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Many medical conditions are named after the physicians credited with first describing them: eponymous syndromes. But who were these physicians, when and how did they make their discovery and how do these affect the breast? We describe some of these, some rarer than others, with illustrations of how they may be recognised on breast imaging.

Rosai Dorfman Syndrome

Sinus Histiocytosis with Massive Lymphadenopathy

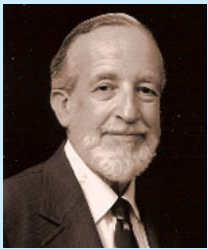
- commonly presents as massive, painless, bilateral lymph node enlargement in the neck with fevers
- Very rare - less than 1000 reported cases
- extra-nodal disease occurs in up to 25% including central nervous system, eyes, upper respiratory tract, skin, head and neck and... breasts.
- cause unknown; usually there is spontaneous regression of symptoms



A 55 year old lady with known Rosai Dorfman disease presented with a painless mass in the upper inner quadrant of her left breast. Mammography & ultrasound confirmed a 10cm irregular mass; T1-W MRI demonstrated an irregular mass with benign enhancement kinetics. Core biopsy confirmed extra nodal Rosai Dorfman disease.

Professor Ronald F Dorfman and Dr Juan Rosai

- Professor Ronald F. Dorfman, South African pathologist came across African boys with 'bull-neck' appearances
- Collaborating with Dr Juan Rosai, Argentine-American pathologist they described 'Sinus histiocytosis with massive lymphadenopathy' in 1969. Both have continued to publish on the subject into the 1990's.



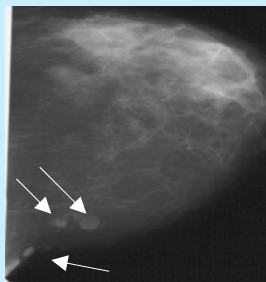
Von Recklinghausen disease

Neurofibromatosis type I (NF-1)

- autosomal dominant condition characterized by multiple benign neurofibromas and cafe-au-lait spots but CNS, peripheral nerve, skeletal, dermatological and cognitive involvement can occur.



Multiple well defined masses on screening mammography corresponded to cutaneous neurofibromata in a woman with Von Recklinghausen's disease



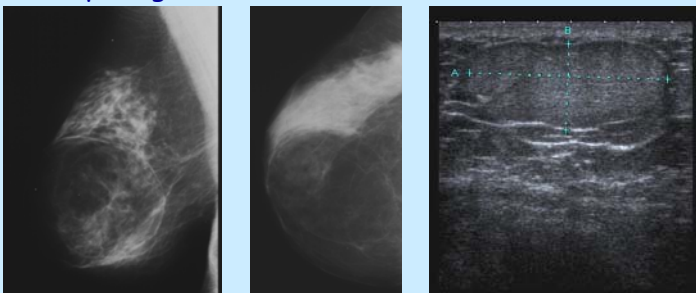
Professor Friedrich Daniel von Recklinghausen (1833-1910)

- German pathologist, studied under Virchow
- also coined the term haemochromatosis first describing the link between that disease & iron accumulation in tissue.
- opposed Robert Koch arguing, to claim that because TB lesions contained Koch's bacillus, therefore the bacillus was the cause of TB was like claiming that the pyramids of horse manure in Strasbourg streets were caused by the sparrows which perched on top!

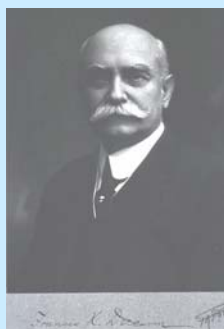
Dercum's disease

Adiposis dolorosa

- characterised by multiple painful lipomas located on the trunk and limbs with sparing of the face and hands but can affect heart or lungs.



