Case 6

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Presentation

- 15-year-old male
- Transferred from the outside area – limited clinical info
- PMH: Autism spectrum disorder, ADHD with no previous hospital admissions

Full Blood Count

Test	Result	Reference Range
Haemoglobin	**105**	127 - 170 g/L
WBC	4.90	3.9 - 9.9 $x10^9$/L
Platelets	**45**	150 – 450 $x10^9$/L
HCT	0.308	0.37 – 0.49 L/L
MCV	**97.9**	77.0 – 95.6 fL
MCH	32.8	25 – 33 pg
RDW	**19.1**	12 – 14.7 %
Neutrophils	3.14	1.7 – 5.7 $x10^9$/L
Lymphocytes	1.60	1.4 – 3.8 $x10^9$/L
Monocytes	0.16	0.15 – 1.28 $x10^9$/L
ESR	2	1 – 8 mm/hr
INR	**1.7**	
APTT ratio	**1.66**	

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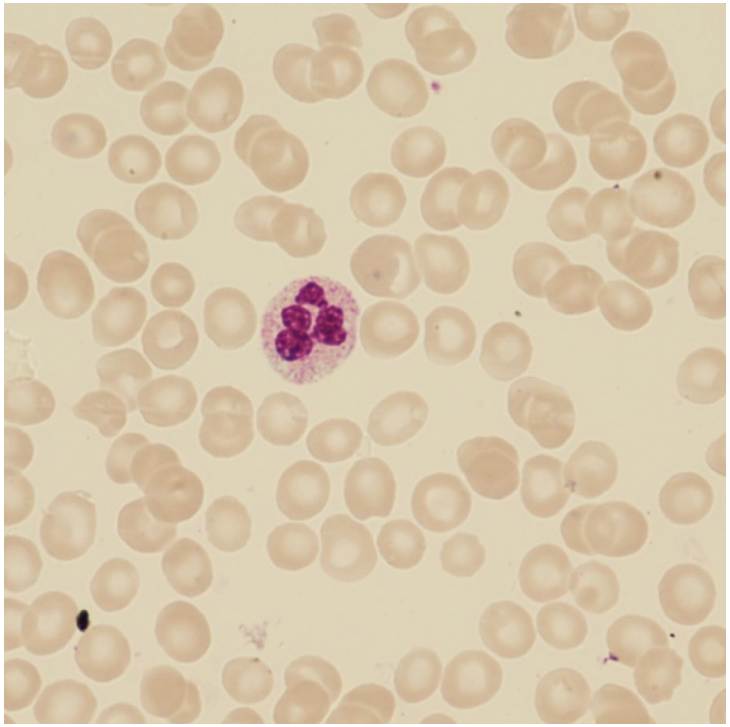
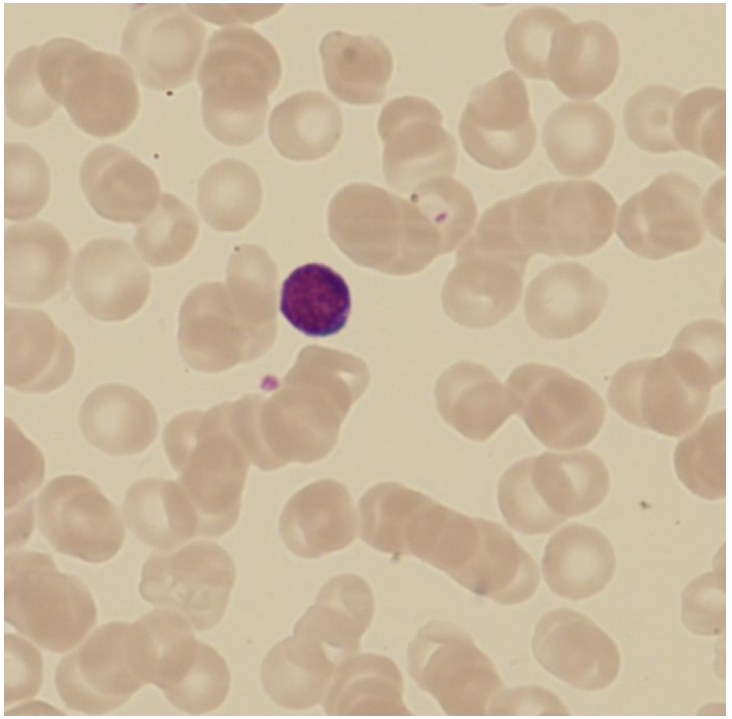
Biochemistry

