Tutorial and Laboratory Visit - Anatomical Pathology

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Learning Objectives

- To observe workflow in an anatomical pathology laboratory (video)

- To understand applications of anatomical pathology tests with examples

Anatomical Pathology

- Surgical Pathology

  (including Frozen Section))
- Cytology

  (including Fine Needle Aspiration)
- Autopsy
- Cytogenetics or Molecular Analysis

Surgical Pathology

- All embedded of small specimens
- Gross examination and block taking from surgical excision specimens
- Tissue processing to produce Haematoxylin – eosin stained sections for microscopic examination

The first step of Surgical Pathology

- All embedded of small specimens

- https://hkuhk-my.sharepoint.com/:v:/g/personal/anycheun_hku_hk/EZGs8sG0eItEjSr3iohaM9cBqVpS8LF0uiZCoMqpSH3iPA?e=Zq1B8H

The first step of Surgical Pathology

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The first step of Surgical Pathology

- Tissue processing to produce Haematoxylin – eosin stained sections for microscopic examination

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Cytology

- **Exfoliative Cytology**

e.g. Cervical cytology, pleural fluid

- **Fine Needle Aspiration Cytology**

Special Techniques

- Special stains
- Immunohistochemistry
- Immunofluorescence
- In situ hybridization
- Electron microscopy
- PCR
- Microsattelite analysis
- Sequencing
- NGS …

History

- F/40
- Known SLE for 20 yr on oral methylprednisolone 10 mg q24h; MMF 1gm q24h for 5 yr;
- Complained for uncontrolled speech; poor sleep; then speechless & somnolence;
- admitted to 2 other hospitals & Rx by steroid, antibiotics, antifungal

Mycobacterial Histiocytosis (Mycobacterial spindle cell pseudotumour)

Lower limb Skin biopsy

Small Round Cell Tumours

- Diverse group of neoplasms
- Primitive or embryonal in appearance
- More common in children, adolescents and young adults
- Involve skeletal system and soft tissue
- Differential diagnostic challenge

Differential Diagnoses

- Lymphoma
- Ewing Sarcoma …
- Melanoma
- Oat (Small) cell carcinoma
- Neuroblastoma
- Rhabdomyosarcoma
- Osteosarcoma
- Synovial sarcoma
- ……..

Diagnostic Parameters

- Demographic features
- Primary site
- Metastatic pattern
- Microscopic features
- Immunohistochemistry profile
- Ultrastructural features
- Genetic findings

Clues

Definitive diagnosis

Molecular Study

- RT-PCR
  - t(11;22)(p13;q12)
  - EWS-WT1

Diagnosis

- **CYSTIC DUCT tumour, Whipple operation:**

  DESMOPLASTIC SMALL ROUND CELL TUMOUR.

Next-generation sequencing of *EWSR1-FLI1*

- Next-generation RNA sequencing of Ewing sarcoma demonstrates in-frame fusion between *EWSR1* & *FLI1* gene products.
- NGS is more specific than *EWSR1* break-apart FISH because it provides detailed information about both translocation partners.

WHO Classification of Soft Tissue and BoneTumours 5th Ed

WHO classification of tumours of haematopoietic and lymphoid tissues 2017

Myeloproliferative neoplasms
Chronic myeloid leukaemia, *BCR-ABL1*–positive 9875/3
Chronic neutrophilic leukaemia 9963/3
Polycythaemia vera 9950/3
Primary myelofibrosis 9961/3
Essential thrombocythaemia 9962/3
Chronic eosinophilic leukaemia, NOS 9964/3
Myeloproliferative neoplasm, unclassifiable 9975/3

Mastocytosis
Cutaneous mastocytosis 9740/1
Indolent systemic mastocytosis 9741/1
Systemic mastocytosis with an associated haematological neoplasm 9741/3
Aggressive systemic mastocytosis 9741/3
Mast cell leukaemia 9742/3
Mast cell sarcoma 9740/3

Myeloid/lymphoid neoplasms with eosinophilia and gene rearrangement
Myeloid/lymphoid neoplasms with *PDGFRA* rearrangement 9965/3
Myeloid/lymphoid neoplasms with *PDGFRB* rearrangement 9966/3
Myeloid/lymphoid neoplasms with *FGFR1* rearrangement 9967/3
Myeloid/lymphoid neoplasms with PCM1-JAK2 9968/3*

Myelodysplastic/myeloproliferative neoplasms
Chronic myelomonocytic leukaemia 9945/3
Atypical chronic myeloid leukaemia, *BCR-ABL1*–negative 9876/3
Juvenile myelomonocytic leukaemia 9946/3
Myelodysplastic/myeloproliferative neoplasm with ring sideroblasts and thrombocytosis 9982/3
Myelodysplastic/myeloproliferative neoplasm, unclassifiable 9975/3

Myelodysplastic syndromes
Myelodysplastic syndrome with single lineage dysplasia 9980/3
Myelodysplastic syndrome with ring sideroblasts and single lineage dysplasia 9982/3
Myelodysplastic syndrome with ring sideroblasts and multilineage dysplasia 9993/3*
Myelodysplastic syndrome with multilineage dysplasia 9985/3
Myelodysplastic syndrome with excess blasts 9983/3
Myelodysplastic syndrome with isolated del(5q) 9986/3
Myelodysplastic syndrome, unclassifiable 9989/3
Refractory cytopenia of childhood 9985/3

Myeloid neoplasms with germline predisposition
Acute myeloid leukaemia with germline *CEBPA* mutation
Myeloid neoplasms with germline *DDX41* mutation
Myeloid neoplasms with germline *RUNX1* mutation
Myeloid neoplasms with germline *ANKRD26* mutation
Myeloid neoplasms with germline *ETV6* mutation
Myeloid neoplasms with germline *GATA2* mutation

Acute myeloid leukaemia (AML) and related precursor neoplasms

AML with recurrent genetic abnormalities
AML with t(8;21)(q22;q22.1); *RUNX1-RUNX1T1* 9896/
AML with inv(16)(p13.1q22) or t(16;16)(p13.1;q22); *CBFB-MYH11* 9871/
Acute promyelocytic leukaemia with *PML-RARA* 9866/
AML with t(9;11)(p21.3;q23.3); *KMT2A-MLLT3* 9897/
AML with t(6;9)(p23;q34.1); *DEK-NUP214* 9865/
AML with inv(3)(q21.3q26.2) or t(3;3)(q21.3;q26.2); *GATA2, MECOM* 9869/
AML (megakaryoblastic) with t(1;22)(p13.3;q13.1); *RBM15-MKL1* 9911/
AML with BCR-ABL1 9912/
AML with mutated *NPM1* 9877/
AML with biallelic mutation of *CEBPA* 9878/
AML with mutated RUNX1 9879/

AML with myelodysplasia-related changes 9895/

Therapy-related myeloid neoplasms 9920/

Acute myeloid leukaemia, NOS 9861/
AML with minimal differentiation 9872/
AML without maturation 9873/
AML with maturation 9874/
Acute myelomonocytic leukaemia 9867/
Acute monoblastic and monocytic leukaemia 9891/
Pure erythroid leukaemia 9840/
Acute megakaryoblastic leukaemia 9910/
Acute basophilic leukaemia 9870/
Acute panmyelosis with myelofibrosis 9931/

Myeloid sarcoma 9930/

Myeloid proliferations associated with Down syndrome
Transient abnormal myelopoiesis associated with Down syndrome 9898/1
Myeloid leukaemia associated with Down syndrome 9898/3

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The videos on anatomical pathology processing are kindly provided by Mr. Samuel Tat-fai LUK, Queen Mary Hospital