Large hyperechoic lipoma in the breast of a patient with multiple trunk and limb lipomata



Professor Frances Xavier Dercum (1856-1931)

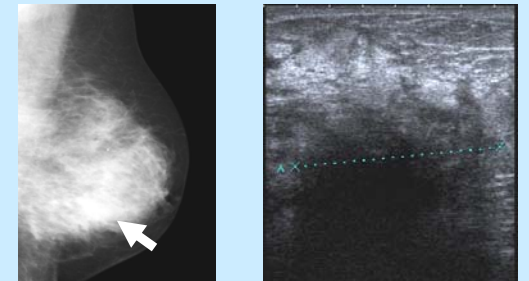
- American neurologist, professor of nervous and mental diseases, Jefferson Medical College
- elected president of the American Philosophical Society
- death at an APS meeting sitting in the "ladder library chair", crafted by Benjamin Franklin, was described in press "Dr. Dercum dies as he presides over scientists".

Erdheim-Chester disease

Polyostotic sclerosing histiocytosis or non-Langerhans-cell histiocytosis.

- lipid storing histiocytes localize to the bones or multiple organ systems with onset in middle age.
- Very rare- 80 cases reported

A 73 year old lady with known Erdheim-Chester disease presented with a painless hard breast mass. Mammography & ultrasound demonstrated an 8cm irregular mass. Core biopsy confirmed features of Erdheim-Chester disease.



First described by Jakob Erdheim and William Chester in 1930. They described two patients who had a distinctive lipidosis with associated bone changes.

Professor Jakob Erdheim (1874-1937)

- Austrian pathologist
- Beside other complex syndromes, the craniopharyngioma, was originally named after him - "Erdheim Tumor".

Paget's disease of the breast

- An eczematous skin condition of the nipple and areola due to infiltration of malignant epithelial (Paget) cells in the epidermis.
- >97% cases are associated with underlying breast cancer, invasive in 90%
- MRI indicated if mammograms are normal



A 74 year old woman presented with an itchy sore nipple. Punch biopsy confirmed Paget's disease of the nipple.



Sir James Paget 1st Baronet (1814-1899)

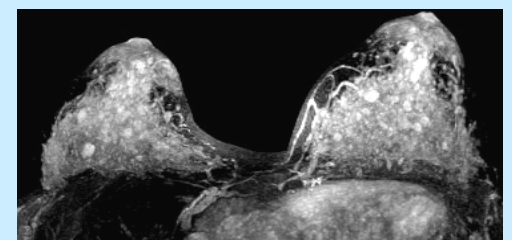
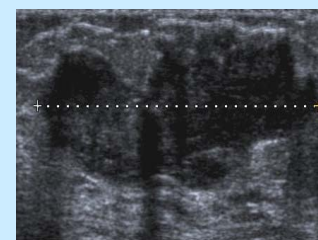
- British Surgeon & one of the founders of modern pathology
- in 1854 became surgeon to Queen Victoria
- Narrowly escaping death from infection following a cut during a post mortem, went on to describe 10 conditions including Paget's disease of bone, Paget's abscess, Paget-Schrotter disease and Paget's sign.

•in 1874 reported a chronic eczematous disease on the skin of the nipple and the areola in 15 women, with associated intraductal carcinoma

Cowden's disease

Multiple hamartoma syndrome

- rare autosomal dominant condition characterised by multiple hamartomata, characteristic skin lesions occur by age 20.
- increased risk of developing cancer, including thyroid, endometrial, brain and renal.
- 25-50% lifetime risk of developing breast cancer and 75% lifetime risk of benign breast conditions - ductal hyperplasia, fibroadenomas, and fibrocystic changes.



A 25 year old woman with characteristic skin lesions & bilateral breast masses

U/S demonstrates one of the well defined lobulated masses - biopsy proven fibroadenoma. MRI demonstrates multiple bilateral well defined fibroadenomas. No malignant tumour was found but the patient opted for risk reduction mastectomies due to the high risk of future malignancy.

Dr Kenneth M Lloyd II

- First described in 1963 by Dr KM Lloyd & Dr M Dennis, American dermatologists
- named the condition after the surname of the index patient.
- Dr Lloyd continues to practise as Chief, Dermatology Service, Youngstown, Ohio .