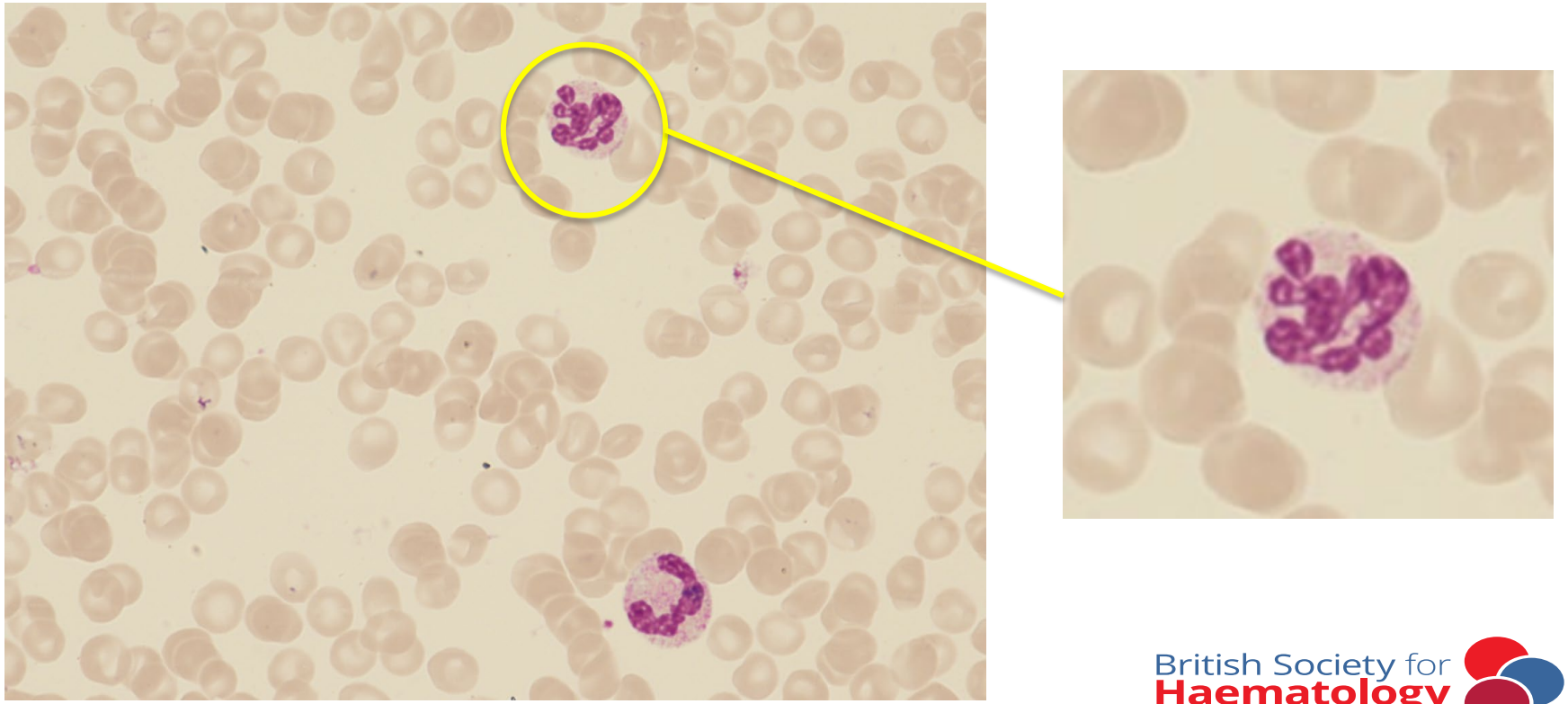
Test	Result	Reference Range
Sodium	139	133 – 146 mmol/L
Potassium	3.8	3.5 – 5.0 mmol/L
Urea	**10.4**	2.5 – 6.5 mmol/L
Creatinine	**197**	47 – 98 µmol/L
Bilirubin	**28**	0 – 21 µmol/L
ALT	**415**	9 – 22 IU/L

