

# Natural history of RDEB: news from PEBLES

EB2017 & EB-CLINET

Salzburg September 2017

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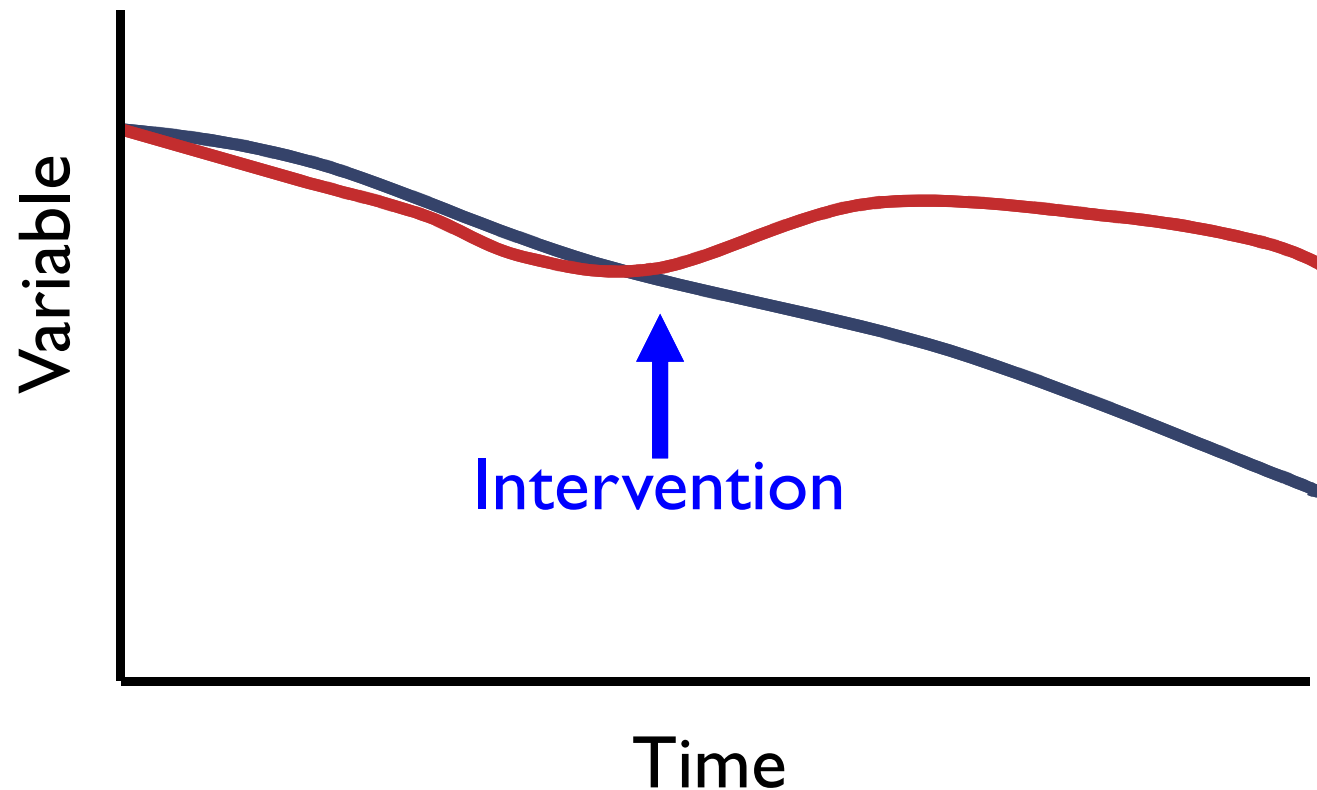
Great Ormond Street and  
St John's Institute of Dermatology  
London

Great Ormond Street Hospital for Children   
NHS Trust



# Natural history of EB

- Clinical trials need endpoints
- Need to know natural history of disease to know what to evaluate



# Natural history of EB

**P**rospective  
**E**pidermolysis  
**B**ullosa  
**L**ongitudinal  
**E**valuation  
**S**tudy





# Natural history of EB

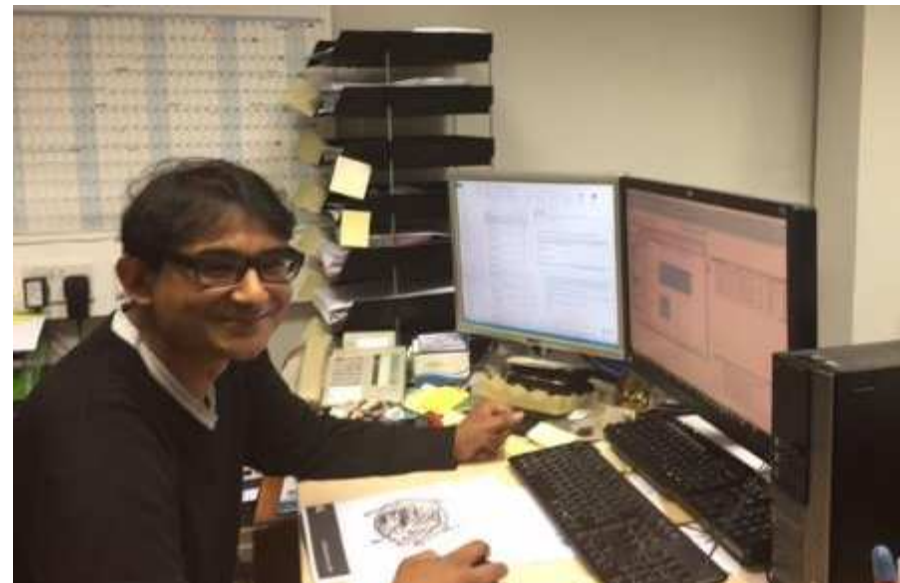
## **P**rospective **E**pidermolysis **B**ullosa **L**ongitudinal **E**valuation **S**tudy

### Recessive dystrophic EB

- Type of RDEB and lab data
- Clinical aspects
- Laboratory results and investigations
- Disease severity scores
- Subjective scores
- Photographs of skin
- Economic costs

# PEBLES

- Company employed to devise data capture tool
- Tablet device with questionnaires
- Up to 2000 possible data entry points per patient





# PEBLES

- Moving to web-based data capture
- More robust to handle volume of data
- Retrieve data via a dashboard to interrogate different variables

# PEBLES

- Data collection repeated
  - every 6 months ( $<10$ y age)
  - every 12 months ( $>10$ y age)





# PEBLES

- Patients/parents sent booklet of questionnaires to complete before review by researcher
- Age appropriate scores e.g. QOL, puberty, pain
- Blister count
- Dressings
- Also sent a camera for standard view photographs



# PEBLES: background and clinical data

- Demographics

- Name and DOB
- Gender
- Type of EB
- IF and mutation results
- Ethnicity
- Family history
- Social history
- School/work

- Skin

- Body map wounds etc
- Blister count

- Photographs

- Standard views on tablet or own camera

- Growth/development

- Ht and Wt
- BMI
- Pubertal stage

# PEBLES: background and clinical data

- Dental

- Caries/missing teeth
- Microstomia
- Dental surgery

- Hands

- Hand contracture/flexion
- Splints
- Previous surgery

# PEBLES: background and clinical data

- Mobility

- Fully mobile
- Limited walking
- Walking around the house
- Wheelchair dependent

- Eyes

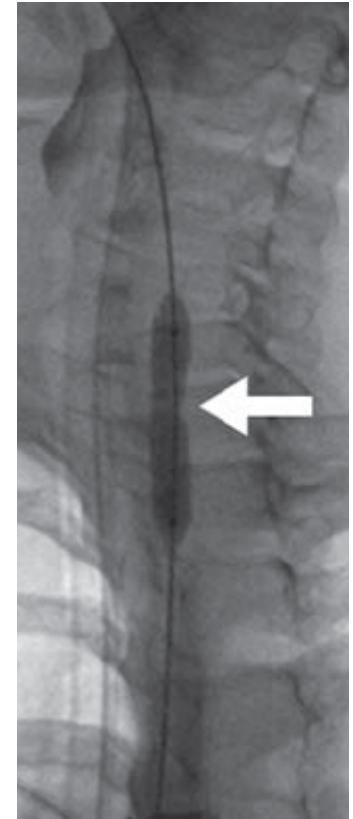
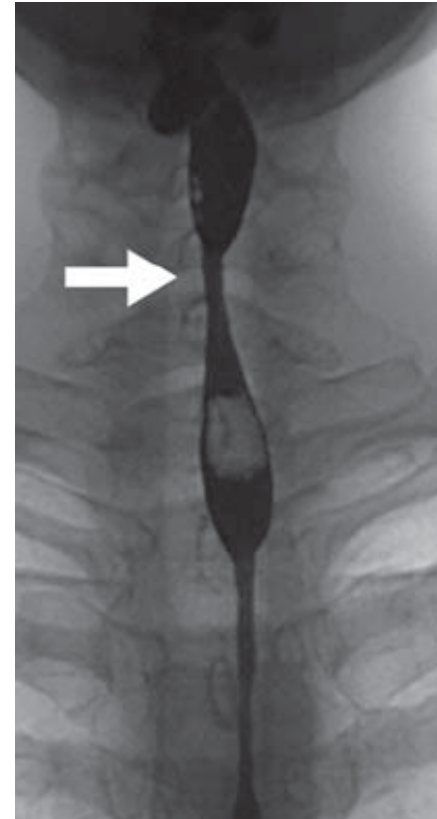
- Symptoms

