

# 與大師對談

許居誠 教授

2021-04

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- 依照臨床時序，請大師模擬一線放射科醫師；於未知診斷，或者有限度臨床線索之情形下，進行閱片及解讀。
- 鑑別診斷為主要，確定診斷為次要。
- 目的在於學習大師之影像判讀邏輯思考。
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- 請大師給予本院影像品質建議：  
： Protocols, techniques,  
etc.

**CASE 1**

**53 Y/O WOMAN**

# Brief history

- **Past history**

Withheld

- **Chief complain**

For routine mammographic screening

# Imaging studies

**Digital mammography (2021.03.17)**

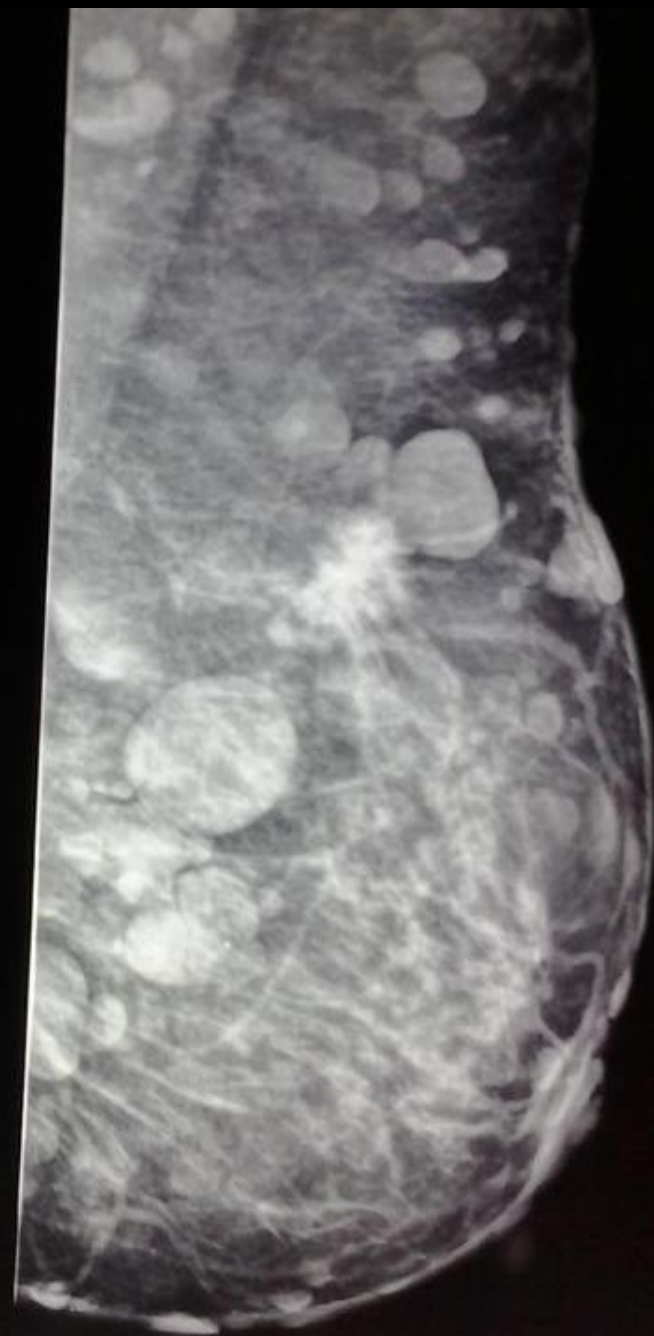
# Imaging findings

- ✓ **Multiple nodules or skin nodules of bilateral breasts and chest wall**

**→ Neurofibromatosis (BIRADS 2)**

# Neurofibromatosis

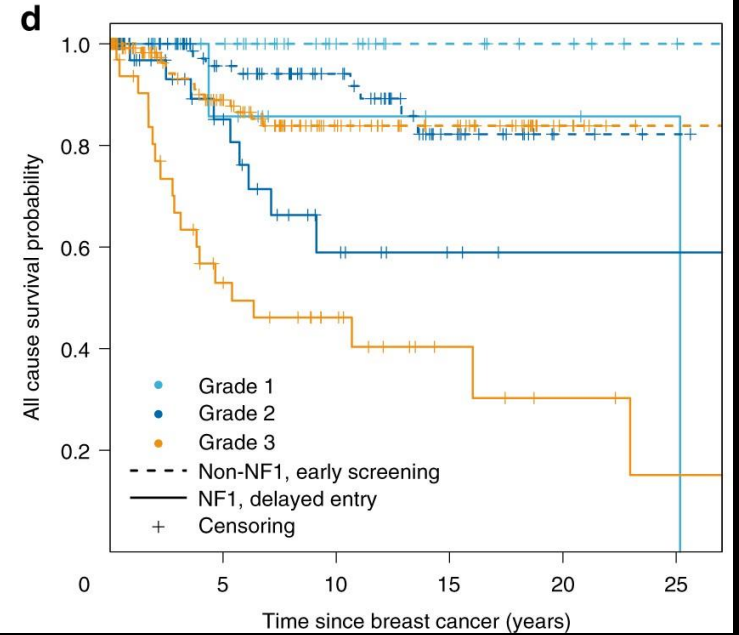
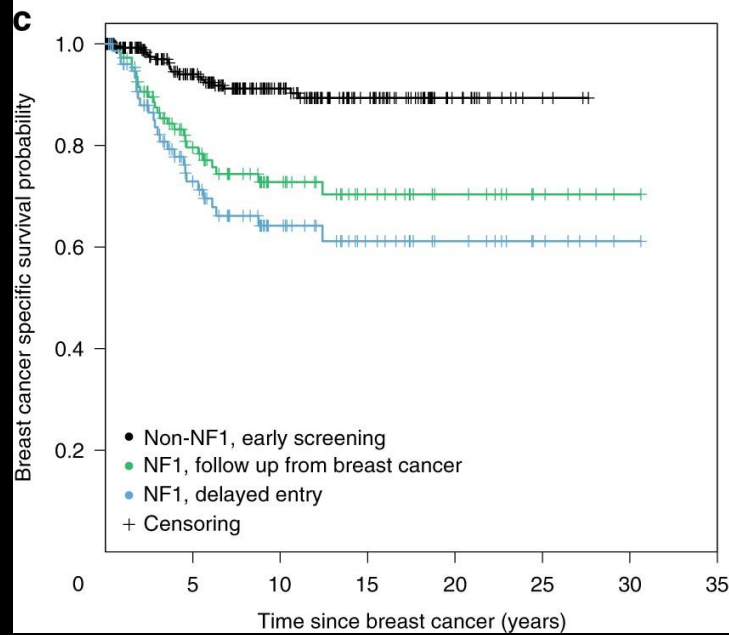
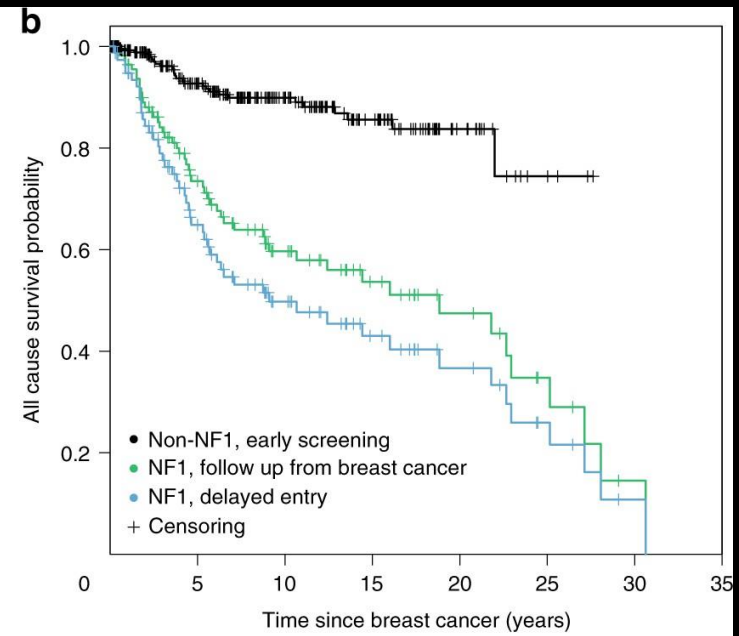
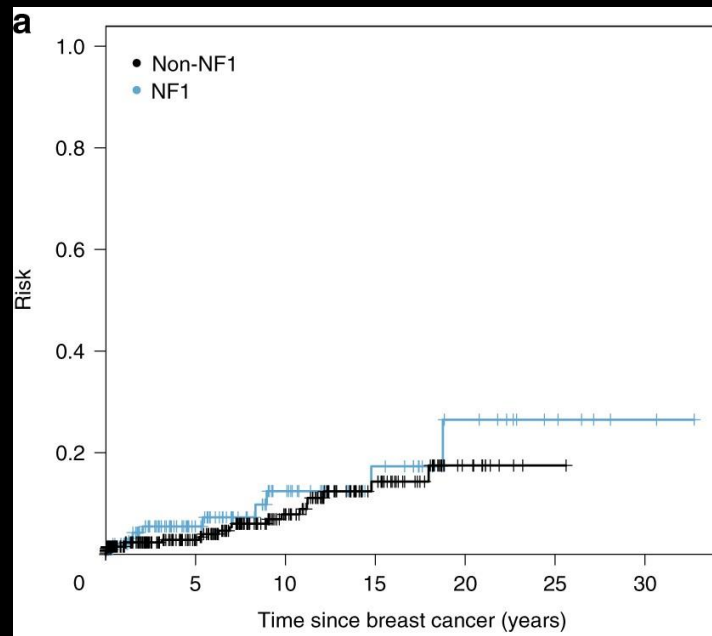
- A.k.a. **von Recklinghausen disease (NF1)**
- **Heterogeneous multisystem neurocutaneous disorders** caused by autosomal dominant mutations in NF genes, which are considered a classical tumor suppressors
- **NF1**: 17q11 locus on NF1 gene (85-90%)  
**NF2**: 22q12 locus on NF2 gene (10-15%)
- **Mammography:**
  - Multiple well-defined benign-appearing masses, classically **peri-areolar** in location. Portions of the outline may be rimmed by air density reflecting their superficial nature
  - Lesions can **mimic and partially obscure breast lesions**





# Neurofibromatosis

- NF1 is associated with **increased risk of breast cancer**
- Both the **NF1** gene & **BRCA1** gene are located in the **long arm of chromosome 17**  
→ interaction between these two genes has been suggested
- Women with NF1 have a **substantial contralateral breast cancer incidence** and **poor survival (20yr SV. 37% vs. 84%)**
- **Early start of breast cancer screening** may be a way to improve the survival.



Evans, D.G.R., *et al.* Breast cancer in neurofibromatosis 1: survival and risk of contralateral breast cancer in a five country cohort study. *Genet Med* **22**, 398–406 (2020).