Blood Film x60





Blood Film x60

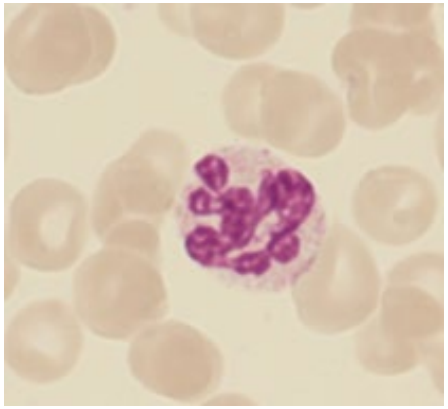


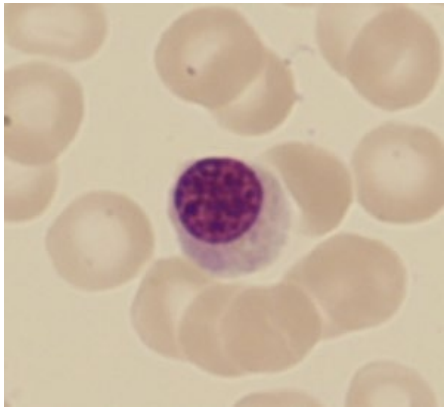
1. What is your suspected diagnosis?

a) Normal
b) Iron Deficiency Anaemia
c) B12 or Folate Deficiency
d) Mixed Iron & B12/Fol Deficiency
e) Need further information

Blood Film Findings

- Occasional hypersegmented neutrophils present
- Thrombocytopenia confirmed
- 1 solitary NRBC
- No blasts
- No red cell fragments





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Further Presentation History

- Initially presented with leg ache and tiredness 3 months earlier
- Unusual gait with increasing difficulty in moving arms and legs with painful calves
- Also, onset scrotal soreness and peeling rash – treated with unknown steroid cream

2. What further tests would you request?

a) Vitamin D
b) B12, Folate & Iron Studies
c) Creatine Kinase
d) All of the above
e) Others not listed

Clinical History contd.

- Worsening vomiting symptoms and slurred speech with unsafe swallow
- Maculopapular rash on legs, feet and bilateral cheeks – occasionally developing on chest and neck
- Patient admitted to PICU and intubated