- ENT

- Symptoms
- Interventions

# PEBLES: background and clinical data

- **Gastrointestinal tract**
  - Dysphagia
  - Aspiration
  - Previous ODs
  - Nutrition
  - Oral/tube feeding
  - Bowel habit
  - Colitis



# PEBLES: background and clinical data

- Cancer

- Age at first SCC
- Numbers/ sites of SCCs
- Local/regional/distant spread
- Therapies
- Other cancers

- Severity scores

- iSCOREB
- BEBS

# PEBLES: laboratory data

- Haematology

- Hb
- Iron
- Ferritin

- Inflammatory status

- CRP
- ESR
- Ferritin
- Platelet count

- Renal

- Urinalysis
- BP
- Renal function

- Serum stored

# PEBLES: laboratory data

- Endocrine/bone health

- Calcium
- Vitamin D
- Spine X-ray
- DEXA scan

- Cardiology

- Echocardiogram

- Nutritional

- Albumin
- Vitamin D
- Selenium
- Zinc



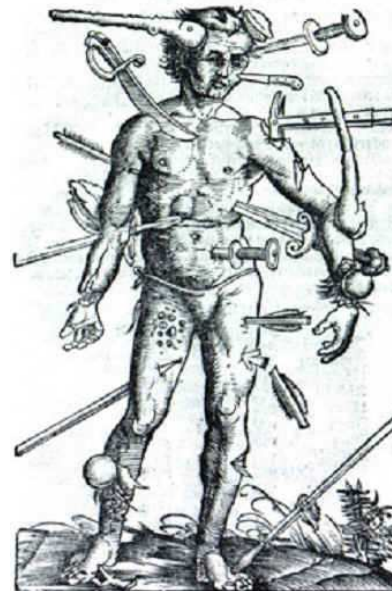


# PEBLES: subjective data

- Pain questionnaire
  - Procedural pain
  - Background pain

- Quality of life score
  - PEDSQL
  - QOLEB

- Itch questionnaire
  - Leuven itch score





# PEBLES: health economic data

- Hospital appointments
  - Numbers
  - Admissions
  - Length of stay
- Medication
  - Types and route



# PEBLES: health economic data

- Dressings
  - Types
  - Sizes
  - Numbers used per week
  - Linked to costs

- Dressing changes
  - Who does them?
  - Paid?
  - Hours/week



## EB Natural History Project Medications

How many PRN medications do you wish to list?

☐ 1    ☐ 2    ☐ 3    ☐ 4    ☐ 5    ☐ 6    ☐ 7    ☐ 8    ☐ 9    ☐ 10    ☐ None

### Regular Medication

Name

Dose

Unit

Route -

☐ Oral    ☐ Per gastrostomy    ☐ Jejunostomy    ☐ Naso-gastic tube    ☐ Trans-dermal    ☐ Nasal spray    ☐ Injection

Frequency

☐ Weekly    ☐ OD    ☐ BD    ☐ TDS    ☐ QDS    ☐ Continuous

Taken regularly?

☐ Yes    ☐ No

### PRN Medication - frequency in 24 hours

Name

Dose

Average number of doses in 24 hours

Unit

Total dose in 24 hours

Route -

☐ Oral    ☐ Per gastrostomy    ☐ Jejunostomy    ☐ Naso-gastic tube    ☐ Trans-dermal    ☐ Nasal spray    ☐ Injection

Spinal fractures since last review? ☐ Yes ☐ No

How many spine fractures?

Thoracic - Site(s) of fractures

T1 ☐ T2 ☐ T3 ☐ T4 ☐ T5 ☐ T6 ☐ T7 ☐ T8 ☐ T9 ☐ T10 ☐ T11 ☐ T12 ☐

Fractures - Thoracic -  
Date of diagnosis

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

Fractures - Thoracic -  
Source of information

☐ Hospital EPR  
☐ Hospital notes  
☐ GP

Lumbar - Site(s) of fractures

L1 ☐ L2 ☐ L3 ☐ L4 ☐ L5 ☐

Fractures - Lumbar -  
Date of diagnosis

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

Fractures - Lumbar -  
Source of information

☐ Hospital EPR  
☐ Hospital notes  
☐ GP

Calcaneal fractures since last review? ☐ Yes ☐ No

Calcaneal fractures since last review? - Right?

☐ Yes ☐ No

Fractures - Calcaneal Right -  
Date of first diagnosis

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

Fracture - Calcaneal Right -  
Source of information

☐ Hospital EPR  
☐ Hospital notes  
☐ GP

☐ Yes ☐ No

Fractures - Calcaneal Left -  
Date of first diagnosis

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

Fracture - Calcaneal Right -  
Source of information

☐ Hospital EPR  
☐ Hospital notes  
☐ GP



# EB Natural History Project SCC

Squamous cell carcinoma (SCC) Skin cancer - Does the patient currently have; or has he/she had an SCC in the past?  
☐ Yes ☐ No

SCC - Date of first diagnosis

DD/MM/YYYY

Total Number of SCCs

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

Surgical treatment (SCC) - Total number of excisions

SCC - Date of diagnosis

DD/MM/YYYY

SCC - Date of excision

DD/MM/YYYY

SCC - Histology (differentiation)

☐ Well ☐ Moderate ☐ Poor

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

Date must be completed in (DD/MM/YYYY) format. Put hashes (#) where date info is not known

SCC - Type of excision

☐ WLE ☐ Mohs

Location of SCC

Size of SCC (length cm's)

Size of SCC (width cm's)

Was PET/CT performed?

☐ Yes ☐ No

SCC - PET/CT results

Was FNA performed?

☐ Yes ☐ No

SCC - FNA results

☐ Positive ☐ Negative ☐ Inconclusive

SCC - FNA results details

SCC Were there any amputations for this case of SCC ☐ Yes ☐ No

Amputations LEFT

Thumb

☐

Index finger

☐

Middle finger

☐

3rd finger (ring)

☐

Little finger

☐

Hand

☐

Arm below elbow

☐

Arm above elbow

☐

Amputations RIGHT

Thumb

☐

Index finger

☐

Middle finger

☐

3rd finger (ring)

☐

Little finger

☐

Hand

☐

Arm below elbow

☐

Arm above elbow

☐



# PEBLES: where are we now?

- 55 consented patients
- 11 children, 44 adults
- 2 withdrawn, 1 lost to follow-up
  - 52 completed data for 1<sup>st</sup> assessment
  - 46 completed 2<sup>nd</sup> assessment
  - 23 completed 3<sup>rd</sup> assessment
  - 5 completed 4<sup>th</sup> assessment
  - 3 completed 5<sup>th</sup> assessment
  - 1 completed 6<sup>th</sup> assessment

# PEBLES: where are we now?

- 55 consented patients
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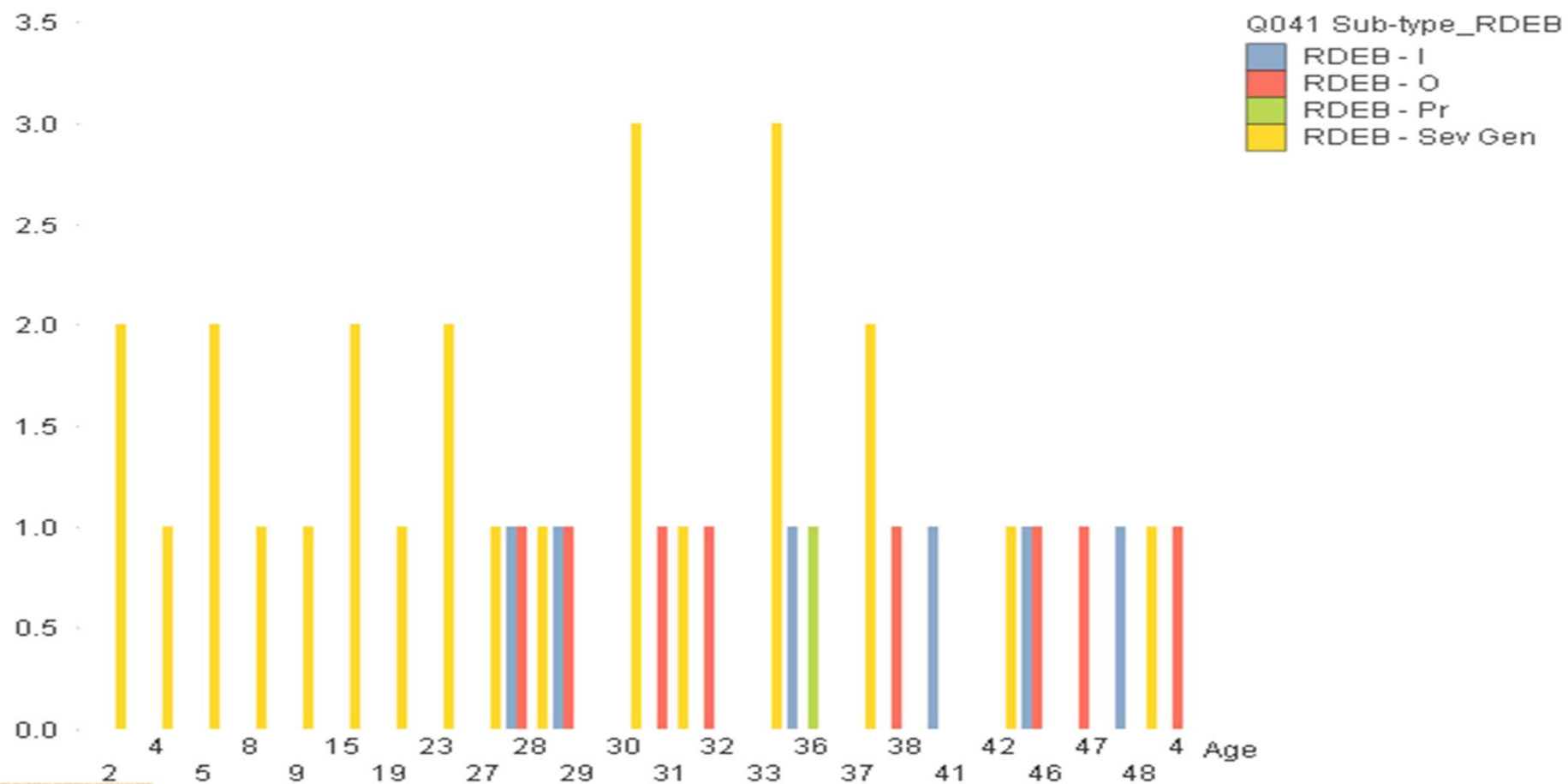
25 GS-RDEB

