perform karyotyping • FISH to identify Down Syndrome, Trisomy 18, CATCH 22, etc. • Molecular testing for cystic fibrosis 	-	Authors mentioned that the written resident performance evaluation form that divides evaluation into 6 core competencies was under development and will be used for all residency programs in the future
The AMP Training and Education Committee; 1999; USA 18	Outlines general goals and recommendations for molecular pathology education in AP/CP residency programs	Molecular pathology	Major areas of molecular pathology that should be covered in rotations – infectious disease testing, molecular oncology, inherited disorders, histocompatibility, identification determination, clinical correlation, laboratory management	<ul style="list-style-type: none"> • Molecular pathology residency programs should include combination of didactic sessions, rotations within molecular pathology labs, participation in conferences, clinical service experiences • Didactic sessions may substitute for practical lab experiences for training institutions that lack molecular pathology service 	Residents should understand: <ul style="list-style-type: none"> • general concepts in basic technologies, genetics, instrumentation in molecular pathology • principles and practical aspects of molecular diagnostic applications • limitations of specific assays and molecular tests, test specificity and sensitivity • costs associated with molecular pathology testing 	<ul style="list-style-type: none"> • Lack of guidance available on important components of molecular pathology training 	<ul style="list-style-type: none"> • Programs that have at least 3 molecular pathology subject areas are likely to have adequate case material for resident education (programs with less than that may have difficulty developing an

							educational program with adequate breadth)
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Abbreviations: AMP – American Board of Pathology, ACGME – Accreditation Council of Graduate Medical Education, AMP – Association of Molecular Pathology, AP – Anatomical Pathology, CP – Clinical Pathology, DTC – Direct-To-Consumer, EMR – Electronic Medical Records, EU – European Union, FISH – Fluorescent In-Situ Hybridization, GWAS – Genome-Wide Association Study, NGS – Next Generation Sequencing, PGY – Post-Graduate Year, QA – Quality Assurance, QC – Quality Control, qPCR – Quantitative (real-time) Polymerase Chain Reaction, RCPATH – Royal College of Pathologists, RISE – Resident In-Service Exam, SNP – Single Nucleotide Polymorphisms, TAT – Turn Around Time