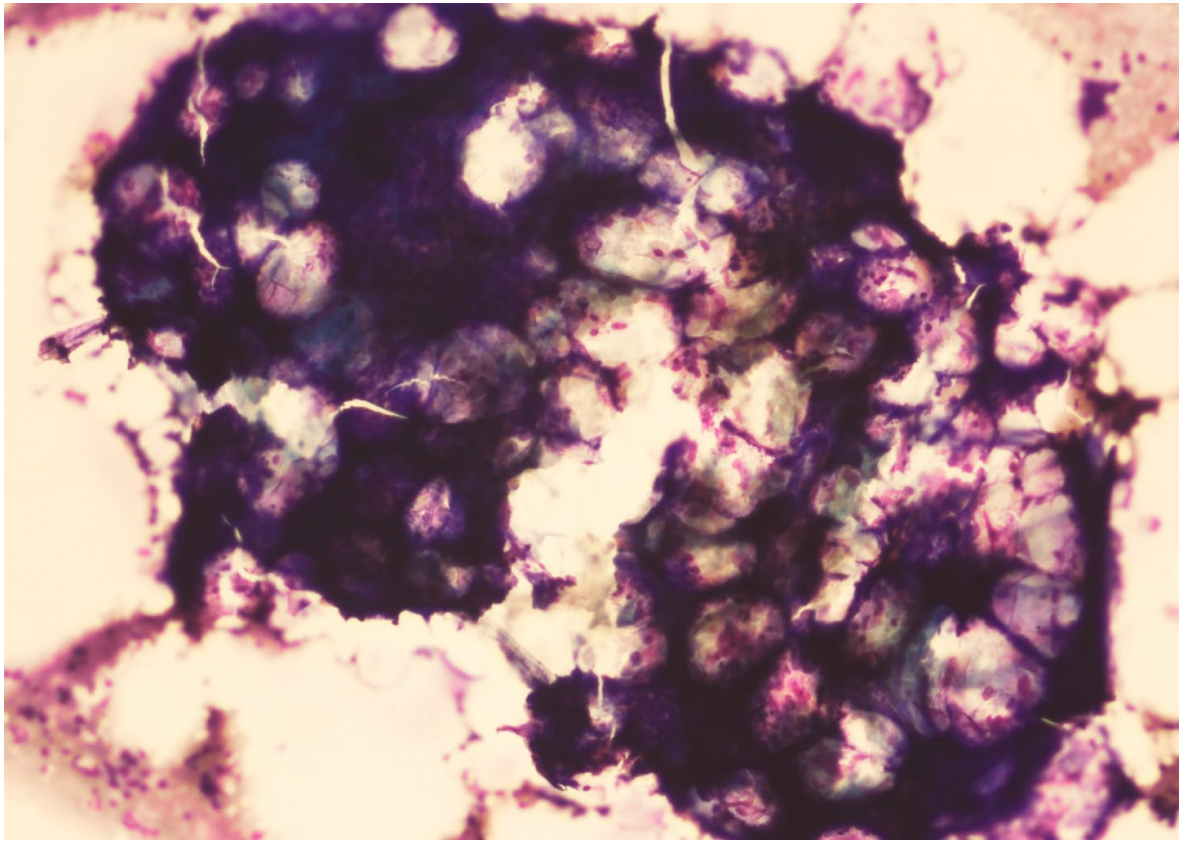
Further Biochemistry

Test	Result	Reference Range
CK	**4824**	90 – 354 IU/L
CRP	**193**	0 – 5 mg/L
B12	**152**	211 – 911 ng/L
Folate	Haemolysed	
Ferritin	**1723**	15 – 200 µg/L
Vitamin D	**<8**	50.1 – 200 nmol/L
Zinc	**7.2**	10.7 – 18.4 µmol/L
Copper	16.7	11 – 24.4 µmol/L

3. What is your revised diagnosis?

a) Sepsis
b) Rhabdomyolysis
c) Nutritional deficiencies
d) Macrophage Activation Syndrome
e) Need further information