18 GI-RDEB

8 RDEB-I

1 RDEB-pru

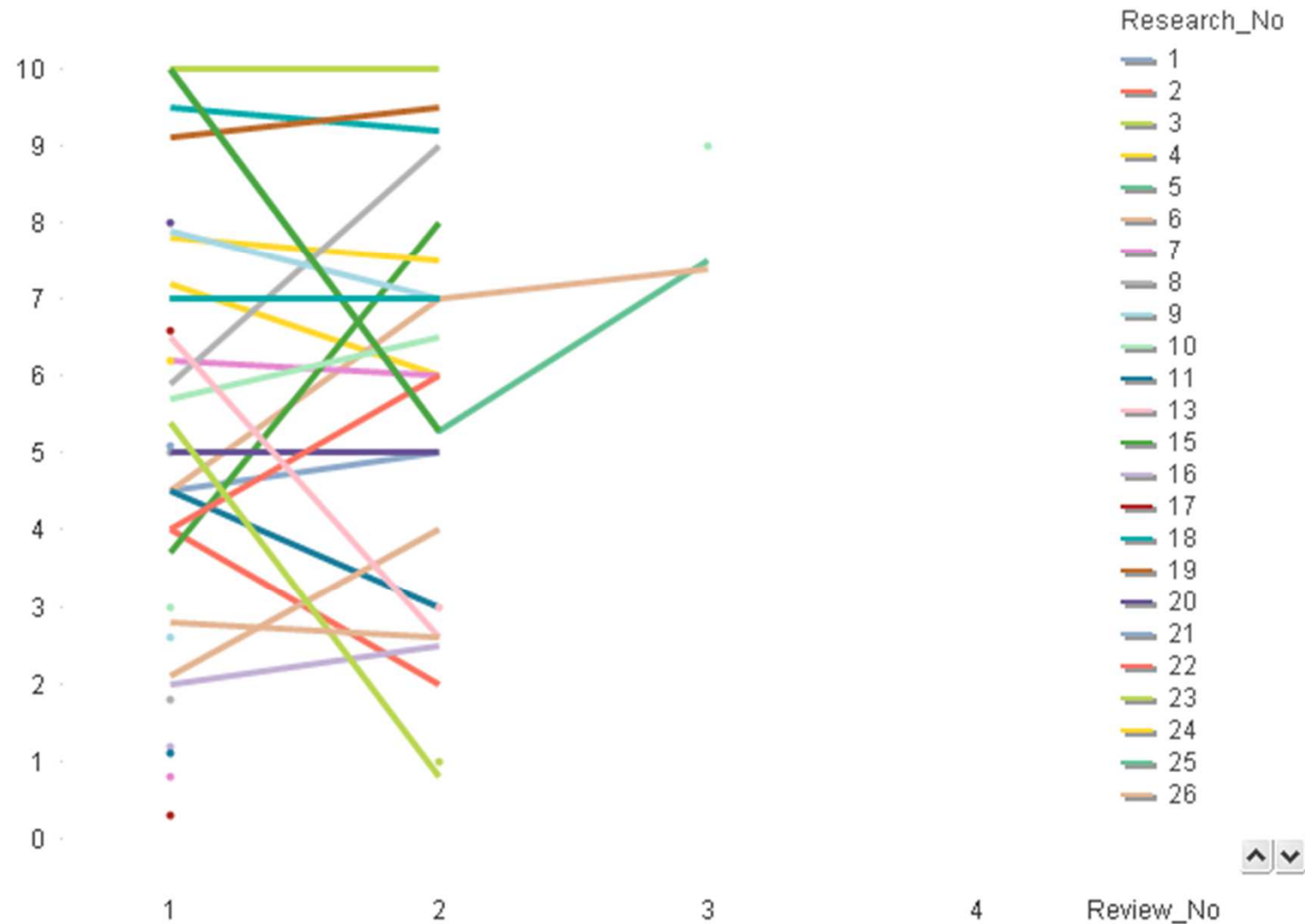
## Age by RDEB type





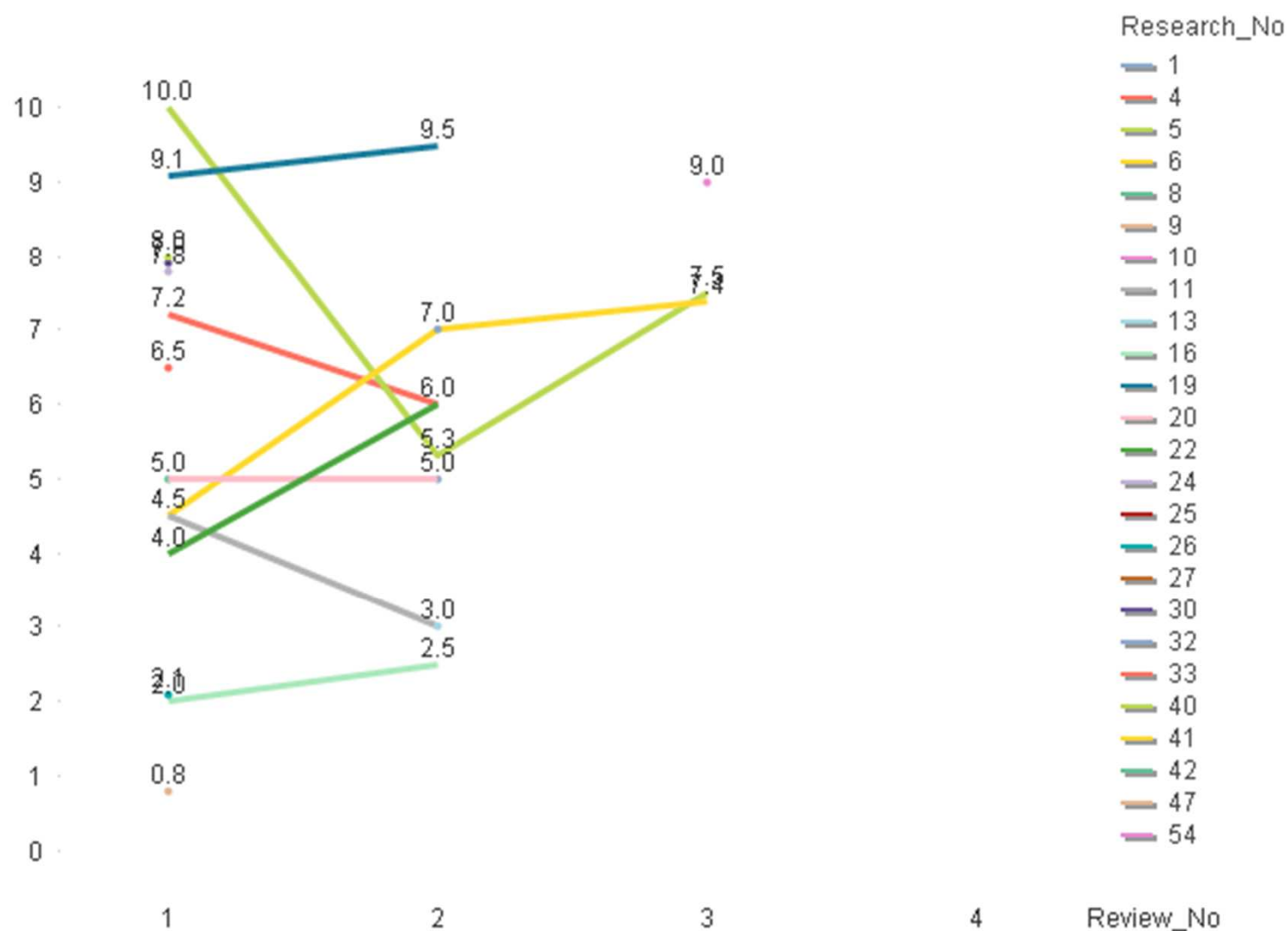
# Itch distress all RDEB types

10. In the past month, how distressing was your itching? (0.0-10.0)

