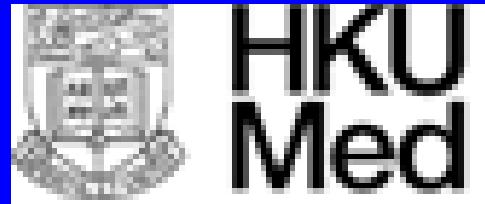


Tutorial and Laboratory Visit - Anatomical Pathology

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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))

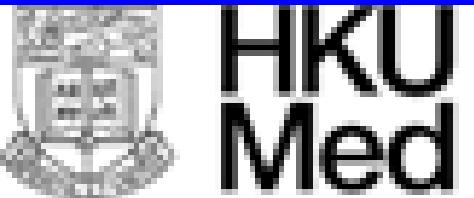
- **Cytology**
(including Fine Needle Aspiration)

- **Autopsy**

- **Cytogenetics or Molecular Analysis**

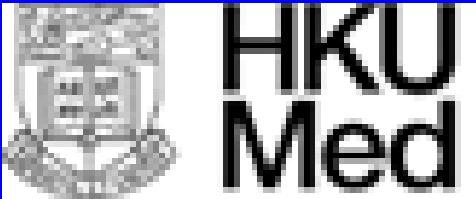
Surgical Pathology

- 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



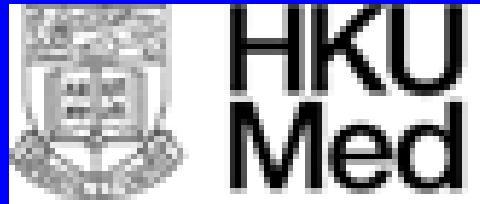
The first step of Surgical Pathology

- All embedded of small specimens
- https://hkuhk-my.sharepoint.com/:v/g/personal/anycheun_hku_hk/EZGs8sG0eltEjSr3iohaM9cBqVpS8LF0uiZCoMqpSH3iPA?e=Zq1B8H



The first step of Surgical Pathology

- Gross examination and block taking from surgical excision specimens
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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

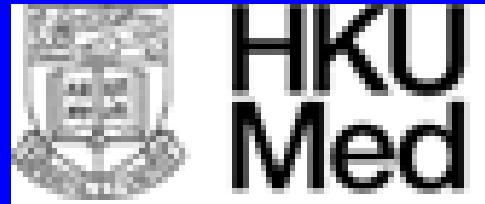
- Exfoliative Cytology
 - e.g. Cervical cytology, pleural fluid
- Fine Needle Aspiration Cytology

Special Techniques

- Special stains
- Immunohistochemistry
- Immunofluorescence
- In situ hybridization
- Electron microscopy
- PCR
- Microsatellite 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

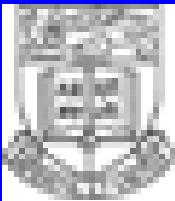


Mycobacterial Histiocytosis (Mycobacterial spindle cell pseudotumour)

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



**HKU
Med**

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 tumours of haematopoietic and lymphoid tissues 2017