Bone marrow aspirate x 10



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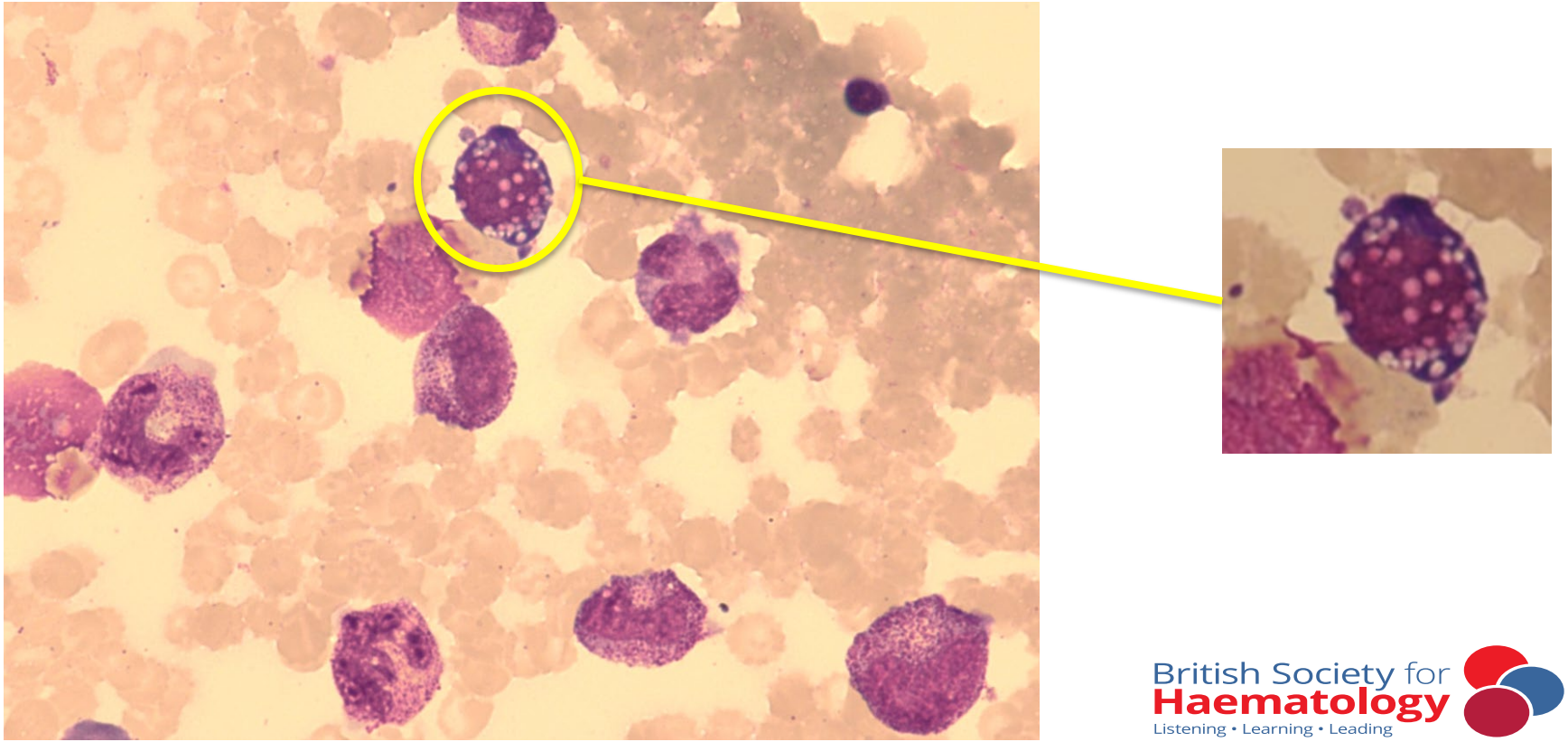
Bone marrow aspirate x 60