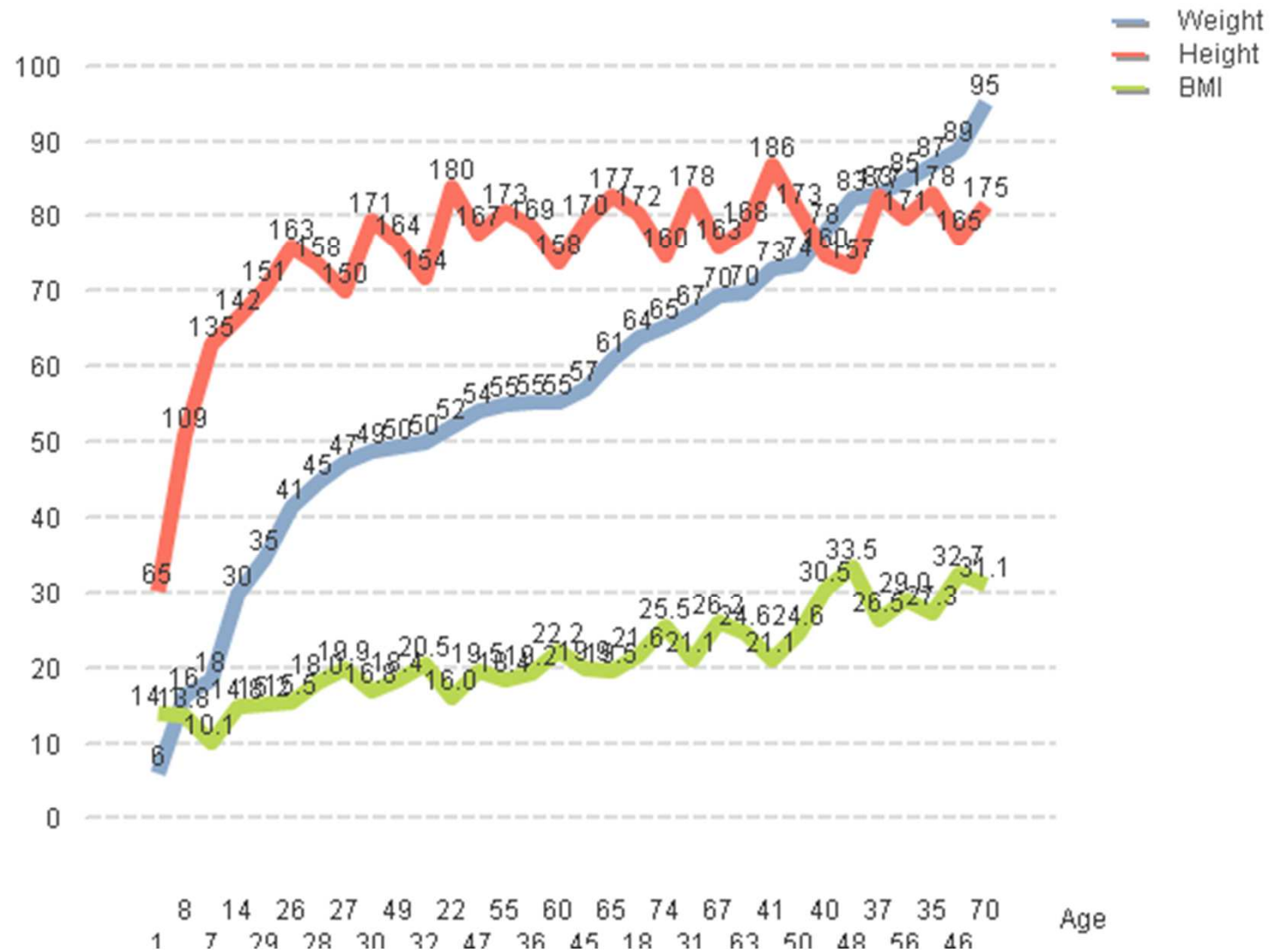


# Itch distress GS-RDEB

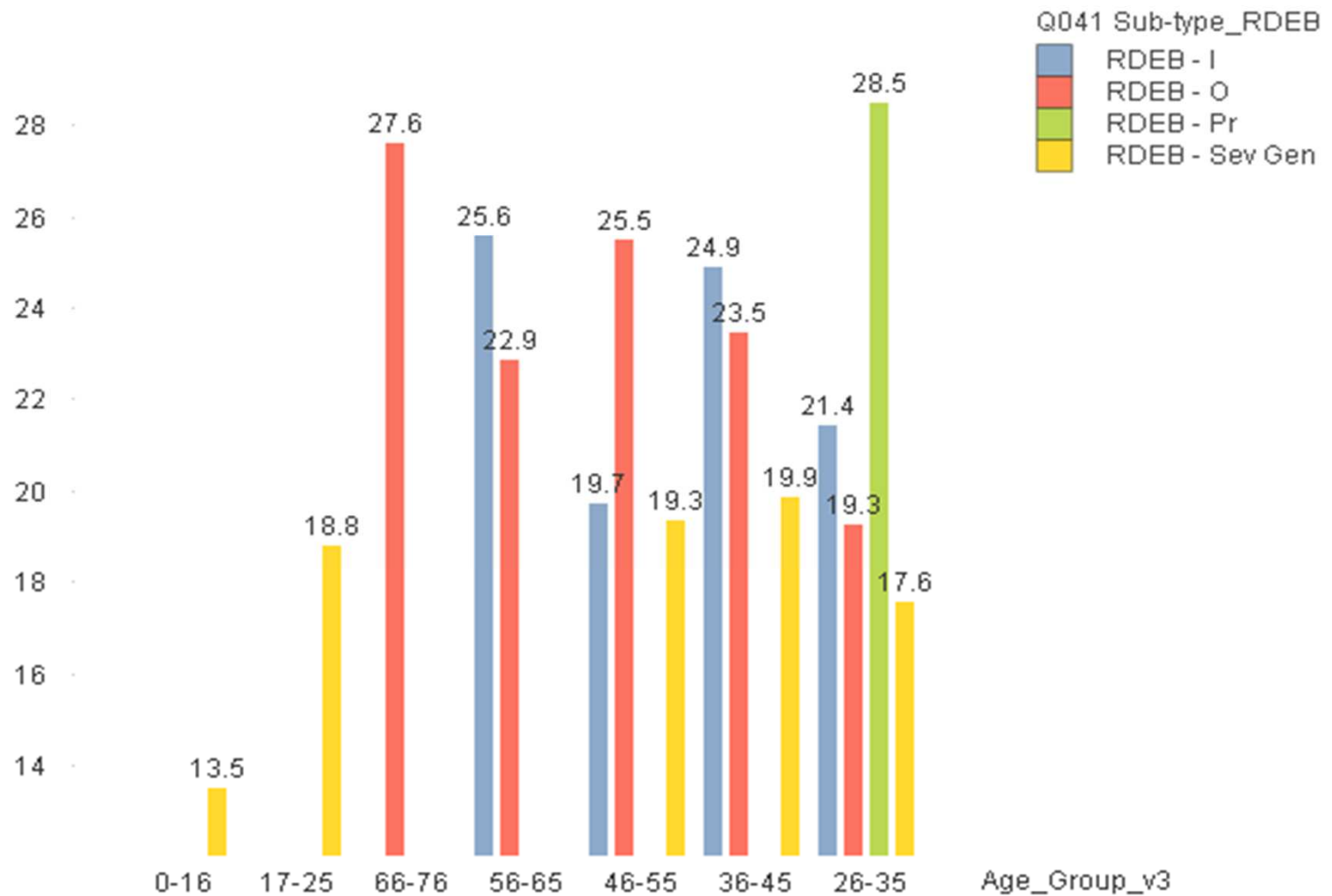
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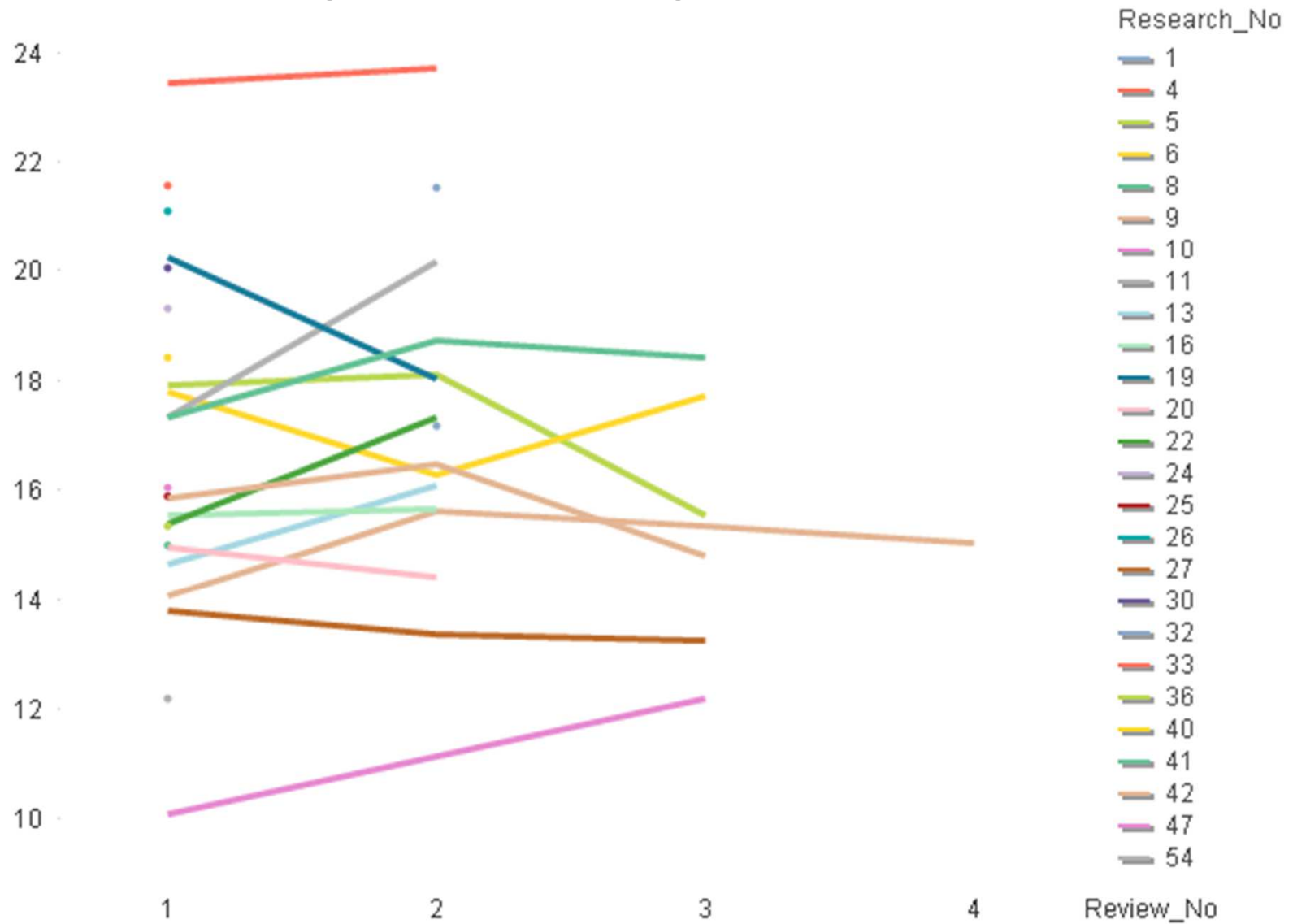
## Height/weight/BMI by age all types RDEB