Myeloproliferative neoplasms		Myeloid neoplasms with germline predisposition
Chronic myeloid leukaemia, <i>BCR-ABL</i> 1-positive	9875/3	Acute myeloid leukaemia with germline <i>CEBPA</i> mutation
Chronic neutrophilic leukaemia	9963/3	Myeloid neoplasms with germline <i>DDX41</i> mutation
Polycythaemia vera	9950/3	Myeloid neoplasms with germline <i>RUNX1</i> mutation
Primary myelofibrosis	9961/3	Myeloid neoplasms with germline <i>ANKRD26</i> mutation
Essential thromocythaemia	9962/3	Myeloid neoplasms with germline <i>ETV6</i> mutation
Chronic eosinophilic leukaemia, NOS	9964/3	Myeloid neoplasms with germline <i>GATA2</i> mutation
Myeloproliferative neoplasm, unclassifiable	9975/3	
Mastocytosis		Acute myeloid leukaemia (AML) and related precursor neoplasms
Cutaneous mastocytosis	9740/1	
Indolent systemic mastocytosis	9741/1	AML with recurrent genetic abnormalities
Systemic mastocytosis with an associated haematological neoplasm	9741/3	AML with t(8;21)(q22;q22.1); <i>RUNX1-RUNX1T1</i>
Aggressive systemic mastocytosis	9741/3	AML with inv(16)(p13.1q22) or t(16;16)(p13.1;q22); <i>CBFB-MYH11</i>
Mast cell leukaemia	9742/3	Acute promyelocytic leukaemia with <i>PML-RARA</i>
Mast cell sarcoma	9740/3	AML with t(9;11)(p21.3;q23.3); <i>KMT2A-MLLT3</i>
Myeloid/lymphoid neoplasms with eosinophilia and gene rearrangement		AML with t(6;9)(p23;q34.1); <i>DEK-NUP214</i>
Myeloid/lymphoid neoplasms with <i>PDGFRA</i> rearrangement	9965/3	AML with inv(3)(q21.3;q26.2) or t(3;3)(q21.3;q26.2); <i>GATA2, MECOM</i>
Myeloid/lymphoid neoplasms with <i>PDGFRB</i> rearrangement	9966/3	AML (megakaryoblastic) with t(1;22)(p13.3;q13.1); <i>RBM15-MKL1</i>
Myeloid/lymphoid neoplasms with <i>FGFR1</i> rearrangement	9967/3	AML with <i>BCR-ABL1</i>
Myeloid/lymphoid neoplasms with <i>PCM1-JAK2</i>	9968/3*	AML with mutated <i>NPM1</i>
Myelodysplastic/myeloproliferative neoplasms		AML with biallelic mutation of <i>CEBPA</i>
Chronic myelomonocytic leukaemia	9945/3	AML with mutated <i>RUNX1</i>
Atypical chronic myeloid leukaemia, <i>BCR-ABL</i> 1-negative	9876/3	AML with myelodysplasia-related changes
Juvenile myelomonocytic leukaemia	9946/3	
Myelodysplastic/myeloproliferative neoplasm with ring sideroblasts and thrombocytosis	9982/3	Therapy-related myeloid neoplasms
Myelodysplastic/myeloproliferative neoplasm, unclassifiable	9975/3	
Myelodysplastic syndromes		Acute myeloid leukaemia, NOS
Myelodysplastic syndrome with single lineage dysplasia	9980/3	AML with minimal differentiation
Myelodysplastic syndrome with ring sideroblasts and single lineage dysplasia	9982/3	AML without maturation
Myelodysplastic syndrome with ring sideroblasts and multilineage dysplasia	9993/3*	AML with maturation
Myelodysplastic syndrome with multilineage dysplasia	9995/3	Acute myelomonocytic leukaemia
Myelodysplastic syndrome with excess blasts	9983/3	Acute monoblastic and monocytic leukaemia
Myelodysplastic syndrome with isolated del(5q)	9986/3	Pure erythroid leukaemia
Myelodysplastic syndrome, unclassifiable	9989/3	Acute megakaryoblastic leukaemia
Refractory cytopenia of childhood	9985/3	Acute basophilic leukaemia
		Acute panmyelosis with myelofibrosis
		Myeloid sarcoma
		Myeloid proliferations associated with Down syndrome
		Transient abnormal myelopoiesis associated with Down syndrome
		Myeloid leukaemia associated with Down syndrome

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*
- AML with inv(16)(p13.1q22) or

t(16;16)(p13.1;q22); *CBFB-MYH11*

- Acute promyelocytic leukaemia with *PML-RARA*

AML with t(9;11)(p21.3;q23.3); *KMT2A-MLLT3*

- AML with t(6;9)(p23;q34.1); *DEK-NUP214*

AML with inv(3)(q21.3q26.2) or

t(3;3)(q21.3;q26.2); *GATA2, MECOM*

AML (megakaryoblastic) with

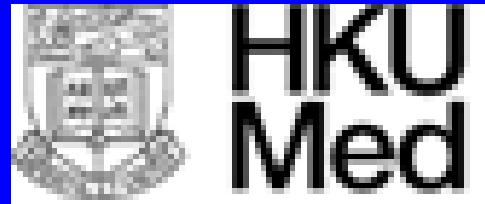
t(1;22)(p13.3;q13.1); *RBM15-MKL1*

AML with *BCR-ABL1*

AML with mutated *NPM1*

AML with biallelic mutation of *CEBPA*

AML with mutated *RUNX1*



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

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