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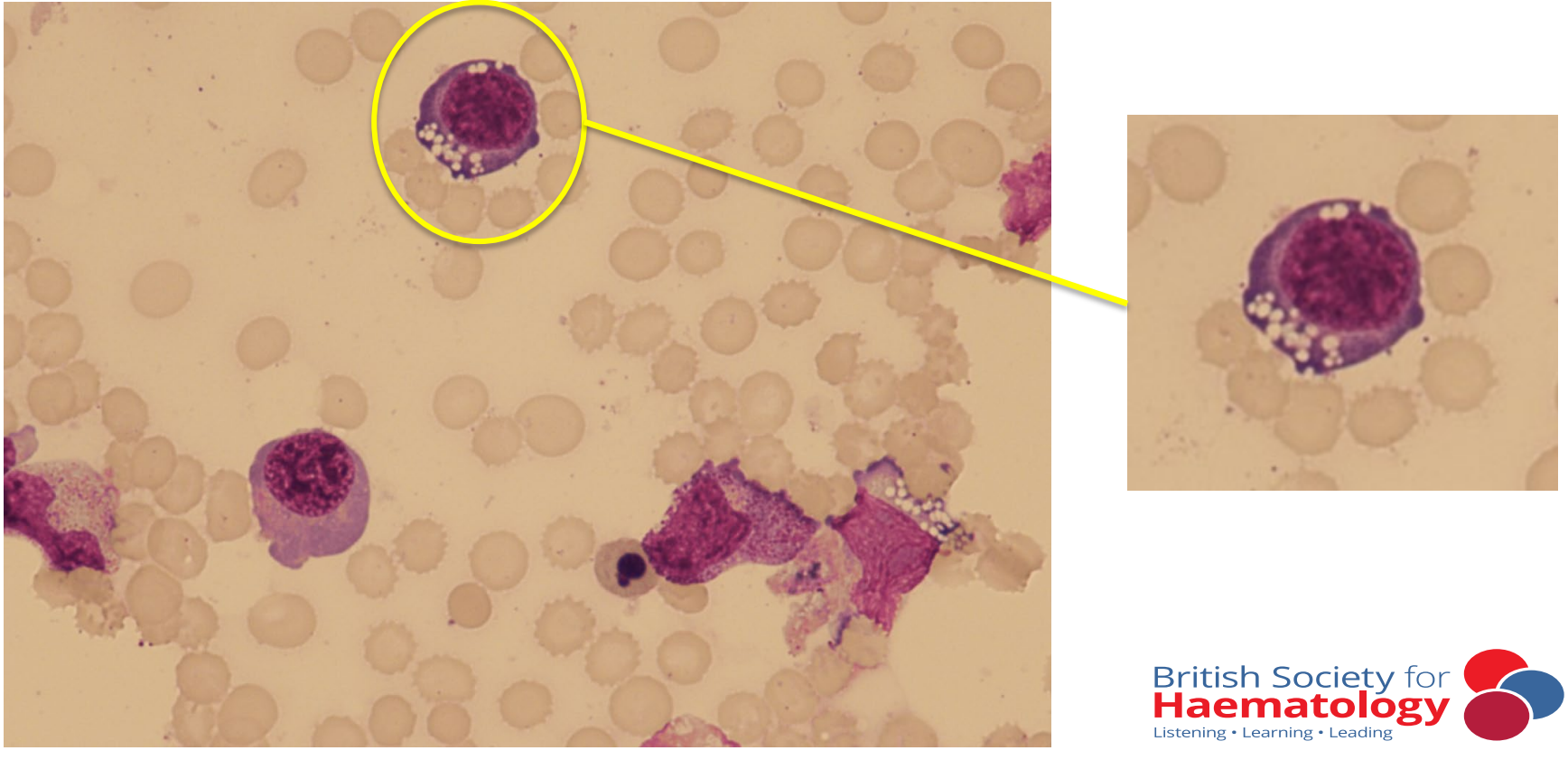
Bone marrow aspirate x60