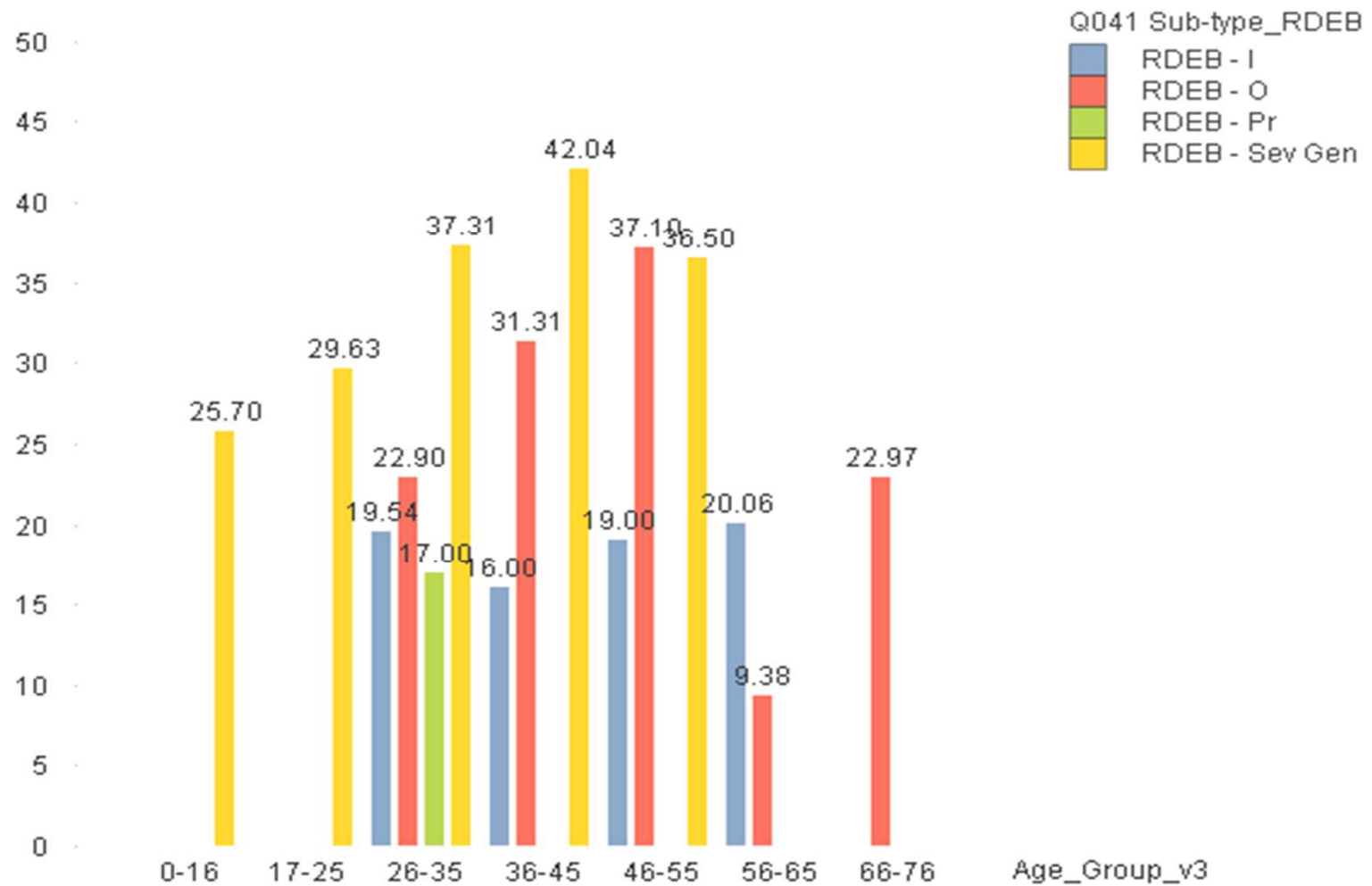
## Average BMI by age group



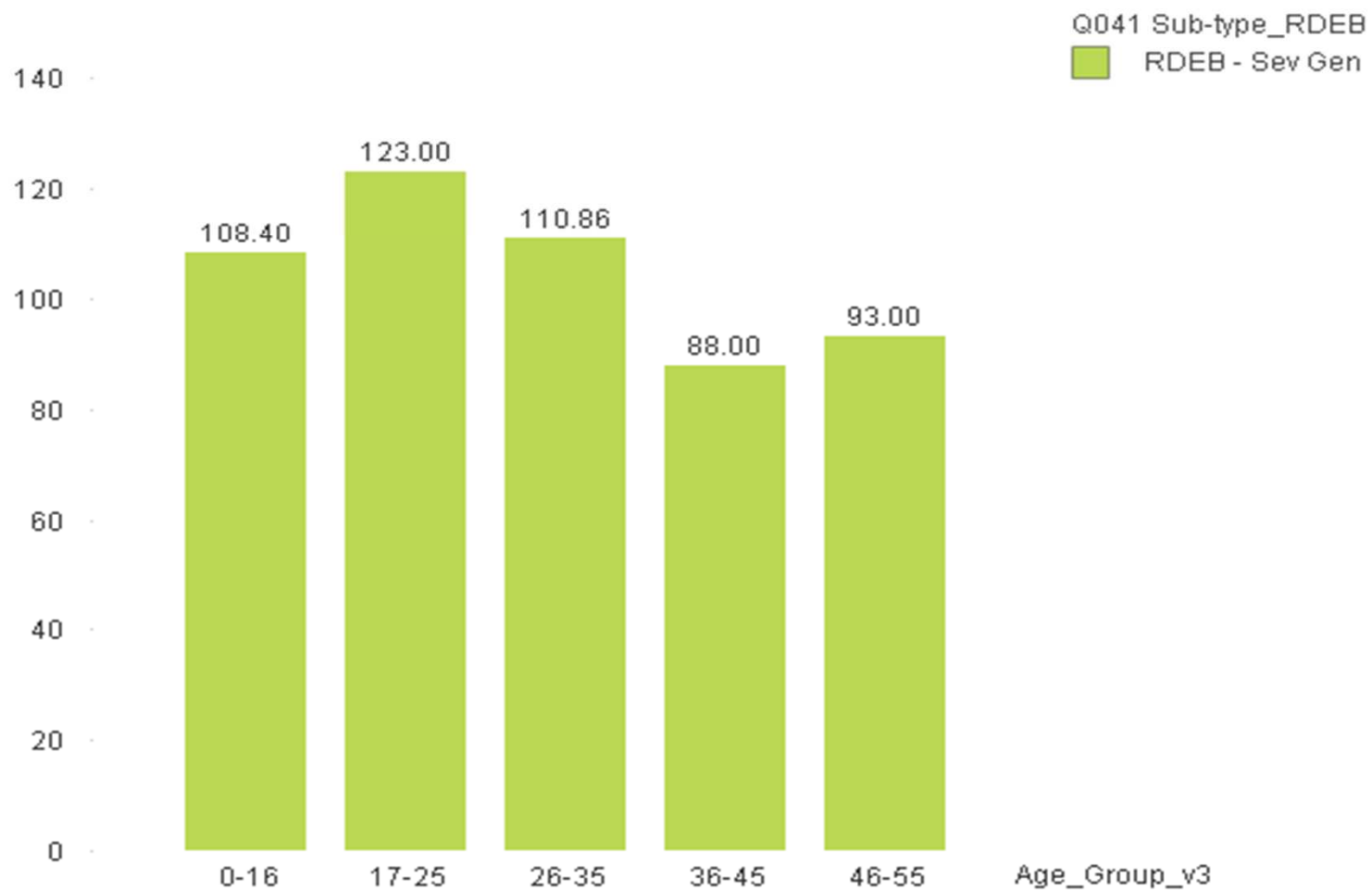
## Longitudinal average BMI GS-RDEB



## Average BEBS score by age group



## Average Hb by age group RDEB-SG

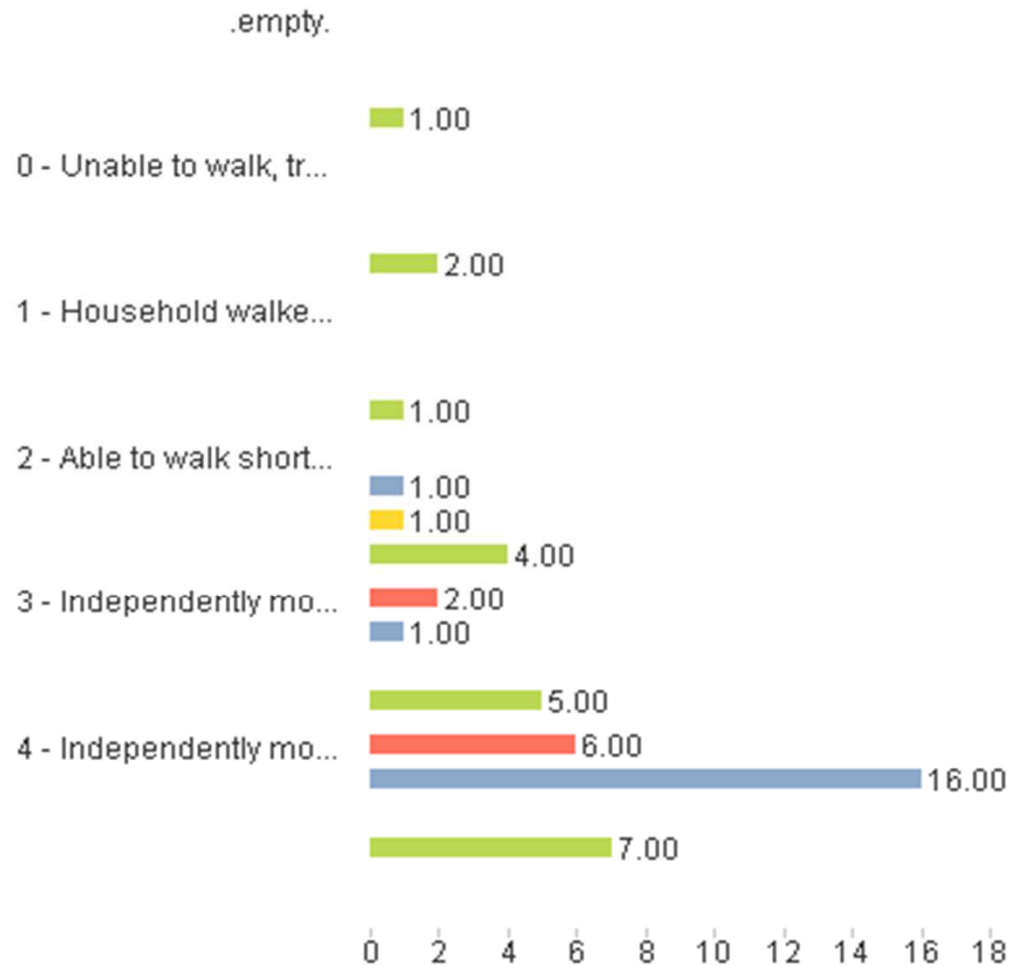
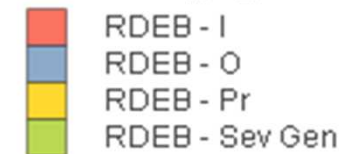




# Mobility by RDEB subtype

Q537 Mob\_Child\_Adults

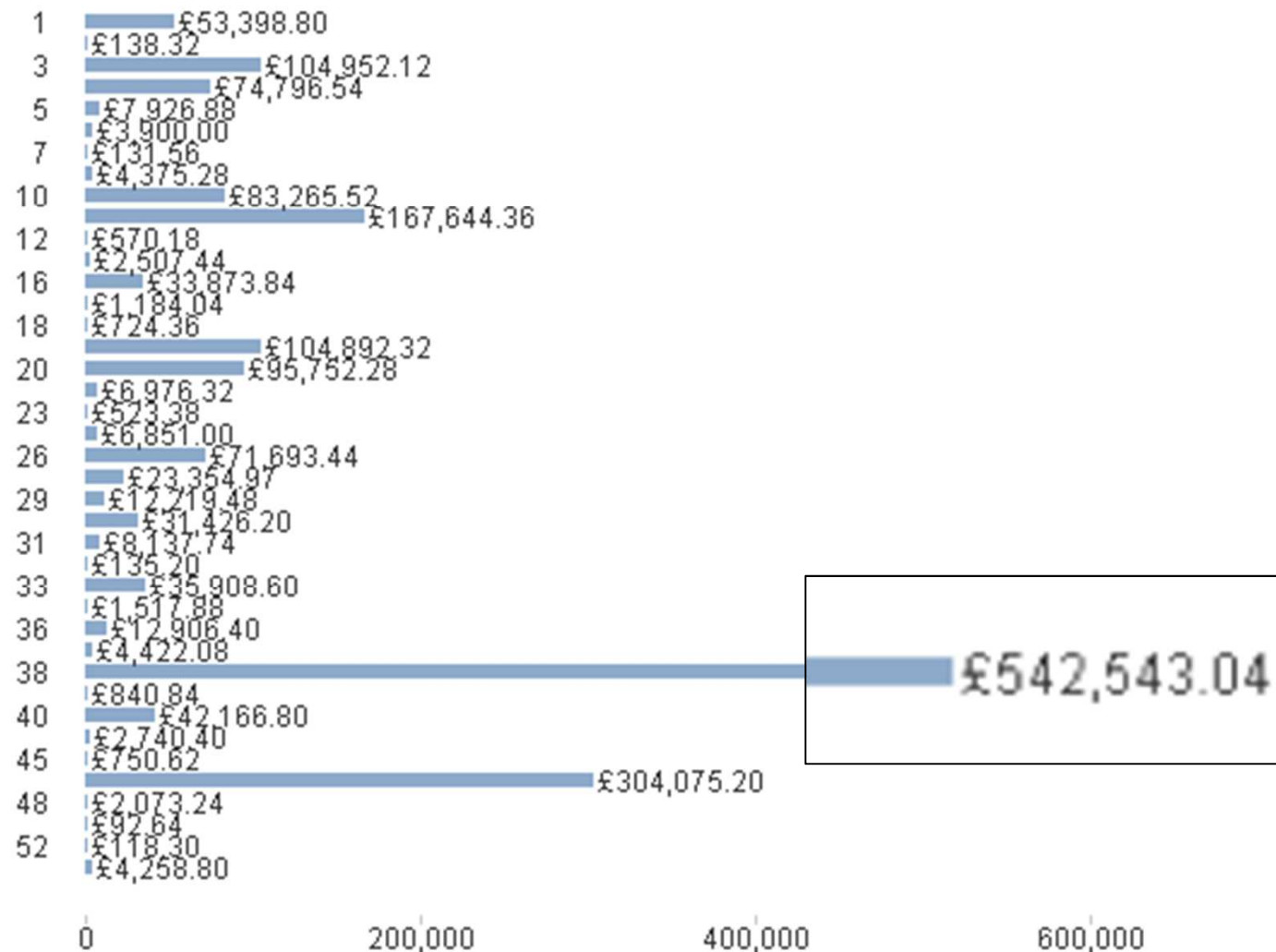
Q041 Sub-type\_RDEB





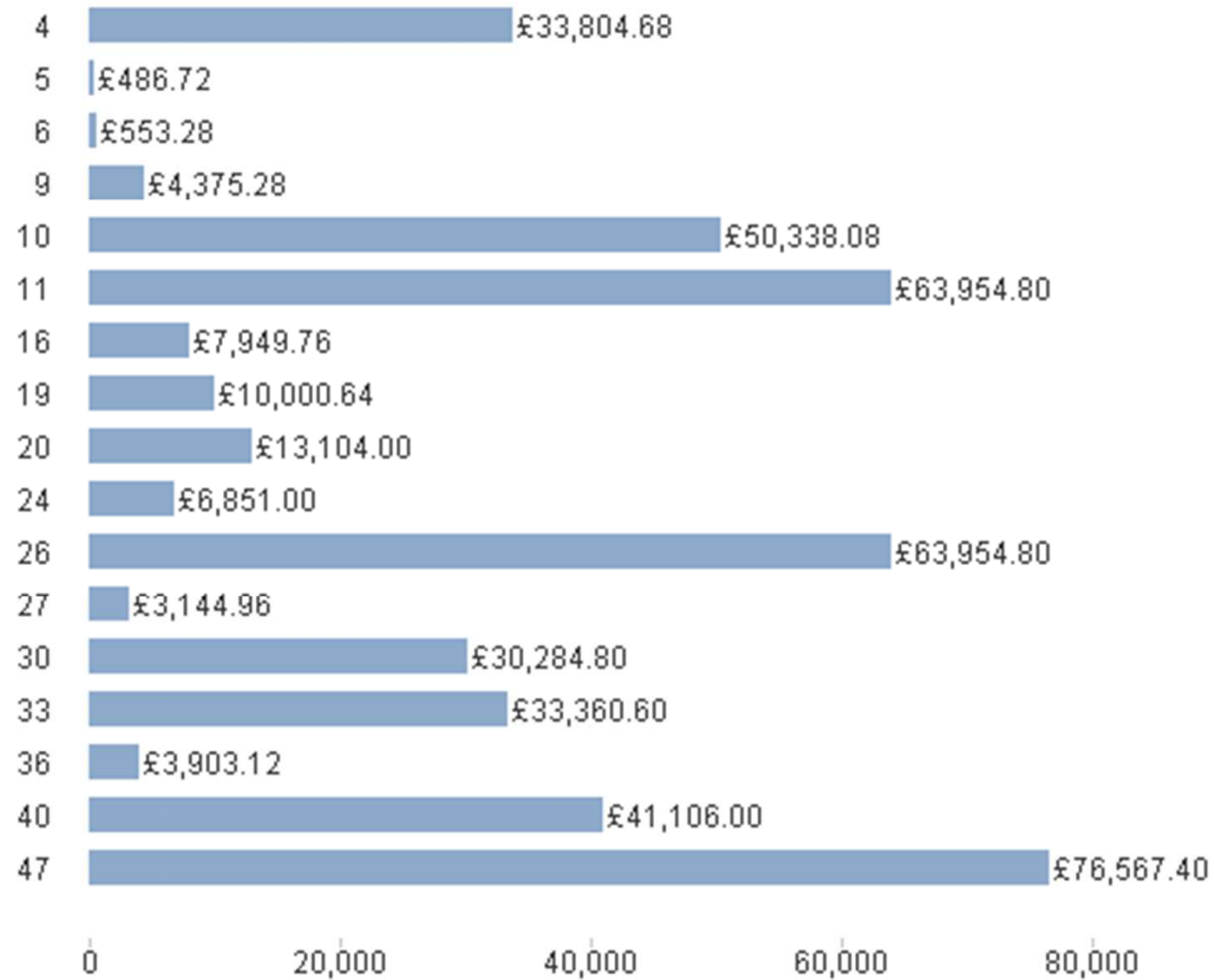
# Dressing costs per year all patients

Research\_No



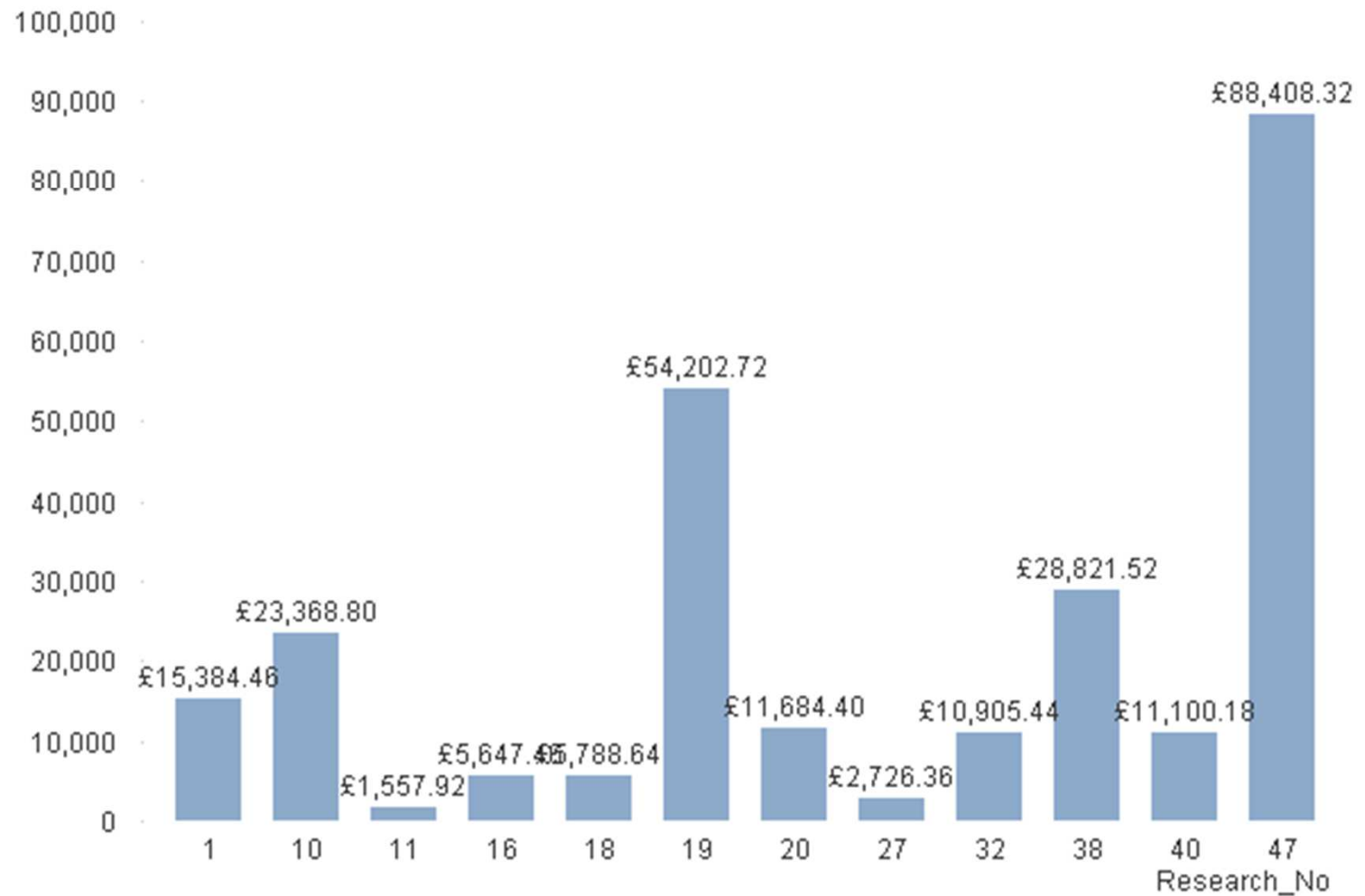
## Dressing costs per year GS-RDEB

Research\_No

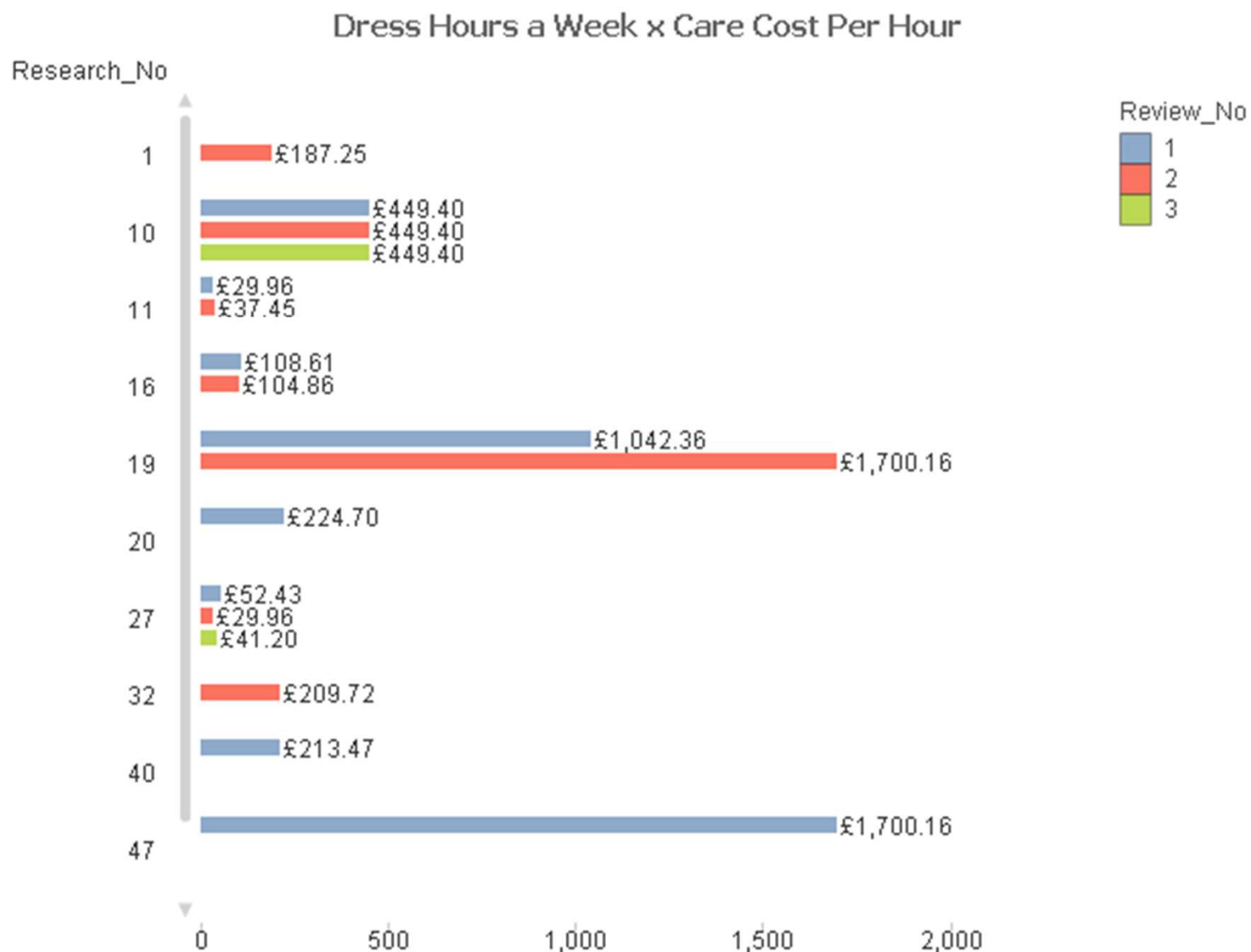


# Care costs per year RDEB-SG

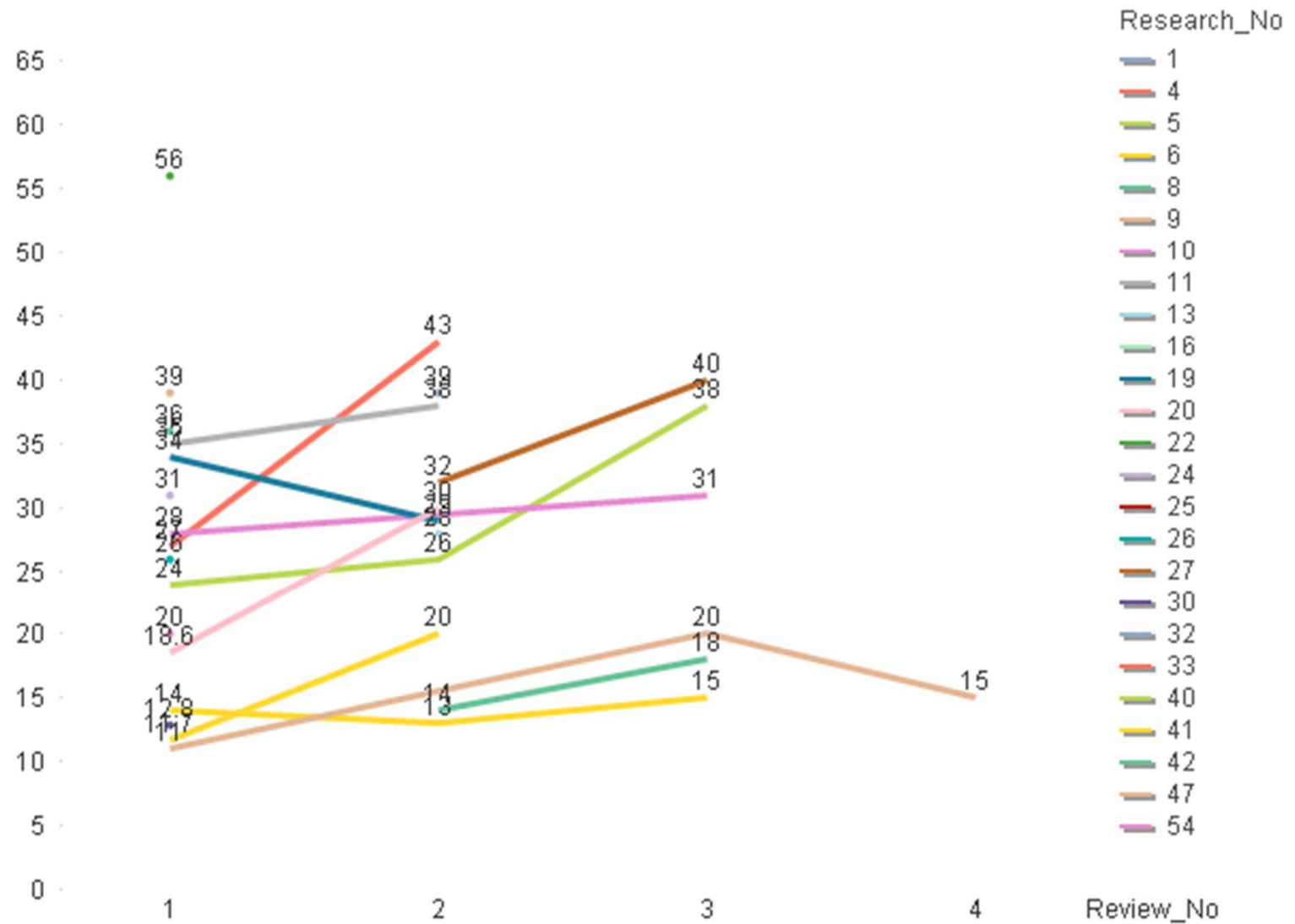