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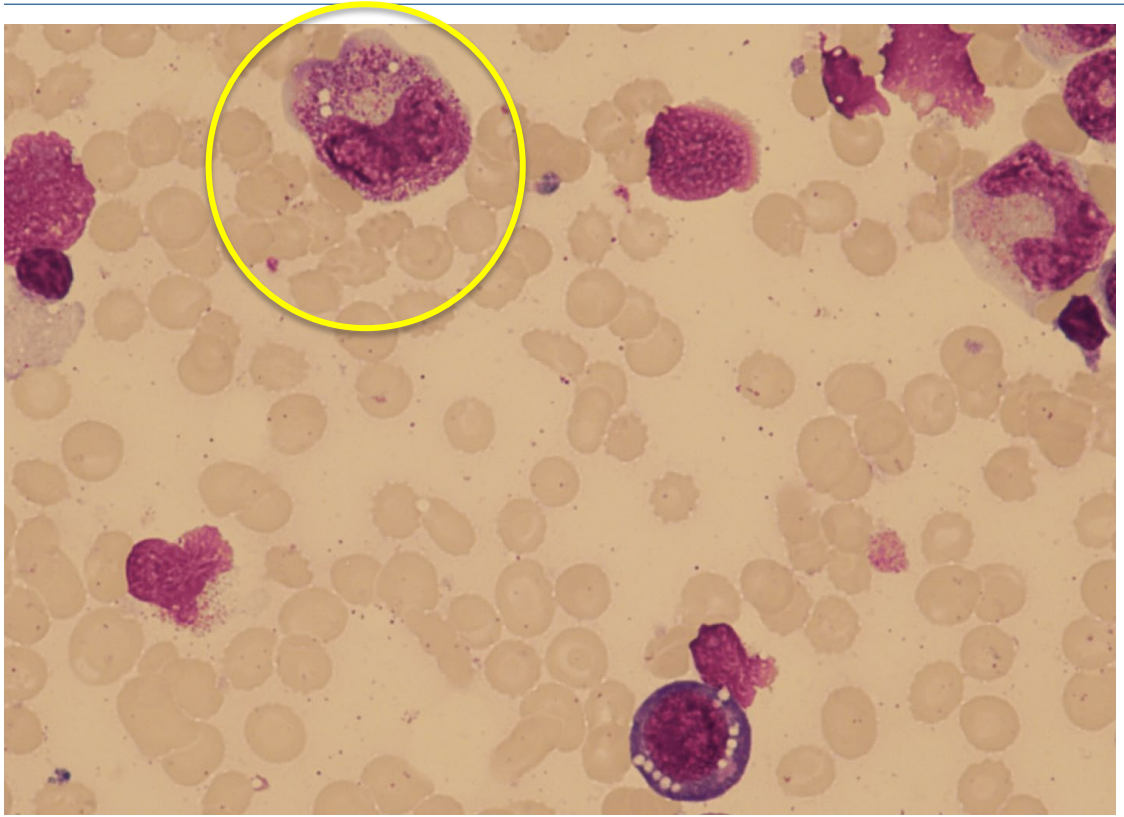
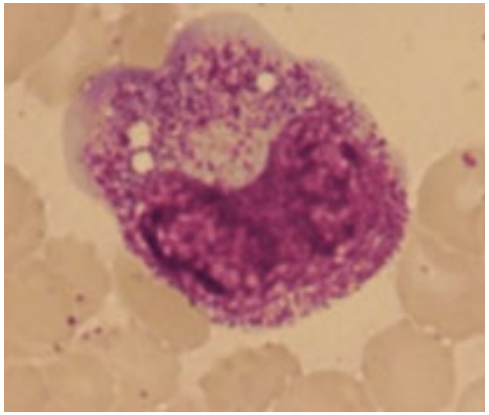
Bone marrow aspirate x60





4. What is your suspected final diagnosis?

a) Nutritional Deficiencies
b) Juvenile Dermatomyositis & Nutritional Deficiencies
c) Viral Myositis
d) VEXAS
e) Macrophage Activation Syndrome