Dress Hours a Week x Care Cost Per Hour x 52 Weeks



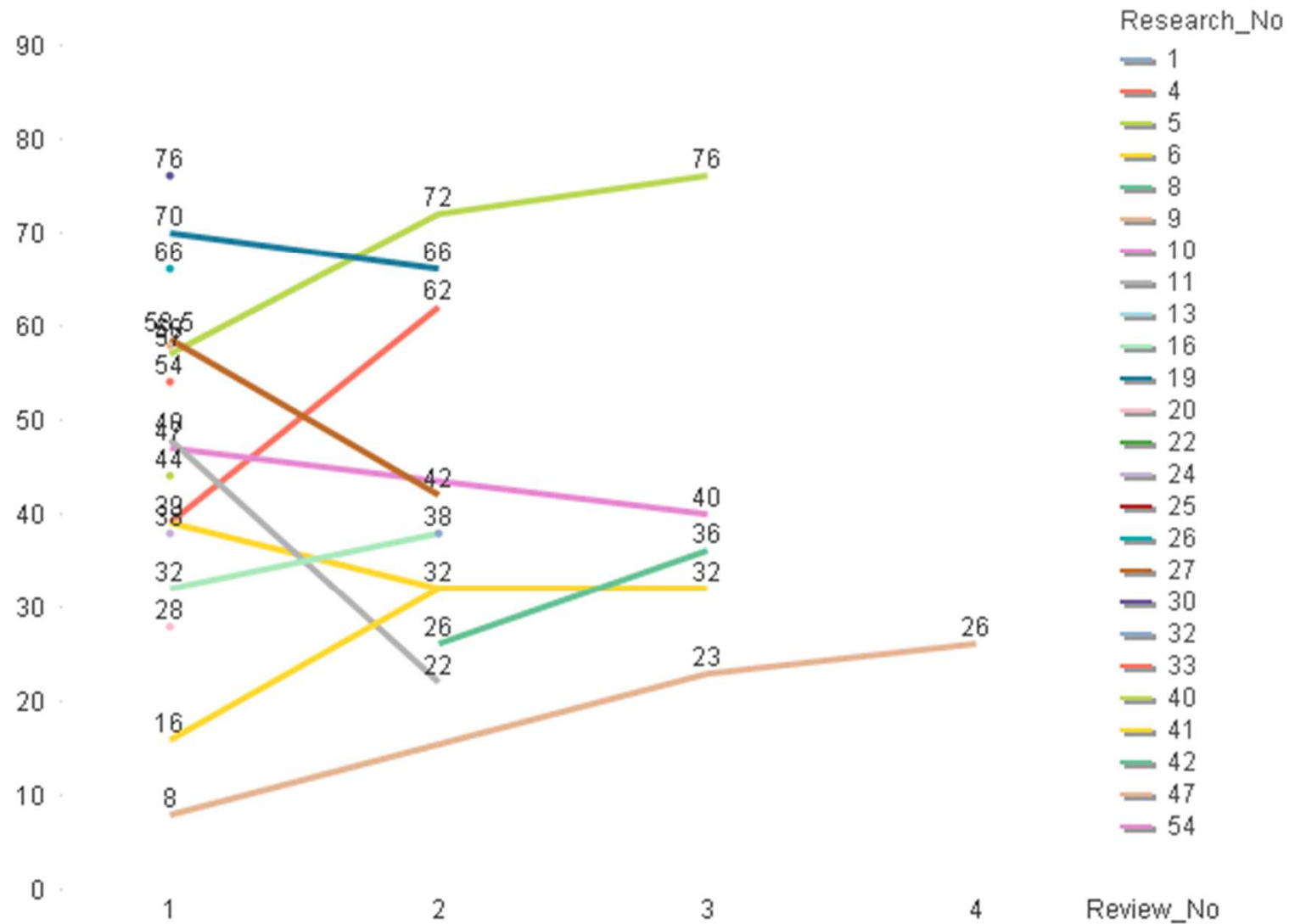
# Longitudinal care costs per week RDEB-SG



# Longitudinal iscor-EB physician scores RDEB-SG



# Longitudinal iscor-EB patient scores RDEB-SG





# PEBLES

- More detailed information about RDEB than previously collected
- Comprehensive picture of all aspects of well-characterised RDEB patients
- Will extend to other centres
- In time, for other types of EB

Identify meaningful endpoints  
for future clinical trials



# Acknowledgements

- Liz Pillay
- Pankaj Patel and DCC
- Jennie Hon
- Manpret Lakhan
- Danielle Greenblatt
- Anna Martinez
- Susan Robertson
- Emma Batten





# PEBLES: natural history of EB

Any  
questions  
or comments?