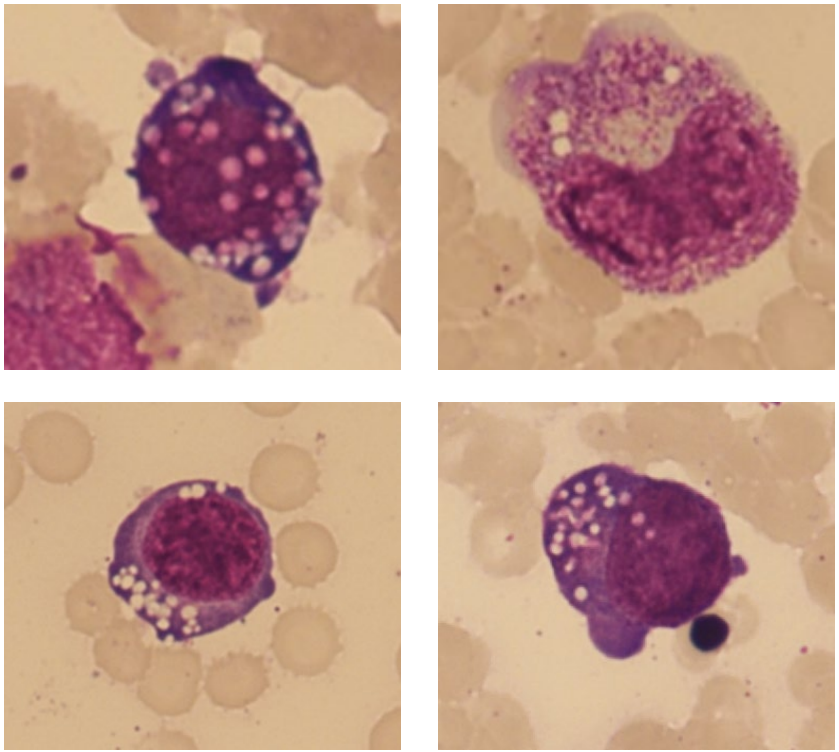
Confirmed diagnosis

- **Juvenile Dermatomyositis with multiple nutritional deficiencies – Anti Ro52 positive confirmed**
- **R14 genetics panel normal**

Note…

- Copper normalized but
  - Can increase greatly in response to infection, injury, chronic inflammatory conditions
  - Consider appropriateness of copper measurement where CRP is significantly elevated
  - If CRP 100-200, there may be a 30% increase in serum copper concentration
    Copper results should be interpreted alongside albumin and CRP results

Bone marrow aspirate x60



- Particulate but relatively hypocellular
- Left shifted with giant metamyelocytes
- Vacuolated erythroid cells
- No evidence of Haemophagocytosis

- In keeping with B12 and Copper deficiency – check genetics to exclude MDS & VEXAS

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Juvenile Dermatomyositis

- Inflammation of the blood vessels, muscles and skin – muscle weakness and skin rashes
- Mean time between the onset of the first symptoms and confirming the diagnosis is 6 months (5 weeks to 2 years) – more common in females
- Standard initial management for children with JDM consists of high-dose corticosteroids
    - IVIG & cyclophosphamide
    - Prednisolone 40mg on weaning dose
    - Gabapentin 600mg – 3 times daily
    - Nutritional deficiencies replaced
    - Physiotherapy & OT

Acknowledgments

- Paediatric team at UHNM
- Clinical Haematology colleagues for the lively Morphology MDT discussions
- Lab Haematology colleagues for the interesting Bitesize Morphology sessions each Friday

Literature

JOURNAL ARTICLE EDITOR'S CHOICE

British Society for Rheumatology guideline on management of paediatric, adolescent and adult patients with idiopathic inflammatory myopathy

Alexander G S Oldroyd, James B Lilleker, Tania Amin, Octavio Aragon, Katie Bechman, Verna Cuthbert, James Galloway, Patrick Gordon, William J Gregory, Harsha Gunawardena ... Show more

Author Notes

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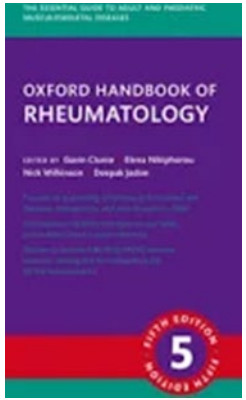

https://juveniledermatomyositis.org.uk/wp-content/uploads/2024/01/MSAMAA-Myositis-antibody-interpretation_24052022-.pdf

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